APPLYING ETHNOGRAPHIC METHODOLOGIES & ECOLOGY TO UNVEIL DIMENSIONS OF SLEEP PROBLEMS IN CHILDREN & YOUTH WITH NEURODEVELOPMENTAL CONDITIONS

by

OSMAN S. IPSIROGLU

MD, University of Vienna, 1985
MAS, Vienna University of Economics and Business Administration, 2003
Doctoral Equivalent (PhD), Medical University of Vienna, 2005
MBA, Vienna University of Economics and Business Administration, 2007

A DISSERTATION SUBMITTED IN PARTIAL FULFILMENT OF THE REQUIREMENTS FOR THE DEGREE OF DOCTOR OF PHILOSOPHY

in

THE FACULTY OF GRADUATE AND DOCTORAL STUDIES

(Interdisciplinary Studies)
[Developmental Paediatrics / Medical Anthropology /Educational Psychology]

THE UNIVERSITY OF BRITISH COLUMBIA
(Vancouver)

January 2016

©Osman S. Ipsiroglu, 2016
Abstract

Willis Ekbom disease/restless legs syndrome is a relatively well-known neurological disorder in adult and paediatric medicine; however, the possibility of its presentation as familial early onset chronic intractable insomnia has not yet been recognized. I investigated the possible causes of intractable insomnia in children and youth with neurodevelopmental conditions. Through my studies of medical anthropology and educational psychology, I learned to apply qualitative methodologies in my clinical explorations, namely:

a) *ethnographic exploration of the ecology of* paediatric intractable insomnia;

b) the operationalization of this *ethnographic exploration* as a *therapeutic emplotment concept* in clinical practice, and

c) the *creation of new methodological tests and tools for structured behavioural observations* as further development of the *therapeutic emplotment concept*.

Application of these concepts led to the development of a *functional sleep and wake-behaviours assessment* model. This model exceeds the current clinical practice of categorical sleep and/or wake diagnoses and the predominantly daytime-focused explanatory models of developmental paediatrics, child psychiatry and mental health.

My research has privileged me with becoming the first physician to identify Willis Ekbom disease/restless legs syndrome in children with neurodevelopmental conditions, who suffer from early onset chronic intractable insomnia, in a methodologically reproducible way. This research also enabled me to demonstrate that unrecognized Willis Ekbom disease/restless legs syndrome leads to cascades of medical diagnoses and medication prescriptions causing iatrogenic harm. Weaning children off medications and increasing their wellbeing and performance after addressing their sleep problems became the proof of concept.
The *Willis Ekbom disease/restless legs syndrome story* in children with neurodevelopmental conditions is a modern parable, illustrating how conventional categorical diagnoses with overemphasis on daytime behaviours can produce systemic gaps in healthcare. While conventional medicine facilitates a spectrum of diagnoses that are applied based on training culture, symptoms that are not in alignment with the standard repertoire are not recognized and diagnoses are missed. Understanding this parable and finding applicable answers for how such systemic errors can be avoided in mainstream medicine in the future has taken me seven years, and is still a work in progress.
Preface

List of Publications Arising from Work Presented in the Dissertation. The manuscripts, which have been used for the partial fulfilment of the requirements of the interdisciplinary PhD in developmental paediatrics, medical anthropology and educational psychology, are presented in Chapters 1, 2 and 3.

- *Challenging Sleep/Wake-Behaviours leading to a Cascade of Diagnoses and Medications: A Case Report* (quoted in Chapter 1; referenced in Chapters 1, 2 and 3). I am the first and corresponding author; co-authors include Elbe, D., and Carleton, B. This manuscript has been prepared for publication [İpsiroğlu, O. S., Elbe, D., & Carleton, B. (2015). Challenging Sleep/Wake-Behaviours leading to a Cascade of Diagnoses and Medications: A Case Report. Manuscript in preparation.].

- *Transkulturelle Paediatrie, Fakten, Hypothesen, Lösungen* [Transcultural Paediatric Facts, Hypotheses, Solutions] was published first in German as a review article in Neuropaediatrie [İpsiroğlu, O. S. (2008a). Transkulturelle Pädiatrische Fakten, Hypothesen, Lösungen [Transcultural Paediatric Facts, Hypotheses, Solutions]. *Neuropädiatrie*, 7, 38-46.] and then published as *Transkulturelle Paediatrie* [Transcultural Paediatrics], a section in the *Epidemiologie – Sozialpädiatrie* [Epidemiology - Social Paediatrics] chapter of the German textbook *Paediatrie* [Paediatrics] (used for my advancement to candidacy, referenced in Chapters 1, 2 and 3). I am the first and only author in the *Neuropaediatrie* article, and the lead and corresponding author responsible for the entire manuscript in the textbook section, and Aksu, F. is the co-author. This textbook section was published by Springer Verlag in the third (2009) and fourth (2013) editions [İpsiroğlu, O. S., & Aksu, F. (2009). Transkulturelle Pädiatrie [Transcultural Paediatrics]. In M. Gahr, & C. Speer (Eds.), *Pädiatrie* [Paediatrics] (3rd ed., pp. 963-967). Berlin, Germany: Springer Berlin Heidelberg.; von Kries, R., Strassburg, H., İpsiroğlu,

- **Cultural Aspects in the Management of Inborn Errors of Metabolism** (referenced in Chapters 1, 2 and 3). This article was published in the Journal of Inherited Metabolic Disease. I am the corresponding and last author; co-authors are Stockler, S., Moeslinger, D., Herle, M., and Wimmer, B. [Stockler, S., Moeslinger, D., Herle, M., Wimmer, B., & Ipsirolu, O. S. (2012). Cultural aspects in the management of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 35(6), 1147-1153. doi: 10.1007/s10545-012-9455-4].

- The literature reviews of sleep problems and our current approach to paediatric sleep has been thematized in three review papers and one consensus paper, which provided the contextual framework for the original research. These three critical literature reviews of sleep problems were spearheaded by my clinical mentor, Dr. James E. Jan, who is the main and corresponding author of the papers (Neurophysiology, BC Children’s Hospital, UBC). In these manuscripts, my role was that of a clinician, critical discussant and partner. These three manuscripts include:
  - **Sleep Hygiene for Children with Neurodevelopmental Disabilities** (used for my advancement to candidacy, referenced in Chapter 1, 2, and 3). This article addresses sleep hygiene principles associated with sleep environment, scheduling, sleep-practice, and physiologic sleep-promoting factors were analyzed for children with neurodevelopmental disabilities


- **Neurophysiology of Circadian Rhythm Disorders of Children with Neurodevelopmental Disabilities** (quoted in Chapter 1; referenced in Chapters 1, 2 and 3). This article outlines the neurophysiological background of circadian rhythm sleep disorders of children with neurodevelopmental conditions was reviewed, and a theoretical concept for delayed sleep onset was developed. It was written by the authors Jan, J.E., Bax, M., Owens, J.A., Ipsiroglu, O.S. and Wasdell, M., and published in the European Journal of Pediatric Neurology [Jan, J.E., Bax, M., Owens, J.A., Ipsiroglu, O.S., & Wasdell, M. (2012). Neurophysiology of circadian rhythm sleep disorders of children with neurodevelopmental disabilities. European Journal of Pediatric Neurology, 16(5), 403-412. doi: 10.1016/j.ejpn.2012.01.002].

- The current approach to paediatric sleep or how sleep problems are currently dealt with and should be dealt with by health authorities was analyzed in How to Approach Paediatric Sleep

- “‘They silently live in terror...’ Why Sleep Problems and Night-time Related Quality-of-life are Missed in Children with a Fetal Alcohol Spectrum Disorder’ (quoted in Chapter 2; referenced in Chapters 1, 2 and 3; full manuscript in Attachment I). I am the first and corresponding author, and the co-authors are W.H. McKellin, N. Carey, and C. Loock; it was published in Social Science & Medicine [Ipsiroglu, O. S., McKellin, W. H., Carey, N., & Loock, C. (2013). “They silently live in terror...” Why Sleep Problems and Night-time Related Quality-of-life are Missed in Children with a Fetal Alcohol Spectrum Disorder. Social Science & Medicine, 79, 76-83. doi: 10.1016/j.socscimed.2012.10.027].

- De-medicalizing sleep: sleep assessment tools in the community setting for clients (patients) with FASD & prenatal substance exposure (quoted in Chapter 1; referenced in Chapters 1, 2 and 3; full manuscript in Attachment 1). This publication operationalizes the initial clinical assessment concept, applied in the years 2010 to 2012. I am the first and corresponding author, and the co-authors are Carey, N., Collet, J.P., Fast, D., Garden, J., Jan, J.E., Kuttner, L., Loock, C., Lucyshyn, J. Schlarb, A.A. and Witmans M.; it was published by the National Organisation for Fetal Alcohol Syndrome – UK (NOFAS-UK) [Ipsiroglu, O. S., Carey, N., Collet, J., Fast, D., Garden, J., Jan, J. E., . . . Witmans, M. (2012). De-medicalizing sleep: sleep assessment tools in the community setting for clients (patients) with FASD & prenatal substance exposure. NOFAS-UK: Fetal Alcohol Forum.

- The assessment concept (referenced in Chapters 2 and 3) was presented as a workshop organized by FASD Deutschland in 2012 and then published in the German book article Sleep & Chronic Care-Management of Children with FASD. I am the first and lead author, a previous developmental paediatrics fellow, D. Veer, the co-author [Ipsiroglu, O. S., & Veer, D. (2012). Sleep & Chronic Care-Management of Children with FASD. In E. Paditz & O. S. Ipsiroglu (Eds.), Facetten eines Syndromes [Facets of a Syndrome]. Dresden, Germany: Kleanthes.].


- “Diagnosis by Behavioural Observation” Home-videosomnography - A Rigorous Ethnographic Approach to Sleep of Children with Neurodevelopmental Conditions (quoted in Chapter 2; referenced in Chapters 1, 2 and 3; full manuscript in Attachment 1). I am the first and corresponding author, and the co-authors are Hung, Y-H. A., Chan, F., Ross, M.L., Veer, D., Soo, S., Ho, G., Berger, M., McAllister, G., Garn, H., Kloesch, G., Barbosa, A.V., Stockler, S., McKellin, W., and Vatikiotis-Bateson, E.; this article was published in Frontiers in Psychiatry [Ipsiroglu, O.S., Hung Y.H., Chan, F., Ross, M.L., Veer, D., Soo, S., Ho, G., . . . Vatikiotis-Bateson E. (2015) "Diagnosis by

- *Structured Behavioural Daytime Observations in Children with Neurodevelopmental Conditions on the Willis Ekbom Disease/Restless Legs Spectrum with Early Onset and Chronic Insomnia* (quoted in Chapter 2; referenced in Chapters 1, 2 and 3; full manuscript in Attachment 1): presents the development of novel daytime focused structured behavioural observation methodology for unveiling the dimension of challenging/disruptive behaviours in children and youth with neurodevelopmental conditions due to WED/RLS. This research became core in understanding the effects of WED/RLS in developmental paediatrics and child psychiatry. I am the first and corresponding author, and the co-authors are Beyzaei, N., Dhalla, S., Wagner, A., Berger, M., Garden, J., and Stockler, S. This manuscript is in preparation for the *European Journal of Paediatric Neurology* [Ipsiroglu, O. S., Beyzaei, N., Dhalla, S., Wagner, A., Berger, M., Garden, J., & Stockler, S. (2015). Structured Behavioural Daytime Observations in Children with Neurodevelopmental Conditions on the Willis Ekbom Disease/Restless Legs Spectrum with Early Onset and Chronic Insomnia. Manuscript in preparation.].

- *Pathways to Overmedication and Polypharmacy: Case Examples from Adolescents with Fetal Alcohol Spectrum Disorder* (quoted in Chapter 2; referenced in Chapters 1, 2 and 3; full manuscript in Attachment 1). I am the first and corresponding author, the co-authors are Berger, M., Lin, T., Elbe, D., Stockler-Ipsiroglu, S., and Carleton, B. and published by Elsevier [Ipsiroglu, O.S., Berger, M., Lin, T., Elbe, D., Stockler-Ipsiroglu S., & Carleton, B. (2015) Pathways to Overmedication and Polypharmacy: Case Examples from Adolescents with Fetal Alcohol Spectrum Disorders. In N. Di Pietro & J. Illes (Eds.), *The Science and Ethics of Antipsychotic Use in Children*. (pp. 125-148). Waltham, MA: Elsevier.].
Sleep Problems in Children and Adolescents with Neurodevelopmental Conditions: Impact on Families and Suggestions on How to Improve Diagnostic Recognition from a Patient/Stakeholder Point of View (referenced in Chapter 3): includes interviews conducted by my research partner of service providing institutions about their current understanding of the dimension of untreated sleep problems. Four core solution strategies were also elucidated. I am the last and corresponding author, and Timler, K. is the co-author. This article is in preparation for the BC Medical Journal [Timler, K., & Ipsioglu, O. S. (2015). Sleep Problems in Children and Adolescents with Neurodevelopmental Conditions: Impact on Families and Suggestions on how to Improve Diagnostic Recognition from a Patient/Stakeholder Point of View. Manuscript in preparation.].

The research undertaken during this PhD endeavour is covered under the UBC Behavioural Ethics Research Board approval H10-03466.
Table of Contents

Abstract ................................................................................................................................. ii

Preface ................................................................................................................................. iv

Table of Contents ................................................................................................................ xi

List of Tables ....................................................................................................................... xiv

List of Figures ....................................................................................................................... xv

List of Abbreviations ......................................................................................................... xvi

Glossary ............................................................................................................................... xvii

Acknowledgements ........................................................................................................... xviii

Dedication ............................................................................................................................. xx

Chapter 1: Introduction to the Main Topics of an Interdisciplinary Dissertation .............. 1

1.1. Presented Concepts and Research Overview: Ethnographic Exploration of Ecologies
   - The Operationalization of Therapeutic Emplotment and Further Development of
     Emplotment-Based Methodologies. .............................................................................. 1
   1.1.1. The Concepts ......................................................................................................... 1
   1.1.2. Research Questions ............................................................................................... 2
   1.1.3. Research Overview ............................................................................................... 5

1.2. Sleep Problems in Children with Neurodevelopmental Conditions: Adoption of an
    Orphan into Medical Concepts or Why We Do need an Ethnographic Exploration of the
    Ecologies of Paediatric Intractable Insomnia? .................................................................. 7
   1.2.1. Etiological and Neurophysiological Aspects ......................................................... 7
   1.2.2. Sleep and Developmental Conditions ................................................................... 10
   1.2.3. Case Vignette #1 ................................................................................................. 11
1.3. Medical Anthropology and Educational Psychology: The Concepts of Ethnography and Ecology for Individualizing Medical Assessments ......................................................... 16

1.3.1. The Concept of Ethnography .................................................................................................................. 17
1.3.2. The Value of Exploration and Bidirectional Communication ................................................................. 22
1.3.3 The Concept of Ecology ............................................................................................................................. 25
1.3.4 Conclusion of Section 1.3 .......................................................................................................................... 29

1.4. Exploring Sleep/Wake Health .................................................................................................................... 30

1.4.1. Cultural Aspects ........................................................................................................................................ 30
1.4.2. Sleep/Wake Health in Paediatrics ........................................................................................................... 33
1.4.3. Child Psychiatry and Developmental Paediatrics Perspectives ............................................................. 35
1.4.4. The Clinical Reality: Life Trajectories ..................................................................................................... 36

1.5. Questions Motivating my Research ........................................................................................................... 37

Chapter 2: Methodology and Results ................................................................................................................ 38

2.1. Research Question #1: “Why have sleep problems remained unrecognized in children with neurodevelopmental conditions?” Application of Medical Ethnography & Ecology Concepts for Understanding the Dimension of the Problem in Everyday Clinical Practice ......................................................................................................................... 38

2.1.1. “They silently live in terror...” Why Sleep Problems and Night-time Related Quality-of-Life are Missed in Children with Fetal Alcohol Spectrum Disorder ................................................................................................................................. 38

2.2. Research Question #2: “How can we optimize our clinical understanding and try to describe the possible dimension of a problem?” Structured Behavioural Observations – Application of Medical Ethnography in Exploration of Movement Patterns................................................................. 59

2.2.1. Structured Behavioural Daytime Observations in Children with Neurodevelopmental Conditions on the Willis Ekbom Disease/Restless Legs Spectrum with Early Onset and Chronic Insomnia ......................................................................................................................... 59

2.2.2. “Diagnosis by Behavioural Observation” Home-vid eosomnography – A Rigorous Ethnographic Approach to Sleep of Children with Neurodevelopmental Conditions ................................................................. 93
2.3. Research Question #3: "What are the consequences of unrecognized sleep problems?" Analysis of Overmedication and Polypharmacy Data..........................112

2.3.1. Pathways to Overmedication and Polypharmacy: Case Examples from Adolescents with Fetal Alcohol Spectrum Disorders.......................................................................................................................... 112

Chapter 3 : Discussion.................................................................................................................................133

3.1. Recognizing and Surmounting Communication Barriers and Cultural Bias ..........135
3.2. Applied Emplotment.....................................................................................................................................141
3.3. How to Deal with Medications or the Phenomenon of iatrogenic Harm..................145
3.4. Closing the Gaps ......................................................................................................................................152
3.5. Highlights on the Horizon ........................................................................................................................155
3.6. Relevance of this Research; Strength & Weaknesses..........................................................156
3.7. Final Remarks .........................................................................................................................................160

Endnotes.........................................................................................................................................................162

References.....................................................................................................................................................173

Appendix A: Patient Assessment Reports....................................................................................................212

A.1. Typical Sleep Consultation Report........................................................................................................212
A.2. Sleep/Wake Behaviour Assessment Report ........................................................................................214
A.3. Abbreviated Immediate Report............................................................................................................219
A.4. Outreach Sleep/Wake Behaviour Assessment Report ......................................................................221
A.5. Immediate Abbreviated Report – Medication History ...................................................................227
List of Tables

Tables in Chapter 2

Table 2.1. The Characteristics of the Participants Summarized using Descriptive Statistics. 45

Table 2.2. Maternal Sleep and WED History Data. 71

Table 2.3. Patient Demographics and Day/Nighttime Clinical Presentations in Paediatric Patients with WED. 71

Table 2.4. WED/RLS Indicators in Paediatric Patients. 72

Table 2.5. Parental Descriptions of Paediatric Sensory Processing Abnormalities. 74

Table 2.6. Patient Profiles (Reference List). 75

Table 2.7. Video Summary. 103

Table 2.8. Overview of Problems. 110

Table 2.9. Patient Demographics and Day/Nighttime Clinical Presentation in Paediatric Patients with FASDs and/or PSE. 120

Table 2.10. Overview of the Prescription Medications, Including: Most Common Medications and First Prescription Medication Given to Patients with an FASD and/or PSE diagnosis. 121
List of Figures

Figures in Chapter 1

Figure 1.1. Medication Use Details for Case Vignette. ................................................................. 14

Figure 1.2. Life-Trajectory Graph for Case Vignette. ................................................................. 15

Figures in Chapter 2

Figure 2.1. Steps of Analysis............................................................................................................. 99

Figure 2.2. Setup Requirements and Domains of Qualitative Analysis. ..................................... 100

Figure 2.3. Mind Map of the Patient Factors. ............................................................................... 101

Figure 2.4. Mind Map of the General Setting.. ............................................................................. 102

Figure 2.5. Home-videosomnography Hypnogram........................................................................ 105

Figure 2.6. Map of BC with Video Equipment Destinations in the Last Period of the Year 2013 – 2014................................................................................................................................. 109

Figure 2.7. Explanatory Model: Impact of Sleep Problems on Day/Nighttime Presentations.... 128
## List of Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>ADHD</td>
<td>Attention deficit hyperactivity disorder</td>
</tr>
<tr>
<td>Autism</td>
<td>Autism spectrum disorder</td>
</tr>
<tr>
<td>CR</td>
<td>Controlled release</td>
</tr>
<tr>
<td>FASD(s)</td>
<td>Fetal alcohol spectrum disorder(s)</td>
</tr>
<tr>
<td>LA</td>
<td>Long-acting</td>
</tr>
<tr>
<td>RLS</td>
<td>Restless legs syndrome</td>
</tr>
<tr>
<td>SA</td>
<td>Short-acting</td>
</tr>
<tr>
<td>SCIT</td>
<td>Suggested Clinical Immobilization Test</td>
</tr>
<tr>
<td>SIT</td>
<td>Suggested Immobilization Test</td>
</tr>
<tr>
<td>WED</td>
<td>Willis Ekbom disease</td>
</tr>
<tr>
<td>WED/RLS</td>
<td>Willis Ekbom disease/restless legs syndrome</td>
</tr>
<tr>
<td>XR</td>
<td>Extended release</td>
</tr>
</tbody>
</table>
Glossary

**Electrophysiological data:** The electrical recording techniques that enable measurement of the flow of ions in biological tissues.

**Hypnagogic hallucinations:** Visual, tactile, auditory, or other sensory events that occur during the transition from wakefulness to sleep.

**Home-videosomnography:** A method for recording sleep and screening for sleep disorders using low-cost video equipment.

**Insomnia:** A sleep disorder characterized by difficulty with sleep onset and/or sleep maintenance.

**Neurodevelopmental conditions:** Disorders characterized by impairments to the growth and development of the brain or central nervous system, e.g. autism spectrum disorders, fetal alcohol spectrum disorders, and Down syndrome.

**Parasomnias:** A category of sleep disorders characterized by abnormal movements, behaviours, emotions, perceptions, and dreams that occur while falling asleep, sleeping, between sleep stages, or during arousal from sleep.

**Polysomnography:** Considered the *gold standard* in sleep medicine; a used test to diagnose sleep disorders by recording brain waves, oxygen levels, heart rate and breathing, as well as eye and leg movements during the study.

**Sudden infant death syndrome:** An unexplained death, typically during sleep, of a seemingly healthy baby.

**Willis Ekbom disease/restless legs syndrome:** A neurological disorder that is characterized by unpleasant sensations, e.g. bugs crawling, in the legs and, occasionally, other body parts. Sufferers typically report an uncontrollable, and sometimes overwhelming, urge-to-move.
I would like to thank all of my patients and their families who supported me with their narratives which were initially unbelievably challenging but coherent. Without their eye-opening stories, needless suffering would continue to be perpetuated.

I would also like to thank all of the professionals who mentored me. Over my academic career, my mentors were Professors Ronald Kurz, Klaus Rosanelli, Hellfried Rosegger, Radvan Urbanek and Arnold Pollak in Austria, and Robert Armstrong, James E. Jan, Kojo Asante, Christine Loock, Stuart MacLeod in Canada. Their trust and constructive criticisms helped me to achieve my otherwise unachievable goals.

My teams of colleagues in Austria and Canada, which consisted of peers, allied health care professionals, and student team members made this work possible. I particularly would like to thank my research assistants over the last five years, Samara Mayer, Michelle L Ross, Amy Hung, and Tracy Pham for their valued input, and Mai Berger for her very much appreciated enthusiastic, trustful, but also critical contributions.

Finally, I would like to thank my colleagues and supervisors, Professors William McKellin, Michael Whitfield and Joseph Lucyshyn, who became over the years of ethnographic exploration and multiple ecology analyses, my friends and partners. Patience and generosity were necessary for making this cumbersome research exercise, which was initially mentored by Robert Armstrong (2007-2011), possible. My supervisors have this patience and generosity and have therefore, enabled my needed academic research space that I have been seeking since I started as a physician within the Canadian health care system. I hope that I can fulfill their high requirements, and that we can all take a step forward to stop the needless suffering of voiceless and vulnerable people – people who can quickly become victims of a professionalized, reproducible, mechanistic, non-
empathetic and non-reflective mainstream medicine, which perpetuates the same errors for decades and generations.
Dedication

This interdisciplinary PhD dissertation, which overstepped the typical University of British Columbia PhD timelines, is dedicated to my parents, Mazhar and Nazan Ipsiroglu, and my partner and spouse, Sylvia Stockler-Ipsiroglu.

My parents have been exemplary role models: my father was one of the first ethnographers in modern art history and, in consequence, some of his work was censored and presentation was prohibited (documentary film: On the Roads of Anatolia, 1958) or published in Turkish 40 years after its first publication in Germany (Ahtamar Kilisesi, Isikla Canlanan Duvarlar, YKY Istanbul, 2003; Die Kirche von Achtamar, Bauplastik im Leben des Lichtes, Kupferberg Verlag, 1963). My father (my mother, my sister and me) were exiled in 1960 for no reason other than for my father’s ethnographic approach to the ancient cultures of Asia Minor and the consequences of his libertine thinking, and amnestied a few years later. Despite his sensitive and warm character, he was perceived as a challenging personality by mainstream people. During all the turbulent years, my mother raised my sister and me with continuous warmth, care and authentic love – a foundation we have both built on during the challenges of academic careers and which has empowered us. My first email was in 1999 to my mother who had already been using the Internet for at least 5 years. My mother has always been an open-minded person to all challenges of the world – she inspired my love of the arts and particularly, music.

In Sylvia, my partner and spouse, I found an authentic character, who is free of mediocrity and false beliefs and is always open for the most challenging thoughts and concepts. Sylvia is also the person who encouraged me to embark on this fairly crazy second PhD endeavour. It was the right decision. This extra academic exercise has been essential in explaining the causes of cul-de-sacs in daily clinical practice in developmental paediatrics and child psychiatry. Over the next years, the presentations of Willis Ekblom disease/restless legs syndrome will become a modern parable.
depicting how the paradigm of evidence-based medicine is applied without reflection, which can restrain our clinical view and understanding, perpetuating systemic gaps.
Chapter 1: Introduction to the Main Topics of an Interdisciplinary Dissertation


1.1.1. The Concepts

Through my daily clinical work as a developmental paediatrician, I have seen an increasing number of children with undiagnosed and untreated sleep problems. I came to the realization that the lack of attention given to these problems by medical care providers resided in the scarcity of explanatory models for reported symptoms. All patients had received various developmental paediatrics and mental health diagnoses based on their daytime presentations (e.g. attention deficit hyperactivity disorder (ADHD), fetal alcohol spectrum disorders (FASDs); autism spectrum disorder (autism)). The effect that possible chronic sleep deprivation had on daytime presentation was either recognized but not investigated further or remained unrecognized and was therefore taken as given or even considered to be part of the condition.

Two key approaches helped me discover the inadequacies of the current clinical explanatory models that ignored children's sleep problems: ethnography and ecology. I learned these approaches while studying medical anthropology and educational psychology, and they became the methodological pillars for my clinical analyzes as well as my clinical research, as demonstrated in this dissertation. The wisdom of bi-directional communication, presented as the concept of ethnographic exploration of ecologies and operationalized as therapeutic emplotment, is derived from both disciplines. Bi-directional communication utilizes listening, watching, exploring
and describing as structured research methodologies. In my clinical work, I listened to patients’ narratives through qualitative interviewing (Greenhalgh, Russel, & Swinglehurst, 2005), which opened my eyes to new explanatory models of sleep problems in children with neurodevelopmental conditions.

The next step was to create a neutral explorative language (to avoid serving the diagnosis on a silver tray, as I teach accompanying trainees) to explore and negotiate clinical symptoms further (Fontana & Frey, 2000), and to develop new tools and methodologies for this exercise. This included watching and describing the presenting symptoms in a more in-depth way and the process of making them reproducible through structured day- and nighttime observations, which resulted in the development of a basic descriptive medicine. I started phenotyping sleep and wake behaviours in collaboration with parents and create explanatory models for some of the previously unexplained challenging/disruptive behaviours (Appendix A: Patient Assessment Reports). The utilization of narratives in clinical history taking (Kleinman, 1988), sharing the collaboratively worked out summary with parents (therapeutic emplotment) (Mattingly, 1994), and the creation of reproducible qualitative and quantifiable methodologies for the documentation of various Willis Ekborn disease/restless legs syndrome (WED/RLS) day- and nighttime presentations (SCIT and home-videosomnography, respectively) are the core parts of this PhD endeavour.

1.1.2. Research Questions

The research presented in this dissertation, as well as in the publications, was inspired by the clinical observation that the sleep problems of children with chronic conditions are frequently undiagnosed and consequently, inappropriately treated. This observation elicited further questioning, and there remained a big ‘Why?’ To answer this question it was necessary to examine the categorical diagnosis-based explanatory models used by physicians to identify the limitations that contribute to a failure to identify sleep problems, and explore the experiences of parents and
health care professionals (e.g. occupational therapists), presented as *eye-opening* descriptions, and the various explanatory models that they had employed. The manuscript ‘“They silently live in terror...” Why Sleep Problems and Night-time Related Quality-of-Life are Missed in Children with Fetal Alcohol Spectrum Disorder,’ which is presented in Chapter 2, section 2.1.1 responds to this core question (Ipsiroglu, McKellin, Carey, & Loock, 2013).

To appropriately recognize the parents and children’s understanding of the symptoms of intractable insomnia, predominantly caused by WED/RLS, I employed the concept of ethnographic exploration of the *family* and *sleep ecology*. *Family ecology* acknowledges the impact of family dynamics, the broader network of social relationships on the experience of the condition, and supports understanding of how the underlying neurodevelopmental condition has impacted the family as the main caregiver (Lucyshyn & Albin, 1993; Lucyshyn, Albin, & Nixon, 1997). *Sleep ecology* acknowledges the impact of the applied existing sleep culture, including routines, perceptions and notions (Worthman & Melby, 2002). Using such explorative strategies for investigation consequently led to the second research question: ‘*How can we optimize our clinical understanding and try to describe the possible dimension of intractable insomnia?*’ The manuscript ‘“They silently live in terror...” Why Sleep Problems and Night-time Related Quality-of-Life are Missed in Children with Fetal Alcohol Spectrum Disorder,’ investigates clinical history taking and the development of a therapeutic relationship (Ipsiroglu et al., 2013). Parallel to this manuscript, I prepared two clinical guideline papers on how clinical best practice should be applied (Ipsiroglu, Carey, Collet, Fast, Garden, et al., 2012; Ipsiroglu & Veer, 2012); both of these manuscripts are not presented in this dissertation. The two manuscripts presented in Chapter 2, section 2.2, examine how structured behavioural observations, as an application of medical ethnography in exploration of movement patterns at daytime (section 2.2.1) (Ipsiroglu, Beyzaei, Dhalla, Wagner, Berger, et al., 2015) and at nighttime (section 2.2.2) (Ipsiroglu, Hung, Chan, Ross, Veer, et al., 2015), can
overcome the current gap in describing and understanding familial WED/RLS, the underlying cause of familial intractable insomnia.

The third research question, which is the first and initial validation of the presented clinical methodologies, became: ‘What are the consequences of unrecognized sleep problems?’ The analysis of clinical data provided the answer to this research question. The core manuscript addressing this research question is found in Chapter 2, section 2.3.1 (Ipsiroglu, Berger, Lin, Elbe, Stockler, et al., 2015). This question is also addressed in one case report, which was prepared for publication (Ipsiroglu, Elbe, & Carleton, 2015) as well as in an additional German article (Ipsiroglu, 2015). These articles are referenced throughout this dissertation.

Applying these methodologies in paediatric sleep medicine exceeded the traditional clinical approach as they (i) explore challenging and disruptive sleep problems or sleep behaviours in context with the patient’s challenging and disruptive daytime behaviours, presented as a narratives by the parents (ii) investigate the functional diagnoses, (iii) consider the medication history alongside the familial sleep history, as well as (iv) the situation or family reality, and family and sleep ecology in a similar manner, which supports the collaborative review of the patients’ and parents’ narratives and explanatory models from a novel developmental paediatrics and child psychiatry/mental health sleep medicine perspective. These four aspects are presented under the medical anthropologic concept of ethnographic exploration of ecologies, operationalized as therapeutic emplotment.

Therapeutic emplotment supports the individually tailored assessment of what parents/caregivers or professionals in the medical or educational system see and define as norm or exceeding the norm (Mattingly, 1994). These various understandings are based on the very specific, individual background, education and training culture, in other words one’s individual culture (Kleinman, 1988; Helman, 2007). Therapeutic emplotment stands here for bi-directional
communication at all levels of explorative communication and for creating a commitment to the therapeutic relationship. In my particular realm of work and research, therapeutic emplotment also stands for the purpose of investigating and re-evaluating previously given clinical diagnoses, medication effects/side effects and adverse drug reactions to overcome previously or currently existing communication barriers in medicine.

1.1.3. Research Overview

This PhD endeavour can be categorized into three phases, which reflect the different stages in my learning experience and the development of the concepts of ethnography and ecology as qualitative methodologies applied to my clinical research. The publication list presented in the Preface gives an overview of these three phases and clarifies any questions regarding intellectual property and my contributions (first, corresponding and/or co-authorship) in the development and application of the described methods, which make up the contents of my dissertation.

Chapter 1 is an introduction to the main topics of the dissertation, and includes a case report describing the medical problem of limited understanding, thus frames the entire research endeavour from a clinician’s viewpoint in sub-section 1.1.2. While learning to apply ethnography and ecology in clinical practice, I used these concepts in four main manuscripts, Transkulturelle Paediatrie, (Ipsiroglu, 2008a; Ipsiroglu & Aksu, 2009; Ipsiroglu, Stockler, & McKellin, 2010; von Kries, Strassburg, Ipsiroglu, & Aksu, 2013) and ‘Cultural Aspects in the Management of Inborn Errors of Metabolism’ (Stockler, Moeslinger, Herle, Wimmer, & Ipsiroglu, 2012) as well as in various talks (including in three Grand Rounds at BC Children’s Hospital). These four manuscripts demonstrate the application and added values of ethnographic exploration and ecology in paediatrics and has been the continuation of a research endeavour I have been involved for the last 15 years (Ipsiroglu, Herle, Spoula, Möslinger, Wimmer, et al., 2005; Ipsiroglu, Jan, Freeman, Laswick, Milner, et al., 2009;

Chapter 2 is presented in three parts, each exploring and responding to a research question:

(i) Part one (2.1) focuses on the application of *ethnographic exploration* of *ecologies* and operationalization as *therapeutic emplotment* in the clinical research and responds to research question #1: (i.e. the big *'Why?'*) *Why have sleep problems remained unrecognized in children with neurodevelopmental conditions* (Ipsiroglu et al., 2013)?

(ii) Part two (2.2) focuses on the medical background and implementation of new clinical explorative methodologies, such as *structured behavioural observations*, which were developed within this PhD research endeavour as well as their clinical application for solving the observed challenges (including several anonymized sleep/wake-behaviour reports) and responds to the research question #2: *'How can we optimize our clinical understanding and try to describe the possible dimension of a problem?'* (Ipsiroglu, 2015; Ipsiroglu, Beyzaei, et al., 2015; Ipsiroglu, Carey, et al., 2012; Ipsiroglu et al., 2013; Ipsiroglu, Hung et al., 2015; Ipsiroglu & Veer, 2012).

(iii) Part three (2.3) explores in the two case series with 17 and 14 patients, respectively, the clinical and social consequences of not investigated sleep problems, and responds to research question #3 *'What are the consequences of unrecognized sleep problems?'* (Ipsiroglu, 2015; Ipsiroglu, Berger, et al., 2015; Ipsiroglu, Elbe, et al., 2015).

Chapter 3 discusses the clinical observation, presented methodologies and findings, which were achieved when these methodologies were applied from medical anthropology, educational psychology and developmental paediatrics/mental health viewpoints, and presents a solution-based strategy, based on the concept of personalizing care.
1.2. Sleep Problems in Children with Neurodevelopmental Conditions: Adoption of an Orphan into Medical Concepts or Why We Do need an Ethnographic Exploration of the Ecologies of Paediatric Intractable Insomnia?

1.2.1. Etiological and Neurophysiological Aspects

Sleep is a neurologic function that is essential for childhood development, physical health, and psycho-mental wellbeing (Mindell & Owens, 2009). Consequently, sleep problems should be of particular concern in children with neurodevelopmental conditions. In the first comprehensive review of sleep habits of children with neurodevelopmental conditions affecting sleep and sleep-wake behaviours, Jan and his colleagues (2008) estimated a 75-80% prevalence rate of chronic sleep problems for these children, in contrast to 30-35% prevalence in otherwise healthy children (Jan et al., 2008). A majority of these sleep problems manifest themselves as insomnia, difficulties in falling asleep and maintaining sleep, resulting in chronic sleep deprivation (Jan et al., 2010; Malow et al., 2012; Wiggs & Stores, 1996; Zucconi & Bruni, 2001).

Sleep deprivation in children generally manifests itself as daytime inattention, hyperactivity, and mood disturbances (Chervin, 2000; Gottlieb et al., 2003); this is also the case in patients with neurodevelopmental conditions (Jan et al., 2008). However, among children with neurodevelopmental conditions, sleep problems are only one of many different types of medical problems and as a result, often go undiagnosed and untreated (Malow et al., 2012). Moreover, the causal role of sleep problems in children’s behaviours and in their entire clinical picture may remain underestimated or unrecognized (Johnson & Malow, 2008). For example, both sleep deprivation and non-restorative sleep result in the same types of daytime hyperactive behaviours and attention deficits as observed among children with ADHD. If some daytime hyperactivity and attention deficit-like behaviours can be the result of an unrecognized sleep problem (Subcommittee
on Attention-Deficit/Hyperactivity Disorder; Management, Steering Committee on Quality Improvement and Management (Sub. Attention-Deficit), 2011) then the current state-of-the-art treatment with stimulants might even exacerbate the underlying sleep problems, as presented in the introductory case report (Ipsiroglu, Elbe, et al., 2015). This may in turn create a Catch-22 situation that produces further deterioration of day- and nighttime behaviours.

**Spectrum of Sleep Disorders.** Most sleep problems encountered in children with neurodevelopmental conditions are due to a myriad of conditions affecting multiple organ systems and functions, which are essential prerequisites for a normal sleep. Conditions include craniofacial malformations (e.g. cleft palate syndromes, high arch palate), neuromuscular disorders (e.g. Duchenne Becker muscular dystrophy), rare biochemical disorders (e.g. mucopolysaccharidoses), obesity, and Down syndrome, and the neurologic disorder, WED/RLS. The underlying pathophysiology includes, upper airway obstructions, muscle weakness impacting respiratory drive, and reduced secretion of sleep related hormones (mainly melatonin), neurotransmitters (e.g. dopaminergic substances) and sensorimotor symptoms. Further, epilepsy (e.g. manifesting as seizures during the night), discomfort or pain, hunger or gastrointestinal tract irritation (e.g. through nocturnal gastrostomy tube feeding), and side effects of pharmacological treatments are additional causes of sleep problems in children with neurodevelopmental conditions. Psychological trauma, mental health disorders (e.g. ADHD), as well as autism spectrum disorder, FASDs, congenital or acquired brain injuries and various forms of cerebral palsy are almost invariably associated with sleep problems. Pathophysiological targets for specific treatments of sleep problems are widely unknown; therefore, therapies mainly focus on interventions targeting daytime symptoms. The research question #2 (‘How can we optimize our clinical understanding and try to describe the possible dimension of a problem?’) reflects the following challenge: ‘Can we better phenotype and differentiate symptoms in order to tailor therapies and target pathophysiological cause?’
Sleep Disordered Breathing. Research on sleep disordered breathing has been mainly been conducted in the domain of lab-based research for the last 35 years. It focused initially on infants with periodic breathing, as well as on central and obstructive apnoea. The focus then shifted to the broader spectrum of sleep disordered breathing, and included hypopnea, upper airway restrictions and flow limitations. Recently, there has been some change in understanding or consensus that in addition to lack of oxygen, desaturation and limb movement-associated arousals can result in a fragmentation of sleep and are the main causes of non-restorative disturbed sleep.

Insomnia in Children. Insomnia in children has been mainly considered a behavioural problem, as almost all children have not only experienced, but also caused or triggered some of their parents’ transient insomnia due to learned behaviours (Ferber, 2006). Successful analyzes of potential causes for behavioural insomnia led paediatricians, psychologists and researchers to focus on sleep hygiene measures, which were investigated and analyzed from various aspects (Mindell & Owens, 2009). The reviews in this area, which framed this research endeavour, were conducted under the leadership of my mentor, Dr. James E. Jan, and impressively demonstrated the limitations in our understanding and the need for more phenomenological research.

Willis Ekbom Disease/Restless Legs Syndrome (WED/RLS). Recently, sleep researchers re-detected and began to re-research WED/RLS symptoms in children and adolescents. The clinical picture of WED/RLS is well known in adults, and was first described by Sir Thomas Willis in 1672 (Coccagna, Vetrugno, Lombardi, & Provini, 2004). Most adults suffering from WED/RLS describe that they first experienced symptoms at a very young age. WED/RLS may cause delayed sleep onset in children with neurodevelopmental conditions and can contribute significantly to childhood insomnia. The discomfort and discomfort-related urge to move most likely also delay pineal melatonin production and secretion.
When I started my clinical work at the Department of Paediatrics at the University of British Columbia, there was not a single paediatric patient diagnosis of WED/RLS in British Columbia, nor was there a paediatric patient with a neurodevelopmental condition and a diagnosis of WED/RLS in the literature. This was a puzzling observation, as WED/RLS typically accompanies sensory processing abnormalities, and many children with neurodevelopmental conditions experience major sensory processing abnormalities (Ipsiroglu, Beyzaei, et al., 2015).

1.2.2. Sleep and Developmental Conditions

Sleep is an integral component of cerebral function and a prerequisite for the development of cognitive, emotional and behavioural skills. The negative impact of sleep problems on child development has been well documented (Wiggs & Stores, 2001; Wiggs & Stores, 2004). Transient sleep problems are highly prevalent across all ages and cause major problems (Gruber et al., 2013); chronic sleep problems have devastating consequences as they adversely affect cognition, emotion, and behaviours (Sivertsen, Posserud, Gillberg, Lundervold, & Hysing, 2012; Wolfson & Montgomery-Downs, 2013).

Up to 20% of children and adolescents live with a chronic physical, neurodevelopmental, behavioural or emotional conditions (Sloper & Beresford, 2006), and up to 80% of these children suffer from chronic sleep problems (Jan et al., 2008; Krakowiak, Goodlin-Jones, Hertz-Picciotto, Croen, & Hansen, 2008; Sivertsen et al., 2012). However, the degree to which early-onset and chronic sleep problems in children with underlying developmental conditions can contribute to (a) aggravation of developmental delay or intellectual disability; (b) mental health disorders; or (c) trigger artificial diagnosis and medication cascades has not been investigated.
1.2.3. Case Vignette #1

The following case vignette is representative of many other cases I have encountered in my developmental paediatrics practice, highlighting the crucial role of sleep in the life trajectory of children with neurodevelopmental conditions. It uses all three of the qualitative methodologies that I have investigated in my dissertation research for diagnosing the underlying neurological disorder, which mimics various clinical presentations and significantly affects neurodevelopment and life trajectory. This case report is presented more in-depth in the manuscript ‘Challenging Sleep/Wake-Behaviours leading to a Cascade of Diagnoses and Medications; Case Report,’ which has been prepared for publication (Ipsiroglu, Elbe, et al., 2015). This case vignette has been visualized with a life trajectory chart (Figure 1.1), depicting the effects of the medication on diagnoses and life events, such as the eventual family breakdown and placement in a group home.

**Case Vignette #1.** A female patient with an FASD diagnosis and insomnia (falling asleep/sleep maintenance problems) reported since childhood, was referred by her psychiatrist for a paediatric sleep assessment. Concerns of speech delay, behavioural control and social interaction were initially raised in kindergarten (age five years) and probability of autism spectrum disorder was noted, and eventually ruled out (by age seven years). At age seven, she was diagnosed with ADHD and partial fetal alcohol effects; co-morbid anxiety, reactive attachment and mixed receptive/expressive language disorders were diagnosed at age nine. She was described to have obsessive compulsive and oppositional defiant-like behaviours at age 11. A total of 11 psychotropic medications were trialed, starting shortly after the initial ADHD diagnosis at age seven to 12 years, with little to no success.

The neurologic/ behavioural/metabolic adverse drug reactions to prescribed medications contributed to two emergency admissions to psychiatry. She was seven years old when first prescribed methylphenidate (Ritalin®), which was ineffective and in fact exacerbated her oppositional behaviour. She was simultaneously trialed on atomoxetine and again methylphenidate (Concerta®), both of which
were “unable to alleviate symptoms of inattention, hyperactivity, and impulsivity” in addition to aggravating her sleep difficulties. She was hospitalized at age nine for a behavioural outburst. At the hospital, she was switched from her previous three ADHD medications to mixed amphetamine salts (Adderall XR®). This proved unsuccessful and concerns regarding “poor appetite and suppression of normal spontaneity and enthusiasm” were raised. At 20 mg of mixed amphetamine salts, she experienced tachycardia. Risperidone was prescribed and proved to be ineffective, wherein her oppositional and disinhibited behaviour increased. In addition, she experienced marked weight loss and appetite suppression from stimulant medication (“while [it is suspected that] her prenatal alcohol exposure is part of the picture, likely her stimulant medications are making things worse”), and was prescribed appetite stimulants such as cyproheptadine as a result.

For falling asleep problems, the patient was medicated with melatonin, which “did not help and induced parasomnias.” Parasomnias are sleep disorders that present as movements (e.g. sleep walking), behaviours (e.g. sleep talking or night terrors), perceptions and dreams (e.g. hypnagogic hallucinations, like in the case of our patient) while falling asleep and/or between sleep stages (American Academy of Sleep Medicine, 2001). In consequence, she was prescribed quetiapine, which caused lipid abnormalities (high low-density lipoprotein; borderline high-density lipoprotein; and thus, an elevated risk ratio and was suggested as contributor to her drastic weight loss. I reference one of her previous physicians, who described her as “one of the sickest children [she] had ever seen.”

The experienced side effects and/or neurologic/behavioural/ metabolic adverse drug reactions contributed to her two emergency admissions to psychiatry (April 2009, August 2009, November 2011); the eventual breakdown in her adoptive (extended) family resulted in her permanent placement in a group home (February 2010). Improvement of her challenging behaviours occurred after diagnosis of her underlying familial neurological condition, WED/RLS, and addressing the WED/RLS-related discomfort and sequelae (insomnia and restless sleep) with gabapentin at the age of 12. Improvements in her clinical presentation are as follows: (a) amount of sleep increased; (b) her sleep
became restful and clinically more restorative as the number of her counted awakenings overnight decreased from approximately thirteen to zero; and (c) after a few months of therapy with gabapentin (Magnus, 1999; Picchietti et al., 2013; Robinson & Malow, 2012), her anxiety decreased and she began to be able to sleep alone. In follow-up, it was retrospectively recognized that her anxiety was caused by hypnagogic hallucinations (occurring at transition from wakefulness to sleep). After 18 months of treatment, teachers describe the patient as resilient, she was able to advance in her math class from grade 3 to 7, and could be sent to school alone on public transportation as she no longer requires one-to-one supervision.

Retrospective analysis reveals that the patient’s sleep problems were evident since early toddler age. However, as she came from a family of insomniacs and sleep deprivation was seen as the norm; therefore, exclusive focus was placed on the challenging and disruptive daytime behaviour that were perceived as sequelae of the FASD diagnosis. As such, the sleep problems had been taken as given, and later on addressed with melatonin and quetiapine. At her current age of 14, the patient is only on neuropathic pain medication and disagrees with the need for psychostimulants to address her daytime ADHD-like behaviours.
Figure 1.1. Medication Use Details for Case Vignette. This figure depicts the medication use order and time courses for the case vignette. Medications for ADHD-like behaviours are depicted in red (and are italicized; for black and white print); atypical antipsychotics and SSRIs are depicted in blue and in purple (and with one star and two stars), respectively; supplements are depicted in black; and medications targeting sleep are depicted in green (and underlined). The numbers preceding the medications represent the chronological order of each medication. Medications that are on-going (e.g. not discontinued) are depicted with an arrow. Abbreviations: short-acting (SA); long-acting (LA), extended release (XR), and controlled release (CR).
This case vignette shows impact of missed sleep problems visualized on a life-trajectory graph. The patient presented with chronic sleep problems since very early childhood caused by WED, which had been (mis-)labelled as ADHD and was diagnosed with multiple mental health co-morbidities (delays noted at age five; FASD and ADHD diagnosed at age seven; anxiety, attachment and language disorders at age nine). In the medical records, sleep problems are noted at age six, and described as associated with starting medications. However, in the caregiver narrative, sleep problems are mentioned as early as 19 months. She was treated for challenging/disruptive daytime behaviours with 11 prescription medications in various combinations, including: hypnotics, stimulants, anti-psychotics, anti-depressants, anti-seizure, and blood pressure medications, and up to 11 medications labelled as ‘food-supplements’. The patient experienced several severe adverse drug reactions with emergency admissions to paediatric or mental health facilities and a significantly impacted family ecology. Improvement of her challenging/disruptive behaviours occurred after diagnosis of the familial WED and treatment of WED-related discomfort with neuropathic pain medication. Abbreviations: short-acting (SA); long-acting (LA), extended release (XR), and controlled release (CR).
Recognition of a sleep problem and its possible aetiology is an important clinical challenge. Due to limited knowledge, symptoms may go unrecognized as sleep-related by clinicians and/or parents. The eventual goal of a clinical assessment is to produce an established diagnosis, as diagnoses enable the development of a concrete action plan and tailored services to support the affected individual. In clinical practice, a professional’s exploratory model of sleep problems is based on conventional clinical diagnoses, which typically present in adult patients and that had been well studied. The missing WED/RLS diagnoses in children with neurodevelopmental conditions is based on this fact – as young children or children with neurodevelopmental conditions are incapable of explaining the obligatory diagnostic criteria of an urge to move in their own words it has simply remained unrecognized and missed. In cases where understanding of the entire clinical presentations is limited, inability to recognise the diagnostic nuances of a sleep problem may result in misdiagnosis, and encourage excessive focus on daytime presentations and diagnoses, as shown in Case Vignette #1 (section 1.2.3). Therefore, the need for a novel approach to overcome the current diagnostic barriers became evident.

1.3. Medical Anthropology and Educational Psychology: The Concepts of Ethnography and Ecology for Individualizing Medical Assessments

In my clinical practice, I noticed that successfully targeting patients’ sleep problems led to an improvement in their well-being, quality of life, and also of the underlying neurodevelopmental condition. This finding is well documented and reproduced in the manuscripts included in this dissertation as well as in the introductory case report (section 1.2.3) and Appendix A. As such, in order to address the underlying foundational question of this dissertation research: ‘How do sleep problems affect the everyday lives of children with neurodevelopmental conditions?’ I had to develop a different diagnostic approach. The observed cause-and-effect interaction between sleep problems, well-being and neurodevelopment made an in-depth phenotyping concept necessary. This concept
had to exceed the current, single-cause-and-single-effect interaction concepts as they had not been sensitive enough to capture the possible underlying problems. In order to make my observations and associated thoughts of diagnostic/medication cascades reproducible, this approach had to be very descriptive.

I had to create a systematic assessment methodology for analysis of paediatric sleep/wake-behaviours to include clinical in-depth phenotyping.

In consequence, the entire clinical encounter and associated follow-up process had to be adapted and individualized to improve my/our clinical understanding, including: (a) the clinical assessment concept; (b) the hypothetic diagnosis generation; and (c) outcome monitoring, which is also quality controlled in close collaboration with the parents/caregivers. This forced me to restructure and exceed the current paediatric sleep assessment concept. After acquiring initial follow-up data, I was able to analyze the validity of these methodologies. The results of this analysis show that the implementation of descriptive qualitative methodologies overcame many of the barriers, which have been related to culture and training-related communication (Garcia-Borreguero et al., 2011), as well as to the limitations associated with the widely criticized technique and data collection-centered western medicine (Illich, 1974). These methodologies furthermore individualize the assessment and treatment process for patients with neurodevelopmental conditions in paediatric sleep medicine. In the following paragraphs, I will give an overview of these emerging concepts, which have been utilized for the in-depth phenotyping exercise.

1.3.1. The Concept of Ethnography

Ethnography is a concept of exploration and observation; the scientific description of peoples and cultures with their customs, habits, and mutual differences (Ethnography [Def. 1], n.d.). Anthropologists Arthur Kleinman and Peter Benson (2006) describe that:
[The ethnographer practices an intensive and imaginative empathy for the experience of the natives—appreciating and humanly engaging with their foreignness [...], and understanding their religion, moral values, and everyday practices [...]. (p. 1674)]

Anthropologists customarily explore cultural phenomena through observation of society from the point of view of the subject of their studies (Powell & Boas, 1887). The motivations, viewpoints and bias in their research have been the subject of various critical discussions (Clair, 2003). As one of the doyens of medical anthropology, Kleinman’s description of ethnography has framed my research. I adapted this concept to observe patients and their families/caregivers, as well as to explore the associated professionals’ thoughts and understanding. The study of societal patterns of action, inter-action and re-action within different cultures has made ethnography the most important methodology of anthropology. The strengths and weaknesses of ethnography were analyzed in Hammersly and Atkinson’s (1983) foundational article series; they suggest that the activities and perspectives of all involved parties have to be portrayed, and multiple data sources in real life or everyday life settings have to prove or negate validity in order to discern the correct manner in which to interpret data and develop a hypothesis without falling prey to misleading preconceptions. This recommendation is based on logical thinking at the hypothesis generation stage in order to create relevant science and includes also a reflection of the viewer’s perspective. The sociologist Emile Durkheim and the philosopher Max Weber used such curiosity driven neutral, and thus, respectful descriptions as the basis for scientific logical thinking. Hence, they were able to exceed the subjectivity of interpretations by overcoming the existing mainstream understanding propagated by religious authorities or the political system (Weber, 1949).

**Empathy and Descriptions.** Empathy is the capacity to put oneself in the position of another, and try to feel or understand what this person is experiencing within their perspective or frame of reference (the English word for the German term *Einfühlung* (Titchener, 1909); this ability
helps to overcome cultural or artificial boundaries. Thus, it offers a more positive 360-degree perspective and a neutral and objective approach as it overcomes the viewer's boundaries. In modern medicine, the term empathy is used fairly often and has been integrated as a communication strategy (Liu et al., 2015; Olufowote, 2015). On the other hand, Gelya Frank (1985), an occupational therapist and anthropologist, explains empathy as a foundational behaviour, actually as a philosophy, and as a contra point to the existing culture of narcissism:

Empathy, in direct contrast, requires the suspension of the self to take the place of the other. It permits a person to leave the self aside for a bit, experience something of another’s life as if it were one's own, and conclude with a return to one's own familiar self. (p. 190)

If we review the recruitment and appointment strategies of the Faculty of Medicine in the University of British Columbia, we realize that the attribute best is used almost in all recruitment postings. Best students are accepted in medicine, and the best doctors are appointed for academic positions. Best as a self-description seems to go along with a more narcissistic self-understanding rather than empathy (Lasch, 1978), which, according to Frank (1985), degrades empathy as a philosophy and makes it superficial.

I reviewed the domains outside of academia in which the concept of objective, explorative observational skills are taught. These teaching or presentations had to fit with Kleinman and Benson’s (2006) description of “intensive and imaginative empathy [...] appreciating and humanly engaging with their foreignness [...] , and understanding their religion, moral values, and everyday practices [...]” (p. 1674); thus, they were curiosity-driven and neutral or empathetic and possibly subjective, but in all cases the interaction had to be respectful. In addition to anthropologists (e.g. Franz Boas and Margaret Mead), many writers (e.g. Fjodor M. Dostojewski and Anton P. Chekhov), and, in a particularly brilliant way (from a medical viewpoint), the novelist Charles Dickens (1812 – 1870) used empathy as a core methodology. Dickens was able to describe the entire dimension of sleep disordered breathing in his book, The Pickwick Papers (serialized in 1836 and published in
book format in 1837), 160 years before the American Academy of Sleep Medicine released their consensus statement about sleep disordered breathing (American Academy of Sleep Medicine Task Force, 1999; Epstein et al., 2009). The Pickwick Papers reads as a 360° depiction of the symptoms and suffering associated with sleep disordered breathing, including the daytime sequelae, impacted emotional well-being, and the social stigma attached to the index case of Joe—the “fat boy” (Dickens, 1837).

Dickens’ empathetic (qualitative) and descriptive case report of sleep disordered breathing utilizes the concept of objective, curiosity-driven and neutral observational skills. It raises the question of how an artist, who was fairly famous for his social milieu studies and had never studied medicine, could describe the entire spectrum of a clinical picture so accurately almost 200 years before contemporary scientists. More than that, it questions the value of research design for most quantitative studies, which reduce complex questions to an artificial model situation with one simplified question (single-cause-and-single-effect interactions). This might explain why the current approach in paediatric sleep medicine can be reviewed critically as it might miss major factors and contexts that contribute to therapeutic success or failure. Kleinman and Benson’s (2006) concept of applying ethnography with “intensive and imaginative empathy” (p. 1674) seems to give a possible answer and a new clinical research direction.

_The question that followed was how to apply “intensive and imaginative empathy” (p. 1674) in paediatric sleep medicine._

Exploration, Observation and Description in Arts. Novelists or anthropologists were not the first to practice such _curiosity driven_ and “intensive and imaginative empathy” (p. 1674) in a professional for the sake of exploring unknown or challenging phenomena (Kleinman, 1988). Starting from ancient Greek mythology, philosophers or authors and composers like Euripides and
Sophocles and later on William Shakespeare, Wolfgang Goethe, Charles Dickens, and Victor Hugo, or Claudio Monteverdi, Wolfgang Amadeus Mozart, and Giuseppe Verdi applied this empathy-based, explorative approach in the arts and described characters from a 360-degree angle. In short stories, novels, theatre and/or opera, the integration of music and theatre, best- and (more often) worst case scenarios were presented exceeding the *white and black or good and evil* thinking that most of us grow up with. Exceeding mainstream understanding and notions, allows the reviewer to overcome personal boundaries and develop new understandings, connotations and notions. My personal understanding is that these timeless art pieces were created not only out of the inspiration and sheer brilliance of these artists, but also from their tremendous capacity for *empathy*, which could convince even censors and oppose the propagated mainstream thinking.

In ancient Greece, the concept of theatre was different than in Shakespeare's time or today. Still, the in-depth studies of wide variety of gods, goddesses, heroes, heroines and mythological creatures were not obedient, but *respectful*, and due to their *empathetic* character, also intense, curiosity driven, and supported the creation of various explanatory models. The *explanatory models* of the presented theatre varied depending on the viewpoint and introduced the concept of relativity. Hot topics in these ancient works ranged from migration (e.g. Medea by Euripides) to vanity or survival strategies of the aging male (e.g. Falstaff by Shakespeare) or love, destiny and death (Orpheus and Eurydice); however, they are presented as a coherent narrative in an empathetic way. The author, librettist and/or composer treats the protagonists always *respectfully*, e.g. Medea, Orpheus and Eurydice or Falstaff; specifically, their lives and thinking are explored full of curiosity, their thinking and acting becomes understandable within a bigger contextual framework, which depending on the presenters (in ancient time, priests, and later, actors) interpretation is influenced each time in a different way. The reader, observer or listener acknowledges them as individuals, their existence and living space, whether they agree or disagree with their acting. Despite their old age, these stories are timeless and still very appealing for
contemporary individuals as they sympathetically deal with essential human problems and challenges, and encourage emotional and rational reflection.

The subject of sleep has been revisited reputedly in the arts (Hobson & Wohl, 2005; Shapiro, Boquiren, Boquiren, & Sherman, 2009); from a sleep medicine perspective, *sleepwalking* is described in Verdi and in Shakespeare’s Macbeth in the Lady Macbeth’s famous sleep-walking scene (Act V, Scene 1). *Sleepwalking* is actually a parasomnia (arousal disorder) but the way it is described (repetitive with the purposeful movement of hand-washing) suggests that it is actually the description of a rapid-eye-movement (REM)-sleep behaviour disorder, a more recently described subtype of parasomnia that occurs during REM sleep. This description again raises the question about the writer’s (librettists) observational skills and the accuracy in his descriptions, which could be identified as a distinct entity approximately 500 years after Shakespeare’s initial description. In *The Sleepwalker* (La Sonnambula), the content encompasses a series of sleepwalking implications and even became the name of Bellini/Romani’s opera. Here the activity of sleepwalking is introduced in a different social and educational context as it challenges the viewers’/listeners’ understanding of what is a *disorder*, a *virtue* or *decision-making* in a more modern and feministic sense.

### 1.3.2. The Value of Exploration and Bidirectional Communication

The concepts of *exploration* and *bi-directional communication* are not new in medicine, and are most definitely not new in the areas of paediatrics, developmental paediatrics or child psychiatry/mental health. However, the application of both of these concepts in the analysis of *culturally obscured sleep behaviours* through structured qualitative methodologies supported the development of a novel phenotyping-focused, *functional assessment model for sleep and wake-behaviours*. This assessment concept incorporates the observations and perspectives of the family and others who deal with challenging and disruptive sleep/wake-behaviours on a daily basis, and
exceeds the typical daytime behaviour-focused categorical diagnoses by acknowledging day-and-nighttime behaviours as a continuum. Further, it has been instrumental in understanding the pathways to inappropriate medications, over diagnosis of uneven developmental profiles and challenging/disruptive behaviours as autism spectrum disorder or fetal alcohol spectrum disorders.

In *The Birth of the Clinic: An Archaeology of Medical Perception*, Michel Foucault (1975) describes the re-discovery of the *individual* in the 19th century. According to Foucault, modern medicine with its foundational autopsy research approach (i.e. focusing on anatomy and pathology), but not philosophy has been instrumental in creating the new discourse of cause-and-effect-interactions, opening the floor for in-depth phenotyping and overcoming the concept of broad hermeneutic interpretations (Ruffing, 2010). The modern, technology-centred medicine that we have all grown up with was built on this in-depth, cause-and-effect investigation (*single-cause-and-single-effect*). Therefore, data collection that describes symptoms with modern technology, e.g. electrophysiological information has been very much appreciated and has thus become more prioritized over time. This approach forces the generation of (*artificial*) model situations. In sleep medicine, electrophysiological information has been fundamental, but it might not explain the patient’s individual suffering in the sense that Kleinman (1988) describes. Modern, western, technique-oriented medicine and its associated tendency to apply categorical diagnoses, which may miss individual suffering, has been critiqued by a number of medical anthropologists (Clark & Mishler, 1992; Helman, 2007). Similarly, existing structures, which are built on this above-mentioned model of situation-based thinking, coin our understanding and decision-making. Ivan Illich (1974) analyzed medicalization and medical centrism, and Foucault dealt with these topics as a philosopher (Foucault, 1994, 2006).
The roots of paediatric sleep medicine’s go back to sudden infant death syndrome research in the seventies and eighties (Kahn et al., 1992). The questions in sudden infant death syndrome research were *what is norm, what exceeds the norm and might lead to (sudden infant) death*. There has always been a strong cultural association between sleep and death: in ancient Greek mythology, sleep (*Hypnos*) and death (*Thanatos*) were twin brothers (Homer, 1990); nighttime prayer traditions were initiated due to a fear of death during sleep (Klug, 2008); and sudden infant death syndrome was, in fact, already mentioned in the Bible (Kings 3:19). Now, modern, technology-based neuropsychological data recording capabilities have opened up a new perspective to the observation of *suffocation* as mentioned in the Bible (Kings 3:19; Paky & Kytir, 2000).

To understand the possible aetiologies of sudden infant death syndrome, sleep labs were created and infant sleep was investigated all over Europe and North America. As a direct result of this some risk factors like obstructive sleep apnoea (Section on Pediatric Pulmonology, Subcommittee on Obstructive Sleep Apnea Syndrome. American Academy of Pediatrics, 2002), prone sleep, and co-sleeping were detected (Task Force on Infant Sleep Position and Sudden Infant Death Syndrome, 2000). Various sudden infant death syndrome prevention campaigns tried to respond to these findings, and the paediatric sleep medicine community began promoting solitary sleep in response to the neurophysiological findings (McKenna, 1996; McKenna, Ball, & Gettler, 2007). This development *medicalized* paediatric sleep and the understanding became that *every* child can learn sleeping (Ferber, 2006), if he/she does not suffer under obstructive sleep apnoea and is not *co-sleeping*.

Over the years, paediatric sleep became a typical example of neurophysiological data collection and the one main question was how to promote/train infants solitary sleep in the best way. On the other hand, a major gap continued to exist between the collected data and reality; cultural background-related perceptions, notions and understandings confound data collection and
in consequence, objective medical understanding (Paky & Kytir, 2000). Confronted with this systemic gap in our clinical understanding and the methodological challenges related to the research question #2 (how can we optimize our clinical understanding of sleep problems ...), I started within this PhD endeavour to investigate single and complex cause-and-effect-interactions in the footsteps of 19th century physicians. In order to do this, I utilized qualitative methodologies for in-depth phenotyping of sleep/wake-behaviours. The first results of my research, presented in this dissertation, support the use of qualitative methodologies in modern 21st century medicine as they add the perspective of self-reflection in (paediatric sleep) medicine and help to overcome the current confounded and technology-focused viewpoint.

1.3.3 The Concept of Ecology

Ecology as a Framework. The Oxford Dictionary defines ecology as the branch of biology that deals with the relations of organisms to one another and to their physical surroundings (Ecology [def. 1], n.d.). The Ecological Systems Theory, developed by Urie Bronfenbrenner in the 1970s, characterizes a child’s development in their core environment (referred to as the microsystem); the individuals interactions with other individuals (referred to as the mesosystem), external factors which impact micro- and mesosystems (exosystem), and the culture in which they live (macrosystem) over a period of time (chronosystem) (Bronfenbrenner, 1974, 1976, 1977, 1979). As such, ecological theories provide a framework from which ethnography and observation can be used to study the context of individual’s distinct development and interactions at various levels over time. In the context of this dissertation, sleep is a phenomenon that everybody deals with to a certain degree. Everybody sleeps through one third of their lives due to physiological reasons; starting with infancy and continuing into old age, everybody has to adjust to this physiological need for sleep according to his/her individual current (culturally-influenced) norms. With the increasing autonomy realized in childhood and adolescence almost everybody tries to change or influence
their sleep patterns due to various perceptions, notions and understandings, which all influence each other. In other words, the norm of sleep is culturally formed and executed, and there is an *epidemiology* of various representations (McKenna, 1996; Sperber, 1985; Steger, 2003; Worthman & Melby, 2002).

**Ecology in Medicine.** The concept of ecology has been used in medicine from various viewpoints: to bring in the factors of a naturalistic setting, e.g. the influence of sun, sea, sand on the human diseases (Yenal, 1960; Yenal, 1973), or to demonstrate how a chronic condition can affect family relations and the family's understanding of the condition (McKellin, 1995). *Family ecology* investigates features of a family's ecology in order to design contextualized family-centered behaviour support plans (Lucyshyn & Albin, 1993; Lucyshyn et al., 1997). The concept offers a framework to enable collaborative, ethnographic exploration of the strengths and experienced challenges of the involved parties. It encourages a *curiosity driven*, family-oriented language that neutralizes the conversation with a *practice [of] an intensive and imaginative empathy*, as the explorative questions are presented *organically* within the flow of the ethnographic exploration, and not a uni-directional questioning experienced as *patronizing*. In the context of sleep/wake behaviour assessments, the family ecology framework enables the assessment of the family's activity settings (routines) and the child’s positive contributions, family strengths, formal and informal resources, sources of social support, sources of stress, and family goals using a neutral, descriptive language and is the model of ecology I have used in my clinical research.

**Family Ecology in Sleep/Wake-Behaviour Assessments and Creating a Safe Space.** Family ecology acknowledges the impact of family dynamics, and the broader network of social relationships on the experience of the condition (Lucyshyn & Albin, 1993; Lucyshyn et al., 1997). The implementation of the family ecology concept in my clinical practice aimed to identify (a) supportive family routines that are challenging or are valued from a sleep health perspective; (b)
prioritizable routines for intervention; and (c) collaboratively generate a family vision of successful routines; after (d) gathering information useful for design of a contextually appropriate behavioural support plan. As the footnote shows, self-medication has been an informal source of support for many families. Self-medication also helps many clients to relax and overcome WED/RLS-related discomfort (Ipsioglu, Elbe, et al., 2015).

An exploration-oriented ethnographic interview is used to understand the family ecology and thus, create a safe diagnostic space, which families need in order to speak of stress and sources of support, and their re-active behaviours. The overall goal of the diagnostic space (our suggested assessment strategies) is to respect the emotional ecology of the counterpart, introduce and implement a sustainable therapeutic relationship. In case it is needed, a long-term behavioural support plan can be created or out of respect to the emotional ecology of the family be postponed. It is evident that this kind of assessment has natural and introductory limits, such the family's willingness to partner and collaborate in this endeavour, as these are foundational aspects. However, in sleep history taking, questions about medication practices, including self-medication, became a core part of the medical interview, which gave eye-opening insights after implementing a bi-directional communication concept. As the footnote shows, self-medication has been an informal source of support for many families. Our observation has been that self-medicating helps many clients to relax and overcome WED/RLS-related discomfort.

Emotional Ecology. The term emotional ecology was first coined by Michalinos Zembylas in 2007; he described that a “teacher's emotional knowledge about teaching and learning is an inextricable part of the ecosystem of teacher knowledge; this is called emotional ecology” (p. 355). Emotional ecology picks up on Karl Jasper's idea of borderline or limit situations (in German, Grenzsituationen) and describes that these experiences, which arise from ordinary experiences, influence thinking, communication and reflection (Jaspers, 1980). In the clinical context, we deal
with the patient's or patient family's emotional ecology, which impacts their action radius; the concept of family ecology operationalizes that in a systematic way (Lucyshyn et al., 1997). However, by applying Jasper's limit situation, Zembylas' emotional ecology framework, and the subjectivity of the viewer concept (Hammersley & Atkinson, 1983; Powell & Boas, 1887) to the research questions presented in this dissertation it becomes clear that we also need also to focus on the emotional ecology of health care professionals. In 1989, four Austrian nurse's aides at Lainz General Hospital in Vienna, the Lainz Angels of Death, confessed to multiple murders and attempted murder charges between 1983 and 1989 (Protzman, 1989). A main reason for the nurses' criminal acts was believed to be due to loss of emotional ecology over the course of their work with feeble and languishing elderly patients (Protzman, 1989). The group killed their victims with overdoses of morphine or by forcing water into the lungs (Protzman, 1989), and were described as “one of the most unusual crime teams in 20th Century Europe” (Wikipedia contributors, 2015).

This significant incident, paralleled by the generational change at the various decision maker levels (from the generations born before and during the Second World War, to the generations born after the war, called baby-boomers) transformed the Austrian health care system from an autocratic, paternalistic, and partly fascistic system to a hierarchically organized matrix system. To achieve that with sustainability a) trans-disciplinary health care management courses were offered in all Austrian Provinces and b) the culture of respectful communication within interdisciplinary teams was established with the help of facilitators/mediators. Both options were seen by many health care professionals as a chance to optimize one's own knowledge and were used with high frequency by employees.

Health authorities particularly targeted professionals working with vulnerable patients in order to achieve a stable working atmosphere. Leadership positions in the health care field asked for a diploma and/or graduate degree in these areas, and many people did that in order to widen
their knowledge and paid for the courses out of their own pockets. Exploration, listening and respect from individual professional’s needs became core elements for the management of professionals’ needs. This enabled a change management and implementation of quality control measures within a short time period and without increasing angst among health care teams, moving the conservative Austrian health care system into a new era. The concept of exploration, listening and respect from individual professional’s needs and peer group discussions are state-of-the-art in British Columbia. However, in contrast to the European facilitated interdisciplinary team meetings (e.g. Balint groups (Balint, 1957; Pinder et al, 2006)), facilitated/mediated discussions are not provided and in general, this concept is unknown. This raises the question of how the health care system deals with the emotional ecology of its own members.

1.3.4 Conclusion of Section 1.3

Through the exploration of medical anthropology literature, which has been foundational to my research (Csordas, 1990; Good, 1994; Scheper-Hughes, 2007; Sperber, 1985), I came to a conclusive realization: In our efforts to create neutral and reproducible medicine, we are overlooking the primary confounding factors caused by our individual cultural background and training and the associated bias or relativism in our diagnoses. Consequently, this meant that using ethnography for analysis of cultural phenomena and the “practice [of] an intensive and imaginative empathy” (Kleinman & Benson, 2006, p. 1674) would provide a strategy to overcome the current training-based bias, similar to what the arts have incorporated in various and changing contextual frameworks for over 2500 years. The medical system was in desperate need of a more evolutionary, reflective and dynamic research design to understand the confounding factors and relativism in collected data. The proof of concept was that we had not recognized familial WED/RLS in children with neurodevelopmental conditions. Similar to the critical self-reflections in anthropology or arts, we needed to learn to acknowledge the subjectivity of our viewpoint and being able to reflect on
that. Further, we needed or we need to recognize how this subjectivity impacts our results and challenge our own research directions as shown in anthropology (Hammersley & Atkinson, 1983). Tailoring the diagnostic approach through the creation of a safe diagnostic space is based on the idea of individualization. Ethnographic exploration and ecological theory are the core methodologies to individualize the assessment of a challenging problem. Further, the concept of ecology frames the experience as a neutral strength/weakness-based assessment rather than a moralizing situation, where the assessing person has the knowledge to judge the counterparts acting as right or wrong, as these black and white terms are again subject of relativism. Besides their utilization in medical anthropology and educational psychology, these concepts have been applied for thousands of years in visual arts and drama, and for hundreds of years in novels and opera.

1.4. Exploring Sleep/Wake Health

1.4.1. Cultural Aspects

The anthropology of sleep can be approached from various perspectives: Worthman and Melby (2002) introduced the concept of sleep ecology, explored sleep and sleeping situations, and showed that group sleep with individual bedding is historically the most prevalent sleep environment in human societies. In a recent article, Worthman (2013) summarizes that “culture drives sleep patterns and quality, sleep is social, and family relationships form the context for sleep” (p. 35), this statement can be most probably generalized for cultures all over the world and not only for the Egyptian families that the authors had studied. Cox (2008) investigates the roots of our perceptions and understandings and concludes that “[a]s both a biological universal and biological necessity, sleep was deeply ingrained in the culture of colonial New England, running a leitmotif through a richly interwoven discourse on religion and morality, society and physiology” (p. 69). Exploring medieval German literature, Klug (2008) analyzed routines, rituals and traditions
and describes the notion of vulnerability and “defenselessness to psychological, spiritual and social levels” (p. 50), which also explains why sleep has been seen as a punishment until recently (Cox, 2008). This understanding might explain why despite focus on, e.g. dietary campaigns sleep behaviours, sleep need, and coping with inadequate sleep has been considered as individual and private and national public health campaigns have not been created, despite the fact that sleep deprivation of youth is debilitating and impairing (Orzech, 2013).

The concept of how environmental factors, lifestyle and cultural background have influenced our sleep/wake cycles was developed with the mentioned sleep health and hygiene literature overviews (spearheaded by Dr. James E. Jan) and influenced by an exceptional eye-opening exhibition, Schlaf und Traum [Sleeping & Dreaming], organized by the Deutsches Hygiene Museum, Dresden, and Wellcome Collection, London, in 2007 in Dresden and in 2008 in London (Dorman, 2007; Dorman, 2007-2008). At the Mini Med School 2008 (organized by me on behalf of BC Children’s Hospital and Child and Family Research Institute) (Ipsiroglu, 2008b) the various sleep perceptions, notions and understandings were reviewed and presented with an inter- and transdisciplinary faculty (www.cfri.ca/docs/news-docs/minimedatcfribackgrounder.pdf?sfvrsn=0). Overall, from a public health perspective, and aside from the anthropology, (developmental) paediatrics and child psychiatry/mental health angles, we can summarize that epidemiologically-focused papers describe a growing tendency to sleep less, and indicate gradual societal and technological changes to be the cause of this.

The invention of electricity has allowed children to stay active later in the evening (Navara & Nelson, 2007). They are exposed to bright light until bedtime, as they sit in front of their computers to conduct various activities, including working on school assignments, watching television, playing video games, talking on their telephone and listening to music (Moore & Meltzer, 2008; Smaldone, Honig, & Byrne, 2007). The biological effect of bright light exposure is a delay of sleep onset (Erren & Reiter, 2009).
The academic environment in many countries has fostered high expectations for success and additional evening activities are expected of many students, which also results in delayed bedtimes. At the same time, many children wake up earlier than previously, as a strategy to deal with their busy schedules (Adolescent Sleep Working Group; Committee on Adolescence; Council on School Health, 2014; Moore & Meltzer, 2008). The medical literature shows school-aged children and adolescents achieve inadequate amounts of sleep and have increased daytime sleepiness (Fredriksson, Rhoades, Reddy, & Way, 2004; Sadeh, Gruber, & Raviv, 2000; Wolfson, Acebo, Fallone, & Carskadon, 2003; Wolfson & Carskadon, 2003; Wolfson, Spaulding, Dandrow, & Baroni, 2007). Although results vary depending on type of survey and magnitude of study, children and adolescent’s self-reported findings indicate that 12% of school-aged children and adolescents experience sleep problems (Ipsiroglu, Fatemi, Werner, Paditz, & Schwarz, 2002), and 87% report that they need more sleep than they get (Wolfson & Carskadon, 1998). Visibly, the increased demand on time has resulted in a decrease in sleep. In conclusion, society approves and encourages the behaviour of hard-working children, and characterizes sleep as an activity of the lazy, dull or sluggish. High activity levels, alertness and liveliness are considered the attributes of hard-working and successful individuals, and this perception has affected human experiences long before electricity.

The Dark Side of the Night. The night has always been associated with the dark and some frightening vulnerable situations (Hobson, 2005). Natural and social enemies attack in the dark and also death, the most extreme form of danger, usually occurs overnight (Klug, 2008). Sleep is even the twin brother of death in ancient Greek mythology as described above. The effects of these long-standing, traditional, epigenetic beliefs, contribute to the current understanding of activity-focused societies, where the successful work but do not sleep (Brunt & Steger, 2008). The medical profession is saturated with this perception and expectations regarding success (Cox, 2008). Despite the fact that healthcare professionals have a general appreciation for the importance of
sleep and the consequences of sleep problems, their actual practices contradict this and reflect these long-standing perceptions.

**Sleep Deprivation Culture in Health Care Professions.** For health care professionals themselves, the endurance of sleep deprivation is required and often celebrated throughout medical training and in everyday practice. This notion is also modeled by senior physicians, which results in a problematic belief system that holds sleep as an existential, but malleable function that is assumed to be *manipulate-able* (Barger et al., 2005; Owens, 2001). As a society, our understanding of sleep issues is affected by the wide-spread perception that *sleep equals inactivity.*

**Cultural Backgrounds Framing Notions of Sleep Health in Patients and Families.** If sleep, and the way we deal with sleep, is affected by our cultural background, the only way to unravel the complexity of diverse perceptions and understandings is by applying ethnography as a core methodology and to follow Kleinman’s suggestion of exploring complex situations, the beliefs of an individual and his/her interactions with the society, to achieve the best outcome (Kleinman & Benson, 2006).

**1.4.2. Sleep/Wake Health in Paediatrics**

**Sleep Hygiene.** The first literature analysis developed by my colleagues (with whom I started this research) and myself significantly framed this research endeavour. It focused on causes and treatment options of sleep disturbances in children with neurodevelopmental conditions (Jan et al., 2008). In addition to a brief review of pharmaceutical and non-pharmaceutical options, we analyzed first-line non-pharmaceutical treatment options, including researched or un-researched clinical best practices to improve sleep quality. The title, *Sleep Hygiene for Children with Neurodevelopmental Disabilities,* reflects our intention and prevention-oriented approach.
In this “first comprehensive, multidisciplinary review of sleep hygiene for children with disabilities” we presented the medical rationale for examining the background of so-called sleep hygiene measures (Jan, Bax, Owens, Ipéroglou, & Wasdell, 2012, p. 403). These measures promote the body's own melatonin production, which communicates sleep time to the brain and various organs (Jan et al., 2012). Through incorporation of such sleep habit measures in clinical treatment, and by outlining "both general and specific sleep-promotion practices" (p. 403), we reviewed and analyzed problem-solving strategies for implementing such measures in clinical practice. We realized that “despite the importance of sleep-hygiene principles, defined as basic optimal environmental, scheduling, sleep-practice, and physiologic sleep-promoting factors” (p. 403), clinicians often lack the appropriate knowledge and skills to implement them. In addition, sleep-hygiene practices may need to be modified and adapted for the population of children with neurodevelopmental conditions as there are additional challenges in successful implementation when compared to their otherwise healthy counterparts (Jan et al., 2012).

Despite the fact the literature was scarce, and that most of our understanding was based on case reports, case series (anecdotal evidence), and health care professional expertise, the publication was published in Pediatrics and was offered by the editors as an open source publication. This illustrates the significance the editors attached to this work, which uses explorative descriptive research.

**De-medicalizing Sleep/Wake Health.** Two years later, we published the paper *Sleep Health Issues for Children with FASD: Clinical Considerations* (Jan et al., 2010). This time, the concept of sleep hygiene was presented as a sleep health notion, highlighting the relationship between sleep and health while avoiding medicalized language and targeting a broader professional audience for the purpose of de-medicalizing sleep. Parallel to this work, I started creating an assessment concept that would not automatically lead to a (categorical) diagnosis, but functional assessment, and could
be conducted by any trained health care professional (Ipsiroglu, Carey, et al., 2012). This opened the door for a brighter and broader discussion about the role of sleep in the populations with FASDs.

Overall, what we, as individuals or group of peers in an academic environment, perceive as a norm is relative and might or might not be the norm. In response to the clear need to increase our understanding of the dimension of sleep problems in children with neurodevelopmental conditions, I started using explorative ethnography in order to understand the family's sleep ecology and the concept of emplotment (sharing reports) as a quality control measure of my activities. This began to include the identification of familial sleep problems in my paediatric setting.

1.4.3. Child Psychiatry and Developmental Paediatrics Perspectives

Sleep is essential to good health and quality of life. Numerous cognitive, and behavioural impairments, as well as increased risk for a variety of disorders or diseases are known to occur with sleep deprivation (Jan et al., 2012). Not enough and/or non-restorative sleep, despite having different aetiologies, can lead to secondary health and behavioural problems, which are summarized in this dissertation under the general term sleep problems. Up to 20-30% of healthy children (Owens, 2000), and 85% of children with specific conditions, have sleep problems (Jan et al., 2008). Therefore, the differentiation of sleep problems, whether they have been caused by environmental factors such as missing sleep health measures, previous sleep hygiene or an organic/non-organic problem, becomes important. The nature of primary sleep problems in infants, as well as in children with genetic conditions and neurodevelopmental conditions, is poorly understood and has not yet been sufficiently investigated or described. These insufficiencies and gaps lead to major challenges in recognizing and differentiating primary from secondary sleep problems. In consequence, treating sleep problems appropriately becomes difficult and a significant
unidentified gap remains in our understanding of how sleep problems and sleep related secondary problems can contribute to challenging behaviours.

1.4.4. The Clinical Reality: Life Trajectories

An exclusive focus on daytime sequelae, and prescription of sedative hypnotics without prior differential diagnosis of the sleep problems, may result in the phenomenon of *chronification*. The underlying cause remains unidentified and develops chronic characteristics that are treated with poly-pharmacy, which may obscure the problem further (Ipsiroglu, Berger, et al., 2015; Ipsiroglu, Elbe, et al., 2015). In particular, pharmaceutical interventions aimed at attenuating challenging/disruptive wake behaviours can aggravate sleep symptoms further, leading to diagnostic overshadowing and can create chronic worst-case scenarios as the presented case vignette shows.

**Case Vignette #2.** *A male adolescent patient with sleep difficulties since birth and the profile of an unidentified neurodevelopmental disability, was diagnosed with ADHD (at the age of two and half years), dysthymia and major depressive disorder (11 years), oppositional defiant disorder (12 years), autism spectrum disorder (13 years) and finally, with static encephalopathy (Alcohol-Related Neurodevelopmental Disorder; four digit code: 1/1/3/3; 16 years)* (Chudley et al., 2005). *He was treated with 7 psychotropic medications between the ages of two and half to 16.5 years, with no success. However, significant medication side effects were seen with almost all medication-based therapies: developmental regression to psychostimulants (prescribed for his ADHD) at the age of 2.5; metabolic effects to atypical antipsychotics (prescribed for his depression) leading to the diagnosis of obesity (body mass index: 34.6 at age 16) and significant sleep disordered breathing. At age 15, reduced tension in the home environment lead to a decrease of oppositional defiant disorder and major depressive disorder symptoms; yet, challenging/disruptive behaviours continued*. (3) *Improvement of his challenging behaviours occurred after diagnosing WED/RLS at age 16, a*
neurologic condition and the underlying reason for his chronic sleep problems, mimicking ADHD-like presentation, and weaning him off from all psychotropic medications in collaboration with his psychiatrist.

1.5. Questions Motivating my Research

This research endeavour has been motivated by my experience with clinical worst case scenarios, as a common denominator in all these scenarios the following questions have come up:

1) ‘How does a health care professional respond to the concerns of patients and their families?’ This question is related to the research question #1: ‘Why have sleep problems remained unrecognized particularly in children with neurodevelopmental conditions?’

2) ‘How may perceptions/understandings in regards to challenging/disruptive sleep/wake behaviour differ, and how to develop a shared language?’ This question is related to the research question #2, ‘How can we optimize our clinical understanding of sleep problems in children with neurodevelopmental conditions and how is the dimension of the problem described?’

3) ‘How does our health care system manage the sleep/wake behaviours of children with complex and chronic health care conditions?’ This question is related to research question #3, ‘What are the consequences of unrecognized sleep problems in this vulnerable population?’

The answers to these questions are presented throughout the methodology and results part of the manuscript-based dissertation. While connecting the answers, the core question for me as a developmental paediatrician became: ‘Why do we focus on special circumscribed clinical domains, rather seeing the patient as a whole?’ and finally, ‘What can we learn from this modern parable?’
Chapter 2: Methodology and Results

I completed this PhD endeavour as a clinician working in a multi-professional setting. The concepts of implementing ethnography and ecology are based on the new knowledge I gained from medical anthropology, educational psychology, and literature reviews. The project protocols and application of the concepts have been developed by me and the research has been conducted in this multi-professional setting with colleagues and research assistants working in my lab. Therefore, in Chapter 2, I use ‘we’ in the presentation of the results in order to be respectful of my research partners. Please refer to the Preface and introductory paragraph listing all authors for any open questions related to intellectual property.


2.1.1. “They silently live in terror…” Why Sleep Problems and Night-time Related Quality-of-life are Missed in Children with Fetal Alcohol Spectrum Disorder


Overview. Children and adolescents with FASDs are at high-risk for developing sleep problems triggering daytime behavioural co-morbidities such as inattention, hyperactivity, and
cognitive and emotional impairments. While daytime morbidities are easily recognized and treated with various psychotropic medications, clinical experience has shown that underlying sleep problems often remain masked and appropriate treatment missed. To understand the lack of recognition of sleep problems in children with an FASD, we (a) conducted qualitative interviews with six parents and seven health care professionals; (b) performed comprehensive clinical sleep assessments in 27 patients together with their caregivers referred to our clinic for unresolved sleep problems; (c) analyzed these patients’ medical records; and (d) used the concepts of narrative schema and therapeutic emplotment to develop treatment strategies. The research was conducted at the Division of Developmental Pediatrics of British Columbia Children’s Hospital in Vancouver, BC between 2008 and 2011. Patients, parents and healthcare professionals came from the province of British Columbia.

**Introduction.** Fetal alcohol spectrum disorders (FASDs) are a paradigmatic case for the study of neurodevelopmental conditions and associated sleep problems. FASDs are the most common form of prenatally acquired brain injury and are due to a toxic substance exposure – the deleterious effects of alcohol on the developing brain of the foetus (Clarren & Smith, 1978; Jones & Smith, 1973). FASDs are a major public and population health problem. The Public Health Agency of Canada (2005) estimates that FASDs affect one percent of the Canadian population. In the United States 0.2-1.5 out of every 1000 live births are affected with an FASD (May et al., 2009). FASD is an *umbrella term* and not a diagnostic category, that encompasses the following diagnoses: fetal alcohol syndrome, the severest form among FASDs; partial fetal alcohol syndrome and alcohol-related neurological disorder, characterized by a range of neurological impairments, but no evident growth deficiency or facial characteristics; and Alcohol Related Birth Defects, which refers to defects in other organs, skeletal abnormalities and vision and hearing problems (Public Health Agency of Canada, 2005; Rasmussen, Andrew, Zwaigenbaum, & Tough, 2008).
Children and adolescents with an FASD represent one of the most vulnerable populations in the spectrum of neurodevelopmental conditions. They, their birth mothers, and even foster parents face social stigma associated with the condition because it is the result of potentially preventable toxic alcohol exposure during pregnancy. The functioning and behaviour of the children and adolescents with an FASD often resembles ADHD; however, it is only partly responsive to pharmacologic treatment (Coles, 2001; Rasmussen et al., 2008). As a consequence these children's behaviours are seen by health care professionals as significantly more challenging to treat than other children with neurodevelopmental conditions or ADHD. Finally, their neurodevelopmental conditions create significant psychosocial burdens on the birth family often resulting in placements in foster families. According to the Institute of Health Economics (2009):

FASDs also affect all other members of the immediate family, including siblings and the extended family. Emotional, financial, and social burdens can be considerable. Indeed, the stress of living with a child affected with FASD may result in family discord or breakup. Adoptive and foster families confront similar issues in dealing with the needs of affected children. Again, proper supports are essential. (p. 8)

Although sleep problems have been recognized as a key symptom of FASDs (Steinhausen & Spohr, 1998; Streissguth, Barr, Kogan, & Bookstein, 1996), and their prevalence has been estimated at 85% by expert opinion (Jan et al., 2010), systematic clinical research to describe the clinical characteristics of FASD-associated sleep problems remained scarce when we began our FASD and sleep research. The few existing papers described children and adolescents with a FASD to be at high-risk for developing sleep problems that trigger daytime behavioural co-morbidities such as inattention, hyperactivity, and cognitive and emotional impairments (Chen, Olson, Picciano, Starr, & Owens, 2012; Hanlon-Dearman, 2003; Stade et al., 2008). However, as demonstrated in the initial case vignette (section 1.2.3) and the case series (section 2.3.1), symptoms of sleep deprivation have
been solely associated with typical daytime diagnosis, such as ADHD and mental health co-morbidities, and therefore, treated with psychotropic medications.

Children with FASDs are similar to children with other chronic disabilities in that they require lifelong care and face common continuity of care management problems (Institute of Health Economics, 2009). In British Columbia, Canada, where the present study took place, children currently receive their diagnoses after assessment by multidisciplinary FASD resource teams funded by the Ministry of Children and Family Development (Streissguth et al., 1996). Afterward, depending on the child’s age and needs, the team’s recommendations are conveyed to the parents and a variety of health care professionals working in the community. These care providers range from pediatricians, child-psychiatrists and family practitioners, to speech-, occupational and physiotherapists, as well as school teachers and social workers. Some of these health care professionals are community based and help the families of patients to navigate through the complex health care, educational, and social services systems. Unfortunately, sleep problems are not a particular issue addressed in these assessments and few of these health care professionals have the experience to decipher the complex relationships among FASD diagnoses, sleep problems, and additional co-morbidities (Ipsiroglu, Andrew, Carmichael-Olson, Chen, Collet, et al., 2011).

Our own practice experience in a sleep clinic for children with complex neurodevelopmental conditions suggests that underlying conditions, like FASD and associated behaviours, such as restlessness, confounded the diagnosis of sleep problems. Although, sleep problems were a common complaint in many of the referred patients, diagnostic assessments and pharmacologic treatments focused primarily on daytime restlessness and related behaviours.

To more effectively address sleep problems in children and adolescents with FASD, we undertook this study to determine the factors and processes that contribute to the apparent lack of recognition and effective treatment of sleep problems in the FASD community. Using semi-
structured, qualitative interviews with parents and health care professionals, our first aim was to identify possible explanatory models to account for the children’s restless day- and nighttime behaviours. Parents’ descriptions of both day- and nighttime activities assisted us in organizing the anecdotal information about sleep problems in patients’ records into coherent narratives of the children’s conditions. These accounts contributed to our final goal, which was to develop a clinical practice strategy that ameliorates any shortcomings in the communication and understanding of a sleep problem among parents and health care professionals.

**Methods.** The research was conducted at the Division of Developmental Pediatrics of British Columbia Children’s Hospital in Vancouver, BC between 2008 and 2011. Patients, parents and health care professionals came from the Greater Vancouver and Southern Vancouver Island Area within the Province of British Columbia. The research consisted of two parts; qualitative interviews of parents and staff, and functional sleep/wake behaviour assessments of a group of referred patients with FASD and their families.

**Qualitative Interviews.** To understand how parents and health care professionals identified and expressed attributes of sleep problems in children with FASD, we (Norma Carey, social worker; and I) conducted semi-structured qualitative interviews with both parents and health care professionals in the setting they preferred (Greenlagh, Russell, & Swinglehurst, 2005). Participants were recruited through the FASD Research Network at the University of Victoria, British Columbia. Health care professionals, who were all members of the network, volunteered to participate in these interviews and also helped to inform parents of children with FASD of the nature and opportunity to participate in our study.

The initial qualitative interviews were conducted with parents of 6 children (birth parents n=1, adoptive parents n=2; foster parents n=3), and 7 health care professionals that were working in the regional service setting for children with FASD (medical doctor, MD, n=1; psychologist n=1;
social workers, SW, n=5). Qualitative interviews with 5 parents were conducted at their homes and with one parent at the Sleep Clinic; qualitative interviews with care providers were conducted at their offices. All participation was voluntary (FASD study-Part 1, Ethics Approval: H07-03126), and all qualitative interviews were recorded and later transcribed.

The discourse structures of the interviewees were analyzed to identify the information thematized by the interviews (Cicourel, 1993; Grimes, 1975). This enabled us to identify the narrative schematic structure of each qualitative interview, interviewees’ interferences, and their underlying schema (Agar & Hobbs, 1985). Sleep problem related key topics as represented by BEARS, a frequently used screening tool (Owens & Dalzell, 2005), were integrated in our open-ended questions and gave interviewees greater freedom to describe their child’s health, social and educational history, as well as sleep problems and their impact on the family in context. Furthermore, in-depth questions about family and family ecology were guided by tools developed by Lucyshyn and Albin (1993) and Lucyshyn et al. (1997).

Accordingly, our qualitative interviews included the following six topics: 1) the child’s biography, e.g. ‘where was he/she born?’ ‘Have there been any changes in primary caregivers?’ 2) Family ecology, e.g. ‘can you give some descriptions related to the child’s strengths and problem behaviour and how these affect the child, you, and your family?’ 3) Child development, e.g. ‘describing his/her development and behaviour?’ 4) Sleep history, e.g. ‘describe sleep patterns and any breathing or sensory problems?’ 5) Sleep treatment, e.g. ‘what efforts have been made to improve sleep?’ 6) Impact of sleep problem on family, e.g. ‘how did your child’s sleep problem impact your life / the life of your family and the life of the child?’

Functional sleep/wake behaviour assessments were performed in 27 children with an FASD who were referred for further assessment of their sleep problems, as well as in 27 parents of these children (birth parents n=9, adoptive parents n=6; foster parents n=12). The assessments
were done in the Sleep Assessment Clinic located in the Division of Developmental Pediatrics of British Columbia Children's Hospital in Vancouver. Clinical examinations and assessments were conducted by myself and at least one additional physician (a resident or fellow), and/or a nurse, who took notes of comments by the parents. Each re-examination started with a review of the existing records to identify the patterns of clinical referrals and investigations. Particular attention was paid to notes made by previously involved clinicians about sleep problems, their accounts of parents’ reports of sleep problems, and perhaps more importantly, symptoms that might be associated with sleep problems, but which were not identified as such by referring clinicians. Guided by Agar and Hobbs (1985) and D’Andrade (1976), we identified the diagnostic inferential schemas, which formed the explanatory models employed by the referring physicians.

Our functional sleep/wake behaviour assessment interview began by requesting parents to describe their main concerns. Parents were particularly encouraged to tell accounts of their child’s sleep history, including examples of the best and worst case scenarios, e.g. of going to bed-situations, nighttime-awakenings. (Interviews in the clinical setting employed the same topics as used in the qualitative interviews.)

From the parents’ narratives, we constructed a structured sleep history using an explorative and descriptive approach. The goal was to understand the presentation of the child’s day- and nighttime symptoms and the ways the families dealt with these challenging situations. The assessment also included standard elements of developmental pediatric clinical history-taking (including a history of the diagnoses and medications), confirmation of the various referrals and the findings of clinical examinations, laboratory tests and imaging, and clinical observations.

Following the physical examination, the clinical assessment was complemented by summarizing the earlier histories and comparing their findings with the findings of the current functional sleep/wake behaviour assessment. The results, including original quotes from the
qualitative interviews and clinical interviews, were synthesized into an emploted functional sleep/wake behaviour assessment summary report. Parents’ narrative descriptions of behaviours, activities, as well as quotes of clinically relevant descriptive features of sleep (e.g. behavioural movement patterns: “gets hyper, before falling asleep”, “sleeps like a windmill”, “soaking pajamas”) were collaboratively integrated into the functional sleep/wake behaviour assessment summary reports by employing the strategy of therapeutic emplotment (Mattingly, 1994, 1998). These reports captured the full range of medical and social factors that might relate to the child’s sleep problems. During this process of co-construction, parents were asked to make changes to create comprehensive, coherent narratives of their children’s conditions. These functional sleep/wake behaviour assessment reports, intended for parents and the referring physicians, were used in follow-up conferences with the parents, physicians and community support team members. This process developed a plan for therapeutic action and facilitated continuity of care. The characteristics of the participants are summarized in Table 2.1. (Ethics Approval H11-01410: Evaluation of Clinical Work; Ethics Approvals: H11-01768: Retrospective Chart Study).

Table 2.1. The Characteristics of the Participants Summarized using Descriptive Statistics.
Note that 22/27 patients had been diagnosed with ‘Suggestive WED/RLS’.

<table>
<thead>
<tr>
<th>Participants of Semi-Structured Qualitative Interviews</th>
<th>Families</th>
<th>Health Care Professionals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parents</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Birth mother</td>
<td>6</td>
<td>Health Care Professionals</td>
</tr>
<tr>
<td>Adoptive parents</td>
<td>2</td>
<td>Psychologist</td>
</tr>
<tr>
<td>Foster parents</td>
<td>3</td>
<td>Social workers</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Participants of Comprehensive Clinical Sleep Assessments</td>
<td>n=27</td>
<td>Diagnoses &amp; Co-morbidities</td>
</tr>
<tr>
<td>Families</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Birth mothers</td>
<td>5</td>
<td>ADHD</td>
</tr>
<tr>
<td>Foster parents</td>
<td>11</td>
<td>Anxiety</td>
</tr>
<tr>
<td>Adoptive parents</td>
<td>12</td>
<td>Affected Daytime Wellbeing &amp; Challenging Behaviour</td>
</tr>
<tr>
<td>Legal guardian</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>Medication</td>
<td></td>
<td></td>
</tr>
<tr>
<td>ADHD</td>
<td>5</td>
<td>Insomnia</td>
</tr>
<tr>
<td>Antipsychotics/-depressants</td>
<td>11</td>
<td>Parasomnias</td>
</tr>
<tr>
<td>Melatonin (quick/slow acting)</td>
<td>20</td>
<td>Difficulty Breathing</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Suggestive WED/RLS</td>
</tr>
</tbody>
</table>
Results. Analysis of the themes in all qualitative interviews of parents and health care professionals, as well as functional sleep/wake behaviour assessments, acknowledged the importance of sleep, and the lack of it, as a major problem. In all in-depth qualitative interviews, the patients’ families identified the significance of sleep problems affecting the overall wellbeing of the child and the impact of the sleep problem on the entire family’s wellbeing or quality of life. Similarly, health care professionals acknowledged sleep problems as a major issue impacting the entire family and their interaction with the health care system. Describing parents’ experiences with sleep deprivation, one physician in the multidisciplinary FASD resource team stated: “They live silently in terror.”

This phrase captured a leitmotif that ran throughout our qualitative interviews. It expressed the impact of a sleep problem on the child and on the child’s family. It was also reflected in statements like the following by a single parent (birth mother) of an 8-year old girl:

*Excuse me. I think last night by the time we fell asleep it was 10:15 but she got up fine, so it’s () it’s hit and miss with her. ...she can stay up until 3 o’clock on a weekend and she’s up at 6:30 and she has an awesome day, ready to go with only 3 and a half hours sleep. Me, I’m like a you know, zombie.*

*... and I’m supposed to be taking that time for ‘me’ time. Well, by the time I get to ’me’ time, there is no time. So there is no ‘me’ time.*

This was also recognized by a social worker from a multidisciplinary FASD resource team:

---

**Participants of Comprehensive Clinical Sleep Assessments**

<table>
<thead>
<tr>
<th>Involved Disciplines Providing Assessment &amp;/or Therapy</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Behavioural Therapy</td>
<td>3</td>
</tr>
<tr>
<td>Mental Health/Child Psychiatry</td>
<td>10</td>
</tr>
<tr>
<td>Psychology</td>
<td>2</td>
</tr>
<tr>
<td>ENT</td>
<td>5</td>
</tr>
<tr>
<td>Occupational/Speech-Language Therapy</td>
<td>5</td>
</tr>
<tr>
<td>Infant Development Program</td>
<td>1</td>
</tr>
<tr>
<td>Pediatric subspecialty programs: (including respirology, developmental pediatrics, neurology)</td>
<td>8</td>
</tr>
</tbody>
</table>
... I think sleep is probably one of the most critical issues and one of the biggest reasons for respite is because a parent is just exhausted…. ...and sleep disturbance is one of the biggest reasons for breakdown in placement (.)

Interviewer: You're referring to foster placement?

Foster placement and placement at home, too! If the child won't sleep at night.... ... so often their placement can break down just because the family haven't been given the support and once the placement breaks down, it's very hard to turn it around and keep that child in the home, (.) or keep that child in a foster home or-or whatever.

Similarly the community-based social workers recognized the impact of the child's sleep problem on the family:

...and it [= sleep problem] aggravates all the other issues, if they had adequate sleep, would be more manageable. It sounds harsh, but it amplifies the possibility of abuse. You know, because, when you're woken up and you're not sleeping, you're more likely to have some stuff happening as a result of that because the coping mechanisms are just not there.

In addition to the leitmotif, we identified three main themes in the qualitative interviews, the first of which was the recognition of “missing knowledge.” The multidisciplinary FASD resource teams as well as health care professionals in the community setting had missed relevant information about sleep problems, preventing them from properly identifying sleep problems and taking steps to mitigate them. The social worker of a multidisciplinary FASD resource team said:

... hyperactive kids have dis- I mean so many children um have disturbed sleep patterns. And the effects are behaviour and I’m not sure that we (.3) they know enough what to do about it or that it's even recognized. (.) You know, I don't think we have a lot of information even now as to why, or I don’t have a lot of information as to why children don’t sleep.
One of the community-based social workers supported the explanation, that sleep problems were not recognized systemically:

*I think just understanding that it is an issue. The system doesn’t recognize it yet, that it’s a key issue...*

The physician also from the FASD multidisciplinary resource team, elaborated further on this lack of recognition:

... *I think it has to do with healthcare, access to healthcare, ... ... sleep is probably not on the radar, ..., it’s not a priority, ... and they’re (health care professionals are) not aware that unless they’ve taken an interest in their own kids or they’ve been in practice a long time...*

In other words, despite realizing the impact in the interview setting, health care professionals did not know how to deal with the challenge of sleep problems, which significantly affected the patient and/or families’ quality of life. A physician stated:

... *and then again, if we don’t ask about it, they don’t tell you about it. It’s like, if it’s not on the exam, then nobody studied for it. So, we don’t ask; they don’t tell... and I was not formally trained to address sleep problems...*

The narrative analysis of the functional sleep/wake-behaviour assessment confirmed the leitmotif of our qualitative interviews concerning the impact of the child’s sleep problem on the family and the first theme, “*missing knowledge*.” The examination of the patient records confirmed that sleep problems were noted by physicians from various disciplines but they were not integrated into the clinical assessment with a structured approach. They did however provide symptomatic pharmaceutical, behavioural and occupational treatment for the children’s sleep problem (see Table 2.1). Prior to the prescription of sleep medication and other symptomatic strategies, none of these children and adolescents was systematically assessed with a relevant screening questionnaire.
(Owens & Dalzell, 2005) or with more formal diagnostic questionnaire (Chervin, 2000; Owens, Spirito, & McGuinn, 2000). Eventually, after the initial medication proved ineffective in treating the sleep problem, patients were referred to our sleep clinic for formal, systematic assessments. This delay took from a few months to eight years. The explanatory model underlying the initial treatment in the community setting did not recognize sleep problems as a distinct category for clinical investigation and diagnosis, despite textbook like presentations of the exhibited daytime symptoms, as the parent (adoptive mother), who is also the aunt of the 11 year old boy, describes:

Oh he - it's like his brain didn’t turn on when you woke him up. It's just like automatically he'll forget when he's in the shower the soap; he won't brush his teeth; he forgets his shoes on his way out the door to school; he forgets his backpack; he forgets his lunch; he forgets... It's just like his brain didn’t turn on when he woke up. Just like when you wake up, you think I gotta do this, this and this during the day...

Interviewer:  Is that related to the quality of his sleep do you think? Or is it related to the amount of sleep?

I think myself it’s how much sleep he gets; how much sleep he gets...Because then if he has a good night’s sleep, he’s cheerful; he’s cooperative; he’s pleasant to be around (.) then ((inaudible)) fun, it’s great to be around him when he wakes up. When he’s not had enough sleep, he’s just the total opposite child; it’s just like night and day almost. You know, you get this kid that hasn’t has enough sleep and is grumpy, he doesn’t listen and then he also has a hard time at school. It’s-that’s the days that we find that he has had an outburst at school, or hasn’t gotten along with the kids at school, hasn’t done his work at school... ... big temper tantrum!!!
Also, the clinical assessments confirmed the first main theme “missing knowledge,” as the parents described textbook symptoms, which appeared to not have been recognized previously; here a foster mother who cares for a 12-year old boy:

.... and his head will be at awkward strange angles to the point where sometimes I will even like, remove a pillow from under his head so that he can-it just looks better for me, ((laughs))

like letting him [sleep]

Interviewer: [Exactly] the way you describe it, can you describe me how is his head is?

Like this. (.) Or like this even. ((chuckles)) Like it’s really weird angles, way far back and seem to be draped over things. ... ((chuckles)) With his butt in the air (.) is probably the weirdest one and he’s fast asleep. Just with his thumbs in the air like he’s kneeling.

The functional sleep/wake behaviour assessments also identified the second main theme, the dominance of the “conventional mainstream view in current medical practice” that hinders recognition and treatment of sleep problems in children with FASD. This theme suggests that by focusing on the features that receive extensive discussion in the standard-of-care, clinicians had missed the big picture. The functional sleep/wake behaviour assessments and reviews of patients’ records verified that symptoms were identified, but not attributed to possible sleep problems. All diagnoses were complicated by multiple co-morbidities (see Table 2.1). In 15 out of 22 medicated patients, FASD related multiple co-morbidities were addressed with pharmaceutical treatment by specialists (pediatricians and/or child psychiatrists) for ADHD 5/22, and for affected daytime wellbeing and challenging behaviour: 11/22, (multiple answers). Most striking was that 20/27 children had been medicated for sleep problems, as already mentioned, without a systematic reproducible assessment. The functional sleep/wake behaviour assessments revealed that many of these children had behaviours not previously reported in the histories, including disordered breathing and WED. These conditions, which had not been investigated previously, probably
contributed to a significant degree to the FASD-related multiple co-morbidities. (Ipsioglu, Andrew, et al., 2011) Thus, we needed to expand the initial health care professionals’ explanatory models, used by both general practitioners and multidisciplinary FASD resource teams, who did not recognize sleep problems as a distinct category that encompassed both daytime as well as nighttime quality of life and behaviour.

The functional sleep/wake behaviour assessments revealed that the provision of a categorical diagnosis of an FASD obscured the need for full functional assessment and intervention. This was also mentioned by the experienced social worker, who coordinates activities of the resource teams, in the qualitative interview:

...I think the support services are uneven: in terms... if support is attached to a diagnostic category, instead of a functional need. ... cause I think it should be according to function, not according to a diagnostic category.

The complex biographies and social histories of these patients contributed to the “communication barriers” which we identified as the third major theme. Of 27 children, 20 had lived in placements made by social services: a minimum of one placement with foster or adoptive parents. Two returned to live with their parents after living with foster or adoptive parents, and some repeated these cycles as many as seven times. (These complex, placement-related, parenting relations might easily contribute to sleep problems.) The increasing number of placements and the multiple referrals often resulted in poor communication about the diagnostic and therapeutic strategies, leading to a fragmented approach to the interconnected symptoms. Because parents play a key role in communication about children’s health (Miller et al., 2009), these children with (multiple) placements lack parents who have consistent oversight of their medical care.

A variation of the third major theme emerged in different contexts from all interviewees: “communication barriers between patients and health care professionals.” These barriers are often
related to the current common medical practice of uni-directional communication, as described byTodd and Fisher (1993). This was most profound in one instance concerning the diagnosis of anFASD (rather than description of a sleep problem), where the single parent (birth-mother of the 8year old girl) describes her own thoughts in regards to a possible FASD diagnosis, as she consumedalcohol during her pregnancy:

...when I mentioned it to the doctor, he said, “Shut up. You'll get her labeled.” So then itbecame something I shouldn’t … What does a label mean … does that mean they come in down and take her away?

Communicational barriers are formed by health care professionals’ assumptions about FASD andthe care needed for the child and support for the parents. “Missing knowledge” and the lack oftraining about functional diagnosis, in addition to the impact of a sleep problem on the child andparent can contribute to parents’ stress. The same mother said:

There was a public health nurse would come whenever because her birth weight was going down and down but my, everything that was always going through my mind was, they’re gonna take her away from me “cause I’m not good enough... ... you gotta listen, they’re the boss!”

Similarly, communication barriers that resulted from a lack of understanding were also seen amongprimary, secondary and tertiary care providers, hindering screening, diagnosis, treatment andcontinuity of care. More generally, complex medical language, missing or delayed reports aboutclinic visits, and missing an appreciation of parents’ descriptions of symptoms contributed tomiscommunication (Todd & Fisher, 1993).

In summary, the three major themes that emerged from our analysis were the recognitionthat health care professionals were “missing knowledge” about the medical, psychological, andsocial approaches to sleep problems. Second, the “conventional mainstream view in current medical
practice” or the application of categorical diagnoses leads health care professionals to “miss the big picture” in complex children with complex life histories and co-morbidities as they followed up on them. And third, these preceding factors aggravated “communication barriers” that hindered continuity of care. All of these issues contributed to the “mainstream diagnostic and prescribing practices of doctors” (Oldani, 2009, p. 134), which may create its own momentum in health care professional’s responses to desperate parents.

Seeing clinical presentations from parents’ viewpoints and giving them voice led us to modify the screening concept of BEARS (Owens & Dalzell, 2005), which asks about bedtime situations, excessive daytime sleepiness, awakenings, regularity, and snoring. In order to address child-specific presentations related to FASDs or other neurodevelopmental conditions, we re-conceptualized excessive daytime sleepiness as excessive daytime behaviours (including symptoms associated with hyperactivity), and snoring as various signs of sleep disordered breathing. Parents are now encouraged to elaborate about their perceptions of stressful situations and restorative sleep in accounts that relate to the well-being of the child (and themselves). This new perspective reveals the significance of parents’ information and understanding about sleep health, habits and disorders together with 1) the previous medical findings, 2) the re-interpretation of these and the results from clinical (e.g. neurological) and functional (e.g. sensory processing) investigations, and 3) medication and possible medication related interferences. This collaborative functional sleep/wake behaviour assessment summary report is the practical application of therapeutic emplotment and helps to reframe the description of symptoms (such as oppositional defiant behaviour) into a more comprehensive explanation (hypnagogic hallucinations causing anxiety). This negotiated understanding documents the mutual commitments of parents and clinician, and transforms narrative emplotment into a script for therapeutic action.
Relevance. As described above, the etiology and the clinical presentation of sleep problems in children and adolescents with an FASD are poorly understood (Chen et al., 2012). Recent studies investigating sleep problems in school-aged children with FASDs have revealed that restless sleep and wake behaviours are generated by triggering factors such as sensory processing abnormalities (Wengel, Hanlon-Dearman, & Fjelsted, 2011), and Willis Ekbom disease (WED; previously Restless Legs Syndrome) (Ipsiroglu, Black, Garden, & Jan, 2011). Both are known causes of significant insomnia, but previously they have not been recognized as potential causes of sleep problems in children with FASDs. As the diagnosis of sensory processing abnormalities and WED has important implications for treatment, and because diagnostic clues in particular for WED reside in the description of symptoms, there is a need to reassess children’s behaviours in collaboration with parents, who are the best observers of their children’s day- and nighttime activities.

This study demonstrates the complexities in assessing sleep problems of children with an FASD or other neurodevelopmental conditions. Even though parents often identify a sleep problem professionals are not equipped with concepts that deal adequately with the symptoms of chronic sleep deprivation, because day- and nighttime symptoms are approached with different explanatory models. The qualitative interviews taught us that parents’ narratives include almost all key behavioural information needed as part of the diagnosis. Using qualitative interviews, we developed therapeutic emplotments in collaboration with parents that bridged the gaps between the different explanatory models. Further, the collaborative functional sleep/wake behaviour assessment summary reports we produced provided a guide for mutually agreed upon treatments and served as shared reference points or “boundary objects” (Star & Griesemer, 1989, p. 387) to develop trans-disciplinary understandings among different health care professionals.

The most striking finding in the qualitative interviews and functional sleep/wake behaviour assessments was the desperation of the families and the effect of sleep deprivation on their quality of life. Inquiries about parents’ sleep problems were not routine, nor were the response to parents’
concerns given appropriate attention. The current understanding of the situation leads to exhaustion, reduces the family’s resilience, and results in the kind of desperation expressed by all interviewees and summarized by the phrase of a physician, which became the title of this publication.

Physicians are informed by FASD standard-of-care guidelines, which do not include an assessment algorithm for sleep problems. Similarly, the standard diagnostic tools to assess challenging daytime behaviours (e.g. psychometric measures and questionnaires for ADHD) do not include a structured screening component for sleep problems. Thus, health care professionals appear to be driven by an FASD explanatory model that over-generalizes the applicability of conventional diagnostic tools. As a consequence, “missing knowledge” about how to deal in a structured way with sleep problems, and the existing “communication barriers” with parents as described above (in addition to many other systemic problems), resulted in fragmented care that did not adequately deal with sleep problems and their short- or long-term sequelae. This produces a standard of practice that misses the big picture about day- and nighttime behaviours and general wellbeing. The fragmented care and the impact of the conventional mainstream view create an overemphasis on the detailed clinical procedures needed for FASD diagnosis. This appears to generate a confirmation bias that perpetuates a diagnostic blindness to sleep problems, and leads to over-reliance on pharmaceutical interventions.

Oldani (2009), in his analysis of prescription strategies of physicians dealing with FASD-related challenging behaviours, refers to this pattern of medicating children to enable them to function as pharmaceutical emplotment. He describes this common practice as creating a new social reality of global psycho-pharmaceutical prescribing for particularly vulnerable patients (Oldani, 2009). Similarly, many of the physicians in our study treated ADHD or anxiety disorders with psychoactive medications even in children younger than 6 years, without a formal sleep assessment.
**Is the System to Blame?** Diagnostic sleep assessment tools and standard-of-care guidelines have been part of physicians’ practice parameters for a decade (Chesson et al., 2000; Morgenthaler et al., 2006; Morgenthaler et al., 2007). However, these tools and guidelines might have been considered by health care professionals as inappropriate for children, or either too broad for application in specialists’ clinics or too specific for generalists’ clinics. The role of sleep problems in the diagnosis of ADHD was recognized early on, discarded, and then recognized by the American Academy of Pediatrics (2011) again very recently. In our study, all interviewed health care professionals wanted the best for their patients, they were aware of the significance of sleep problems, and they readily admitted that families faced challenges as long as the sleep problem was not solved, but they also recognized that they were missing knowledge about sleep problems. Almost all parents stated that their descriptions of sleep problems were not heard or recognized during the clinical assessments. We initially considered this finding as part of the “communication barriers,” which we could over-come with qualitative interviews and narratives. However, we soon realized the desperate need for a screening concept (e.g. BEARS), which explores not only the child’s day- and night-time behaviours, but also their nighttime quality of life, and the family’s wellbeing and/or ecology (Ipsiroglu, Carey, et al., 2012). Such an approach might help to ground the clinical findings in the family’s real-life situation, overcoming the limitations of current standards.

**Why was Nighttime Quality of Life Missed?** A major critique of health care professionals in western medicine is that medical training is too narrowly focused (Foucault, 1975; Illich, 1974). This limited scope of perception and practice may lead health care professionals to miss the bigger picture of patients’ health problem, or it results in an inaccurate framing of the challenges faced by the patient. Our findings concerning the inattention to nighttime related quality of life, reflect this critique as they reside within a contextual framework of missing the family’s wellbeing and/or ecology in clinical practice (e.g. the effects of a main function like sleep problems on the entire family) and missed priority setting in health care system management (e.g. differentiation between
the needs of acute and chronic care or missing simply chronic sleep problems) (Ipsiroglu, Jan, Freeman, Laswick, et al., 2009; Mitton & Donaldson, 2002; Sloper & Beresford, 2006).

We believe that there are two basic explanations for the insufficient attention to nighttime quality of life: First, though health care professionals have a general appreciation of the importance of sleep and the consequences of sleep problems, their actual practice belies this. Throughout their training and in everyday practice, the endurance of sleep deprivation as well as its modeling by senior physicians results in a problematic understanding that sleep is an existential, but malleable function, which is assumed to be manipulate-able (Barger et al., 2005; Owens, 2001). Second, for the most part, health care professionals work under the premise that chronic care management is still based on acute care concepts with one or two underlying causative agents for each health problem (Miller et al., 2009; Sloper & Beresford, 2006). This conventional acute care-based let us fix the problem mentality, which treats isolated symptoms, competes with the emerging understanding of life-long impairments, where many factors contribute to the co-morbidities of patients and family members, e.g. siblings, and where families’ input regarding quality of life may be essential (Bluebond-Langner, 1997). This understanding requires a different conceptual approach: namely, a functionally focused approach to quality of life, which helps to build a shared language among family members (e.g. parents) and health care professionals. In other words, health care professionals need a broader view that does not miss the big picture, but is aware of patients’, social and family ecologies (Lucyshyn et al., 1997; McKellin, 1995). The acceptance and application of this conceptual approach requires health care professionals to re-examine and reconstruct their own perceived medical and social positioning at each assessment, based on collaboration with parents. Such a collaboration results in bi-directional communication with each side contributing to the construction of therapeutic emploiment.

**Perspectives and Suggested Solutions.** Our response to the systemic problems of identifying and conceptualizing sleep problems in these patients was to involve parents and other
family members in gathering additional information within an ecologically-driven framework, about the child’s day- and nighttime activities in which sleep problems became embedded. The descriptive framework helped us to engage parents in re-conceptualizing the anecdotal information into a more coherent understanding of the sleep problem and treatment using therapeutic scripts, based on our mutual understanding. By engaging parents as partners, we formalized their inclusion in their child’s care. We re-directed attention from generalized treatment strategies that focused only on daytime behaviours, to a perspective that encompasses both day-and nighttime-related quality of life of the child and their family.

**Summary.** With this manuscript we have shown that sleep problems in patients with FASD are under-diagnosed, owing in large part, to health care professionals’ inadequate explanatory models. In addition, health care professionals are aware of this situation, but are constrained by categorical diagnosis derived from acute care diagnostic and therapeutic concepts. Our approach, employing therapeutic emplotment as a bi-directional communication concept, identifies sleep problems as a major functional theme and overcomes the limitations of categorical diagnosis in chronic care conditions. We believe that this family- and social-ecology approach can transform current practice and improve care as we showed in the management of families whose children suffered from inborn errors of metabolism (Stockler et al., 2012), and help to establish a more functionally-based approach to assess sleep problems or other challenging burdens in chronic care management.

---

*With our enhanced understanding of the breadth of training culture-related communication barriers contributing missed sleep problems in children with FASDs, in the next section below, I studied a group of families suffering from another neurological condition, namely WED/RLS. As shown in this manuscript, 22/27 (81%) patients had been diagnosed with ‘suggestive’ WED/RLS; therefore, it was clear that we needed to develop further methodologies in order to speak of ‘probable’ or ‘definitive*
2.2. Research Question #2: “How can we optimize our clinical understanding and try to describe the possible dimension of a problem?”

Structured Behavioural Observations – Application of Medical Ethnography in Exploration of Movement Patterns.

2.2.1. Structured Behavioural Daytime Observations in Children with Neurodevelopmental Conditions on the Willis Ekbo Disease/Restless Legs Spectrum with Early Onset and Chronic Insomnia


Overview. Chronic insomnia, falling asleep and sleep maintenance difficulties, is a core problem of children/adolescents with neurodevelopmental conditions. Clinical observations of their daytime presentations suggest hyperactive-like behaviours and an inability to keep still. We investigated presentations of familial WED/RLS, one of the most frequent neurologic causes of chronic insomnia with a bi-directional language development (therapeutic emplotment) concept. 28 paediatric patients with early onset/chronic insomnia and neurodevelopmental conditions, as well as their mothers, were assessed at a sleep-/wake-behaviour clinic, in addition to clinical assessments, with structured behavioural observations during a SCIT. In order to have a coherent patient population with familial WED/RLS, we focused only on patients whose mothers had been
previously diagnosed with WED/RLS (n=28/31). All patients presented with various sensory processing abnormalities; the most common sensory domain was tactile sensitivities (97%) with a shifted pain threshold (69%), defined as less reactivity to pain. Quotations from parents illustrate the experienced clinical symptoms from a narrative-based perspective, and a case report of a 10-year-old boy frames the clinical picture. Our current understanding is that early onset/chronic insomnia caused by familial WED/RLS has to be investigated further with qualitative methodologies and standardized behavioural observations, as symptoms may start at a very young age and maybe overlaid with neurological, behavioural and psychiatric disturbances, which also impact neurodevelopment, leaving lasting, chronic effects. The SCIT standardizes for the first time the description of restless daytime behaviours and might be helpful, as a supportive diagnostic criterion of familial WED/RLS.

**Introduction.** One of the most common causes of insomnia in adults is Willis Ekbom disease/Restless Legs syndrome (WED/RLS) (Allen et al., 2005; Allen et al., 2014), a neurologic disorder characterized by discomfort (up to pain) of feet, legs, hands, and/or other body parts. This discomfort often worsens during periods of rest and towards the night, and is relieved by movement. Brain iron deficiency and dopaminergic dysfunction have long been regarded as potential mechanisms, (Restless legs syndrome: detection and management in primary care. National Heart, Lung, and Blood Institute Working Group on Restless Legs Syndrome, 2000) and treatment with iron and dopamine agonists is based on this understanding. (Nagandla & De, 2013) The majority of cases are caused by an autosomal dominant genetic trait with variable penetrance (Lavigne & Montplaisir, 1994; Vilarino-Guell, J, & Lin, 2008). Symptoms of WED/RLS can also be aggravated during particular periods of life, e.g. pregnancy (Hübner et al., 2013; Srivanitchapoom, Pandey, & Hallett, 2014; Yeh, Walters, & Tsuang, 2012).
In children and adolescents, the prevalence of WED/RLS ranges from 2% to 4% with 0.5% to 1% experiencing moderate-to-severe WED/RLS based upon results of the United Kingdom and United States-based Peds REST study (Pichietti et al., 2007). Other epidemiological studies have shown similar prevalence rates (Kinkelbur, Hellwig, & Hellwig, 2003; Yilmaz, Kilincaslan, Aydin, & Kor, 2004). However, the dimension of WED’s impact on sleep problems has not been fully recognized in the paediatric population; there is, to our knowledge, not a single publication reporting WED/RLS in children with neurodevelopmental conditions.

There are currently five essential diagnostic criteria for the diagnosis of WED/RLS in children and adolescents, developed in 2013 by a paediatric WED/RLS panel in collaboration with the International Restless Legs Syndrome Study Group. These criteria include: (1) an urge to move the legs, usually accompanied by uncomfortable or unpleasant sensations in the legs; (2) symptoms begin or worsen during rest; (3) symptoms are relieved by movement; (4) symptoms occur mainly in the evening/night; and (5) differential diagnostic thoughts, that symptoms cannot be solely accounted for by another medical condition (Picchietti et al., 2013).

The first four diagnostic criteria of WED/RLS in children are built on verbalized self-report and observation. While the diagnosis relies on the child’s perception and understanding of the dimension of their own symptoms (Picchietti et al., 2011), symptom description becomes a major challenge in young patients and/or children with neurodevelopmental conditions, who are unable to express themselves or use the right word (Tilma, Tilma, Norregaard, & Ostergaard, 2013). This phenomenon has already been acknowledged in adults, who may experience language barriers and cultural differences as a diagnostic limiting factor when describing their symptoms (García-Borreguero et al., 2011). Additionally, children may be missing a reference point due to the early onset, and impacts of chronic disease. For such cases, the International RLS-Study group has suggested the use of supportive criteria such as familial history of WED/RLS, periodic limb movements in sleep, and behavioural observations (Picchietti et al., 2013). As such, while
interviewing parents about their sleep history is possible in the clinical setting, the assessment of period limb movements in sleep requires polysomnography in a sleep lab. Furthermore, there are currently no diagnostic protocols to guide healthcare professionals in screening for or collecting behavioural observations of WED/RLS in children with neurodevelopmental conditions.

The significant role of sensory processing in WED/RLS was recognized very recently (Winkelman, Gagnon, & Clair, 2013). Sensory processing abnormalities can cause major insomnia as demonstrated in children with an FASD diagnosis (Wengel et al., 2011), but the impact of sensory processing abnormalities in insomnia of children with/without neurodevelopmental conditions has not yet been described sufficiently. In response to all these unresolved questions we have developed a structured behavioural approach for the clinical assessment of WED/RLS that includes parents’ narratives regarding their children’s sleep problems, a clinical immobilization test, which encourages patients (and parents) to describe their urge to move as well as questions and observations related to sensory processing disorder and/or sensory processing abnormalities. Here, we describe the application of this approach in children with neurodevelopmental conditions assessed in our clinic suffering from early onset chronic and intractable insomnia. We focus on a cohort of children with challenging sleep/wake behaviours whose mothers had been diagnosed with WED/RLS at some point in their life, confirming the familial aspect of this condition.

Methods

Setting. Assessments were performed between 2010-2014 at the Sleep-/Wake Behaviour Clinic of BC Children's Hospital (University of British Columbia), a sub-specialty clinic assessing patients with neurodevelopmental conditions and chronic sleep problems, whose sleep problems could not be diagnosed and treated by community-based paediatricians. Patients were assessed in approximately 1 to 3 hour sessions depending on the complexity of the patient’s sleep problems and whether the patient needed a break (e.g. run around at the playground before being able to
continue with the assessment. Assessments included (1) clinical sleep/wake-behaviour history, facilitated by parents’ narratives about their children’s sleep problems; (2) an extended family sleep history; (3) a SCIT, which encouraged patients and/or parents to describe their urge to move in a playful clinical test situation. Clinical observations and direct quotations by parents and patients (narratives) were documented by a second clinician (usually a trainee) throughout the duration of the assessment. The end product of the assessment was a sleep/wake behaviour report, which consisted of (1) a detailed description and summary of sleep/wake behaviours (including excerpts of original quotations by the patients and their parents); (2) our interpretations, which incorporated the parents’ narrative in a medical language; and (3) recommendations for parents, and involved community-based support teams. Additional areas elucidated in the reports included medical and functional diagnoses of co-morbidities; on-going therapies; medications (in particular psychotropic medications); and scales for subjective assessment of severity and impacted wellbeing of the child and caregivers. The sleep/wake-assessment reports presented the understanding of both, the parents and patients, and various involved professionals, (Lawlor & Mattingly, 1998; Mattingly, 1994) using inclusive language (i.e. medical terminology with explanations (reading level: grade 5).viii (Ipsiroglu et al., 2013) Parents received the final version of the report and were asked to review them to avoid any misunderstandings or miscommunications. Complex cases were followed-up on and discussed with involved community-based paediatricians and therapy teams.

Assessments

Clinical Sleep/Wake Behaviour History. Conducted as a semi-structured interview using the concept of therapeutic emplotment (Mattingly, 1994), which in the clinical setting we refer as a bi-directional language development exercise. It incorporates interconnected topics, including behaviours at bedtime, overnight awakenings, as well as resting activities over the day. Parents are encouraged to describe the sleep and wake-related behaviours of their child in their own words and
in the context of their everyday routines (narrative schemata) (Ipsiroglu et al., 2013; Lawlor & Mattingly, 1998). BEARS domains (bedtime, excessive daytime sleepiness, awakenings, regularity/routines and snoring) are explored with standard questions such as ‘How often?’ and ‘Since when?’ with special emphasis on recognized urge-to-move patterns in the first four domains (Owens & Dalzell, 2005). To further support the clinical assessment and grasp a more comprehensive clinical picture, some adaptations were made to the BEARS screening concept (Ipsiroglu, Carey, et al., 2012): (i) Excessive daytime sleepiness was altered to excessive daytime behaviours, as hyperactive-like behaviours were explored ex aequo. (ii) Situations which positively facilitated the patient’s ability to fall asleep (i.e. ‘how long does it take him/her to fall asleep in the stroller or during a car ride?’), and particular movement patterns, including prior to falling asleep, immediately after falling asleep and during resting situations when awake (i.e. ‘how still can he/she be in the car seat, can you describe his/her movement patterns?’) are explored in-depth. (iii) In the domain routines and regularity (e.g. hours of sleep), special focus is given to transitioning situations (i.e. from movement to rest and vice versa, e.g. during dinner or at school), in addition to sleep health measures. (iv) Snoring was changed to sleep disordered breathing and signs of sleep disordered breathing, such as open mouth posture, signs of non-restorative sleep (restless/sweating), and problems in getting up in the morning, were screened. In positive cases, symptoms are further investigated with the Pediatric Sleep Questionnaire (Chervin, 2000).

Extended Sleep/Wake Behaviour Family History. This includes questions addressing sleep problems and WED-related symptoms (i.e. exploration of going to bed and falling asleep situations; behaviours during TV watching, as an example for restful activity; quality of sleep, e.g. deep or light sleeper; and getting up situations) as well as history of iron deficiency. Caregivers present during the assessment are asked questions regarding their sleep quality (restful/restless), their level of relaxation during periods of rest (e.g. when they watch TV in the evening), history of iron deficiency, particularly during/after pregnancy for mothers, and whether they had received a
diagnosis of WED at some point in their lives. Not only birth parents, but also adoptive and foster parents are asked these questions in order to develop a shared language and receive their full support in the interpretation of behavioural observations during the SCIT.

**The Suggested Clinical Immobilization Test (SCIT).** This is an adaptation of the laboratory-based Suggested Immobilization Test (SIT; a 40 minute test), which is used during the montage of polysomnography LEDs, (Michaud, 2006) lasts approximately 2-5 minutes, and allows for standardized clinical observations of behaviours and movements. The SCIT is administered to both the child and the parent(s). It comprises of the following steps: (1) removal of socks and shoes; (2) getting up, jumping around and shaking-out; (3) sitting down on an appropriately low chair and remaining motionless in a relaxed position with both bare feet placed on the floor; and (4) describing any experienced sensorimotor symptoms. When the instructions for the SCIT are provided, two chances for winning are offered to create a competitive and fun-filled atmosphere for patients/parents, (i) you win if you don’t move and (ii) you win, if you move and explain why you have moved, and one chance to lose (iii) you lose if you move and don’t explain why you moved. In cases where the SCIT cannot be administered (e.g. due to lack of comprehension, behavioural compliance or motor ability), clinical observations of the child with shoes and socks removed, moving around, coming to rest periods and then again starting movements in the examination room are used instead (informal version). Explaining the observations from the SCIT to the parents usually triggers additional narratives of related information about similar situations at home. Patients’/parents’ descriptions of sensations and clinical observations are also documented in the detailed report, which as mentioned above, are reviewed by parents as a quality control of the observations.

**Exploration of Sensory Processing.** During a segment of the sleep/wake behaviour assessment parents are asked to identify: (1) If their child had experienced any sensory processing
abnormalities (‘Does your child have any sensory processing challenges?’ and ‘Have sensory processing challenges been mentioned by any health care professional?’). (i) If yes, the types of experienced sensory processing abnormalities were further explored with narrative examples provided by the parent(s) (e.g. ‘He/she must wear clothes with the labels removed.’). The pain threshold of the child and affected family members is further explored in each case. (ii) If no, sensory challenges were further explored by specific questions which addressed inability to integrate and respond to sensory stimuli appropriately: ‘Does your child show any responses to touch or auditory stimuli which you consider as different from your other children or his/her peers?’. (2) Whether a formal sensory assessment had been conducted for their child by an occupational therapist trained in assessing sensory problems. Of the patients who had a formal occupational therapy assessment, one patient was selected as a case study to frame the dimension of the discussion (not presented in this dissertation). The child’s Sensory Profile™ scores were depicted visually using a radar chart; the length of each spoke is proportional to the magnitude of the child’s sensory processing score in that domain, as compared to a typically developing child. (Dunn & Westman, 1997)

**Patients and Data Analysis.** This qualitative sleep/wake-behaviour assessment has been applied in our clinic since 2010. We retrospectively analysed the sleep/wake behaviour assessment reports of patients seen in our clinic between 2010 and 2014 who fulfilled the following criteria (see below), and the results were categorized into sleep/wake behaviours and sensory processing abnormalities domains in our phenotyping database. (1) Completed clinical sleep/wake behaviour assessment as described above; (2) Evidence of familial WED spectrum through maternal history of a formal diagnosis of WED or reported experience of WED-related discomfort either continuously or at some point in their lives, e.g. during pregnancy, as well as a self-reported history of chronic or pregnancy-related iron deficiency and/or anaemia (Wagner, 2015). (3) Evidence of WED-related insomnia, specified for patient-based expression of the urge-to-move in his/her own words or if the
patient showed urge-to-move behaviours during the assessment, which were supported by sleep/wake behaviour narratives reported by parents.

**Data analysis.** (1) Reports were analysed to capture clinical history and quotations about sleep/wake behaviours and sensory processing as well as behavioural observations, including the SCIT, made during the assessment. (2) Information about sensory processing was categorized in the main domains (i) auditory, (ii) tactile, (iii) visual, and (iv) oral. Parent and patient quotations regarding sensory processing were organized within the four sensory domains. The selection of descriptive sub-categories was guided by the Sensory Profile™, a standardized questionnaire completed by caregivers and teachers to assess a child’s sensory processing patterns. (Dunn & Westman, 1997) (3) For the SCIT analysis, quotations from the assessment were organized into 3 categories; clinician’s observations of (i) sitting position, (ii) leg/feet/toe movements, and (iii) patient’s descriptions of sensations during formal/informal SCIT. Particular focus was given on descriptions of the patient’s experienced sensations and observed associated behaviours in association with the three essential WED criteria (a) the urge to move the feet (hands, legs or the body in general), (b) their change and development (evolution) during rest; and (c) relieving movement patterns. The experienced sensations, behaviours and clinical observations of leg/feet/toe movement and clinicians’ observations of sitting positions were summarized in the report.

**Results**

**Demographics of Patients.** Out of 463 patients seen in the Sleep/Wake-Behaviour Clinic during the given time frame (2010-2014), 31 patients met the inclusion criteria. Three out of the 31 cases were excluded due to missing accurate information (abbreviated report, language barriers due to missing interpreter during the assessment), leaving a remaining 28 patients for analysis. Of
the 28 children/adolescents, the mean and median age of the cohort was 9 years, with ages ranging from four years to 17 years; 22 were males and six were females.

**Maternal Results. Willis Ekbom Disease:** All mothers had been diagnosed with WED and iron deficiency at some point in their lives, 12 mothers (43%) of them during pregnancy. All mothers recalled the characteristic discomfort in their legs when WED was introduced as a possible explanatory model for insomnia; however, only 18 mothers (64%) complained of insomnia. Half of the population (50%) had other sleep disorders, mainly sleep disordered breathing, and 8 (29%) mental health co-morbidities. SCIT: The test related information was positive in all cases; 27 (96%) participated actively in the formal test and had positive results using various descriptions for the core *urge-to-move* symptom. The remaining one case was from the transitioning period where observations and descriptions led to the introduction of the formal test in clinical practice and were based on the in-depth observation based descriptions from the report (informal test, as done with children who could not actively participate). Table 2.2 provides an overview of the maternal data.

**Patient Results. Presentation:** All patients presented with insomnia, disordered circadian sleep rhythms and parentally reported and observed sensory processing abnormalities. In addition, amongst the 28 patients, there were 38 neurodevelopmental and 45 mental health presentations with typically more than one presentation per patient. Table 2.3 provides a summary of the neurodevelopmental and mental health diagnoses of the 28 children/adolescents; the most common neurodevelopmental presentations were global developmental delay (n=14) understand the *gestalt* of the behaviours, and (c) identify periods of interest, which will be the focus of sub and autism spectrum disorder (n=11). The most common mental health co-morbidities were externalizing disorders or disorders of disruptive challenging behaviours (n=27), with attention deficit hyperactivity disorder (ADHD) being the most common presentation within the *challenging behaviours* group 57% (16) presented with internalizing behaviours, 50% (14) with confirmed
anxiety or anxiety under investigation. All patients suffered from insomnia, 26/28 (93) had falling asleep problems and 25/28 (89%) sleep maintenance problems. All patients fulfilled the criteria for circadian sleep rhythm disorders, 50% had reported parasomnias, and 20/28 presented with signs of sleep-disordered breathing.

**SCIT:** the formal and informal test related information was positive in all cases. **Formal test:** 17 (61%) participated actively in the formal test; 16/17 (94%) patients reported various descriptions of urge to move and showed positive signs of involuntary movements of legs/feet/toes (see Table 2.4). Patient six, a boy younger than six years old with insomnia (treated with clonidine 0.1 mg/daily at nighttime), parasomnia and anxiety disorder (treated with fluoxetine mgs/daily), was the only individual in our cohort who did not show / report motor signs and sensory discomfort during the formal. In the remaining 11 (39%) cases, patients could not participate actively in the test; therefore, the observation based involuntary motor movements at random rest situations were utilized as an informal test from the reports. Sensory Processing Abnormalities were stratified based on the type of parental reports and observations: 96% (n=27) of the patients had a tactile sensitivity; 69% (n=20) presented with a shifted pain threshold, and auditory, visual and/oral sensitivities were reported in 21% (n=6) of the patients.

**Additional Explanatory Information**

**Sleep Wake/Behaviour Narratives through Qualitative Exploratory Interviewing.** Quotations by patients/parents were assigned to the following categories of WED symptomatology: motor and sensory, as well as representative descriptive behaviours during day and nighttime. Daytime motor and behaviour characteristics included descriptions such as *always on the go, motor driven, and fidgety* were reported in all patients. Nighttime motor and behaviour characteristics included *restless sleep* and *kicking movements* and were noted in 72% and 93% of the patients. Most
representative keywords and exemplary descriptive quotations are shown in Table 2.4; Table 2.6 has the original quotations.

**Behavioural Observations during the Assessment.** Patient reported description of symptoms or parent reported (triggered) descriptions from similar *resting* situations during the SCIT have been summarized in formal and informal tests in Table 2.4.

**Descriptions of Sensory Processing Abnormalities.** Parents described a variety of experiences and examples regarding their child’s sensory issues, which have been summarized in Table 2.5. Of the 27/28 patients who experienced tactile sensitivity, the most common descriptive categories were a shifted pain threshold (n=19); sensitivity to clothing tags, closed shoes and socks, as well fabrics (n=14); and *other* tactile-seeking behaviours (n=14). Eleven patients had tactile sensitivities that fell into two or more of the aforementioned categories, within a single sensory domain. Within the *other tactile-seeking behaviours* category, parents described their children biting their own hands and arms, stubbing their toes on purpose, banging and thrashing their head against the wall, and picking at sores. Within the ‘*shifted pain threshold*’ category, some parents told stories of their child getting an injury, such as a “broken arm” or “sliced hand”, and not noticing or reacting appropriately to the incident. A total of six patients fell within the auditory sensitivity domain; one of the commonly reported sensory behaviours was a heightened sensitivity to loud or unexpected sounds (n=5). Five patients also had difficulty focusing in noisy environments, particularly with multiple different sounds occurring at the same time. Within the visual sensitivity domain, parents of four children reported a heightened sensitivity to bright lights, particularly fluorescent lights or sunlight. Parents also described children having difficulty finding objects in competing or complex backgrounds (n=2). Within the oral sensitivity domain, all children had varying degrees of difficulty with the taste, texture and smells of their foods (n=6).
Table 2.2. Maternal Sleep and WED History Data. (n=28)

<table>
<thead>
<tr>
<th>Demographics of Mothers of the Patient Cohort with WED/RLS (n=28)</th>
<th>No. of Mothers of Patients with Confirmed Diagnosis, n (%)</th>
<th>No. of Mothers of Patients with Suspected Diagnosis (Under Investigation), n (%)</th>
<th>Total No. of Confirmed and Suspected in Cohort, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sleep Disorders</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Insomnia</td>
<td>13 (46)</td>
<td>5 (18)</td>
<td>18 (64)</td>
</tr>
<tr>
<td>Restless sleeper and/or light sleep (interpreted as periodic limb movements in sleep)</td>
<td>22 (79)</td>
<td>N/A</td>
<td>22 (79)</td>
</tr>
<tr>
<td>Other Sleep Disorders</td>
<td>12 (43)</td>
<td>3 (11)</td>
<td>15 (54)</td>
</tr>
<tr>
<td>WED diagnosis/SCIT Done in Assessment (+ result)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Willis-Ekbom disease (WED)</td>
<td>28 (100)</td>
<td>0</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Positive SCIT results</td>
<td>25 (89)</td>
<td>N/A</td>
<td>25 (89)</td>
</tr>
<tr>
<td>Positive formal SCIT result</td>
<td>16 (57)</td>
<td>N/A</td>
<td>16 (57)</td>
</tr>
<tr>
<td>Positive informal SCIT result</td>
<td>23 (82)</td>
<td>N/A</td>
<td>23 (82)</td>
</tr>
<tr>
<td>Negative SCIT results</td>
<td>0</td>
<td>N/A</td>
<td>0</td>
</tr>
<tr>
<td>No information on SCIT</td>
<td>3 (11)</td>
<td>N/A</td>
<td>3 (11)</td>
</tr>
<tr>
<td>Maternal Medical History/Pregnancy</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Iron deficiency (at some point in their life)</td>
<td>25 (89)</td>
<td>3 (11)</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Iron deficiency (during pregnancy)</td>
<td>11 (39)</td>
<td>1 (4)</td>
<td>12 (43)</td>
</tr>
<tr>
<td>Increased Discomfort WED/RLS Symptoms during pregnancy</td>
<td>5 (18)</td>
<td>N/A</td>
<td>5 (18)</td>
</tr>
<tr>
<td>Constant/Increased movements of foetus during pregnancy</td>
<td>5 (18)</td>
<td>N/A</td>
<td>5 (18)</td>
</tr>
<tr>
<td>Teratogenic exposures in utero (drugs, alcohol, smoking)</td>
<td>2 (7)</td>
<td>N/A</td>
<td>2 (7)</td>
</tr>
<tr>
<td>Other pregnancy-related complications</td>
<td>8 (29)</td>
<td>N/A</td>
<td>8 (29)</td>
</tr>
<tr>
<td>Mental health diagnosis</td>
<td>10 (36)</td>
<td>1 (4)</td>
<td>11 (39)</td>
</tr>
</tbody>
</table>

(1) No information on SCIT: Two mothers were not present during the assessment of their child, one mother does not have record of the SCIT being performed during the assessment, and one mother could not be assessed, as the assessment was conducted via Telehealth.

Table 2.3. Patient Demographics and Day/Nighttime Clinical Presentations in Paediatric Patients with WED. Patients in the cohort often had more than one diagnosis (both confirmed and/or under investigation), contributing to their complex presentations. (n=28, mean9y/median9y; min 4y; max 17y).

<table>
<thead>
<tr>
<th>Demographics of Patient Cohort (n=28, mean9y/median9y; min 4y; max 17y).</th>
<th>No. of Patients with Confirmed Diagnosis, n (%)</th>
<th>No. of Patients with Suspected Diagnosis (Under Investigation), n (%)</th>
<th>Total No. of Confirmed and Suspected Diagnoses in Cohort, n</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neurodevelopmental Conditions</td>
<td>28 (100)</td>
<td>10 (36)</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Autism Spectrum Disorder (ASD)</td>
<td>10 (36)</td>
<td>3 (11)</td>
<td>13 (47)</td>
</tr>
<tr>
<td>Developmental Delay/Intellectual Disability (including Down Syndrome)</td>
<td>18 (64)</td>
<td>4 (14)</td>
<td>22 (79)</td>
</tr>
<tr>
<td>Motor Disorder (Developmental Coordination Disorder, repetitive movements, Tourette syndrome, etc.)</td>
<td>4 (14)</td>
<td>6 (21)</td>
<td>10 (36)</td>
</tr>
<tr>
<td>Alcohol-Related Neurodevelopmental Disorder</td>
<td>2 (7)</td>
<td>0</td>
<td>2 (7)</td>
</tr>
<tr>
<td>Mental Health Co-Morbidities</td>
<td>11 (39)</td>
<td>19 (68)</td>
<td>23 (82)</td>
</tr>
<tr>
<td>Externalizing Disorders or Disorders of Disruptive Challenging Behaviour</td>
<td>10 (36)</td>
<td>13 (47)</td>
<td>22 (79)</td>
</tr>
<tr>
<td>ADHD</td>
<td>8 (29)</td>
<td>12 (43)</td>
<td>20 (71)</td>
</tr>
<tr>
<td>Oppositional Defiant Disorder,</td>
<td>3 (11)</td>
<td>0</td>
<td>3 (11)</td>
</tr>
<tr>
<td>Conduct Disorder</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Attachment Disorder</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Neurobehaviour Disorder</td>
<td>1 (4)</td>
<td>1 (4)</td>
<td>2 (7)</td>
</tr>
</tbody>
</table>
### Demographics of Patient Cohort

(n=28, mean 9y/median 9y; min 4y; max 17y).

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>No. of Patients with Confirmed Diagnosis, n (%)</th>
<th>No. of Patients with Suspected Diagnosis (Under Investigation), n (%)</th>
<th>Total No. of Confirmed and Suspected Diagnoses in Cohort, n</th>
</tr>
</thead>
<tbody>
<tr>
<td>Internalizing Disorders</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anxiety Disorder</td>
<td>8 (29)</td>
<td>7 (25)</td>
<td>14 (50)</td>
</tr>
<tr>
<td>Depression</td>
<td>2 (7)</td>
<td>1 (4)</td>
<td>3 (11)</td>
</tr>
<tr>
<td>Hyperphagia</td>
<td>1 (4)</td>
<td></td>
<td>1 (4)</td>
</tr>
<tr>
<td><strong>Sleep Disorders</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Insomnia</td>
<td>28 (100)</td>
<td>9 (32)</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Falling asleep problems</td>
<td>26 (93)</td>
<td>0</td>
<td>26 (93)</td>
</tr>
<tr>
<td>Sleep maintenance</td>
<td>25 (86)</td>
<td>0</td>
<td>25 (89)</td>
</tr>
<tr>
<td>Restless sleep (interpreted as periodic limb movements in sleep)</td>
<td>25 (61)</td>
<td>1 (4)</td>
<td>26 (93)</td>
</tr>
<tr>
<td>Circadian Sleep Rhythm Disorder (CRSD)</td>
<td>28 (100)</td>
<td>0 (0)</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Clinical sleep-disordered breathing</td>
<td>24 (86)</td>
<td>2 (7)</td>
<td>26 (93)</td>
</tr>
<tr>
<td>Parasomnias</td>
<td>17 (61)</td>
<td></td>
<td>17 (61)</td>
</tr>
<tr>
<td><strong>WED diagnosis &amp; SCIT Results (+ result)</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Willis Ekbom disease (WED)</td>
<td>28 (100)</td>
<td>N/A</td>
<td>28 (100)</td>
</tr>
<tr>
<td>Positive formal SCIT result</td>
<td>17 (61)</td>
<td>N/A</td>
<td>17 (61)</td>
</tr>
<tr>
<td>Positive informal SCIT result</td>
<td>12 (43)</td>
<td>N/A</td>
<td>12 (43)</td>
</tr>
<tr>
<td>No information on SCIT</td>
<td>2 (7)</td>
<td></td>
<td>2 (7)</td>
</tr>
</tbody>
</table>

#### Table 2.4. WED/RLS Indicators in Paediatric Patients

The table structures the core symptom ‘urge to move’ (A) by parental report at day- and nighttime; (B) patient descriptions of sensations and self-reflected behaviours during the formal and informal SCIT; (C) clinical observations during formal and informal SCIT; (D) experienced insomnia symptoms, and (E) the repetitive behaviours worsening towards the night.

<table>
<thead>
<tr>
<th>WED/RLS Indicators</th>
<th>Quoted by patients/parents/clinician n= 28 (%) patients</th>
<th>Keywords &amp; examples of representative quotations from the patient, parents &amp;/or assessing teams observations; original quotations from the reports</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>(A) Urge to Move (parental report)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Motor daytime</td>
<td>14 (50)</td>
<td>Motor driven (parental report and observations of assessing team)</td>
</tr>
<tr>
<td>Motor nighttime</td>
<td>24 (86)</td>
<td>Restless sleep, kicking movements (parental report)</td>
</tr>
<tr>
<td><strong>(B) Urge to Move During (formal/ informal) SCIT:</strong> Patient Descriptions of Sensations &amp; Associated Behaviours (n=25)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sensations in feet/toes</td>
<td>5 (20)</td>
<td>- ‘... feels like a force of air is trying to push [his] feet to move’, (Patient #18, own wording)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- He describes that at school his feet are always ‘going nuts’ and while he can choose to remain still, he prefers not to and reports that his legs become ‘less nuts’ when he sits on them.” (Patient # 10, own wording)</td>
</tr>
<tr>
<td>Sensations in legs</td>
<td>5 (20)</td>
<td>- ‘She gets ‘pins and needles,’ describes that her ‘whole leg feels funny,’ and has a sensation that ‘comes up until above [her] knees.’ (Patient #2, own wording)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ‘He feels ‘energy’ in his legs and body and like that ‘something’ is moving through his legs and ‘controlling’ his body.” (Patient # 19, own wording)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ‘She describes that her legs ‘tingle’ and they feel ‘cold’ and ‘numb’ when she tries to keep them still and she feels as if there is ‘pressure on her legs,’ and it feels ‘weird,’ especially in her ‘calves.’” (Patient #16, own wording)</td>
</tr>
<tr>
<td>Difficulties sitting still</td>
<td>25 (100)</td>
<td>- ‘He likes to sit on his legs and occasionally purposely stubs his toe on the ground because it feels good.’ (Patient #11, own wording and observations)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- ‘He pressed his feet against the wall when lying down, and needed constant tension throughout his body, especially in his legs.” (Patient #22, observations)</td>
</tr>
<tr>
<td>WED/RLS indicators</td>
<td>Quoted by patients/parents/clinician n= 28 (%) patients</td>
<td>Keywords &amp; examples of representative quotations from the patient, parents &amp;/or assessing teams observations; original quotations from the reports</td>
</tr>
<tr>
<td>---------------------</td>
<td>--------------------------------------------------------</td>
<td>----------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>(C) Urge to Move Associated Behavioural Observations</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clinician’s Observations of positions in Formal SCIT/Observations of Positions in Informal SCIT (n=25)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sitting Position:</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
| at edge of chair | 5 (20) | - “He sat on the edge of a table with pressure on one foot (folded and pressed against the end of the table).” (Patient #22, observations)  
- “He sat on the edge of his seat tipping his chair on its front legs with only the tips of his toes touching the ground.” (Patient #10, observations) |
| w/ legs/feet crossed | 11 (44) | - “She always had her legs crossed (e.g. crossed at the ankles, crossed with one ankle on her knee, or crossed at the knees).” (Patient #7 and observations) |
| w/ legs swinging/kicking | 12 (48) | - “When sitting, P#24 preferred to continuously swing her legs back and forth.” (Patient #24, observations) |
| on lower leg/feet | 8 (32) | - “P#10 ‘always sits with one leg under’ and has ‘sat like that for years’, which his school reported as well.” (Patient #10; parental description) |
| in abnormal positions | 7 (28) | - “At one point during the assessment, he sat awkwardly with his right leg hanging over the right arm rest. We observed him swinging his right leg.” (Patient #19; observations) |
| Leg/Feet/Toe Movements: |  | |
| stretching/constant movements | 17 (68) | - “Her legs and feet were constantly moving, i.e. continuously shaking her leg and feet at a quick pace, circling her foot in the air, bouncing her feet up and down with only her toes touching the ground.” (Patient #16, observations)  
- “She continuously moved her feet and legs and walked around with her feet curled in.” (Patient 26; observations of assessment team) |
| repetitive movements | 7 (28) | - “He moved his legs/feet (e.g. tapping feet up and down, flexing and un-flexing toes, rocking forward on toes and burrowing his toes inside of a shoe). (Patient #3, observations) |
| rubbing together/clenching to increase tension | 14 (56) | “He would also rub his foot against the chair leg. When moving around in the chair, he kept his feet pressed together to increase tension.” (Patient #13, observations)  
“With her shoes and socks removed, we observed that P#20’s toes infrequently twitched and she occasionally clenched her toes/feet together; P#20 explained that it ‘feels good’ when she clenches her feet.” (Patient #20; own wording and observations) |
| raising heels | 17 (68) | - “When his feet were on the ground, he would put pressure on his toes or heels to increase the tension in his legs, or lean back by pressing on his toes.” (Patient #3, observations) |
| (D) Urge to Move at Night |  | |
| Falling asleep problems | 26 (93) |  |
| Sleep maintenance problems | 24 (86) |  |
| (E) Repetitive Behavioural Patterns: Increasing Towards Bedtime and/or Worsening at Rest |  | |
| Challenging daytime behaviours: Excessive daytime sleepiness and enhanced hyperactive like behaviours towards nights | 28 (100) | - “When Aaron becomes increasingly hyper during the day, he will use a small trampoline to get rid of some of his hyperactivity, which will allow him to concentrate better on his school work.” (Patient 18; parental descriptions) |
| Challenging nighttime behaviours: Secondary behavioural insomnia/limit setting insomnia | 24 (86) | - “Refuses to go to bed every night, and it is reported that he does not seem anxious at bedtime. He tends to be highly active and restless before falling asleep, and is very reluctant to remain in his bed; he play[s] on [his] iPad or watches television, which reduces his restlessness so he can sit relatively still; [A parent] must stay with him during the falling-asleep period, as he often becomes distressed (e.g. ‘freaks out’) if [left alone], often screaming or crying.” (Patient 23, parental descriptions) |
### Table 2.5. Parental Descriptions of Paediatric Sensory Processing Abnormalities. Quotations taken from Sleep/Wake Behaviour Assessment Reports. (N=28, mean9y/median9y; min 4y; max 17y). Parents have received and reviewed original copies of the assessment reports from which quotations of their own wording have been taken. *Refer to Table 2.6 for patient profiles.*

<table>
<thead>
<tr>
<th>Sensory Domain (% of Children with SPA)</th>
<th>Examples of Parent-Reported Descriptions of Child’s Sensory Processing Abnormalities (child’s age, gender, patient ID*)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Tactile (n=27, 96%)</strong></td>
<td></td>
</tr>
<tr>
<td>Shifted pain threshold (N= 19)</td>
<td>- “He has an extremely high pain tolerance. He barely even cried when he broke his arm.” (7 y, male, 2)</td>
</tr>
<tr>
<td></td>
<td>- “He has a high tolerance to pain. He sliced his hand down to the bone and barely cried for 20 minutes.” (7 y, male, 3)</td>
</tr>
<tr>
<td></td>
<td>- “Picks a sore until it bleeds, and won’t let anyone else touch it.” (14 y, male, 19)</td>
</tr>
<tr>
<td></td>
<td>- “[He’s] constantly touching objects and has no awareness of personal space. He can’t feel when his face is dirty and has a reduced awareness of pain.” (14 y, male, 19)</td>
</tr>
<tr>
<td>Sensitivity to clothing tags, shoes and fabrics (N= 14)</td>
<td>- “He must wear clothes with the label removed and hates ‘closed things’ like closed toe shoes.” (9 y, male, 5)</td>
</tr>
<tr>
<td></td>
<td>- “Only likes to wear sweatpants and refuses to wear jeans. He only started wearing shoes outside house instead of slippers this year.” (6 y, male, 6)</td>
</tr>
<tr>
<td></td>
<td>- “Prefers to wear comfortable clothes such as fleece, sweats, and shorts, and does not like tags or pants with cords.” (10 y, male, 13)</td>
</tr>
<tr>
<td></td>
<td>- “Sensitive to touching certain textures. Started off a bit apprehensive to touch damp washcloth and other textures.” (10 y, male, 13)</td>
</tr>
<tr>
<td>‘Other’ tactile-seeking behaviours (N = 14)</td>
<td>- “He has a grand, unusual fascination with texture. He has a fascination with rubbing the carpet.” (7 y, male, 15)</td>
</tr>
<tr>
<td></td>
<td>- “He has many sensory seeking behaviours. He enjoys very close hugs (i.e. deep pressure) and at night, these hugs need to be especially firm.” (6 y, male, 22)</td>
</tr>
<tr>
<td></td>
<td>- “He stubs his toes on purpose because he says it ‘feels good’” (11 y, male, 11)</td>
</tr>
<tr>
<td></td>
<td>- “He needs more input through his tactile system to get the same level of awareness as others. Constantly touching objects and has no awareness of personal space.” (14 y, male, 19)</td>
</tr>
<tr>
<td><strong>Auditory (N=6, 21%)</strong></td>
<td></td>
</tr>
<tr>
<td>Heightened sensitivity to unexpected or loud noises (N=5)</td>
<td>- “He has heightened sensations. If he hears a loud thump – he can’t stop thinking about it.” (10 y, male, 4)</td>
</tr>
<tr>
<td></td>
<td>- “He holds his hands over his ears for sudden noises. He will cover his ears and scream [if he hears] a dog barking.” (10 y, male, 13)</td>
</tr>
<tr>
<td>Is distracted/has trouble functioning if there is a lot of noise around (N=5)</td>
<td>- “He has difficulty participating in group activities where there is a lot of talking. [He] frequently holds his hands over his ears to protect [himself] from the sounds.” (9 y, male, 12)</td>
</tr>
<tr>
<td></td>
<td>- “He keeps a special noise distracting machine by his bedside to help him sleep.” (11 y, male, 18)</td>
</tr>
<tr>
<td></td>
<td>- “He’s upset by loud sounds and has trouble focusing in a noisy environment. [He] often doesn’t respond to his name.” (14 y, male, 19)</td>
</tr>
<tr>
<td><strong>Visual (N=6, 21%)</strong></td>
<td></td>
</tr>
<tr>
<td>Bothered by bright lights after others have adapted (N=4)</td>
<td>- “Very bothered by sunlight and bright lights.” (10 y, male, 13)</td>
</tr>
<tr>
<td></td>
<td>- “Fluorescent lights seem to agitate her and she needs to leave room” (5 y, female, 24)</td>
</tr>
<tr>
<td>Difficulty visualizing objects in complex/crowded background (N=2)</td>
<td>- “Has a hard time finding objects in competing backgrounds, such as shoes in a messy room. He misses written or demonstrated directions more than other students in the classroom.” (9 y, male, 12)</td>
</tr>
<tr>
<td><strong>Oral (N=6, 21%)</strong></td>
<td></td>
</tr>
<tr>
<td>Avoids certain food tastes/smells/textures that are typically part of children’s diets (N=6)</td>
<td>- “He is very sensitive to the smell and texture of his food. He gets hungry around 8pm...but will only eat a specific meal of popsicles, 3-6 peanut butter chocolate granola bars, and some fruit. He has been seeing an Occupational Therapist since the age of 4 to work on his oral sensory sensitivities.” (10 y, male, 13)</td>
</tr>
<tr>
<td></td>
<td>- “Only eats one type of food that he likes for about 3 days at a time.” (6 y, male, 6)</td>
</tr>
</tbody>
</table>
Table 2.6. Patient Profiles (Reference List). Patient name, gender, confirmed NDC/psychiatric diagnoses, sleep problems and SPAs mentioned by parent during the Sleep/Wake Behaviour Assessment. All mothers were on the spectrum of WED and/or had iron deficiency during pregnancy. Note: * indicates a high pain tolerance, included within the tactile domain. Patient identification assigned randomly.

<table>
<thead>
<tr>
<th>ID</th>
<th>Age Range (&lt;6, 7-12, &gt;12), Gender</th>
<th>Confirmed NDC &amp;/or Psychiatric Diagnoses</th>
<th>Sleep Problems in addition to WED &amp; insomnia</th>
<th>Sensory Processing Abnormalities as reported by Parent</th>
<th>Formal (F1/ Informal (I)2 SCIT</th>
<th>SCIT (+/-)</th>
<th>Behavioural Observation Quotes</th>
<th>Sensorimotor Sensations Quotes</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>&lt;6, male</td>
<td>Copy number variant 12q11 of unknown significance, Autism spectrum disorder, developmental delay</td>
<td>Parasomnias, Suspected sleep-disordered breathing</td>
<td>Tactile</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A - He was very active and intent on climbing furniture, and would move his feet regularly even while sitting with a toy. (Insufficient information from report)</td>
<td>N/A (no descriptions documented)</td>
</tr>
<tr>
<td>2</td>
<td>7-12, male</td>
<td>Mild intellectual disability, ADHD</td>
<td>Sleep-disordered breathing (obstructive sleep apnoea), sleep-related movement disorder, periodic limb movements, parasomnias</td>
<td>Tactile*</td>
<td>I +</td>
<td>&quot;on his tiptoes to increase tension in his body&quot;; urge to move</td>
<td>Mother reports, &quot;he does not complain of leg pains but feels that &quot;he might not have the language to articulate this.&quot;</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>7-12, male</td>
<td>Anxiety disorder</td>
<td>Sleep-disordered breathing, periodic limb movements, parasomnias</td>
<td>Tactile*, Auditory, Visual</td>
<td>F +</td>
<td>&quot;immediately moved his toes and stretched his feet after he sat down&quot;; his feet are &quot;relaxing quiet&quot; and are not bothering him as much;</td>
<td>&quot;he experiences pain and/or discomfort in his legs almost every night</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>7-12, male</td>
<td>ADHD, Disorder of disruptive challenging behaviour</td>
<td>Sleep-disordered breathing, parasomnias</td>
<td>Tactile, Auditory</td>
<td>F +</td>
<td>&quot;stretched his legs/feet after approx. 7 minutes&quot;; &quot;minor tingling sensations at the end of his toes&quot;</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>7-12, male</td>
<td>Disorder of disruptive challenging behaviour</td>
<td>Periodic limb movements during sleep/wakfulness</td>
<td>Tactile</td>
<td>F +</td>
<td>&quot;feet twitching&quot;; &quot;feels like [he] must do some activity&quot;, explaining the reason for his constant shifting and kicking of his feet; he could not control [his feet] and that [the twitches] usually happen automatically&quot;</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>&lt;6, male</td>
<td>Social/Generalized/Separation Anxiety, parasomnias</td>
<td>Insomnia, parasomnias</td>
<td>Tactile*, Visual, Oral</td>
<td>I -</td>
<td>&quot;very calm and relaxed, not &quot;He responded appropriately&quot;</td>
<td></td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range (&lt;6, 7-12, &gt;12), Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F1/Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
</tr>
<tr>
<td>----</td>
<td>----------------------------------</td>
<td>--------------------------------------</td>
<td>---------------------------------------------</td>
<td>--------------------------------------------------</td>
<td>----------------------------</td>
<td>----------</td>
<td>--------------------------------</td>
<td>----------------------------------</td>
</tr>
<tr>
<td>7</td>
<td>7-12, female</td>
<td>Developmental Delay, Neurofibromatosis Type 1, cerebral dysgenesis with migrational abnormality, Hyperphagia</td>
<td>Parasomnias (including confusional arousals), periodic limb movements in sleep/wakefulness</td>
<td>Tactile*</td>
<td>F +</td>
<td>“showed a significant amount of fidgeting”</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>7-12, male</td>
<td>Trisomy 21, Autism Spectrum Disorder</td>
<td>Parasomnias, periodic limb movements wakefulness/sleep, suspected sleep-disordered breathing</td>
<td>Tactile*</td>
<td>I +</td>
<td>“like a yogi, showing incredible body control”; “bending his body around the room on his toes (increase the tension in his legs)” ; “sitting with legs crisscrossed with his feet underneath his butt”; “sitting on his lower legs”; “sitting with his legs wide apart”; “sitting with only his bum touching the floor and lifting his legs off the ground” ; “sitting with his arms clenched to the sides of his body” “bending his body forward and over his legs, sometimes with his bum slightly raised above the ground”; “sat on his lower leg while facing his father, on his lap” “walked on his toes and periodically clenched his feet” “walking around the room in circles (on his toes)”</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range (6, 7-12, &gt;12), Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F)1/Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
</tr>
<tr>
<td>----</td>
<td>--------------------------------</td>
<td>------------------------------------------</td>
<td>---------------------------------------------</td>
<td>-------------------------------------------------</td>
<td>--------------------------------</td>
<td>------------</td>
<td>--------------------------------</td>
<td>--------------------------------</td>
</tr>
<tr>
<td>9</td>
<td>&gt;12, male</td>
<td>Autism Spectrum Disorder, ADHD</td>
<td>Periodic limb movements during wakefulness, suspected sleep-disordered breathing</td>
<td>Tactile* F</td>
<td></td>
<td>+</td>
<td>&quot;has difficulty sitting relaxed.&quot;</td>
<td>&quot;needed to concentrate really hard to keep his feet flat on the ground without fidgeting or moving&quot;; &quot;legs were getting really warm&quot;; &quot;legs were uncomfortable and were a little shaky but that he was 'not really sure' how he felt exactly&quot;; described that at school, his feet are always &quot;going nuts&quot; and while he can choose to remain still, he prefers not to and reports that his legs become &quot;less nuts&quot; when he sits on them.</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>&lt;6, male</td>
<td>Anxiety disorder</td>
<td>Parasomnias, periodic limb movements during wakefulness and sleep, hypnagogic hallucinations, sleep-disordered breathing</td>
<td>Tactile* F</td>
<td></td>
<td>+</td>
<td>&quot;clenched his feet and twitched his toes. When asked to sit still in a relaxed and seated position, [he] increased the tension in his feet and his toes continued to twitch.&quot;</td>
<td>N/A (no descriptions documented)</td>
</tr>
<tr>
<td>ID</td>
<td>Age Range &amp; Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F1/Informal (I)2 SCIT)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
<td></td>
</tr>
<tr>
<td>----</td>
<td>-------------------</td>
<td>----------------------------------------</td>
<td>---------------------------------------------</td>
<td>--------------------------------------------------------</td>
<td>--------------------------------</td>
<td>-------------------------------</td>
<td>--------------------------------</td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>7-12, male</td>
<td>ADHD</td>
<td>Periodic limb movements during sleep</td>
<td>Tactile*</td>
<td>F+</td>
<td>&quot;continuous twitches in his toes during the assessment (periodic limb movements during wakefulness) and his leg 'banging' / 'pushing'; &quot;sitting on the edge of his seat tipping his chair on its front legs with only the tips of his toes touching the ground; sitting on his lower legs while playing with Lego on the floor&quot;; &quot;[he] occasionally swung his legs back and forth over the edge of his seat. At one point during the assessment, [he] sat on the edge of his seat tipping his chair on its front legs with only the tips of his toes touching the ground.&quot; &quot;sits on his legs and occasionally, and purposefully stubs his toe on the ground.&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>7-12, male</td>
<td>ADHD, Anxiety disorder, Tourette syndrome, partial complex epilepsy</td>
<td>Periodic limb movements in sleep and wakefulness, sleep-disordered breathing</td>
<td>Tactile*, Auditory, Visual, Oral</td>
<td>I+</td>
<td>&quot;says that he &quot;moves a lot&quot; in bed.&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>13</td>
<td>7-12, male</td>
<td>ADHD, Separation anxiety</td>
<td>Periodic limb movements in wakefulness, sleep-disordered breathing, parasomnias</td>
<td>Tactile*, Auditory, Visual, Oral</td>
<td>F+</td>
<td>&quot;immediately transferred pressure to his toes (to increase tension); &quot;flickering movements in his toes&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F)1/Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
</tr>
<tr>
<td>-----</td>
<td>-----------</td>
<td>------------------------------------------</td>
<td>-----------------------------------------------</td>
<td>------------------------------------------------------</td>
<td>---------------------------------</td>
<td>-----------</td>
<td>-------------------------------</td>
<td>--------------------------------</td>
</tr>
<tr>
<td>14</td>
<td>7-12, male</td>
<td>Autism spectrum disorder, seizure disorder, global developmental delay</td>
<td>Periodic limb movements during sleep, sleep-disordered breathing, parasomnias (night terrors, sleep speaking, teeth grinding)</td>
<td>Tactile</td>
<td>F</td>
<td>+</td>
<td>&quot;sat leaning forward on the front edge of his seat; &quot;sat with his feet not completely flat on the floor (either his toes or heels were touching the ground)&quot;</td>
<td>N/A (no descriptions documented)</td>
</tr>
<tr>
<td>I</td>
<td>+</td>
<td>&quot;constantly moving and ‘on the go’ throughout the assessment, from bouncing up and down on his lower legs on the floor to climbing up the assessment bed and jumping up and down. When sitting in a chair, [he] sat leaning forward on front edge of his seat; ‘constantly shuffled and rubbed his feet against the floor, when he was sitting in a chair as well as when he was walking around the room’; periodically rubbed or squeezed [his] legs during the assessment and he would stop kicking and moving and appeared more relaxed. For the majority of the assessment, Judah sat hunched over playing with his iPad while sitting on his lower legs on the floor or on the assessment bed. [His mother] reported that sitting on his lower legs is [his] favorite position.</td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range (&lt;6, 7-12, &gt;12), Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormal-ities as reported by Parent</td>
<td>Formal (F1) / Informal (I) SCIT</td>
<td>SCT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
</tr>
<tr>
<td>----</td>
<td>----------------------------------</td>
<td>------------------------------------------</td>
<td>---------------------------------------------</td>
<td>--------------------------------------------------</td>
<td>---------------------------------</td>
<td>----------</td>
<td>-------------------------------</td>
<td>----------------------------------</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Tactile</td>
<td></td>
<td></td>
<td></td>
<td>At one point when [he] was on the floor sitting on his lower legs, he lowered down on to his back and rocked his legs up.</td>
</tr>
<tr>
<td>15</td>
<td>7-12, male</td>
<td>Autism spectrum disorder, Developmental delay</td>
<td>Parasomnias (nightmares, sleep talking and possible sleep walking), suspected sleep-disordered breathing, suspected hypnagogic hallucinations</td>
<td>Tactile I</td>
<td></td>
<td>+</td>
<td>&quot;toes were continuously flexed&quot;; &quot;toes were always under tension&quot;; &quot;showed various feet/leg positions that could be interpreted as uncomfortable. (Standing with weight only on one foot with both feet arched); &quot;toes clenched and knees slightly bent&quot;; &quot;sitting with legs wide apart&quot;</td>
<td>N/A (no descriptions documented)</td>
</tr>
<tr>
<td>16</td>
<td>&gt;12, female</td>
<td>Diffuse idiopathic pain syndrome, depression</td>
<td>Suspected sleep-disordered breathing, periodic limb movements during sleep/wakefulness</td>
<td>Tactile F</td>
<td></td>
<td>+</td>
<td>&quot;experienced difficulties when asked to sit still in a relaxed position, with her feet flat on the floor.&quot;</td>
<td>&quot;frequently and stretching her arms and legs periodically. Throughout the assessment, her legs and feet were constantly moving (i.e. continuously shaking her leg and feet at a quick pace, circling her foot in the air, bouncing her feet up and down with only her toes touching the ground). Maia sat for the majority of the time with her legs crossed at her ankles or knees, with her hands positioned under or between her thighs; she switched the positions of her legs frequently. At one point, Maia sat with her entire body hunched over and bent at the knees, with only her toes touching the seat of the chair (with her entire body weight feet &quot;tingle&quot;, &quot;feel cold&quot;, and &quot;numb&quot;; &quot;feels as if there is pressure on her legs&quot;; &quot;especially during the falling asleep time, she experiences a &quot;burning, itching feeling&quot; in her foot&quot;; During the night, she reports that her &quot;legs get itchy&quot; and her knees hurt&quot;;</td>
</tr>
<tr>
<td>ID</td>
<td>Age Range</td>
<td>Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormal -ities as reported by Parent</td>
<td>Formal (F1/Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
</tr>
<tr>
<td>----</td>
<td>------------</td>
<td>--------</td>
<td>----------------------------------------</td>
<td>---------------------------------------------</td>
<td>----------------------------------------------------------</td>
<td>---------------------------</td>
<td>-------------</td>
<td>---------------------------------</td>
</tr>
<tr>
<td>17</td>
<td>1-6, female</td>
<td>Autism spectrum disorder</td>
<td>Suspected periodic limb movement disorder</td>
<td>Oral</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A (no assessment was conducted)</td>
<td>N/A (no assessment was conducted)</td>
</tr>
<tr>
<td>18</td>
<td>7-12, male</td>
<td>ADHD (suspected, not yet confirmed)</td>
<td>Parasomnias (sleepwalking), periodic limb movements in sleep and wakefulness</td>
<td>Tactile*, Auditory</td>
<td>F</td>
<td>+</td>
<td>&quot;very uncomfortable when asked to sit still&quot;; &quot;restless in his legs and his hands.&quot;</td>
<td>&quot;it really hurts in his feet and legs if he does not move his legs&quot;; &quot;feels like a force of air is trying to push my feet to move&quot;; &quot;feels that he has never been very good at sitting still.&quot;</td>
</tr>
<tr>
<td>19</td>
<td>&gt;12, male</td>
<td>Complex neuropsychiatric phenotype with ADHD, learning disability, motor disorder, depression, anxiety disorder</td>
<td>Sleep disordered breathing (on nCPAP)</td>
<td>Tactile*, Auditory</td>
<td>F</td>
<td>+</td>
<td>&quot;We observed him flexing his toes and shifting around (he was unable to keep still for very long).&quot;</td>
<td>&quot;felt 'energy' in his legs and body&quot;; &quot;felt something' is moving through his legs and 'controlling' his body&quot;; &quot;felt like moving&quot;; &quot;flexing his toes and shifting around&quot;</td>
</tr>
<tr>
<td>20</td>
<td>7-12, female</td>
<td>ADHD, written and verbal output disorder, generalized anxiety disorder, disorder of disruptive challenging</td>
<td>Mild periodic limb movements during wakefulness</td>
<td>Tactile*</td>
<td>F</td>
<td>+</td>
<td>&quot;toss infrequently twitched and she occasionally clenched her toes/feet together.&quot;</td>
<td>N/A (no descriptions documented)</td>
</tr>
</tbody>
</table>

---

81
<table>
<thead>
<tr>
<th></th>
<th>ID</th>
<th>Age Range (&lt;6, 7-12, &gt;12), Gender</th>
<th>Confirmed NDC &amp;/or Psychiatric Diagnoses</th>
<th>Sleep Problems in addition to WED &amp; insomnia</th>
<th>Sensory Processing Abnormal-ities as reported by Parent</th>
<th>Formal (F1/I2) SCIT</th>
<th>SCIT (+/-)</th>
<th>Behavioural Observation Quotes</th>
<th>Sensorimotor Sensations Quotes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>behaviour, motor disorder, non-verbal learning disability</td>
<td>I</td>
<td>&quot;sat in the chair with her feet not completely flat on the ground and with only her toes touching the floor and her legs bent slightly back beneath her seat&quot;; &quot;periodically shook her legs; however, Amanda sat for the majority of the assessment quite still and in a position with high tension in her hands kept in her lap as she played with her fingers&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>21</td>
<td>&gt;12, female</td>
<td>Alcohol-Related Neurodevelopmental Disorder, mild conductive hearing loss</td>
<td>Sleep-disordered breathing</td>
<td>Tactile*</td>
<td>F +</td>
<td>&quot;rub feet together while trying to keep them still&quot;</td>
<td>&quot;she feels she can’t sit for long, and if she would sit for a longer time, then she gets pins and needles&quot;; &quot;his whole legs feel funny, and that sensation comes up until above her knees&quot;</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>I +</td>
<td>&quot;Increasing the tension in her legs to sit still&quot;; &quot;reports that she will &quot;accidentally kick people in front of her&quot; while sitting at school.&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>22</td>
<td>&lt;6, male</td>
<td>Autism spectrum disorder (non-verbal)</td>
<td>Circadian sleep rhythm disorder, sleep disordered breathing</td>
<td>Tactile*, Oral</td>
<td>I +</td>
<td>&quot;stretching his legs out and tip-toeing to increase tension&quot;; &quot;sat on the edge of a table with pressure on one foot (bended and pressed against the end of the table); &quot;his other leg swinging and dangling below; &quot;He needed constant tension throughout his body and especially in his legs&quot;; &quot;observed his toes to be flexed upwards when not on the floor&quot;</td>
<td>&quot;he feels pain when his feet are flattened&quot;</td>
<td></td>
<td></td>
</tr>
<tr>
<td>23</td>
<td>&lt;6, male</td>
<td>Autism spectrum disorder (non-verbal)</td>
<td>Tactile*</td>
<td>I +</td>
<td>&quot;flex and twist his legs&quot;; &quot;crossing and uncrossing his legs/feet while standing&quot;; &quot;squeezing and</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range (&lt;6, 7-12, &gt;12), Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F1/ Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
<td></td>
</tr>
<tr>
<td>----</td>
<td>---------------------------------</td>
<td>------------------------------------------</td>
<td>-------------------------------------------</td>
<td>-------------------------------------------------</td>
<td>-------------------------------</td>
<td>-----------</td>
<td>-------------------------------</td>
<td>---------------------------------</td>
<td></td>
</tr>
<tr>
<td>24</td>
<td>&lt;6, female</td>
<td>Alcohol-related Neurodevelopmental disorder (1/2/3/4)</td>
<td>Sleep-disordered breathing, periodic limb movements in sleep, parasomnia (sleep talking and nightmares)</td>
<td>Tactile*, Visual</td>
<td>I</td>
<td>+</td>
<td>&quot;cannot sit for more than 10-15 minutes&quot;; &quot;She is &quot;constantly on-the-go,&quot; and even &quot;needs to stand up and eat at the dinner table,&quot; and her &quot;feet and/or hands are always moving.&quot;; &quot;When sitting, Cecelia preferred to continuously swing her legs back and forth.&quot;; &quot;can't slow her down.&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>25</td>
<td>7-12, female</td>
<td>Trisomy 21</td>
<td>Sleep-disordered breathing, periodic limb movements in sleep</td>
<td>Excluded</td>
<td>I</td>
<td>+</td>
<td>&quot;yogi-like movement patterns (e.g. kicking her leg up onto the wall into an almost split position); while sitting, she would stretch her legs far out in front of her, with her toes flexed upwards; &quot;pull her legs up into a crossed position&quot;; While walking, [she] either shuffled her feet on the carpeted floor or kicked her legs up high. We observed one occasion where she had kicked her leg in an almost split-like position against the wall&quot;; &quot;we did not observe any twitching movements in her feet or toes&quot;; &quot;loves to dance on her tiptoes.&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>ID</td>
<td>Age Range (&lt;6, 7-12, &gt;12), Gender</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F)1/ Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
<td></td>
</tr>
<tr>
<td>----</td>
<td>----------------------------------</td>
<td>------------------------------------------</td>
<td>-------------------------------------------</td>
<td>-------------------------------------------------</td>
<td>---------------------------------</td>
<td>-----------</td>
<td>-----------------------------</td>
<td>-----------------------------</td>
<td></td>
</tr>
<tr>
<td>26</td>
<td>&lt;6, female</td>
<td>Significant prenatal alcohol exposure, suspected FASD (1124)</td>
<td>Possible sleep-disordered breathing</td>
<td>Excluded-maternal depression</td>
<td>F</td>
<td>+</td>
<td>&quot;constantly moved her feet and legs and walked around with her feet curled in&quot; &quot;when playing on the floor, she had her knees bent with her feet to the side of her body&quot; &quot;she would occasionally shift the positions of her legs, sometimes extending one leg with the other bent, or both bent and turned in different directions&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
<tr>
<td>27</td>
<td>7-12, male</td>
<td>Developmental delay, Williams syndrome</td>
<td>Tactile</td>
<td>I</td>
<td>+</td>
<td>&quot;constantly kicking his feet and clapping his hands&quot; &quot;he moved his feet a lot and never sat still&quot; &quot;playing with his feet, and increasing tension by pushing his legs back and forth&quot; &quot;observed him feeling uncomfortable during the assessment, and started arching his neck and back, and coughing&quot; &quot;constantly playing the piano with his toes, and doing complex movements that are not usually present at infants his age&quot;</td>
<td>N/A (no descriptions documented)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>28</td>
<td>&gt;12, female</td>
<td>Dysautonomia, history of concussion</td>
<td>Possible sleep-disordered breathing</td>
<td>Excluded</td>
<td>F</td>
<td>+</td>
<td>when she had her legs uncrossed, a twitch in her toes started (small, uncontrollable, toe, flexion); &quot;witching in her upper leg&quot; &quot;increased tension in her feet, especially arching of her feet&quot; &quot;When she had her legs crossed, we observed minimal leg and feet movement, although she frequently fidgeted with her hands (e.g. tapping her fingers on the</td>
<td>vibrating/twitching feeling in her upper legs and knees and an urge to cross her legs&quot;</td>
<td>N/A (no descriptions documented)</td>
</tr>
<tr>
<td>ID</td>
<td>Age Range</td>
<td>Confirmed NDC &amp;/or Psychiatric Diagnoses</td>
<td>Sleep Problems in addition to WED &amp; insomnia</td>
<td>Sensory Processing Abnormalities as reported by Parent</td>
<td>Formal (F1/ Informal (I)2 SCIT</td>
<td>SCIT (+/-)</td>
<td>Behavioural Observation Quotes</td>
<td>Sensorimotor Sensations Quotes</td>
<td></td>
</tr>
<tr>
<td>----</td>
<td>-----------</td>
<td>----------------------------------------</td>
<td>---------------------------------------------</td>
<td>-----------------------------------------------</td>
<td>---------------------------------</td>
<td>-----------</td>
<td>---------------------------------</td>
<td>-------------------------------</td>
<td></td>
</tr>
<tr>
<td>29</td>
<td>&gt;12, male</td>
<td>Autism spectrum disorder, mucopolysaccharidosis type 3C, developmental delay, visual/hearing impairment</td>
<td>Sleep-disordered breathing, periodic limb movements in sleep/wakefulness</td>
<td>Tactile*</td>
<td>I</td>
<td>+</td>
<td><em>slouched over with his legs crossed at the ankles</em>; <em>kicked and moved his legs around</em>; <em>twitching in his body</em>; <em>occasionally moved his feet from side to side</em>; <em>jerked up suddenly, from the examination bed, and refused to go back down</em>; <em>noticed hand jerking/twitching</em></td>
<td><em>some sort of discomfort in his body</em></td>
<td></td>
</tr>
<tr>
<td>30</td>
<td>&gt;12, male</td>
<td>Autism spectrum disorder, intellectual disability, urea cycle disorder</td>
<td>Periodic limb movements in wakefulness/sleep</td>
<td>Tactile, Visual, Oral</td>
<td>I</td>
<td>+</td>
<td><em>regular toe twitching and feet shaking</em>; <em>frequently touched his ears, lips and side of his face</em>; <em>We believe that although Seroquel® manages his agitation, it increases the discomfort he experiences, causing him to need to be moving continuously.</em>; <em>Throughout the assessment, he was constantly pacing around the room</em>; <em>sat slightly hunched in his chair and continuously shuffled his feet, shook his legs and moved his hands; when his feet were not in motion, they were either crossed at his ankles, positioned with only the side of his feet touching the ground, or with only his toes touching the ground and his feet not completely placed in his shoes.</em></td>
<td>N/A (no descriptions documented)</td>
<td></td>
</tr>
</tbody>
</table>
**Relevance.** In this study, we are describing reported and observed symptoms of children and youth with neurodevelopmental conditions and insomnia most probably due to familial WED/RLS. In order to have a coherent patient population, we confined our study to patients whose mothers had been previously diagnosed with WED/RLS. Challenging sleep/wake behaviours are frequently encountered in individuals with neurodevelopmental conditions, and have typically been attributed to concomitant ADHD (Coles et al., 1997; Walters et al., 2008), melatonin deficiency (Gringras et al., 2012; Jan et al., 2012) or the underlying disorder *per se* (Chen et al., 2012; Jan et al., 2008). WED/RLS and particularly familial WED/RLS, one of the most frequent causes of sleep problems in the general adult population, has not been considered as a possible cause of insomnia in early childhood or paediatric patients with neurodevelopmental conditions until recently (Ipsioglu, Black, et al., 2011; Tilma et al., 2013). This applies even in obvious cases were the mothers were on the WED/RLS spectrum and suffered from insomnia or disturbed restless sleep. We speculate that inadequate awareness of WED/RLS as a paediatric condition and challenges in diagnostic assessment, specifically the restriction due to the essential criteria ‘self-reporting’, have been the principal reasons for the neglected or missed connection (Picchietti et al., 2013; Tilma et al., 2013).

**The Assessment Concept.** We have applied several assessment strategies to overcome this situation and have stratified our behavioural observations in a structured approach. This endeavour started with a basic qualitative exploratory (clinical ethnographic) interview of parents.
The interview was operationalized using narratives co-constructed by the family and health care professionals to ensure a common joint understanding (therapeutic emplotment) (Mattingly, 1994). Then we broadened our assessment strategy of exploration by the implementation of a standardized test not only for the patients, but also for the mothers and accompanying fathers and siblings. As the patient’s subjective experience is needed for a clinical diagnosis in paediatric sleep medicine (Picchietti et al., 2011; Picchietti et al., 2013), both the family and the patient are essential counterparts in any medical investigation, and should be involved in bi-directional communication. For validating our diagnostic understanding of typical signs and symptoms of probable WED/RLS in children with neurodevelopmental conditions, we use a plain language in our reports and ask parents for their review and any edits in order to avoid misunderstandings in our diagnostic approach (Ipsiroglu et al., 2013).

As a common denominator of this cohort, which was heterogeneous in terms of clinical presentations, we selected children whose (birth) mothers had already been diagnosed with WED/RLS and had experienced symptoms themselves as an unambiguous trait of familial occurrence. Using Vancouver Polar-BEARS as a qualitative exploratory interviewing approach (Ipsiroglu, Carey, et al., 2012) and the SCIT as an assessment strategy we found several indicators for WED/RLS, besides intractable insomnia.

Qualitative exploratory interviewing, using a semi-structured combined sleep/wake-behaviour focus, captured quotations by parents, which resembled textbook citations for WED. The following is such a quotation, embedded in the medical report, by a mother of a 5-year-old patient (P#24) with an FASD-diagnosis about restless day-time behaviours due to familial WED/RLS: She is “constantly on-the-go,” and even “needs to stand up and eat at the dinner table.” Her “feet and/or hands are always moving.”
Overall, there were at least two typical characteristic descriptions of significant restlessness, divided in day- and nighttime motor and behavioural presentations compatible with WED/RLS in all 28 children as shown in Table 2.3: (i) Restlessness or inability to rest over the day in any situation associated with rest, and (ii) restlessness at the falling asleep situation contributing to significant bedtime problems and requiring interventions, like melatonin application; in addition all children, except two were described as restless sleepers 26 (93%) of them with kicking movements, described as “even when she is sleeping you can’t slow her down.” (P#24)

The SCIT. In order to investigate the restlessness presentations, the necessity to create a standardized resting test became evident so we adapted the lab-based, long-standing SIT for application in everyday clinical practice (Michaud, 2006). The standardized SCIT provoked a rest situation allowing behavioural observations as a part of the clinical exam, during which typical WED symptoms arise. As an unexpected and fun activity during the assessment, this test positively supported the interaction with the patient and the family. Administering the test to all family members present during the assessment gave information on affected family members, triggered reporting of familial anecdotes and speaking about previously made observations; thus, facilitated the critical development of a shared language with the patient and attending family members. Subjective patient/caregiver descriptions and subjective clinician observations were gathered during the assessment via the SCIT and exchanged with the patients and caregivers. We observed that the SCIT was eye-opening for some parents and supported the acceptance of WED/RLS-related discomfort as a plausible explanation for their child’s challenging/disruptive behaviours. Through the use of the SCIT, we also recognized relief in most patients, even in those with significant intellectual disability. The spotlight, which was typically focused on them as the cause of their challenging/disruptive behaviours, was suddenly taken away and the familial explanatory model was presented as the possible cause of their behaviours. The following original quote is from the report of a patient with a complex chronic condition, including global developmental
delay/intellectual disability, a significant craniofacial condition, autism, significant gastroesophageal reflux, and iron deficiency. The observation opened our professional eyes:

**Formal SCIT:** Ms. [mother] was asked to stand up, shake out and after sitting down give her legs a movement break (to sit with her legs completely still). She found this very difficult and felt an urge to move her legs and create tension in her seating position. Most interestingly, during Ms. [mother]'s assessment, [patient] became interested, watched the assessment and stopped giving moaning sounds when we looked at him; more than that we could see him visibly relaxing, and in consequence he also did not reject the initial phase of the clinical examination.

The narrative based approach in junction with the *familial SCIT* motivated children with neurodevelopmental conditions, similar to their parents and siblings, to describe their symptoms in their genuine own words. 61% of the patients in this cohort had the ability to describe the perceived symptoms in their own wording while performing the SCIT. The self-reported symptoms of the verbal patients matched with parental reports and our observations, and supported the assessment concept for behavioural observations utilizing the formal and in cases the patients could not participate actively the informal test. Therefore, we conclude that in our attempt to understand insomnia of children with neurodevelopmental conditions, who are too young or non-verbal, focusing on behaviours during resting situations at the clinical assessment, and standardizing descriptions and observations in the structured resting situation of the formal or informal SCIT became a foundational part of our otherwise narrative based clinical assessment strategy.

**Unravelling Challenging/Disruptive Behaviours.** In this cohort of children with insomnia all had concomitant evidence of sensory processing abnormalities. Sensory processing abnormalities are a disrupted ability to process, integrate, and respond to internal and external
sensory stimuli (Laverdure-Dupont, Lavigne, Montplaisir, & Soja, 2009; Schaaf & Miller, 2007; Soja, 2007). Since the brain does not filter all sensations during sleep, sensory processing deficits can impact sleep behaviours (Hennevin, Huetz, & Edeline, 2007). Parent reported observations and SCIT based explanations mainly referred to tactile sensitivities, which were observed in 96% (27/28) of the children/adolescents who scored positive for sensory processing abnormalities. Some of the behaviours described within the tactile sensitivity domain are self-harming behaviours (e.g. biting their own hands and arms), suggesting a heightened pain tolerance in these children; therefore, the 19 (68%) confirmed patients might represent an underestimation of the true number with a shifted pain threshold. The SCIT was particularly helpful in connecting sensory processing abnormalities with WED/RLS, as shown by this quotation from the medical assessment report:

(Pat. #3 during informal SCIT):

We observed that Patient 3 sat at the edge of his chair, could not sit still, and constantly moved his legs/feet (e.g. tapping feet up and down, flexing and un-flexing toes, rocking forward on toes and burrowing his toes inside of a shoe). When his feet were on the ground, Patient 3 would put pressure on his toes or heels to increase the tension in his legs or lean back by pressing on his toes.

The following excerpt from a medical report, demonstrates the patients’ wording about the sensations during the SCIT, the parental descriptions of sensory problems, and our behavioural observations.

Pat. #19’s description from the medical report:

When asked to sit in a relaxed manner, patient described that he felt “energy” in his legs and body (he felt that “something” is moving through his legs and “controlling” his body). Parental description: ‘...picks at sores until it bleeds, and won’t let anyone else touch it... He is also upset by loud sounds and has trouble focusing in a noisy environment...’
Clinical observation: We observed him flexing his toes and shifting around, he was unable to keep still ...

While the quotations in this study might not reflect the entire complexity of the child’s sensory profile, we believe that given the diversity of our patient cohort, our study provides significant, preliminary insights into paediatric early onset and chronic WED/RLS, which goes along with sensory processing abnormalities.

**Co-Morbidities and their Link to WED/RLS.** Similar to adults with WED/RLS, children with WED/RLS present with a high occurrence of comorbid psychiatric conditions, including externalizing (e.g. Attention Deficit Hyperactivity Deficit, ADHD; aggression) and internalizing behaviours (e.g. anxiety; depression) (Allen et al., 2005; Allen et al., 2014; Pullen, Wall, Angstman, Munitz, & Kotagal, 2011). ADHD has been mainly in the focus, because of the associations with restless daytime behaviours (Picchietti et al., 2013; Pichietti & Pichietti, 2010; Walters et al., 2008). In our cohort 97% of the patients had a diagnosis of externalizing (43% confirmed; 54% under investigation) disorders such as ADHD, oppositional defiant, conduct, and/or attachment disorder and 50% had a diagnosis internalizing behaviours (25% confirmed; 25% under investigation) such as anxiety and depression. None of the patients had an in-depth sleep assessment prior to establishment of the respective diagnosis. Based on our clinical observations, we speculate that the tendency to use well-known daytime focused explanatory models for diagnosis, as well as the missing diagnostic protocols for WED/RLS-related *behavioural observations*, might be the reason for why WED/RLS-related familial insomnia in children goes un-or under diagnosed.

**Limitations and Strengths.** Our study has several limitations, which need to be taken into account in the planning of subsequent clinical studies and in the development of a standardized assessment protocol for WED/RLS in children with neurodevelopmental conditions. First, this study has a proof of principle character and is based on the understanding that in developmental
paediatrics and child psychiatry, parental agreement to a medical explanatory model is the first step of validation and quality control. Larger cohorts of patients with evidence of familial occurrence of WED/RLS, stratified according to the type and severity of their co-existent neurodevelopmental conditions need to be tested in order to determine the prevalence of familial WED/RLS in children with neurodevelopmental conditions and early onset chronic and intractable insomnia. Second, comparison of the SCIT as a familial assessment strategy, against the established diagnostic WED/RLS criteria and the sleep lab-based SIT is required for validation of this promising clinical tool in verbal patients and their parents. Finally, a systematic investigation of sensory processing abnormalities in patients with proven WED/RLS will elucidate whether and which category of sensory processing abnormalities is part of the WED/RLS spectrum. Until now in paediatric WED/RLS the role of sensory processing abnormalities has not been fully investigated, raising the question whether we all have been focussing just on the tip of the iceberg and focusing on patients who could explain the urge to move in appropriate words. All of these questions might be answered in utilizing a concept of therapeutic emplotment and structured behavioural observations as an addendum. The new insights will result in a better understanding of the pathophysiology and thus, of treatment options for WED/RLS-related sleep/wake behaviour disturbance. The study's strength relies on taking a heterogeneous cohort with neurodevelopmental conditions suffering from major insomnia where causalities have never been investigated from a familial WED/RLS viewpoint. The sleep history and clinical observations are consistent with the adult and paediatric literature showing psychiatric comorbidities, but almost 1/3 of the cohort were not in the situation to explain the experienced sensations – the structured behavioural observations using the formal or non-formal SCIT helped to overcome the gap in our current understanding.

**Summary.** Through the use of qualitative methodology and developing a *behavioural observation* protocol, we could demonstrate that the insomnia experienced by children with
neurodevelopmental conditions is most probably caused by familial WED/RLS. This diagnosis goes along with sensory processing abnormalities, and restless challenging/disruptive sleep and wake behaviours. The presentations occur on a wide spectrum and on a chronic basis, significantly impacting daily functioning and wellbeing. Current diagnostic and therapeudic approaches to children with neurodevelopmental conditions and challenging/disruptive day and nighttime behaviours are mainly daytime focused, and have not included the concept of WED/RLS-related discomfort in our medical explanatory models of disorders with challenging disruptive behaviours (i.e. ADHD, oppositional defiant disorder, conduct disorder, and/or attachment disorder). Therefore, we suggest that restlessness over the day and night, as well as sensory processing abnormalities, may serve as useful clue for behavioural observations indicating that a child should be investigated for WED/RLS.

This research was about structuring daytime-related behavioural observations in patients and accompanying family members in order to be able to start a WED/RLS assessment; the next step was to structure nighttime-related behavioural observations, which will be presented in the next section, below.

2.2.2. “Diagnosis by Behavioural Observation” Home-videosomnography – A Rigorous Ethnographic Approach to Sleep of Children with Neurodevelopmental Conditions

Overview. Advanced video technology is available for sleep-laboratories. However, low-cost equipment for screening in the home setting has not been identified and tested, nor has a methodology for analysis of video recordings been suggested. We investigated different combinations of hardware/software for home-videosomnography and established a process for qualitative and quantitative analysis of home-videosomnography-recordings. In addition, the case vignette described in the full manuscript (home-videosomnography analysis for a 5.5-year-old girl with major insomnia and several co-morbidities) demonstrates how methodological considerations were addressed and how home-videosomnography added value to clinical assessment.

We suggest an ideal set of hardware/software that is reliable, affordable (~$500) and portable (=2.8 kg) to conduct non-invasive home-videosomnography, which allows time-lapse analyzes. The equipment consists of a net-book, a camera with infrared optics, and a video capture device (Sloper & Beresford, 2006). We present a home-videosomnography-analysis protocol consisting of three steps of analysis at varying replay speeds: (a) basic overview and classification at 16× normal speed; (b) second viewing and detailed descriptions at 4–8× normal speed, and (c) viewing, listening, and in-depth descriptions at real-time speed (Jan et al., 2008). We also present a custom software program that facilitates video analysis and note-taking (Annotator©), and Optical Flow software that automatically quantifies movement for internal quality control of the home-videosomnography-recording. The case vignette demonstrates how the home-videosomnography-recordings revealed the dimension of insomnia caused by restless legs syndrome, and illustrated the cascade of symptoms, challenging behaviours, and resulting medications.

The strategy of using home-videosomnography, although requiring validation and reliability testing, opens the door for new “observational sleep medicine,” which has been useful in describing discomfort-related behavioural movement patterns in patients with communication difficulties presenting with challenging/disruptive sleep/wake behaviours.
**Introduction.** Although sleep problems have been reported in the vast majority of children with neurodevelopmental conditions (Jan et al., 2008), their role in chronic disease morbidity is rarely recognized, complicating appropriate diagnosis, and treatment (Jan et al., 2012; Jan et al., 2008; Sloper & Beresford, 2006). A structured and rigorous approach is needed to reduce the significant health economic burden for this population (Newachek, Inkelas, & Kim, 2004). A major reason for this shortcoming is the multifaceted clinical appearance of sleep problems in this population; they may go unrecognized, especially if the parent/caregiver-reported presentation does not match the well-known diagnostic criteria such as sleep apnoea (Jan et al., 2008; Jan et al., 2012). Sleep lab assessments do not reveal the full spectrum of possible causes for an underlying problem. A medical ethnographic approach (Kleinman, 1988; Kleinman & Benson, 2006; Sanker, 1986), using the observational skills of parents and therapists, has enabled us to better understand reasons for unspecified insomnia (Ipsiroglu, Black, et al., 2011; Ipsiroglu et al., 2013) and connect novel observations such as sensory processing abnormalities (Wengel et al., 2011), and WED/RLS (Picchietti et al., 2013), which are particularly missed in standard clinical assessments and treated for their daytime sequelae.

As previously presented WED/RLS is a frequent neurologic disorder, which can significantly impact sleep/wake-behaviours from early infancy (Allen et al., 2014; Tilma et al., 2013), mimic ADHD (Picchietti et al., 2013), and aggravate mental health problems (Ipsiroglu, Berger, et al., 2015). Current diagnostic criteria are based on patient-reported urge to move and sensory discomfort/pain, mainly in the legs, which typically occur during rest and at bedtime. For children, who are non-verbal and/or unable to express these symptoms in their own words, the International Pediatric RLS-Consensus Group recently has suggested “diagnosis by behavioural observation” as part of the diagnostic criteria (Picchietti et al., 2013, p. 1257). However, behavioural observations at bedtime or overnight are particularly challenging, as they are not accessible to observation in the typical clinical working hours.
To facilitate *behavioural observation* in situations that are not amenable to clinical settings, we have developed a home-videosomnography recording system that allows for observations and data collection in the natural setting where sleep occurs (Kloesch & Ipsiroglu, 2014). Videography has been used in clinical work, e.g., observing sleep/wake-behaviours of infants, for more than 35 years (Anders & Sostek, 1976), including time-lapse home-videosomnography-recordings made in the home setting (Anders & Keener, 1985; Baddock, Beckers, Talor, & Bolton, 2004; Mwenge, Uquccioni, & Arnulf, 2013; Ottaviano, Giannotti, & Cortesi, 1991; Sivan, Kornecki, & Schonfeld, 1996). However, there have been no suggestions for the rigorous analysis of time-lapse and/or other video recordings captured by home-videosomnography. Our system operationalizes an ethnographic explorative approach, combining narratives with real-time video recording and allowing observations with analyzes of high-quality motion data.

The goal of this section is to present the methodology of a home-videosomnography system, which we have developed to observe bedtime and sleep in individuals with neurodevelopmental conditions and undiagnosed sleep problems. Our considerations of choice of hard and software for generating, reviewing, and analyzing sleep video recordings will help investigators to customize technical equipment for their individual research settings. A case vignette shows the utility of our home-videosomnography system in screening for WED/RLS associated sleep problems.

**Methodology and Results.** The detailed methodology and results explaining the selection of hard-and bit-and compression software, as well as the quantitative analysis of home-videosomnography, are presented in the manuscript (Ipsiroglu, Hung, et al., 2015).

**Qualitative Analysis of Home-videosomnography Recordings.** A home-videosomnography-recording has to enable: (i) identification of patient behaviours (e.g., movements, patient-generated sound, and awareness), attributes (e.g., positioning of the body and different parts of the body), and interactions. (ii) Description of the general setting, including time
of the day, sleep environment (e.g., room, bedding situation, lighting, ventilation), and agents (e.g., parents, pets). (iii) The analysis methodology should be considerate of the attention span of the video analyzers, which may change over long periods of viewings, and cause major inconsistencies in the analysis. Solution: Originally, notes were taken using Microsoft Excel™ with a time stamp. In order to reduce the viewing time and optimize the time-consuming procedure (which also caused reproducibility problems as stopping, writing down and switching between files became competing tasks), we developed, in collaboration with the Austrian Institute of Technology and the Sleep Lab of the Department of Neurology, Medical University of Vienna, customized analysis software, which allows segments of interest to be marked directly. In addition to the marking capabilities of this software, the Annotator® also allows for viewing and re-viewing of home-videosomnography-recording in real time and at various higher speeds (2–32 times fast forward), flagging, searching for marked clips, archiving, as well as clipping and archiving video clips in patient files (Garn et al., 2013).

**Data Analysis.** The home-videosomnography-recording are reviewed at various levels of qualitative and quantitative analysis to ensure comprehensive and efficient data extraction and, later on, visualization of this data. Each level of review follows an individualized yet coherent methodology that is tied together by the main clinical hypothesis. Manual coding of information (i.e., notes or scoring taken during the analysis) can be stored in Annotator© software. The process of reviewing the video, performing qualitative and quantitative analyses, and visualizing the data currently takes ~3–4 h. The use of the Annotator© software for data processing (viewing, annotating, cutting, archiving) has reduced the time commitment by ~30–40%.

**Qualitative Data Analysis.** Identification of key features, including the patient behaviours, general setting, and sleeping arrangements; key features set the framework for further analyzes and are addressed during repeated viewings. An excerpt of the key features from the home-
videosomnography-recording of the case vignette is presented in Figure 2.1. (i) Behaviours. In addition to movements and attributes (positioning of the body and different parts of the body), the patient’s interactions with parents, tiredness, and anxious facial expression could be viewed and registered. (ii) General setting and sleeping arrangements: The time is shown digitally on the home videosomnography-recording and allows for orientation; the environment, e.g., room, bedding situation, lightening, and interacting partners, is categorized and presented in Figures 2.2-4. Steps of qualitative analysis obtain information at three levels, (i) overview, (ii) detailed, and (iii) in-depth. The single steps are demonstrated using an excerpt depicted in Figure 2.1. (i) Overview. First viewing, basic overview and classification: this first overview explores and describes the content of the home-videosomnography-recording. The video is played at 16x normal speed from the beginning to end to (a) obtain an overview and identify the key features, which may interfere with sleep; (b) understand the gestalt of the behaviours, and (c) identify periods of interest, which will be the focus of subsequent viewings. Minimal notes inform about the periods of observable qualities of sleep (e.g., restless, restful), awakenings, transitioning periods not easily characterized from the recording, and the behaviours during these waking times. This stage of review helps to develop the main hypothesis (e.g., rhythmic movement disorder or confusional arousals) and thus, subsequent review of the video can be done in an efficient way to support the hypothesis.
Figure 2.1. Steps of Analysis. This figure shows the analysis process for completed home-videoosomnography recordings. The annotated notes describe a sample of the case vignette home-videoosomnography-recording. The video clip, presented at three viewing speeds, corresponds to the “Transition 2” section of the annotated notes.
Figure 2.2. Setup Requirements and Domains of Qualitative Analysis. This figure gives a graphical overview of the suggested setting for home-video-somnography-recordings and the targeted key features, general settings, sleeping arrangements and the patient garments.
Figure 2.3. Mind Map of the Patient Factors. This figure is a visual representation of the recommended framework for describing the patient environment and behaviour during the home-videosomnography-recording. The patient’s actions are categorized by types of sleep/wake behaviours and sleep positions.
Figure 2.4. Mind Map of the General Setting. This figure is a visual representation of the recommended framework for describing general setting during the home-videosomnography-recording. The patient's environment is categorized by the external factors that can influence sleep.
After fully reviewing the home-videosomnography-recording, information about total sleep time, sleep efficiency, and estimates of restful- and restless sleep amounts is recorded. The video summary of the case vignette is presented in Table 2.7. (ii) ‘Detailed.’ Second viewing and detailed descriptions: the video recording is reviewed more slowly at 4–8× normal speed for more in-depth explorations. At this stage, more detailed notes of movement patterns and behaviours are taken, and the identified periods of interests are verified, i.e., the falling asleep situations, movements, which lead to possible sleep stage changes (arousals). If there are too many periods of interests, they are categorized depending on the clinical diagnosis (hypothesis), e.g., in WED/RLS, the patterns are analyzed before and immediately after falling asleep, followed by all body-position changes, including how they start and what happens after a body-position change. We use a simplified qualitative analysis of movements derived from the Gross Motor Function Classification System (Palisano et al., 1997).

Table 2.7. Video Summary. This table provides the quantitative summary of the case vignette home-videosomnography-recording. Video summaries provide information about sleep/wake times, sleep onset, and total sleep period, and allow calculation of sleep efficiency.

<table>
<thead>
<tr>
<th>Video Summary</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>LENGTH OF VIDEO/</td>
<td>19:51 – 5:51 (10 HOURS, 0 MIN)</td>
</tr>
<tr>
<td>Total Recording Time</td>
<td></td>
</tr>
<tr>
<td>WENT TO BED</td>
<td>19:51 (already in bed)</td>
</tr>
<tr>
<td>LIGHTS OUT</td>
<td>20:39 (lights fully turned out after already asleep)</td>
</tr>
<tr>
<td>SLEEP ONSET</td>
<td>20:05</td>
</tr>
<tr>
<td>Transition from wake to sleep.</td>
<td></td>
</tr>
<tr>
<td>SLEEP LATENCY</td>
<td>14 MIN (note that patient was already in bed)</td>
</tr>
<tr>
<td>Total time between going to bed and falling asleep.</td>
<td></td>
</tr>
<tr>
<td>WAKE TIME</td>
<td>22:48, 00:13 – 00:16, 03:00 – 03:10, 03:40 – 03:43 (27 MIN)</td>
</tr>
<tr>
<td>Total time in minutes that is scored awake occurring between sleep onset and final wake-up.</td>
<td></td>
</tr>
<tr>
<td>TOTAL SLEEP PERIOD</td>
<td>20:05 – 5:51 (9 HR 46 MIN)</td>
</tr>
<tr>
<td>Period of time measured from sleep onset to final awakening. In addition to total sleep time, it is comprised of the time taken up by arousals and movement time until wake-up.</td>
<td></td>
</tr>
<tr>
<td>TOTAL SLEEP TIME (TST)</td>
<td>9 HR 19 MIN</td>
</tr>
<tr>
<td>Amount of actual sleep time in a sleep period; equal to total sleep period less movement and awake time.</td>
<td></td>
</tr>
<tr>
<td>SLEEP EFFICIENCY (SE)</td>
<td>93.2%</td>
</tr>
<tr>
<td>Proportion of sleep in the period potentially filled by sleep-ratio of total sleep time to time in bed.</td>
<td></td>
</tr>
<tr>
<td>RESTFUL SLEEP</td>
<td>5 HR 7 MIN</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>
In the case vignette, the identified periods of interests included the falling asleep situation, the first twenty minutes after falling asleep, 10 minutes before falling asleep, and around each awakening. Focus was also placed on the patient’s limb movements and body-position changes, as they can fragment sleep. (iii) In-depth. Third viewing, listening and in-depth descriptions: the periods of interests are viewed in real time; in-depth notes are taken and checked for consistency with previous notes. Patient-generated sounds during the points of interest are reviewed in real time, and described in-depth. Listening to and reviewing the patient-generated sounds supports discrimination between inspiratory and expiratory snoring. Review of the case vignette home-videosomnography-recording through the Annotator software, which visualizes sound digitally and supports discrimination of crying and snoring, revealed missing snoring episodes. This more in-depth level of analysis confirms that the developed clinical hypothesis (e.g., viewing a patient with WED/RLS and fragmentation of sleep) has not changed and the initially identified periods of interest were valid for further in-depth analysis.

**Descriptive data summary** is presented as a semi-quantitative home-videosomnography-recording report (Table 2.7). This summary report includes information about the length of the video (went to bed; lights out; sleep onset; wake time; left the bed; came back, falls asleep). Manual coding information (e.g., notes and/or scoring during analysis) is conducted using the Annotator© software, enable a graphical visualization of the sleep as a home-videosomnography-hypnogram (i.e., manual sleep state related coding information on the y-axis and time on the x-axis; Figure 2.5).
Figure 2.5. **Home-videosomnography Hypnogram.** This diagram shows the final hypnogram resulting from analysis and annotation of the case vignette home-videosomnography-recording. Time is displayed on the horizontal axis (total period of one night); sleep state (indicated by the solid black line) is displayed on the vertical axis. Sleep events are labelled according to the embedded legend.
Relevance. For almost 40 years, time-lapse video recordings have inspired physicians; however, technical complexity requiring intensive labour and bulkiness of equipment have prevented their widespread popularity and usage in medical practice (Anders & Sostek, 1976; Sitnick, Goodling-Jones, & Anders, 2008). In paediatric sleep medicine, videos have been used for research and to exclude artefact-related data during sleep and/or detect respiratory events (Sivan et al., 1996). Despite an increasing interest in videosomnography for REM sleep behaviour disorders and epilepsy in adults (Ferini-Strambi, 2012; Mahowald, Cramer Bronemann, & Schenck, 2010; Zucconi & Ferini-Strambi, 2000), their use for sleep/wake-behaviour analyzes and their association with analyzing ADHD and/or RLS presentations in children has been limited (Silverstri et al., 2009). Actigraphy has been suggested as the current gold standard of sleep/wake-rhythm assessments (Sadeh & Acebo, 2002; Sadeh, Haurie, Kripke, & Lavie, 1995). Although actigraphy shows more than 80% agreement with overnight polysomnographic laboratory-based studies (Sadeh et al., 1995), actigraphy has poor agreement for detecting nocturnal awakenings compared to video observations (Sadeh & Acebo, 2002; Sadeh et al., 1995). Actigraphy can miss the clinical significance of major movements such as getting up or jumping on the bed (which are typical movement patterns of children with ASD and WED/RLS) (Ipsiroglu, Hung, Soo, Ho, Barbosa, et al., 2012; Sitnick et al., 2008). Home-videosomnography provides all necessary information, and has the advantage that any marked periods of interest on the hypnogram can be visually re-reviewed and analyzed further.

Cardiorespiratory-video or Level III home PSG's (Morielli, Ladan, Ducharme, & Broilette, 1996; Nixon & Brouillette, 2002) have been trialed in various settings in typically developing children (Moss et al., 2005), and in some children with neurodevelopmental conditions (Veer, Chan, Loock, Matthew, & Ipsiroglu, 2011). However, the frequency of artefacts due to the loss of leads has been considered as a significant challenge, not to mention poor tolerability by children with
challenging/disruptive behaviours and the challenges parents have to face during the data collection process.

We initially developed home-videosomnography to verify caregivers’ narratives about sleep problems in their child’s natural setting with an anthropological ethnographic approach (Ball, Ward-Platt, Heslop, Leech, & Brown, 2006; Hooker, 2001; St James-Roberts, Roberts, Hovish, & Owen, 2015; Teti & Crosby, 2012). We have been able to improve our understanding of the dimensions of sleep fragmentation through generalized body movements. The case vignette demonstrates the dimension of chronic and early onset WED/RLS-related insomnia, negatively impacting developmental coordination, sensory processing, mental health, causing anxiety, and mimicking ADHD-like behaviours. Home-videosomnography-recordings not only enabled us to assess the dimension of the falling asleep and sleep maintenance problems but it also gave us insight in the patient's burden of suffering as demonstrated by her distressed facial expressions, and her help-seeking interactions with her parents. Interestingly, the home-videosomnography-recordings also revealed information on the familial nature of the patient’s and her parents’ RLS presentations. Further, home-videosomnography prompted us to understand the impact of trivial phenomena, e.g., scratching, which is likely not mentioned in most sleep studies. Scratching can be an expression of nocturnal paroxysmal arousals with motor behaviours during sleep (Zucconi & Ferini-Strambi, 2000) or can be associated with RLS-related discomfort as demonstrated, but has never been investigated in a systematic way in the sleep of children.

Complex movements, associated with changes in body position, are proxies for typical neurophysiological arousals (Miano, Parisi, & Villa, 2012), which are detected by full polysomnography including EEG recording. In contrast to neurophysiological information retained by complex sleep lab equipment, the use of a simple home-videosomnography-recording gives basic and limited, but valuable, clinical information. In the context of clinical WD/RLS research, the discrimination of restless and restful sleep at the first viewing level allows for most valuable
information about the degree of restorative or non-restorative sleep. Further detailed and/or in-depth analyses of the movement patterns, mainly counting of body-position changes and how they are initiated, presents more valuable information at the next levels of viewing.

Due to its completely non-invasive character, home-videoesomnography has particular advantages in children with autism or FASDs, who do not tolerate the attachment of any instrumentation on their body due to their sensory processing abnormalities. Home-videoesomnography might also have a value in adult patients (e.g., with dementia or after a stroke), who are unable to express their sensations. Portable and robust hardware and user-friendly video analysis software make our system practical for use in various home settings. We were able to send our equipment across the entire province of British Columbia, which bridged the gap between enormous geographical distances (see Figure 2.6). Our phone and online service to assist parents with installation of the home-videoesomnography system was used in the initial stages; however, after some adjustments, the need for these services has decreased.
Figure 2.6. Map of BC with Video Equipment Destinations in the Last Period of the Year 2013 - 2014. This figure shows the distribution and frequency of video equipment destinations throughout British Columbia, Canada. There were 74 deliveries in total over the last year, 47 of which were within the Greater Vancouver Area and Lower Mainland (distributed by municipality as follows: Coquitlam: 5, Vancouver: 8, Surrey: 9, Burnaby: 6, Abbotsford: 4, New Westminster: 2, Chilliwack: 1, Delta: 3, North Vancouver: 3, Port Coquitlam: 1, Port Moody: 1, South Surrey: 1, Fort Langley: 1, Langley: 1, Maple Ridge: 1). Original map: royalty-free image ©Bruce Jones Design Inc. 2009.
The main problems encountered in our experience have been due to soft and hardware incompatibilities or unintentional changes of netbook default setting. These are problems that can usually be solved by research assistants or sleep technicians through the remote access sync folder without major challenges. Return rate, demolition, and damage of equipment are other factors that have to be taken into account when medical devices are delivered into patients’ homes. As shown in Table 2.8, in our experience these problems were insignificant. Finally, security and confidentiality of data obtained from home settings are an issue that needs to be tackled carefully. Customized consent forms and hospital-based data security support helps overcome this potential hurdle.

### Table 2.8. Overview of Problems. This table shows the history of recording errors with the home-videosomnography system. The main problems with home-videosomnography-recordings were caused by software and/or hardware incompatibilities, or accidental changes of netbook default setting by parents during the set-up.

<table>
<thead>
<tr>
<th>Year</th>
<th>Equipment Setup</th>
<th>Number of Times Equipment was Sent Out</th>
<th>Problems with Video Equipment Type</th>
<th>Count</th>
<th>Team</th>
</tr>
</thead>
<tbody>
<tr>
<td>2010</td>
<td>Changing software</td>
<td>31</td>
<td>Inconsistent frame rates: 'stuttering phenomena' [system/set-up unsuitable for internal quality control]</td>
<td>N/a</td>
<td>One dedicated research assistant, a 'technical enthusiast finding solutions'</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>No setup problems or damages occurred</td>
<td>N/a</td>
<td></td>
</tr>
<tr>
<td>2011</td>
<td>Webcam©</td>
<td>36</td>
<td>Inconsistent frame rates: 'stuttering phenomena' [system/set-up unsuitable for internal quality control]</td>
<td>N/a</td>
<td>Several dedicated research assistants, still high dropout rate because of technical difficulties</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Videos repeated due to technical issues, e.g. software crash</td>
<td>12</td>
<td></td>
</tr>
<tr>
<td>2012</td>
<td>Virtualdub©</td>
<td>45</td>
<td>No setup problems</td>
<td>43</td>
<td>System/set-up suitable for internal quality control with optical flow; no need for 'technical enthusiasts.'</td>
</tr>
<tr>
<td>2013-present</td>
<td>Virtualdub©</td>
<td>56</td>
<td>Netbook crashes while recording</td>
<td>N/a</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Video only records for a few minutes</td>
<td>N/a</td>
<td>System/set-up suitable for internal quality control with optical flow; no need for 'technical enthusiasts.'</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Infrared does not activate</td>
<td>N/a</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Video records onto netbook but file is corrupt/does not play</td>
<td>N/a</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Broken Equipment</td>
<td>7</td>
<td></td>
</tr>
</tbody>
</table>
At its current stage, home-videosomnography serves as a screening tool providing observational information from situations not amenable to routine clinical sleep assessments and supports the collection of in-depth ethnographic information (Ball et al., 2006; Hooker, 2001; St James-Roberts et al., 2015; Teti & Crosby, 2012). Expansion of its utility as a diagnostic and monitoring tool for sleep disturbances, particularly when associated with abnormal body movements, is feasible. The standardization of the technical settings and of the video analysis procedures described here, will allow for validation studies needed to determine its sensitivity and specificity as well as strengths and weaknesses compared to established methods such as polysomnography and actigraphy.

The customized Annotator© software (Garn et al., 2013), which in our experience has reduced home-videosomnography-analysis time from 4–5 h for a trained individual to 2–3h, will enable testing of larger numbers of individuals and accomplish further methodological evaluation such as intra- and inter-rater variability and establishment of movement patterns during sleep in a normal, age-stratified population.

**Summary.** While traditional sleep lab studies inform about neurophysiological, cardio-circulatory, and respiratory surrogate markers, the home-videosomnography allows for explorative clinical description of the symptoms associated with a sleep problem, such as going to bed/falling asleep behaviours, nighttime awakenings, sleep-related movements and the associated family interactions. Advanced digital image processing methodologies will enable semi-automatic detection of featured movement patterns and will support automated analysis (Barbosa et al., 2010; Nakatani et al., 2013; Okado et al., 2008), opening the floor for observational sleep/wake-behaviour research in infants, children, youth and adults with movement disorders, neuromuscular conditions and various behavioural conditions. In clinical practice, home-videosomnography will have its place for phenotyping challenging and disruptive behaviours, which are not easily characterized upon clinical sleep assessments and not captured by traditional lab-based studies.
We also expect that home-videosomnography will particularly be useful in the recognition of so-called restless sleep caused by WED/RLS and periodic limb movements as well as for monitoring of interventions.

This research was about structuring nighttime related ethnographic behavioural observations in patients in order to be able to understand challenging/disruptive sleep and wake behaviours due to WED/RLS. In turn, I assess the impact of unrecognized sleep problems on the medical management of children with neurodevelopmental conditions, which will be presented in the next section.

2.3. Research Question #3: “What are the consequences of unrecognized sleep problems?” Analysis of Overmedication and Polypharmacy Data.

2.3.1. Pathways to Overmedication and Polypharmacy: Case Examples from Adolescents with Fetal Alcohol Spectrum Disorders.


Overview. Due to their challenging/disruptive daytime behaviours, children and youth with fetal alcohol spectrum disorders (FASDs) are at high risk for multiple medication prescriptions. Here we describe how undiagnosed chronic sleep problems paved the pathway for overmedication and polypharmacy in adolescents with FASDs: prescription of multiple, off-label, and concurrent pharmaceutical medications.
We analysed the challenging/disruptive sleep and wake behaviours and medication history of 17 adolescent patients with a pharmacotherapy timeline, capturing: (1) the medications and order of prescriptions, and (2) the age at the time of first prescription. An in-depth case report demonstrates how unrecognized sleep problems led to a diagnosis and medication cascade, impacting the life and development of the patient.

All patients presented with chronic insomnia and fulfilled the diagnostic criteria for familial Willis Ekbom disease (restless legs syndrome). 11/17 had additional clinical signs of sleep disordered breathing, and 14/17 showed excessive daytime behaviours (sleepiness and/or hyperactive-like behaviours to fight fatique/sleepiness).

The medication analysis revealed two patterns in prescription strategies: (a) targeting sleep problems with melatonin, second-generation antipsychotics, and/or combination of both (10/17); and (b) targeting hyperactive-like daytime behaviours with a psychostimulant (7/10). In addition, many medications were prescribed in combination and at alarmingly young ages.

Based on our findings we suggest assessment of sleep before any assessment of challenging/disruptive daytime behaviours and prescription of psychotropic medications. This observation raises the question to what degree children with neurodevelopmental conditions are subject to overmedication due to an unrecognized underlying condition.

**Introduction.** Prescription drugs are one of the most important clinical treatments. With the wide spectrum of pharmacologic therapies currently on the market, it can be difficult to select a drug most likely to benefit a specific patient. This is particularly difficult in children or youth with neurodevelopmental conditions such as FASDs and/or prenatal substance exposure (PSE) diagnosis, due to the complex, heterogeneous and multi-factorial nature of their clinical presentations. FASDs are the most common form of prenatally acquired brain injury and are due to toxic substance exposure – the deleterious effects of alcohol on the developing brain of the foetus.
(Clarren & Smith, 1978; Jones & Smith, 1973). The Public Health Agency of Canada (2005) estimates that FASDs affect one percent of the Canadian population. Depending on methodology and magnitude of studies, prevalence rates will change; in the United States, 0.2-1.5 out of every 1000 live births are affected with an FASD (May et al., 2009). FASDs encompass a spectrum of diagnoses ranging from fetal alcohol syndrome, the severest form among FASDs, including growth deficiency or facial characteristics; to partial fetal alcohol syndrome and alcohol-related neurodevelopmental disorder, characterized by a range of neurological and behavioural impairments; and alcohol-related birth defects, which refers to defects in other organs, skeletal abnormalities and vision and hearing problems (Chudley et al., 2005; Public Health Agency of Canada, 2005; Rasmussen et al., 2008).

In this section, we present our results taken from our collaborative clinical and pharmaceutical work to demonstrate how chronic sleep problems in children and youth with complex neurodevelopmental conditions such as an FASD can become the gateway to treatments with multiple, off-label, and concurrent pharmaceutical medications over time. We refer to this dangerous practice as poly-pharmacy. We suggest that in children the use of medications such as antipsychotics and stimulants to treat challenging/disruptive daytime behaviours can be reduced or even eliminated after treating underlying sleep disorders. Our aim is to highlight how an alternative intervention approach can be used to stop the trend towards overmedication of children, which leads to poly-pharmacy at adolescent age. In other words, our aim is to lessen the need for pharmacological interventions in patients suffering from neurodevelopmental conditions and comorbid behavioural presentations such as conduct disorder or attention deficit hyperactivity disorder (ADHD). We do not suggest that pharmaceutical treatment is unnecessary; on the contrary, many individuals with neurodevelopmental conditions will benefit better from medication after identifying and targeting underlying sleep problems.
In this article, we analyze prescription practices in adolescents with an FASD diagnosis, as this population is prone to multiple prescription medications (Chudley et al., 2005; Paley & O’Connor, 2009; O’Malley & Nanson, 2002). Our findings suggest that tackling sleep problems as a primary treatment target starting at a young age, reduces the trend to overmedication and polypharmacy and improves outcomes in adolescents with neurodevelopmental conditions and challenging day and night time behaviours. The results from our case series analysis represent a first step towards practice-based evidence. We believe that our finding and recommendations can be applied within the broader context of children and youth who receive treatment with psychotropic medications for a variety of mental health conditions.

Sleep Problems in Children with Neurodevelopmental Conditions. The importance of sleep is well documented in regards to cognition, emotional and physical resilience, and wellbeing or quality of life. In adults, addressing sleep problems has a long tradition originating in ancient cultures (Brunt & Steger, 2008). Sleep problems have been recognized as a key symptom of neurodevelopmental conditions (Bruni & Novelli, 2010; Wiggs & Stores, 1996, 2001; Zucconi & Bruni, 2001). However, in spite of their documented negative impacts on psychomotor development and cognitive and behavioural functioning (Jan et al., 2012); sleep problems are not systematically integrated into current assessment and treatment practice. Lack of training and assessment tools, as well as a siloed sub-specialist approach, hinders the recognition of sleep problems as a significant contributor to illness and reduced quality of life (Ipsiroglu et al., 2013; Jan et al., 2008). Furthermore, the cause of an underlying sleep problem is difficult to identify as nighttime behaviours are not accessible to direct clinical observation outside a sleep lab or hospital based environment.

In our clinical work, we have observed a high prevalence of Willis Ekbom disease/restless legs syndrome presentations in children and adolescents with an FASD and/or PSE diagnosis.
Ipsiroglu et al., 2013). WED/RLS is an example of a treatable neurologic disorder that impacts sleep and is under-diagnosed in children. WED/RLS is prevalent in 5-10% of the general population, and is associated with major sleep problems in 2-3% in adults (Allen et al., 2005). Prevalence rates of WED/RLS that impacts sleep are similar in children (Pichietti et al., 2007). The challenge is that WED/RLS diagnosis is based on the patient’s subjective expression of discomfort (Picchietti et al., 2011), and there are currently no diagnostic protocols allowing for recognition of WED/RLS in children who are unable to express themselves (Picchietti et al., 2013). As WED/RLS is associated with both night- and daytime restlessness, it is often misdiagnosed as ADHD (Picchietti et al., 2013; Walters et al., 2008). Treatment with typical ADHD medications (psychostimulants) often results in behavioural or other adverse effects, perpetuating the challenging sleep and daytime behaviours. Accurate diagnosis of WED/RLS is of clinical relevance as it allows for disease-specific treatment options such as iron supplementation, levodopa and gabapentin (Magnus, 1999; Picchietti et al., 2013; Robinson & Malow, 2012).

Sleep problems are a key symptom of individuals with FASDs (Steinhausen & Spohr, 1998; Streissguth et al., 1996) but, due to overriding behavioural comorbidities, they often remain undiagnosed (Ipsiroglu et al., 2013; Jan et al., 2012). Thus, knowledge about the phenotype of fetal alcohol spectrum disorder-related sleep problems is limited. In addition to our work (Ipsiroglu et al., 2013), the association between sleep problems and daytime behaviours has been presented in only two other studies: 1) Chen et al. (2012) showed significant sleep problems in a population with FASD referred for behavioural intervention, and 2) Wengel et al. (2011) demonstrated that unidentified sensory processing abnormalities contribute to behavioural problems and insomnia (falling asleep, sleep maintenance problems) in patients with FASDs.

**Fetal Alcohol Spectrum Disorders, Overmedication and Polypharmacy.** Children with an FASD represent one of the most vulnerable populations in terms of psychotropi
prescription. Their birth mothers, and even foster parents, face social stigma associated with the condition because it is the result of potentially preventable toxic alcohol exposure during pregnancy (Hill, Hegemier, & Tennyson, 1989; Oldani, 2009; Ryan & Ferguson, 2006). The social stigma might lead caregivers to seek out medications and physicians to prescribe them for attenuating the impact that FASDs have on cognitive functioning and behaviour (Oldani, 2009). The functioning and behaviours of children with a FASD typically resembles ADHD; however, research has shown that they are only partly responsive to pharmacologic treatments aimed at addressing ADHD symptoms (Coles, 2001; Rasmussen et al., 2008). Literature suggests that in complex clinical presentations, multiple medications should be trialed in order to find the right treatment (O’Malley K., 1997; O’Malley, Koplin, & Dohner, 2000; O’Malley & Nanson, 2002; O’Malley & Streissguth, 2003; Wilens, Biederman, & Spencer, 1996). At our follow-up clinic, observation has shown a high prevalence of behavioural adverse drug reactions upon treatment of ADHD-like behaviours with stimulants and antipsychotics, leading to a cascade of additional diagnoses (interpreted as FASD comorbidities) and treatments, and eventual break down in family situations resulting in placements as depicted by the case vignette presented in the introduction (section 1.2.3).

**Medication History in 17 Children with a Fetal Alcohol Spectrum Disorder and Sleep Problems.** We retrospectively analysed medical records to capture the medication history of patients with FASDs/ PSE who had been assessed for their challenging/disruptive sleep/wake behaviours at our clinic. We were mainly interested in (1) the drugs used and the order in which they were prescribed, and (2) the age of the patient at the time of their first prescription.

During the time period between 2010 and 2014, 20 paediatric patients with a FASD or prenatal substance exposure diagnosis above the age of 10 had been referred to our clinic by community paediatricians for assessment of their sleep problems. In 17 patients (age range: 10-17 years, median age: 13 years) we created a pharmacotherapy timeline (see Figure 1.1 for an example). Patients
included in our analysis had the following sleep/wake behaviours and neurodevelopmental conditions:

- All 17 patients fulfilled the diagnostic criteria for WED/RLS and/or even familial WED/RLS and showed evidence for disordered circadian rhythm sleep disorders, caused by insomnia and a non-restorative sleep quality.
- 17 (100%) patients had falling asleep problems; 10 patients (59%) had sleep maintenance problems, and 1 had early morning awakenings.
- 11 patients (65%) also had clinical evidence of sleep-disordered breathing and 5 (29%) had regular washroom awakenings over the night.
- 14 (82%) patients showed excessive daytime behaviours (sleepiness and/or hyperactive-like behaviours to fight fatigue/sleepiness) and 8 (47%) were increasingly fidgety towards the evening and/or at bedtime.
- 16 patients (94%) also presented with one or more coexisting neurodevelopmental conditions, such as autism spectrum disorder (n=2; 12%), attention deficit hyperactivity disorder (n=15; 88%), or mental health disorders, such as anxiety disorders (n=11; 65%), oppositional defiant disorder (n=7; 41%), or mood disorders (n=6; 35%).

The medication history of patients in our sample included the following highlights (see Tables 2.9 and 2.10 for further details):

- In total, 120 medication trials were prescribed across the 17 patients, most of them in sequential order over a period of time. There was only one medication-naïve patient who was assessed for sleep/wake-problems (at the age of 13).
- The maximum number of medication trials prescribed to a patient over the time was 21 (this patient was 14 years old at the time of our assessment and was diagnosed with an Fetal Alcohol Spectrum Disorder and PSE, mood disorder; moderate to severe ADHD, and anxiety.
- All patients, except the medication-naïve patient, were treated with medications for their sleep problems (94%; in total, 39 medication trials with prescription medications or over the counter sleep medications). Sleep medications topped the prescription list when we further analyzed median and max number of medications prescribed or bought over-the-counter.

- ADHD medications were most commonly prescribed for daytime behaviours (76%; in total 29 prescription trials) followed by first- and second-generation antipsychotics (59%; in total 20 prescription trials), selective serotonin reuptake inhibitors (47%; in total 12 prescription trials), and anticonvulsants (24%; in total 8 prescription trials) for challenging/disruptive daytime behaviours.

- Over a third of patients were also treated with prescription and over-the-counter medications categorized as others (including: iron supplements, lorazepam, chlorpromazine, propranolol, lithium, imipramine, docusate, and gabapentin (35%; in total 12 prescriptions). For example, 7 (41%) patients received over-the-counter melatonin for their underlying sleep problems as their first medication, either by the recommendation of their physician or by their own discretion. The youngest patient was 3 years old when melatonin was first given.

- 7 (41%) patients were prescribed their first medication at age 5 years or younger.

- 7 (41%) patients were first prescribed psychostimulants and ADHD medications to target hyperactive-like daytime presentations. The youngest patient was 2.5 years of age when methylphenidate was first prescribed; this patient did not tolerate methylphenidate, and after one week the medication was changed to dextroamphetamine, which was also not tolerated either and had to be stopped.
- 2 patients (12%) received second-generation antipsychotics, prescribed for sleep problems, as the first medication, the youngest patient was 5 years old at the time of the first prescription.
- 1 patient (6%) received a selective serotonin reuptake inhibitor (SSRI) as the first medication at the age of 5 years.

Table 2.9. Patient Demographics and Day/Nighttime Clinical Presentation in Paediatric Patients with FASDs and/or PSE. This table gives a detailed overview of the sleep/wake-behaviour assessment results, the neurodevelopmental conditions and co-morbidities.

<table>
<thead>
<tr>
<th>Patients with an FASD/PSE Diagnosis &amp; Associated Medical Conditions</th>
<th>Diagnoses</th>
<th>Percentage of Patients (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sleep disorders</strong></td>
<td>Excessive daytime behaviours (sleepiness and/or hyperactive-like behaviours to fight fatigue/sleepiness)</td>
<td>82</td>
</tr>
<tr>
<td></td>
<td>Circadian rhythm sleep disorder</td>
<td>100^1</td>
</tr>
<tr>
<td></td>
<td>Sleep disordered breathing</td>
<td>65^2</td>
</tr>
<tr>
<td></td>
<td>Periodic limb movements</td>
<td>53</td>
</tr>
<tr>
<td></td>
<td>Insomnia</td>
<td>100</td>
</tr>
<tr>
<td></td>
<td>Parasomnias</td>
<td>76</td>
</tr>
<tr>
<td><strong>Neurodevelopmental conditions</strong></td>
<td>Attention deficit hyperactivity disorder</td>
<td>88</td>
</tr>
<tr>
<td></td>
<td>Autism spectrum disorder</td>
<td>12^3</td>
</tr>
<tr>
<td><strong>Mental health or psychiatric co-morbidities</strong></td>
<td>Anxiety disorder</td>
<td>65</td>
</tr>
<tr>
<td></td>
<td>Oppositional defiant disorder</td>
<td>41</td>
</tr>
<tr>
<td></td>
<td>Mood disorder</td>
<td>35</td>
</tr>
<tr>
<td></td>
<td>Reactive attachment disorder</td>
<td>35</td>
</tr>
<tr>
<td></td>
<td>Obsessive compulsive disorder</td>
<td>11</td>
</tr>
<tr>
<td><strong>Neurologic co-morbidities</strong></td>
<td>Willis Ekbom disease /restless legs syndrome</td>
<td>100^4</td>
</tr>
<tr>
<td></td>
<td>Sensory processing abnormalities</td>
<td>94^5</td>
</tr>
<tr>
<td><strong>Summary</strong></td>
<td>All patients presented FASDs and/or PSE, as well as sleep disorders. 94% were diagnosed with other neurodevelopmental conditions besides FASDs and PSE, the most common being attention deficit hyperactivity disorder (ADHD, 88%). There was also a high incidence of psychiatric disorders such as anxiety disorder (65%) and oppositional defiant disorder (ODD, 41%).</td>
<td></td>
</tr>
</tbody>
</table>

(1) 14/17 patients had confirmed CRSD; 3/17 patients had highly suspected CRSD  
(2) 8/11 patients had confirmed SDB; 3/11 patients had suspected SDB  
(3) 2/17 patients had suspected ASD due to their clinical presentation, but the diagnosis was not confirmed after formal assessment  
(4) 11/17 patients had confirmed WED; 6/17 patients had suspected WED  
(5) 16/17 patients had confirmed SPAs; 1/17 patients had suspected SPAs
Table 2.10. Overview of the Prescription Medications, Including: Most Common Medications and First Prescription Medication Given to Patients with an FASD and/or PSE diagnosis.

<table>
<thead>
<tr>
<th>Medication Categories</th>
<th>All Medications</th>
<th>Sleep Medication (clonazepam, clonidine, chlorpromazine, dimenhydrinate, diphenhydramine, lorazepam, melatonin, quetiapine, tramadol)</th>
<th>ADHD Medication (amphetamine, methylphenidate (Concerta®, Ritalin®), dextroamphetamine, ginsect, hydroxybutyric acid, mixed amphetamine salts,</th>
<th>Second-generation Antipsychotics (daytime) (aripiprazole, olanzapine, quetiapine, risperidone)</th>
<th>SSRI (citalopram, fluoxetine, paroxetine, sertraline)</th>
<th>Anti-convulsants (lamotrigine, topiramate, sodium valproate, valproic acid)</th>
<th>Other (docusate, gabapentin, imipramine, iron, lithium, propranolol)</th>
</tr>
</thead>
<tbody>
<tr>
<td>N=17</td>
<td>Mean Age: 13y</td>
<td>Median Age: 13y</td>
<td>Range: 10-17y</td>
<td>% Patients Prescribed Medication</td>
<td>94%</td>
<td>94%</td>
<td>76%</td>
</tr>
<tr>
<td>Number of Medications per Patient (Mean/Median)</td>
<td>7.1/6</td>
<td>2.5/2</td>
<td>1.71/1</td>
<td>1.18/1</td>
<td>0.70/1</td>
<td>0.47/0</td>
<td>0.47/0</td>
</tr>
<tr>
<td>Max/Min Number of Medication Prescribed per Patient per Drug Class</td>
<td>21/0</td>
<td>5/0</td>
<td>5/0</td>
<td>4/0</td>
<td>2/0</td>
<td>4/0</td>
<td>2/0</td>
</tr>
<tr>
<td>Most Commonly Prescribed Medication per Drug Class</td>
<td>melatonin</td>
<td>melatonin</td>
<td>Concerta®, dextroamphetamine</td>
<td>risperidone</td>
<td>Flusoxetine</td>
<td>lamotrigine, topiramate, sodium valproate, valproic acid</td>
<td>iron</td>
</tr>
<tr>
<td>% Patients Prescribed Most Common Medication per Drug Class</td>
<td>35%</td>
<td>35%</td>
<td>41%</td>
<td>40%</td>
<td>50%</td>
<td>75%</td>
<td>38%</td>
</tr>
<tr>
<td>% First Medication Prescribed</td>
<td>N/A</td>
<td>41%</td>
<td>41%</td>
<td>0%5</td>
<td>6%6</td>
<td>0%</td>
<td>0%</td>
</tr>
<tr>
<td>Mean Age at First Prescription Overall</td>
<td>6.8y</td>
<td>7.9y</td>
<td>7.8y</td>
<td>6y</td>
<td>5y</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Youngest Age at First Prescription Overall</td>
<td>2.5y</td>
<td>3y</td>
<td>2.5y</td>
<td>5y</td>
<td>5y</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Age at First Prescription Per Drug Class (Mean/Median)</td>
<td>N/A</td>
<td>8.4/7y</td>
<td>7.5/7</td>
<td>9.6/8y</td>
<td>9/9y</td>
<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

Summary
Sleep medications were the most commonly prescribed drugs of this cohort, and made up the majority of the first prescribed medications (94%). ADHD medications were shown to be prescribed at the youngest age overall (2.5y), while SSRIs were prescribed at the youngest mean age at first prescription (5y).

(1) This medication is regarded as an antipsychotic; however, chlorpromazine was used in 2 patients at low dosages for targeting sleep problems.
Relevance. Internationally, there is increasing concern regarding the prescription of psychotropic drugs to children. In 2012, Canada had the highest prescription rates for psychostimulants and antipsychotic medications for children in a multijurisdictional comparative study (Zhang et al., 2013). Complementing the recommendations of the Canadian Alliance for Monitoring Effectiveness and Safety of Antipsychotics in Children (CAMESA) and the position statement released by the Canadian Paediatric Society, there are applicable prescription guidelines for children and adolescents (Pringsheim, Doja, Belanger, Patten, & the CAMESA guideline group, 2011; Pringsheim, Panagiotopoulos, Davidson, Ho, & the CAMESA guideline group, 2011). These guidelines emphasize the importance of initial evaluation and ongoing monitoring, limiting concurrent prescriptions, careful selection of first-line interventions, and adherence to evidence-based applications. Why haven’t these guidelines been applied? Why are there still such high prescription rates?

Children with neurodevelopmental conditions and concurrent sleep problems are at particularly high likelihood for psychotropic drug prescriptions because of the intertwined relationship between their challenging day- and nighttime behaviours. As we were analyzing the medical histories of patients referred to our sleep/wake-behaviour clinic, it was evident that sleep problems were a prominent concern in all cases. Prior to referral, patient sleep problems (mainly insomnia) had already been targeted with melatonin, which topped the list of medications.
Melatonin, clonidine, clonazepam, antihistamines, quetiapine, and trazodone all addressed insomnia but insufficiently, accounting for the number of different medication trials; thus, medications targeting sleep led the prescription list when we further analyzed median and maximum number of medications bought over-the-counter or prescribed. Medications targeting ADHD had the first ranking among the medications prescribed for daytime behaviours, making up more than three-fourths of the patient prescriptions, followed by the antipsychotics with almost two-thirds of the patient prescriptions. Second-generation antipsychotics were the most common antipsychotic class prescribed for the patient cohort. Only two patients were taking the first-generation antipsychotic chlorpromazine. Approximately half of the patients were taking SSRIs.

In our recent work, which was based on parent and health care professional descriptions and analyzes of 27 patient histories in children and youth, we conjectured that sleep problems reported by parents were probably taken as given during clinical assessments and due to missing assessment tools, lack of training and insufficient infrastructure, not followed up further (Ipsiroglu et al., 2013). In addition, daytime-focused medication cascades did not improve the sleep problem and may have even aggravated the underlying condition. Therefore, the sequelae of medication cascades were a main reason for why the patients were eventually referred to our clinic after an odyssey of assessments and treatment trials, like the presented case vignette. The in-depth analysis of prescription strategies in the presented 17 patient histories of adolescents (defined here as children older than 10 years) and the case vignette demonstrate the dimension of this sleep perspective-based conclusion for the first time. As shown, diagnosis and treatment cascades can be triggered if sleep problems are missed in the assessment of wake behaviours and in consequence, iatrogenic harm can occur. WED/RLS and WED/RLS-like presentations deserve special attention as they manifest with both restless daytime behaviours mimicking ADHD, and with restless night and sleep behaviours mimicking behavioural insomnia; together they resemble challenging/disruptive
(externalizing) behaviours, which justify prescription strategies exceeding current best practice guidelines (Canadian Attention Deficit Hyperactivity Resource Alliance (CADDRA), 2011).

Diagnosis of WED/RLS is based on subjective symptoms reported by the patient and, in the absence of a disease specific biomarker, diagnosis of WED/RLS is challenging, particularly in children. Sensory discomfort and a shifted pain threshold, which is mostly associated with the typical motor hyperactivity, exacerbate abnormal behaviours. We hypothesize that there are two likely causes for difficulties with expressing subjective suffering in children. One is simply that they are too young or do not have the language capacity. The second one is that sensory processing abnormalities start at early age and a reference point, which enables for appropriate realization and description of sensations, is missing. In fact, 82% of the patients from the case series had sensory processing abnormalities. The Wengel study (2011) supports our explanation, and suggests for a more in-depth explorative and observational assessment strategy as recommended by the International WED Study Group (Picchietti et al., 2013). The recently published description of early onset familial WED/RLS (Allen et al., 2014; Tilma et al., 2013), which also supports the need for more in-depth explorative and observational assessment strategy and additional criteria such as assessment of parents and other family members before starting to focus exclusively on daytime behaviours. In our assessments, we have been using (in cases where it was possible) positive family history and signs of involuntary movements in a clinically adapted version of the SIT in the child and the affected parent to establish the diagnosis (Michaud, 2006).

As demonstrated in our case vignette, patients with WED can have severe adverse reactions to psychostimulants and/or antipsychotics. Based on our observations from this case series, treating neurological adverse effects, presenting as challenging/disruptive behaviours, with additional stimulants and/or antipsychotics triggers new symptoms and act as a Catch-22, leading to further deterioration. After treating WED with gabapentin, the patient’s clinical presentation and life trajectory changed. Both her sleep problems and excessive daytime behaviours, including
ADHD-like presentation and anxiety improved. While her anxiety, as well as her need for one-to-one supervision, had been exclusively associated with prenatal alcohol related injury, the observed behavioural and mental health changes were perceived as very surprising for the entire care team, who were convinced that her behaviours were caused by prenatal alcohol exposure and its impacts on the developing brain. This transformation in clinical presentation highlights to what degree WED had aggravated, and even possibly caused, the mental health co-morbidities, and that treatment with sleep helped her brain to recover to a novel degree perceived by the care team as unexpected.

Prescription Strategies and Challenges. The analysis of the 17 patients’ first medications reveals two patterns in prescription strategies: (a) Targeting sleep problems with melatonin, second-generation antipsychotics, and/or combination of both (10/17; 59%); (b) Targeting hyperactive like daytime behaviours with a psychostimulant (7/17; 41%).

The Challenge with Melatonin. Melatonin was trialed in all patients, excluding the one medication-naïve patient, and was partly efficient in 14/17 (82%) cases for sleep initiation, but not for sleep maintenance. Since 2004, melatonin has been sold in Canada as a licensed natural health product, with some review of safety and efficacy but is a prescription medication in most European countries. It is available over-the-counter in any drug store in Canada as a licensed natural health product, but from a clinical point of view has to be considered as unregulated. In this case series, melatonin dosages of up to 30 mg/day were applied and in some cases, medication started already at very young age (3 years). Melatonin is a zeitgeber (an external cue to entrain circadian rhythms), but not a sedative. If given unselectively, aspects of the individual patient’s sleep problems such as obstructive sleep apnoea/hypopnea, sensory discomfort in WED, periodic limb movements during sleep and other potential reasons for sleep problems such as pain, hunger, gastrointestinal discomfort (in children with night time G-tube feeding) may not be tackled and time to find the
right treatment is lost. This might be the reason for contradicting literature, which praises and criticizes the effects of melatonin from various viewpoints (Buscemi & Witmans, 2006; Gringras et al., 2012; Jan et al., 2008; Jan et al., 2012). Our data shows that in the absence of a sleep/wake behaviour assessment and follow ups, melatonin is a *starter drug* in children with neurodevelopmental conditions e.g., melatonin was first used in 41% percent of the 17 cases reported here) and sleep problems, and significantly contributes to the medication cascade.

**The Challenge with Psychostimulants / ADHD Medications.** Children with FASDs are diagnosed with ADHD six times more frequently than those without an FASD diagnosis (Rasmussen et al., 2008; Streissguth et al., 1996). Despite indications that the complexity of behavioural problems in this population cannot be explained by an ADHD-related pathophysiology alone (Coles, 2001; LaFrance et al., 2014), and despite variable responses to medications (e.g., lack of therapeutic response and/or adverse drug reactions) (Doig, McLennan, & Gibbard, 2008; O’Malley & Nanson, 2002; Rowles & Findling, 2010), ADHD-like symptoms are a frequent target for pharmacological interventions with psychostimulants. Indeed, 88% of our cohort presented with the co-morbid condition ADHD. One patient from our cohort is a boy who was diagnosed with ADHD at age 2.5 years and was immediately trialed with two stimulants (methylphenidate and dextroamphetamine, each for approximately one week), with major behavioural adverse drug reactions. This case and another case seen at our clinic, where more than two psychostimulants had been prescribed at a time, demonstrate how desperate the parents and the prescribing physician were to find a solution for challenging/disruptive hyperactive behaviours (at that time a sleep clinic investigating sleep/wake behaviours had not yet been established in BC). Guidelines for the prescribing of drugs for ADHD suggest they not be administered until the patient is at least 4 years of age (Aagaard & Hansen, 2011; CADDRA, 2011; Centers for Disease Control and Prevention, 2013; Sub. Attention-Deficit, 2011). Furthermore, our data reveals that 41% of the cohort was prescribed stimulants as their first medication, and that 38% were prescribed stimulants before the age of 6 years. Despite
the fact that there are insufficient long-term studies and a lack of information on the effects of combinations of ADHD medications, the American Academy of Pediatrics expanded the age ranges for ADHD diagnosis to children as young as 4 years. (Sub. Attention-Deficit, 2011) As a consequence, we may see treatment with psychopharmacology at even earlier ages to become more commonplace in the near future. Stepping back and observing from the lens of a medical anthropologist, (Oldani, 2009) we realize that prescription of ADHD medications, despite the lack of confidence in positive medication effects, became a strategy of action for relieving all involved parties.

By analyzing the ADHD prescription strategies, we can conclude that current prescription practices for children with an FASD and/or PSE diagnosis are complicated by complex clinical presentations, which overwhelm clinicians. As non-restorative sleep quality typically results in difficulties in waking up and leads to excessive daytime behaviours (sleepiness and/or hyperactive like behaviours to fight fatigue/sleepiness) most likely the high number of ADHD diagnoses (82% of all adolescents) were/are not correct. Therefore, a re-evaluation of all the ADHD diagnosis becomes necessary. This hypothesis is further supported by the fact that none of the WED and/or Willis Ekbom-like presentations (which also resemble ADHD) had been diagnosed before the ADHD diagnosis. Figure 2.7 give an overview of our understanding of how sleep problems affect day-/nighttime behaviours presentations.
The Challenge with Second-generation Antipsychotics. As described above, clinical circumstances where there has been little or no therapeutic success, often lead to deviations from guidelines and/or common prescription practices such as prescribing atypical antipsychotics to children younger than 6 years, which is generally not recommended, though recent studies show that the rate of prescribing in children has been growing at a substantial rate (Findling et al., 2011; Ronsley et al., 2013; Therapeutics Initiative, 2009). In our population, almost 60% had been prescribed a second-generation antipsychotic at some time over their life trajectory, and almost half of the patients were prescribed an SSRI. These drugs were used in children as young as 6 and 5 years, respectively. In addition to ADHD, our cohort presented with a high co-morbidity of psychiatric disorders, such as anxiety disorder (65%) or oppositional defiant disorder (41%), also a finding that has been consistently noted in the literature (Hellemans, Sliwowska, Verma, & Weinberg, 2010; O’Connor & Paley, 2009). However, the case vignette shows that anxiety, which prevented our patient from being able to sleep alone, was caused by sleep deprivation triggering hypnagogic hallucinations. Hypnagogic hallucinations belong to the sleep disorder category of
parasomnias and, like all other parasomnias, are triggered by sleep deprivation, medications, and irregular sleep schedules. Treatment of WED enabled the patient to be able to fall asleep and achieve a *good night sleep* (restorative sleep), resulting in cessation of the hypnagogic hallucinations and over time, her anxiety also disappeared.

Adverse drug reactions from second-generation antipsychotics, such as the ones described in our case vignette, have been well documented. Moreover, increasing numbers of children may be receiving antipsychotic or antidepressant medications for reasons not supported by scientific evidence, many without any licensed approval (Findling et al., 2011; O’Connor & Paley, 2009) or off-evidence use (Elbe, McGlanaghy, & Oberlander, 2015). The CAMESA guidelines aim to frame this discussion of how antipsychotic medication in paediatric patients should be managed (Ho et al., 2011; Pringsheim, Doja, et al., 2011; Pringsheim, Panagiotopoulos, et al., 2011).

**The Challenge of Behavioural Adverse Drug Reactions.** Beard and Lee (2006) define an adverse drug reaction as an “... unwanted or harmful reaction experienced after the administration of a drug or combination of drugs under normal conditions of use and suspected to be related to the drug” (p. 1). In contrast to adverse drug reactions that cause clinical presentations, which require immediate emergency admission such as Stevens-Johnson syndrome (Del Pozzo-Magana, Lazo Langner, Carleton, Castro-Pastrana, & Rieder, 2011), and metabolic or neurological adverse drug reactions, which require close monitoring, (Pringsheim, Doja, et al., 2011; Pringsheim, Panagiotopoulos, et al., 2011) the conceptualization that adverse drug reactions can significantly affect behaviour has not yet been established in paediatrics. Interestingly, the terminology of behavioural adverse drug reactions was not used in any of the charts of the 17 analyzed patients, although this was the likely reason for the frequent medication changes and thus the high number of prescriptions. Instead, terminology such as *allergy, intolerance* and *parents’ concerns* were used when discussing impacts of medications. A 16-year-old patient from our cohort who was described
as experiencing reduced amounts of sleep and also non-restorative sleep ("falling asleep can take him from one to two hours") had a severe adverse drug reaction to mixed amphetamine salts (Adderall XR®). This adverse drug reaction encompassed twitches in his limbs and entire body, shivering and restlessness for hours, followed by emotional breakdown with uninterrupted sleep, and finally amnesia. Following this event, medical staff in the hospital diagnosed the patient with Tourette syndrome and an “allergic reaction” to mixed amphetamine salts. He and his family were not counselled about the possibility of similar symptomatic episodes with future use of psychostimulants. This case illustrates that even the concept of neurological adverse drug reactions such as akathisia (a feeling of internal restlessness) (Keller et al., 2013; Miller & Fleischhacker, 2000), which present clinically as fairly dramatic effects, are not established in paediatric and child psychiatry as adverse drug reactions.

The Challenge with the Accurate Diagnosis of a Sleep problem and its Treatment. In current clinical practice, professionals’ exploratory models are based on previously established clinical diagnoses and bias towards categorical daytime-diagnoses. Integration of sleep and wake phenotypes in behavioural, neurological and mental health assessments is not currently the standard of care. Screening tools for assessing sleep/wake behaviours in children were initially introduced a decade ago (Owens & Dalzell, 2005); however, systematic research to describe the clinical characteristics of sleep problems in children with neurodevelopmental conditions started very recently with the development of specific clinical tools for in-depth assessment of children/adolescents with neurodevelopmental conditions (Blankenburg et al., 2013; Galland, Meredith-Jones, Terrill, & Taylor, 2014; Ipsiroglu, Carey, et al., 2012).

The case vignette #2 (section 1.4.4) demonstrates the problem effectively; sleep problems were existent since early toddlerhood in the narratives and were typically mentioned later in the medical records after diagnosing challenging/disruptive daytime behaviours, mainly ADHD but also
oppositional defiant and obsessive-compulsive disorders were mentioned to the family (but not formally diagnosed). At our clinic, in order to refine our clinical assessment concepts (Ipsiroglu et al., 2013), we use qualitative methodology (therapeutic emplotment) and a home-based video recording system (i.e., videosomnography) to capture sleep behaviours and movement patterns. Therapeutic emplotment allows patients and caregivers to tell their story and explain their perspective. This information is then transformed into a medical report, which is shared with the patient and checked for correct understanding of their observations. In addition, videosomnography acts as an extended eye during the sleep situation; allowing us to record and analyze movement patterns and has been instrumental in approaching and understanding the sequelae of WED/RLS (Galland et al., 2014). In summary, we are consequently applying the clinical skills of listening, observing, exploring and describing, like physicians have always done; however, nowadays time constrictions make this practice fairly challenging.

**Conclusion and Outlook.** This analysis shows how desperate clinical circumstances often lead to deviations from common prescription practices, such as prescribing more than two psychostimulants for one patient at a time or inappropriately prescribing ADHD medication to a toddler or starting with off-label prescribing of antipsychotics at the age of five. Analyzes of such worst case scenarios and tracking various treatment attempts are a clinical necessity, which help us to develop a better understanding and alertness in regards to prescription practices in association with sleep problems, their co-morbidities, sequelae and associated complications, as well as missed treatment opportunities (American Academy of Sleep Medicine Task Force, 1999). In our clinic, which has been developed into a clinical research unit over the past 5 years, the application of qualitative methodology on top of medical assessment standards allowed us to acquire the necessary in-depth understanding of current prescription practices. We can conclude the following: (1) Current prescription practices are complicated by patients who present with multiple medical conditions; (2) The variety of pathophysiology and aetiologies causing a sleep problem and our
increasing understanding of the strong interdependence between sleep and wake behaviours in children with neurodevelopmental conditions requires new assessment methodologies, targeted treatment strategies and monitoring of treatment effects and adverse effects; (3) Monitoring has to include observations and tracking of prescription and over-the-counter medication effectiveness/adverse effects including the changes in sleep/wake behaviours, in order to overcome the current overmedication cycles leading to polypharmacy. For this purpose, we have to develop new monitoring concepts (e.g., goal attainment scales and/or use of digital technology); (4) Finally, we suggest the implementation of a clinical database to assess practice-related outcomes as a powerful tool for quality of care improvement, as the Cancer or Cystic Fibrosis Foundation Registries have proven (Cystic Fibrosis Canada, 2011; Statistics Canada, 2012). A similar database for children with FASDs or other neurodevelopmental conditions, who require medication could be a powerful vehicle to establish functional sleep/wake behaviour assessment screening before starting medications and support the implementation of evidence based criteria as suggested by multiple research groups (Di Pietro, & Illes, 2014; Ho et al., 2011; Pringsheim, Doja, et al., 2011; Pringsheim, Panagiotopoulos, et al., 2011), updates to the CAMESA guidelines are available at www.camesaguideline.org].

Paediatric sleep medicine spans a matrix of organ-specific conditions and, as a theme, involves most paediatric sub-specialities. The retrospective analyzes of the data from the patients I have seen and investigated in-depth case reports, show that unrecognized and/or uninvestigated sleep problems lead to a cascade of diagnoses and medication. This can be interpreted as ‘iatrogenic harm’. In the next section (Chapter 3: Discussion), I will analyze why and how this culture of ‘sleep neglect’ has evolved in general and in particular in British Columbia.
Chapter 3: Discussion

This dissertation is about the need for a traditional clinical phenotyping-based understanding in paediatric sleep medicine. I engaged in this research endeavour in response to the reality that sleep problems have been poorly recognized as a determinant of health and wellbeing in children and youth with neurodevelopmental conditions (Jan et al., 2008; Newachek, Inkelas, & Kim, 2004).

From population-based epidemiological to genome-based association studies, there are many ways to shed light on unrecognized phenomena in health and disease. In my research, I used ethnographic exploration and the concept of ecology to create new understandings of intractable insomnia in children with neurodevelopmental conditions (Ipsiroglu et al., 2013). The paediatric working group of the International Restless Legs Study Group recommends behavioural observations in children with neurodevelopmental conditions, who might suffer from WED/RLS (Picchietti et al., 2013). The operationalization of these qualitative methodologies, which were merged as therapeutic emplotment in clinical practice, led to new insights and in consequence, to the creation of new methodologies (Ipsiroglu, Beyzaei, et al., 2015; Ipsiroglu, Hung, et al., 2015). I started with structured behavioural observations, which supported the exploration and explanation of challenging/disruptive sleep and wake behaviours in children with neurodevelopmental conditions from a novel perspective. In conclusion, my proposed assessment model recommends function-based diagnostic explanatory models (Cipani, 2011), which exceed the current practice of paediatric sleep medicine that operates within the realm of categorical diagnoses (Ipsiroglu, 2015; Ipsiroglu, Carey, et al., 2012; Ipsiroglu & Veer, 2012).

Kleinman suggests the application of ethnography to bridge challenging situations, which might be interpreted differently by each of the involved parties, through my work and research; I followed up on Kleinman’s recommendation (Kleinman, 1988, 2008; Kleinman & Benson, 2006).
The clinical groundwork of my research is built on the real life clinical cases that I have encountered over the last decade, and the worst-case-scenarios that were investigated more in-depth and presented in this dissertation (Ipsiroglu, 2015; Ipsiroglu, Berger, et al., 2015). Karl Jaspers suggests the discourse about Grenzsituationen as a foundational experience each individual has to deal with (Jaspers, 1980); the theoretical and rational basis for this work was laid by this discourse. In consequence, I attempted to create the space for a constructive transparent and demedicalized discussion and find allies and partners in order to run this discussion collaboratively in public (Miller, Recsky, & Armstrong, 2004).

Many children referred to my clinic had previously received child psychiatry/mental health diagnoses, which were partly or in some cases even completely resolved after treatment of the underlying cause of their sleep problems (Ipsiroglu, 2015; Ipsiroglu, Berger, et al., 2015; Ipsiroglu, Elbe, et al., 2015). From a public health perspective, appropriate diagnosis and treatment of sleep problems, will (hopefully) result in the reduction medication prescriptions cascades, causing iatrogenic harm in children with complex neurobehavioural symptomatology, as we have previously observed and reported.

The question of iatrogenic harm caused by missed diagnosis of underlying causes of sleep problems, impacted my own emotional ecology and initiated a critical discourse on how health care is managed at a micro- and macro-system levels. The comprehensive analyses of the observed worst-case-scenarios require(d) detective-like investigations, which explain why I developed a desire to understand the existing shortcomings of the health care system and particularly, those in British Columbia. Further, it became necessary to find strategies for how to overcome the current inefficiencies in paediatric sleep medicine, as the existing methodologies did not meet the needs of patients with developmental paediatrics and child psychiatry/mental health diagnoses (Kleinman, 2008). My previous training in health care management, (Ipsiroglu, 2003, 2007; Ipsiroglu & Stockler-Ipsiroglu, 1996; Ipsiroglu & Stockler-Ipsiroglu, 1999) and work, which included the
accreditation of the first paediatric sleep lab in Austria (at the Department of Paediatrics, Medical University of Vienna) as well as the design and solicitation of the Viennese Secure Sleep Campaign, (Ipsiroglu, Lischka, Sacher, Stögmann, & Pollak, 2000; Ipsiroglu et al., 1999; Ipsiroglu, Stekel, Pollak, & Kaspar, 2000; Ipsiroglu, Urschitz, & Pollak, 2003) became instrumental in this (still ongoing) discourse.

The first stages of my research aimed to answer the question: ‘Why have sleep problems remained unrecognized in children with neurodevelopmental conditions?’ with the aim of finding solutions to overcome the identified shortcomings. This query was closely tied to the question of how health care professionals respond to the concerns of patients and their families.

3.1. Recognizing and Surmounting Communication Barriers and Cultural Bias

The research presented in this dissertation was initially motivated by the recognition of training based cultural bias as a confounding fact in assessments and, thus obvious communication barriers hindering the recognition of sleep problems as an important morbidity in children with complex developmental disabilities. Communication challenges due to language differences are a well-known cause of such barriers. Insufficient information, differing education concepts, diverse conceptions of life, and dissimilar value systems of the involved parties are additional contributing factors (Stockler et al., 2012; Wimmer & Ipsiroglu, 1999, 2001). Hindrances to communication are well described in medical anthropology and linguistics literature (Bluebond-Langner, 1997; Clark & Mishler, 1992; Kleinman, 1988; Kleinman & Benson, 2006; Wodak & Lalouschek, 1997). The discrepancy between what the professionals ask in order to make a diagnosis and what the individual patient has experienced constitutes the main gap (Kleinman, 1988).

Qualitative interviews and analyzes of professionals’ systematic inattention to the sequelae of sleep problems, revealed that this practice was due to an insufficient understanding of the
interconnections between nighttime problems with daytime behaviours. As shown in the publication (section 2.1.1) (Ipsiroglu et al., 2013), the awareness is present but training is missing (i.e. the interviewed physician shared the statement: *I was not formally trained to address sleep problems*). This testimonial reveals a culture of separating between *practically experienced* and *academically proven knowledge* when it comes to making clinical decisions. Interpretations of sleep-related symptoms and decision-making in clinical practice are negatively impacted by *categorical diagnoses* and the complexity of situations in which these diagnoses present; therefore, symptoms are not well-understood, and the expectations from involved parties (e.g. parents), including macro-and meso-systems (e.g. legal or school system, respectively) might bias further notions and understandings.

Samara Mayer, a medical anthropology honours student who conducted her research in my clinic for her Bachelor of Arts Honours thesis, could reproduce this finding in a case series of patients with various neurodevelopmental conditions and intractable insomnia (Mayer, 2012). In addition, she also demonstrated how communication errors in general (e.g. missing shared reports) and in particular (e.g. missing descriptions of symptoms) contributed to missing sleep problems as an underlying cause for impacted daytime behaviours and well-being. Parents are partly excluded from the discussion, as very few clinicians’ share reports, and they typically need to ask their paediatrician (whom they see after some waiting time) for a copy of the report (Mayer, 2012).

*Analyzing the communication deficits and optimizing our understanding about various perceptions and understandings in regards to challenging/disruptive sleep/wake behaviours, I adapted the concept of therapeutic emplotment as a shared language strategy in my clinic. This was the first step of a multistep strategic approach.*
Solution I: Parents as Partners. The critique of a parent advocate (see endnote viii), which was provided in the protected environment of a participatory research seminar attended by healthcare professionals and parent advocates, highlighted that the conventional clinical reporting method reflected in our traditional communication style, was unidirectional and thus inefficient. In response to this critique, I started to address parents directly in all reports (we also thank the parents for their visit at our facility). This act has been the formal documentation of the respect to parents as first line-treatment providers. More than this, the commitment to present the symptoms that parents had reported in their own words was fulfilled by incorporating direct quotations into the reports and implementing a patient-based quality assurance strategy. After a very short period of time, it became clear that our clinical reports had opened up a new in-depth perspective to unexplored sleep and wake behaviours of the referred patients. Parents and assessing professionals were describing (for the first time in many cases) the same (for clinicians’ not well-recognized) phenomena using a shared language, and collaboratively exploring the iceberg-like hidden behind the intractable sleep problem of their child.

In consequence, it became foundational to use original quotations from parent/caregivers narratives when the results of the assessment were communicated verbally and later on, as a written emploted summary report. Specifically, parents’ descriptions of behaviours, activities, and quotes of clinically relevant descriptive features of sleep and wake behaviours were highlighted (e.g. movement patterns: “fidgets in her bed, before falling asleep”, “develops anxiety at bedtime”) (Appendix A: Patient Assessment Reports) (Mattingly, 1994, 1998).

The plain and transparent language for describing patients’ behaviours, and patient/family interactions avoids inaccessible medical jargon and has been referred to multiple times as respectful, (i.e.: sentences about parents are started by using their names (never refer to them in third person, as he or she) and/or use wordings you would like to get when people speak about you, e.g. do not use words signalizing missing respect (e.g. he denies that his daughter snores); always
address parents/caregivers with their last names parents/caregivers (Ms. Goldfinger and Mr. Bond, and not with their first names Sarah and James). Hence, the reports became a document of shared understanding making the concept of therapeutic emplotment to the foundational framework of my research.

**Solution II: Physicians as Strategic Allies.** In an atmosphere of economic productivity protected space for discussions between physicians and other health care professionals is scarce. I had to introduce my own findings into the practice of paediatric sleep medicine, and convince the referring physician that considering the underlying cause of the sleep problem as a primary cause of disease might reverse mental health diagnoses such as ADHD (Ipsiroglu, Berger, et al., 2015), and/or developmental paediatrics diagnosis such as autism spectrum disorder (Ipsiroglu, 2015). For example, I had to explain that the FASD diagnosis alone might not explain the challenging and disruptive behaviours, and in fact, an underlying sleep problem, such as WED/RLS, might cause these stereotypical repetitive discomfort or pain associated behaviours (e.g. stomping with the feet or kicking walls). In other words, in patients with FASD or ASD and intractable insomnia, WED/RLS as a cause of discomfort and/or pain and the effects of sleep deprivation were offered as a contributor to the challenging and disruptive daytime behaviours. I viewed this as a window of opportunity: treatment for the sleep problems could be applied, and the deleterious effects of alcohol on the developing brain (Clarren & Smith, 1978; Jones & Smith, 1973) would not be the only explanatory model for all challenging/disruptive sleep and wake behaviours of the patient. My perspective became that behaviours could be changed and that, in some situations, even the diagnoses of FASDs or autism could be challenged (Ipsiroglu, 2015; Ipsiroglu, Berger, et al., 2015).

Such unconventional explanatory models had to compete with current concepts in order to convince colleagues and parents. Sleep(y) Rounds (2007-10), Complex Chronic Care Rounds (2010-2012) and later on Pharmacology Rounds (2013-ongoing) and the British Columbian Cluster
Project (2014-ongoing), the latter two now supported by NeuroDevNet, a Pan-Canadian Centers of Excellence Networks’ Strategic Planning Initiative grant created the necessary academic space for protected discussions with physicians and therapists.

The second stage of my research was aimed to find ways to expand the clinical understanding of sleep problems in children with neurodevelopmental conditions in collaboration with their parents/caregivers. With increasing understanding, parents/caregivers and I started to challenge categorical daytime-based diagnoses, which had been given without assessing possible underlying sleep problems.

The Environment or Ecology of the Paediatric Sleep. Paediatric sleep medicine spans a matrix of organ-specific conditions and, as a theme, involves most paediatric sub-specialities. The establishment of paediatric sleep services is a direct response to the increased need to address organic sleep problems; the establishment of interdisciplinary sleep services a response to organic and non-organic sleep problems in a structured way, in order not to medicalize all sleep problems. In paediatrics, sleep labs have typically investigated the causes for sudden infant death syndrome (Paky & Kytir, 2000). In addition, the breathing problems of preterm infants were initially a major concern and research topic, leading the establishment of traditional sleep labs parallel to the expansion of respiratory medicine (Ipsiroglu et al., 2003; Poets, von Bodman, & Urschitz, 2003). In Europe, as well as in Canada, most of the sleep services operate at a tertiary hospital level and had a predominantly respiratory focus, while some of the labs developed an interdisciplinary approach in order to meet the needs of the patients (Fleetham et al., 2011; Ipsiroglu, Jan, Freeman, Laswick et al., 2009; Ipsiroglu & Saletu, 2001). Despite the major contributions of neurologic and psychiatric sleep labs to adult medicine, the number of paediatric sleep labs and sleep services led by a neurologist or psychiatrist (or developmental paediatrician) is significantly low (or zero, respectively) (Katz et al., 2014). In consequence, the concept of clinical descriptions as an approach to understand phenomena and thus, the impact of a phenomenological approach, has been limited.
**Phenotyping Sleep Problems in Unexplored Situations.** As already shown, failure to recognize sleep problems in children with neurodevelopmental disabilities is due to missing training and understanding. In September 2012, one of my mentors, Bruce Carleton, the founder of the Pharmacogenomics Program at BC Children’s Hospital and I organized a Peter Wall Exploratory Workshop, *Phenotyping Sleep Behaviours*, an academic space, which enabled us a trans-disciplinary brainstorming discussion at the University of British Columbia with Canadian and international guests (http://www.sleep-phenotype.pwias.ubc.ca/). The testimony of this workshop was to highlight the need for phenotyping sleep and wake behaviours in collaboration with parents and community based partners. In November 2014, clinical and benchmark researchers came together at the National Institutes of Health to discuss the missing pieces of paediatric sleep medicine. Again an identified core element was missing phenomenology and phenotyping, despite the avid collection of neurophysiological data. All case reports and the case series presented in this dissertation reproduce and respond to this foundational critique of missing phenomenology and phenotyping from a developmental paediatrics and child psychiatry/mental health perspective. They demonstrate how recognition of the sleep problem and addressing the underlying cause of the sleep problem led to a significant change in cognitive skills and emotional wellbeing, supporting the argument of missing phenomenology and phenotyping. When children with neurodevelopmental conditions advance in age their sleep problems usually increase due to the additional secondary behavioural, somatic and psychological co-morbidities. As such, identifying the cause of the sleep problems becomes increasingly challenging, but also more important.

However, as causes of sleep problems have multiple aetiologies, and functional assessments are not well established, it is almost impossible to create a strict logical model and analyze cause-and-effect interactions. New methodological approaches to gather data on clinical phenotypes of sleep disorders are needed. This might be the reason why, until recently, some of the sequelae or
mental health co-morbidities in children with chronic neurodevelopmental conditions have not been recognized as sleep-related.

The first major achievement in my clinical research was unravelling WED/RLS as a frequent cause of challenging/disruptive sleep and wake behaviours, intractable insomnia and ADHD-like behaviours, respectively.

3.2. Applied Emplotment

The application of therapeutic emplotment supported phenotyping in my clinical practice, and resulted in the establishment of a functional sleep and wake behaviours assessment concept Ipsiroglu (Ipsiroglu, 2015; Ipsiroglu, Elbe, et al., 2015; Ipsiroglu & Veer, 2012). Using this algorithm, I began to characterize and describe WED/RLS as a potential cause of intractable insomnia in children and adolescents with FASD (Ipsiroglu, Andrew, et al., 2011; Ipsiroglu, Black, et al., 2011; Ipsiroglu, Elbe, Witmans, Berger, Garden, et al., 2014; Ipsiroglu, Loock, & Carleton, 2014). At that time, we knew that WED/RLS could present at a young age, but the dimension of the problem was not clear and fairly hypothetical as the assessment methodologies were not yet established (Lee-Chiong, 2007). The main clinical challenge was how to recognize WED/RLS in patients who were not able to describe the WED/RLS-related discomfort in their own words, e.g. nonverbal or very young patients.

The emplotment approach, which explores possible WED/RLS-associated phenomena and supports building of a shared language with parents and referring physicians, was further developed with the home-videosomnography and the SCIT for structured night-and-daytime observations.

Home-videosomnography. Inspired by parents who use their smartphones to present clips of their child’s challenging/disruptive sleep/wake behaviours, we developed an affordable
home-videosomnography recording system utilizing commercially available hard-and-software. Using video recordings was the ideal inspiration for almost 40 years; however, the technical complexity, including the requirement of a lab and the bulkiness of the equipment, has prevented their widespread popularity and usage in medical practice (Anders & Sostek, 1976; Sitnick et al., 2008).

Currently, the gold standard for diagnosing sleep-related movement disorders is a lab-based sleep study (i.e. polysomnography) including electromyographic recordings to detect sleep-related movement disorders, such as period limb movements in WED/RLS. The concept of home-videosomnography bridged the clinical gap in our understanding between the narrative-based description of symptoms occurring during daytime/prior to falling asleep and movements occurring during sleep, which are normally not reported by the patients. This fostered the development of a phenomenology of disturbed sleep, mainly presenting as restless sleep with kicking movements, which was not based on assumptions but on concrete, in-depth observations. The home-videosomnography recordings facilitated qualitative and quantitative analysis of limb and body movements. In my clinical practice, this has been instrumental in characterizing WED/RLS in children and adolescents with neurodevelopmental conditions, such as ADHD, autism spectrum disorders, FASDs and cerebral palsy (Hill, Parker, Allen, Paul, & Padoa, 2009; O'Connell & Vannan, 2008).

Until now, the main tool for collecting data in behavioural sleep research has been actigraphy, which collects wake/sleep information but does not reveal important clinical information, and has not been validated in children with neurodevelopmental conditions, e.g. cerebral palsy. In the absence of better tools, it has been proposed as the current gold standard of sleep/wake-rhythm assessments (Sadeh & Acebo, 2002; Sadeh et al., 1995). Actigraphy also misses the clinical significance of major movements such as getting up or jumping on the bed (which are typical movement patterns of children who are on the autism spectrum and experience intractable
insomnia) as the research team directed by Dr. Thomas Anders, professor of neurology and psychiatry, has shown (Ipsioglu, Hung, et al., 2012; Sitnick et al., 2008).

Towards the end of this dissertation research, my understanding has become that using actigraphy follows the trend of collecting quantitative data, i.e. it has been useful to prove concepts in larger epidemiological studies, but not in explorative study settings. Actually, actigraphy has obscured the practice of looking for the phenomenon itself and understanding the causes of a problem represented in the quantitative data set.

Interestingly, thanks to the structured discussion of the professional associations, actigraphy is even billable. Actigraphy became a typical research tool which enthusiastic clinical researchers use for creating data, but are not familiar with the strength or weaknesses of the methodology and therefore miss important information (O’Donnell, 1997). In contrast, home-videosomnography is instrumental in understanding the individual problem, including the dimension of the insomnia (i.e. falling asleep and maintaining sleep). It also provides insight into the patient’s burden as demonstrated by the distressed facial expressions of the patient in the case vignette and her help-seeking interactions with her parents.

**SCIT.** Based on parent narratives and home-videosomnography results, I soon realized that almost all patients who presented with restless sleep, characterized as *kicking movements* and *thrashing around*, also had daytime presentations suggestive of hyperactive-like behaviours, including an inability to keep still and various sensory processing abnormalities. Therefore, the next step was to develop a concept for reproducible behavioural observations of daytime behaviours of parents and their children, as presented in section 2.2.2. The decision to adapt and develop the lab-based SIT for clinical purposes was made at the Peter Wall Exploratory Workshop in 2012 based on the discussion between the attendees. The SIT is a 40-minute long test that was
created to collect structured information about WED and periodic limb movement in the wake phase of the adult patient (Michaud, Paquet, Lavigne, Desautels, & Montplaisir, 2002; Montplaisir et al., 1997). In this context, adaptation meant that it had to be done in a clinical setting which provided a child friendly atmosphere, could be reproduced with ease, and was a manageable amount of time to complete. First and foremost, it had to deal directly with the typical daytime challenge of sitting still under controlled circumstances. As such, we developed the SCIT, which prompted the associated reactions in a diagnostic and therapeutic setting for the patient and his/her accompanying family members. We changed the focus from the patient (subject of the investigations) to the family (all family members as subject of intervention), which became an enjoyable part of the assessment. Children begin to laugh, giggle and joke with their parents about their own symptoms as well as their parents. Even children with severe disabilities enjoy the test as a kind of unexpected activity associated with fun activity. xv

The first four diagnostic criteria of WED/RLS are built on verbalized self-report (Picchietti et al., 2013), but can also be adapted for self-or 3rd-party observations. The original diagnostic criteria rely on the patient's perception and understanding of the dimension of their own symptoms, which is accompanied by a number of major challenges in verbalization and description for paediatric patients and/or children with neurodevelopmental conditions, as well as adult patients (Allen et al., 2014; Picchietti et al., 2013). Even in adults, language barriers and cultural differences act as a diagnostic limiting factor when describing their symptoms (Garcia-Borreguero et al., 2011).

The SCIT, as I applied in my clinical assessments, enables the structured, observational assessment of the first three self-reported essential criteria: (1) An urge to move the legs, usually accompanied by uncomfortable or unpleasant sensations in the legs; (2) Symptoms begin or worsen during periods of rest; (3) Symptoms are partially or totally relieved by movement. This way the discussion of the 4th
and 5th criteria was also cued in a structured way. In addition, a foundation of understanding had been laid (symptoms occur mainly in the evening/night and symptoms cannot be solely accounted by another medical condition).

Similar to Garcia-Borreguero's observation, in our manuscript we could show that only 78% (21/27) the mothers were able to describe sensory symptoms (Ipsiroglu, Beyzaei, et al., 2015). Given the fact that all mothers had been diagnosed previously with WED/RLS, and the SCIT was conducted just for the verification of the previous diagnosis, we can conclude that aside from language also the perception of sensory symptoms vary. Additionally, patients who might be missing a reference point due to early the onset and impacts of chronic disease were tested in a structured, reproducible situation. For these cases, the International Restless Legs Syndrome Study group had suggested the use of supportive criteria such as familial history of WED, periodic limb movements in sleep, and behavioural observations, without clarifying what and how behavioural observations had to be done (Picchietti et al., 2013).

The SCIT enabled us to conduct these behavioural observations in the clinical setting in the context of therapeutic emplotment. Further, this bridged the gap between traditional paediatric sleep medicine, which did not have answers for patients with neurodevelopmental conditions, and facilitated the creation of a structured explorative (ethnographic) sleep/wake-behaviour assessment concept.

3.3. How to Deal with Medications or the Phenomenon of Iatrogenic Harm

Through the application of therapeutic emplotment in context with the familial sleep history I realized a very special pattern: WED/RLS presentations were aggravated by psychotropic medications, and, in some patients, stopping trials of such medications had been seen as a solution by parents and many professionals (Ipsiroglu, Berger, et al., 2015).
The next level of achievement was the detection of the association of behavioural or neurologic adverse drug reactions to psychostimulants as a frequent cause of challenging/disruptive sleep and wake behaviours in patients with WED/RLS.

This exercise helped me to learn which drugs were tolerated, which ones were not tolerated (e.g. ADHD medication, which put children and teenagers under major stress (one child called their medications “crying pills”), and what had provided some relief (in adolescents self-medication with marijuana or even alcohol, provided by parents, before bedtime). The parents shared my fascination about the various WED/RLS presentations and began to speak about their self-medication strategies to treat their WED/RLS-related discomfort. Further, I began to understand that some mild adverse drug reactions to the medications agitating the child and hindering from falling asleep, had been attributed to the neurodevelopmental condition and subsequently treated with additional medications (Ipsioglu, Elbe, et al., 2015). Within a very short time, I realized the similarity of most familial narratives: usually they were coherent (e.g. people who did not tolerate methylphenidate also did not tolerate coffee) and added, despite sometimes being presented as a side note of the story, significant further value (e.g. “after starting crawling he started climbing”). The parents of these children and sometimes their siblings had also frequently been diagnosed with ADHD, but did not tolerate ADHD medications, and were suffering from not recognized and not treated intractable insomnia as well as day- and nighttime restlessness due to WED/RLS. The main reoccurring theme was the inability to deal with ADHD medications, but having a positive response to alcohol, marihuana or antihistamines. The side effects, which lead to a change of medications, were usually fairly dramatic, and unique, but shared two main clinical presentations: a) the drugged zombie and b) agitated devil-like. While the first description, i.e. drugged zombie, was fairly reproducible –it was about sedation and drugged behaviours — the second phenotype was not as simple. Asking pharmacologists and pharmacists for their help, I realized that the latter were
various presentations of akathisia, a neurological adverse drug reaction presenting with a state of inner restlessness and worsened behaviours (Keller et al., 2013; Miller & Fleischhacker, 2000).

The next observation was, depending on the advocacy skills of the parents, sometimes the medications were changed immediately; in others, a second or third medication was given to overcome behaviours, which were in fact neurologic symptoms as described in the case vignettes and case series. Without generalizing birth parents and parents of children with autism spectrum disorder or ADHD had higher advocacy skills, than adoptive and foster parents, who followed recommendations of professionals in a more strict (compliant) way as they wanted or had to follow the fixed mentality.

The presented case studies and in-depth analysis of life trajectories demonstrate how unrecognized sleep problems lead to iatrogenic harm, and how categorical diagnoses can mask iatrogenic harm for a long time. Sleep problems were taken as given and not investigated further, which caused the daytime-focused medication cascades. This not only affected quality of live, but also intruded in the individual life story and autonomy, becoming an issue of fate leading parents to desperateness.

In my clinical practice, I observed that not only treating the cause of the sleep problem, but also weaning the patient off medications, resulted in an increase in individual and family's wellbeing and quality of life, which supported my observation based hypothesis. Stopping daytime medications, and treating the underlying neurologic disorder, e.g. WED/RLS, completely changed the clinical presentation and life-trajectory.

Michael Oldani, a medical anthropologist, who worked in his first life as a pharmaceutical representative, has shown how doctors, despite not trusting medications, utilize prescription medications (mainly stimulants) for children with an FASD diagnosis. (Oldani, 2009) Typically, the medications were used as an urgent solution to pressing problems that arise within their time
limited assessments. FASD and medication literature shows that children with FASDs frequently do not tolerate medications (stimulants and antipsychotics) and multiple trials are necessary before finding a good medication regimen (O’Malley, 1997; O’Malley et al., 2000; O’Malley & Nanson, 2002; O’Malley & Streissguth, 2003; Wilens et al., 1996). The need for more time in order to optimize assessments is clear, but not provided, thus non-existent. Therefore, based on Oldani’s and our observation based research we can conclude that the witnessed problem of overmedication and poly-pharmacy in the population with FASDs is the specific demonstration of a general and systemic problem. To prove this conclusion, epidemiological studies are necessary.

_Vulnerable patient groups, who need better service, are at particular risk for iatrogenic harm due to the gaps in our clinical understanding and missing time for in-depth investigations and reflection of that what we are doing._

We could prove this observation recently in another study. Individuals with Down syndrome are to have high sleep disordered breathing prevalence; depending on the study, the sample cohort and magnitude of the study prevalence rates vary between 50% and 80%, approximately (de Miguel Diez, Villa-Asensi, & Alvarez-Sala, 2003; Dyken, Lin-Dyken, Poulton, Zimmerman, & Sedars, 2003; Waldman, 2009). The data from the _British Columbia Down Syndrome Needs Survey_ show that 85% (n=221/316) individuals with Down syndrome have symptoms of sleep disordered breathing while only 22% (n=71/316) received the clinical diagnosis of a sleep disorder (Ipsiroglu, Beyzaei, Tse, & Marwaha, 2015). I believe that there are several factors, which contribute to such a discrepancy. The major issues seem be the culture of separating between _practically experienced_ and _academically proven knowledge_, prompted and elicited by _missing time_ (for reflection) and _categorical diagnoses_, which are not further explored by symptoms, and the common perspective of the clinician: ‘_I was not formally trained to address sleep problems_’ (Ipsiroglu et al., 2013), which might support the desire for _self-protection_. As described by Oldani, missing
assessment and follow-up time constitutes an organizational communication barrier and systemic failure and in addition, liability-related concerns might support being ‘conventional’ and less open and courageous to accept new concepts.

Most alarmingly, these factors lead to iatrogenic harm. Despite not believing in medications, perceptibly they might be considered one important chance for therapeutic intervention. In our case series of children with FASDs or autism spectrum disorder (Ipsiroglu, 2015; Ipsiroglu, Berger, et al., 2015), many medications were prescribed in combination and at very young ages. Case vignette #2 (section 1.4.4), is the youngest patient with an ADHD diagnosis, and was medicated with two different stimulants at the age of two and half. The youngest patient with ASD was medicated at the age of two years and 4 months for insomnia with second-generation antipsychotics (i.e. Seroquel® (quetiapine) as well as medicated with Depakote® (divalproex sodium) typically prescribed for seizures and Remeron® (mirtazapine) typically prescribed for depression without investigating the cause of the insomnia. In contrast to the first patient who received his ADHD diagnosis at age two and a half (in an age where one cannot diagnose ADHD), the latter patient was diagnosed with pervasive developmental disorder (on the autism spectrum) and recurrent major depressive disorder at 30 months.

**A Special Remark on ADHD, WED/RLS and Sleep Problems.** There is an overlap between ADHD, WED/RLS and the sleep problems associated with both conditions. A diagnostic ascertainment bias is likely depending on the applied diagnostic tools and on the medical subspecialist performing the assessment.

WED/RLS is a neurologic condition causing major day-and-nighttime restlessness. While sleep-related nighttime symptoms are well recognized, the effects of this condition on daytime behaviour are less well-described. As a matter of fact, patients with WED/RLS often report that their initial diagnosis was ADHD. Comparably, the vast majority of our patients were given the
diagnosis ADHD, which inevitably lead to a cascade of comorbidities most likely triggered by intolerance to psychostimulants (Ipsiroglu, Berger, et al., 2015). ADHD is a functional rather than a categorical diagnosis, which is given based on experienced symptoms, and indicates a position on a spectrum (Heidbreder, 2015). Therefore, the described symptoms, which create a spectrum, should be considered as a “starting point for further testing rather than being an endpoint of diagnosis that results in pharmacological treatment that may or may not be the optimal therapy” (Heidbreder, 2015, para. 1). These findings are in accordance with Miano and her colleagues (2012) who analyzed the literature for sleep phenotypes of ADHD and identified five phenotypes among the 25-50% of children and adolescents with ADHD experiencing sleep problems.

The American Academy of Pediatrics (2011) recommends considering sleep problems prior to making an appropriate ADHD diagnosis. However, common ADHD questionnaires, particularly those published before 2000, do not screen systematically for primary or secondary sleep problems. Tools published after 2000 have questions that explore excessive daytime sleepiness and sleep disordered breathing and possible WED/RLS, but in an unsystematic way (Ipsiroglu, Hung, Mayer, Collet, & Carleton, 2012).xviii

**Based on the confounded patient population that I have seen in my clinic, I believe that all patients with an ADHD diagnoses and who do not tolerate psychoactive medications should be considered at risk for WED/RLS.**

In British Columbia, ADHD and mental health diagnoses are a growing epidemic. The Waddell Report speaks of a prevalence rate of 2.5 and 16,600 children and youth between 4-17 with an ADHD and 3.8% for anxiety 25,300 (Waddell, Shepherd, Schwartz, & Barican, 2014). Recently, Ms. Erin Ellis, journalist at Vancouver Sun, reported that at their (actually, her) request “ministry staff checked the province’s prescription tracking system, PharmaNet, and found that...
312,304 prescriptions to treat ADHD were filled in 2011, while 94,251 were filled in 2000” (Ellis, 2012).

These two different, actually, divergent facts and associated justifying viewpoints (see endnote xix) confuse me as a clinician and might confuse parents and other professionals even more. The only consistency between the two statements is that sleep problems, as a possible cause of ADHD-like presentations and/or WED/RLS, were not mentioned. On the other hand, despite the known high ADHD-prevalence, there was not a single child who had been diagnosed with WED/RLS in British Columbia when I started with paediatric sleep assessments.

In 2012, a consortium of researchers led by Richard Morrow, including employees of the BC Ministry of Health presented a cohort study involving 937,943 children in British Columbia. The study investigated the absolute and relative risk of receiving a diagnosis of ADHD and a medication prescription for treating ADHD. A relative age effect was revealed: children born in December were at higher risk than if they were born in January (Morrow et al., 2012). Morrow and colleagues (2012) conclude their findings with the following statement:

... raise concerns about the potential harms of overdiagnosis and overprescription. These harms include adverse effects on sleep, appetite and growth, in addition to increased risk of cardiovascular events. (p. 761)

The two presented studies and the fact checking conducted by a journalist demonstrate ‘how perceptions/understandings of challenging/disruptive sleep/wake behaviours may differ’ and that our health care system does not have a concept for managing the challenging/disruptive sleep and wake behaviours of children with and without complex and chronic health care conditions.
3.4. Closing the Gaps

My work with children with unrecognized sleep problems not only motivated my research, but also revealed the enormous gaps in regards to our current understanding in paediatric sleep medicine in general, and, particularly, in regards to the services offered in British Columbia (Bull, 2011).

**Understanding-related Gaps.** The *ethnographic exploration of the ecology* of paediatric intractable insomnia; the operationalization of this *ethnographic exploration* as a *therapeutic emplotment concept* in clinical practice, and the *creation of new methodological tests and tools for structured behavioural observations* as further development of the *therapeutic emplotment concept* are the strategies in order to close the gaps in paediatric sleep medicine from a developmental paediatrics and child psychiatry/mental health viewpoint.

**Service-related Gaps.** Canadian provinces, such as Alberta, Ontario, and Quebec are leading the national discussion about sleep deprivation and its consequences with high quality research and have thus shaped the clinical sleep services (Fleetham et al., 2011; Katz et al., 2014). Compared to some of the recent recommendations of the American Academy of Pediatrics (Bull, 2011), paediatric sleep medicine-related services offered in British Columbia are not currently fulfilling the mandate of state-of-the-art services. BC Children's Hospital is the only site in the province that hosts a paediatric sleep lab. It offers services to a minority of patients with sleep problems who are primarily seen for respiratory problems and has a limited capacity. There is no specialized service offered for children with neurodevelopmental conditions and sleep disorders. The clinical services that I am providing are funded, for the most part, by donations and research grants.

These service-related gaps have been summarized in a consensus paper published seven years ago, which was authored by the interested sleep medicine professionals at BC Children's
Hospital in collaboration with invited guests from other faculties and provinces (Ipsiroglu, Jan, Freeman, Laswick, et al., 2009). In this paper, we outline suggested clinical decision-making procedures for various tier service levels and recommend how offer screening and prevention-based solutions starting at the public health level (Ipsiroglu, Jan, Freeman, Laswick, et al., 2009). This paper was followed by a position paper that structured this discussion further (Ipsiroglu, Jan, & Freeman, 2009). Involved decision-makers at the administrational level have expressed interest, but the agenda has still not moved forward.

There are always multiple reasons and causes for not being able to move an agenda forward. From a cultural perspective, particularly in achievement-oriented societies, the connotation of sleep is akin to the perception described in the bible where “excessive sleeping was regarded as being unacceptable as it produces laziness and could subsequently lead to poverty” (Cox, 2008; Klug, 2008; Thorpy, 2010, p. xxiii). From a work culture perspective, the discrepancy between applied empathy as a philosophy as suggested by Frank (1985), which contributes to increased alertness in systemic decision-making, and a professional strategy managed by the best, which follows indifferently political parameters of saving money, might contribute to this discussion.xv From a health-policy decision-making point of view, professionals make decisions, e.g. a diagnosis, based on their assessment tools and communicate these to the patients (Ipsiroglu, Stekel, et al., 2000); this process has supported a uni-directional communication culture in medicine (Kleinman & Benson, 2006; Mishler, 1986), but also in health care administration with top-down decision making concepts. These pathways have troubled other colleagues before me (Bruni, Laupacis, & Martin, 2008): Drs. Whitfield and Chessex (2010, p. 573) presented “an argument in support of the need for public involvement in decisions made about health care resource allocation.” Drs. Miller, Armstrong and Recsky (2004) suggest that specific actions on a number of fronts including work with media and advocacy groups and systematic efforts to ensure that family-centred care or the interdisciplinary team work are incorporated into policies and practices for
their care. One can conclude that the desperateness about “… systemic gaps and inefficiencies that are driving increased health care costs…” is apparent (Stanbrook, 2001, p. 741).

Ultimately, decision-making is often subject to the influences of the right momentum, triggered by serious incidents, or politicization or the emotionalization of the discussion by the creation of an empathetic environment by strong advocacy. Children with neurodevelopmental conditions and sleep problems, as I have referred to in my research, usually do not have advocates, as parents and caregivers are most overwhelmed with the care of the child or, as often seen in the case of FASD, are having mental health issues themselves. In a study among service providing institutions about their current understanding of the dimension of untreated sleep problems, interviewees stated that after a certain “tipping point,” families are overwhelmed by the imbalance between the treatment results, and insufficient financial and medical / community support resources (Timler & Ipsiroglu, 2015), finding themselves in emergency situations or crises (Caplan, 1961; Graham, 2015; Lucyshyn & Albin, 1993; Lucyshyn et al., 1997; Poal, 1990).

The evaluation report of our Interior outreach activity confirms these results (Graham, 2015). In regards to sleep, interviewees clearly stated that the current concept at BC Children’s Hospital with waiting lists of six to 12 months is insufficient (Graham, 2015). In a capturing the status-quo study, all interview partners, who were asked for short- and long-term solutions, unanimously highlighted the significance of knowledge dissemination, increased awareness and advocacy which are best achieved by (1) medical partnerships, (2) medical homes, and (3) care guidelines (Timler & Ipsiroglu, 2015). Overall, a team-based healthcare delivery model led by a health care professional that provides comprehensive and continuous medical care has been the proposed solution (Timler & Ipsiroglu, 2015). This solution is analogous to the recommendations of the American College of Physicians and American Academy of Pediatrics, which has been seen as a management strategy for complex care patients by British Columbian health authorities (American

When I initially reported my concerns of iatrogenic harm particularly for children and youth with FASDs to clinical and administrative chief decision makers in the province, the response was ‘we need more research.’ With the research presented here, the testimonies of patients and parents, and the open public discussion, which was started in 2012 at the Peter Wall Workshop Meeting, I trust that enough momentum has been generated to move the agenda forward at the political decision-making level.

3.5. Highlights on the Horizon

There are several opportunities on the horizon that may be instrumental in creating an interdisciplinary sleep program in British Columbia. The Annual Sleep Science Conference of the Thompson River University traditionally presents sleep disorders from a 360-degree perspective. Several articles recently published in the media show that sleep is garnering increasing public attraction.xxxii Finally, polypharmacy and psychotropic overmedication are attracting public interest, and unrecognized sleep problems are an important trigger for this problem, as demonstrated in children with neurodevelopmental conditions within this dissertation framework (see Chapter 2).

These activities are supported by ongoing research, which aims to further implement the presented results into clinical practice. I have translated findings of my research on sleep phenotypes, polypharmacy and adverse reactions to psychotropic medications in a downloadable application which facilitates a standardized sleep assessment for use in community-based practice. Funding from the BC Government and from NeuroDevNet (http://www.neurodevnet.ca/) has been secured to validate and broadly disseminate this tool. Funding from TIDE-BC (www.tidebc.org) and
international collaborations, e.g. with the Austrian Institute of Technology (www.ait.ac.at) will enable an advanced algorithm for automated home-videsomnography analysis, and numerous international collaborations will support further validation of its use in research and clinical practice.

The implementation of the comprehensive sleep assessment protocol has been initiated through collaborations with community-based paediatricians, the BC Paediatric Society, the Burnaby-based Down Syndrome Research Foundation, and Thompson Rivers University in Kamloops. The recent Apps4Kids Hackathon, which my team and I co-organized with the Department of Electrical and Computer Engineering at UBC and with professionals from the Child Family Research Institute, attracted over 80 students to work on apps that support children with chronic care conditions (Berger et al., 2015; McAllister et al., 2015). A Capstone project team developed an EMG App to objectively record limb movements as an observation-based expansion of the SCIT described in this dissertation (https://www.ece.ubc.ca/courses/capstones) (Geisler et al., 2015; Saad et al., 2015); this idea has been picked up at the Apps4Kids Hackathon and is currently under refinement. The link of apps will generate clinical research data through registries; information that can be used in the future for comparing outcomes, evaluation, and thus, quality improvement in collaboration with service providing non-governmental organizations.

3.6. Relevance of this Research; Strength & Weaknesses

**Strengths.** *First,* the sample of patients and their families is large and representative of the population of children and youth with neurodevelopmental conditions across British Columbia. Recruitment is based on referrals from physicians rather than active or direct recruitment of participants; referrals are not coming from densely settled locations and milieus. In addition, the referred patients and their families come from various social-economic groups, ethnicities and cultural backgrounds found in British Columbia. *Second,* the data collection occurred over the
course of several years, which should mitigate any seasonal or calendar-related effects on sleep patterns. Third, the research utilizes self-reported narratives of parents, extended family members and, if possible, the patients themselves. The concept of therapeutic emplotment was implemented in my research as the gold standard of history taking as it is obviously quality controlled by parents and involved caregiver teams through shared reports. Fourth, our interviewing concept allowed parents to share their thoughts and concerns in regards to sleep problems with the security of not getting accused or blamed for any of their behaviours or explanatory models (Graham, 2015). Fifth, we developed two methodologies to unravel the different dimensions of challenging/disruptive sleep-and-wake behaviours. In order to understand the obscured dimension of challenging/disruptive sleep-and-wake behaviours we developed the home-videosomnography recordings and, in order to cue narrative-based descriptions of best- and worst-case scenarios going along with discomfort up to pain, we developed the SCIT as a standardized individualized behavioural observation strategy. Sixth, most patients were followed up by the referring physician and their own caregiver teams and thus, had further assessments and monitoring of their therapies by third parties as an objective (but not standardized) quality control. Seventh, the coherency of the descriptions in the adverse drug reactions to psychotropic substances proves that despite the variability and/or unknown cultural factors the reported observations were reproducible. This, actually, proves that narratives are the gold standard of clinical history taking. Last but not least, the strength of this assessment concept is that it is based on descriptions and supports phenotyping, which can be done by any interested health care professional and can add significant value in phenotyping the various presentations of early onset and chronic sleep disorders impacting brain development (see endnote ii).

Limitations. First, the partly prospective and partly retrospective style of the clinical assessments is a limiting factor. Learning and applying methodologies includes different levels of skills and might have confounded results. Second, despite of or because of close monitoring and
follow-up by me as a clinician, a second independent researcher was not involved until very recently, which might have caused a confounded view. Third, while numerous clinicians have validated the clinical assessment results, physicians whom I could not convince with my diagnoses and concepts might have not given a negative feedback and contributed again to election bias and further confounded our view (Graham, 2015). Fourth, the scoring of the home-videosomnography is, at best, heuristic and the interpretation of the SCITs is exploratory and has not been validated by other researchers. Validation of the home-videosomnography and the SCIT methodologies has recently started and has not been included in this dissertation. Fifth, the limited measure of sleep quality and lack of objective validated measures of sleepiness (which will follow after validation of the home-videosomnography methodology) is a limiting factor. Similarly, the absence of structured pre-and post-mental or physical health assessments in most children and adolescents limited our ability to link sleep behaviours to functional outcomes in a structured way in this sample. Finally, case series studies, despite being the start point of clinic evidence and research, are typically reviewed as low-level evidence according to evidence-based medicine criteria (Sackett, Straus, Richardson, Rosenberg, & Haynes, 2000).

The hot topic of this research, which came up towards the end of the dissertation research and will be one future perspective, is based on the application of the concept of ecologies. The concept of family ecology did not only help to review stress and distress of parents (Stockler et al., 2012), but the application of the ecology concept also helped to review stress and distress of professional staff. I identified the missing protected space for reflection of complex cases and objective external review, which includes patient advocates and an ombudsman, as the main factor perpetuating the current vicious cycle (Jenkins, Hoube, Smith, & Mitton, 2007). Further analysis showed that the voice of patients was not only, not heard in clinical sleep history taking, but also not in the discussion or evaluation of clinical services. This is similar to the situation of doctors, also their voices are not heard; as a result of this discovery, my recommendation is that professionals
should collaborate with parent advocates and media in order to achieve systemic changes. I am following up on the recommendation of the previous head of the Department of Pediatrics at the University of British Columbia (Miller et al., 2004).xvi, xiv

These are significant, possibly frightening observations and results, which need further investigations; however, they are all beyond the scope of this dissertation research. In Chapter I, I referred to the concept of emotional ecology. Using empathy as a professional concept in my ethnographic investigations, I realized my own burn-out experience as a clinician. Reviewing one’s own situation, including the experienced emotions, is a basic research concept in qualitative methodology (Hammersley & Atkinson, 1983) and it is also recommended practice in psychology, psychotherapy (Balint, 1957), and, recently, in medicine. However, the way I dealt with own emotional burden might be reviewed by some readers as a weakness. On the other hand, it would be inappropriate to speak about emotional ecology and to censor the discussion about one’s own or professionals’ emotional ecology (Kropiunigg, 2005). This is a topic that might be discussed controversially in the dissertation defence – I am looking forward to this discussion.

In my clinical practice, and for my own emotional ecology, the solution was to overcome the set boundaries with clinical research – the Interdisciplinary PhD endeavour provided the perfect setting for that and catalyzed the change of focus from the initial transcultural paediatrics theme to the current medical anthropology and educational psychology-based applied clinical research. This endeavour provided a protected academic space to review the situation from a distance and to create methodologies for collecting new and more grounded data (Martin & Turner, 1986; Strauss & Juliet, 1994). The breakthrough in my clinical research came from the analysis of the worst-case scenarios, which I would describe as borderline or approaching the limit situations according to Jaspers’ philosophy (Jaspers, 1980).xv Suddenly, the need for new methodologies that support phenotyping at a community-based clinical level and the generation of better clinical-academic hypotheses (and better support genetic or pharmacogenomics investigations) became evident. This
personal experience raises the question of how our system deals with worst cases or with limit situations. The answer to this will show whether we have a learning system or an indolent and ineffective system.

3.7. Final Remarks

As referenced in the introduction, Michel Foucault (1975) described in his book *The Birth of the Clinic: An Archaeology of Medical Perception* that the methodology of autopsies was foundational to the development of modern medicine, and led to a new individualistic approach using the analysis of ‘single-cause-and-single-effect’ interactions, exceeding traditional religion and politically dominated holistic thinking. This cause-and-effect interaction model fostered the medicalization of holistic medicine with our current modern critical understanding of evidence-based medicine and also facilitated let-us-fix-the-problem thinking. The let-us-fix-the-problem thinking is based on the tremendous success of antibiotics and survival rates, and works best in acute care situations. With the high survival rates of patients from neonatology, biochemical diseases, cystic fibrosis and oncology the need for developing a more holistic and also grounded (patient-focused) understanding (exploring, developing a shared language and avoiding communication barriers) became evident (Niethammer, 2007).

Modern medical anthropology picked up this as a core topic (Helman, 2007; Kleinman, 1988). Essential themes, like accompanying patients in end-of-life situations, (Kaufman & Morgan, 2005), autonomy and personal space of patients or like in my research exploring patients with challenging/disruptive sleep and wake-behaviours, who come with significant developmental paediatrics and child psychiatry/mental health diagnoses, made the need for a more reflective framework evident. Aristotle postulated that ‘philosophy begins in wonder’ (Rosenberg, 1989). Modern philosophers of the 18th (Kant, 1974) and 19th centuries (Jaspers, 1980) and anthropologists (Clark & Mishler, 1992; Kleinman, 1983, 1988; 2008; Kleinman & Benson, 2006),
who have investigated phenomena and perceptions, notions, and understandings, respectively, from various viewpoints, have utilized explorative thinking to understand ecologies. In response to the ongoing discourse and experienced challenges in modern medical care management, the World Health Organization suggested to focus on the development of a joint understanding that acknowledges health as a value. This recommendation stresses the importance of creating a shared goal-oriented language and value system, which, in consequence, will become the foundation for all interactions with patients, including the creation of prevention and intervention focused informational activities (Clark & Mishler, 1992; Kleinman, 1988; Kleinman & Benson, 2006; Lau, Hartman, & Ware, 1986). This implies the integration of patients in the decision-making process; in situations where patients cannot be integrated, it suggests the implementation of an ombudsman system to overcome autocratic inappropriate or wrong decision-making on behalf of patients.

My research has revealed major systemic challenges in paediatric sleep medicine impacting developmental paediatrics, child psychiatry/mental health diagnoses in general and, particularly, in the British Columbian health care system. This research also revealed several windows of opportunity for addressing these gaps with concrete qualitative methodologies. Further discussions regarding paediatric sleep medicine and health care management go beyond the realm of this dissertation research. The questions that were brought to light in this discussion, including: ‘How do we further develop concepts of health care management integrating ethics, solidarity, subsidiary, and resource allocation?’ ‘How do we implement them at all levels of political decision making in biomedicine (Chambers & Kurz, 1999; Sass, 1995)?’ and ‘How do we incorporate the patients’ and the family’s interests?’ (Miller et al., 2004; Whitfield & Chessex, 2010) are questions that will be answered in the near future with the help of innovative qualitative concepts like ethnography or ecology, using bi-directional communication models.
Endnotes

i Modern anthropologists, such as Robin Patric Clair, discuss ethnography and the subjective viewpoint of those who apply ethnography fairly critically (Clair, 2003). This kind of reflection is necessary, and my personal subjectivity, including my philosophical background, will be analyzed in this dissertation.

ii Recently, I received a referral form at my clinic that was filled out by both a community-based paediatrician and an occupational therapist. Both were asking for the consultation of the same child who was exhibiting some ADHD-like daytime behaviours at school and was suffering from non-restorative sleep. The paediatrician asked for assessment of 'sleep apnoea,' while the occupational therapist reported her mainly descriptive finding of 'sensory-seeking behaviours relieved by movement,' which is a textbook-like description of WED/RLS. My understanding after the assessment of the child was that the WED/RLS had been causing the non-restorative sleep in this patient, which presented as ADHD-like challenging/disruptive behaviours at school, and the description (phenotyping) provided by the occupational therapist was ideal.

iii Topics include the most challenging real-life situations as previously described, which deal with love, jealousy, generosity or self-medication and sleep disorders in an empathetic way. Composers like Gaetano Donizetti, Vincenzo Bellini or Giuseppe Verdi created unique individual characters by using music to present the experiences of the protagonists in an ecological setting and to develop situations and portrait characters. By using both chorus and soloists, they are able to describe the dynamic interplay between society's and the protagonist's thinking, notions, and emotions from 360-degree perspective. A slightly different interpretation of the music and/or staging may change the entire narrative, and opens up a new interpretation of the well-known and un-changing plot and characters from the conductor and staging directors' more or less empathetic perspective,
highlighting personal subjectivity of each presenter and their very individual impact on the narrative.

iv The latter question became the topic of the Vienna Secure Sleep Sudden Infant Death Prevention Campaign, which had a unique design (Ipsiroglu et al., 2003; Ipsiroglu, Fatemi, Rabl, Klupp, Roll, et al., 2002). Under the name ‘Secure Sleep,’ positive prevention-related information was disseminated, e.g. I love co-sleeping (we differentiated between sleeping on the same mattress and co-sleeping and particularly emphasized the positive effects of co-sleeping with parents within the first year of life) (Forster, Ipsiroglu, Kerbl, & Paditz, 2003; Ipsiroglu et al., 2003; Kerbl, 2003).

v The following quote is out of an assessment report of two young adults who do not fit the typical patient type for clinical assessment at my clinic. I was very interested in seeing both of these boys due to the clinical diagnosis of familial WED in their step-sister (biological cousin) and both parents, as well as the anecdotal information that both of the boys were diagnosed with ADHD, and have reported adverse reactions to stimulant medication.

[Patient 1] was diagnosed with ADHD, in grade 11, prior to his brothers’ ADHD diagnosis. Both boys were initially prescribed 18 mg of Concerta® (stimulant), but experienced side effects, when the dosage was increased to 27 mg the side effects worsened. Both patients described that utilizing the stimulant medication was a “bad experience” because it made them “too hyped up” and they became very “jittery”, with their hands constantly shaking uncontrollably. The medication caused their hearts to race and “set [their] heart off” by beating really fast; additionally, the boys experienced anxiety-like symptoms and sweated immensely when on this medication. [Patient 1] described the stimulant medication experience similar to “drinking 5 cups of coffee” or “drinking energy drinks right after another.” [Patient 2] also added that drinking coffee (caffeine) also produces the same stimulant-like effect and causes his heart rate to speed up rapidly. Other medications: Both
young men have taken melatonin; this may have helped [patient 2] more than [patient 1]. Both young men have also experimented with self-medication and realized that: 1) alcohol may improve the falling asleep situation and make them feel more tired and ease their falling asleep situation, but it does not improve their sleep maintenance and they wake up earlier; 2) cannabis may help to improve their falling asleep situation as well as sleep maintenance. They describe that cannabis use facilitates a more “calm” and “restful” situation, with more “comfort,” which reduces the tossing and turning in bed and enables them not only to fall asleep with more ease, but also have a more restorative sleep. During a recent family vacation, (where the ability to self-medicate was obviously more restricted) nighttime awakening were up to five to six times per night.

vi Autocratic, paternalistic, partly fascistic does not sound like a typical scientific description found in a PhD. However, the personal experience does not have to be scientific. Seeing that students were asked to leave the exams as they were not dressed appropriately (black and brown suits were seen as appropriate, blazer with grey pants or a grey suit not), hearing descriptions of tumor cells (comparing with a Jewish assembly) might be considered as a single professors’ statement. On the other hand they were given this action radius and in addition were partly the only professors who were teaching and examining. In the seventies and eighties, the political background and autocratic, paternalistic, partly fascistic behaviour of Austrian medicine (and law) professors has been subject of many research projects and newspaper titles.

vi As a senior physician working in the Intensive Care Unit, I was accustomed to working 48 hour weekend shifts (starting on Saturday morning and ending Monday morning). Although we were allowed to go home immediately on Monday mornings, it was a habit of most senior physicians to stay longer and accomplish work or join rounds. This was done with the aim of creating an atmosphere of pro-active work in a busy department and being a role model for residents and
In 2009, in a seminar, which I was hosting for review of my clinical research with Dr. Judy Owens, Professor of Paediatrics at Brown University and other colleagues from UBC, a parent advocate from ACT-Autism Community Training, asked me why a typical report from my clinic addresses the referring physician and not directly the parents. This question opened up a discussion on how we collect information, and how and with whom we share this information. The parent advocate’s critique was as follows: parents, despite being the central sources for important information and the main supervisors of therapy implementation, were not even addressed. Instead, I directly addressed the physicians that might be seen after a waiting period of three months, and were not responsible for overseeing the critical at-home therapy measures, e.g. the challenges in the application of the suggested sleep health measures.

The German philosopher and psychiatrist Karl Jaspers frames *borderline* situations (*Grenzsituationen*) as a core element, which drives humans; reviewing and reflecting the own vulnerability, which affects his/her communication and interaction with others (Jaspers, 1980).

Samara Mayer’s (2012) study was conducted as part of the Evaluation Study of the Sleep Clinic at Sunny Hill Health Centre for Children. Five patients with neurodevelopmental disabilities and a chronic sleep problems (at least six months) requiring complex chronic care, from at least five sources were selected. The charts were reviewed; accompanying network diagrams were developed to understand the structure of information dissemination. Semi-structured interviews were conducted to understand parental perceptions of their child’s health care management. Networks were composed of five-33 interconnected care sources as dependent on the patient chart. The network diagrams (1) revealed the complexity of each patient’s care network, (2) visualized informational gaps by revealing directionality and frequency of information dissemination. The semi-structured interviews revealed that some parents had to take on managerial and advocacy
roles to coordinate their child’s care. The narratives contextualized and validated the results of the network diagrams, highlighted institutional deficits namely (1) the fragmentation of care through informational discontinuity by barriers e.g. parents did not have any concerns regarding confidentiality if the flow of information was effective, (2) though they expressed the desire to be more involved in the care planning process, they did not receive the reports. The findings suggest the need to (1) consider the capacity of families and provide them with their child’s information (2) develop a shared charting system that provides better clarity of pertinent clinical history where up-to-date reports are distributed on a regular basis.

xi A recent survey-based study conducted in Canada by Katz and colleagues (2014) found that there are no paediatric sleep physicians or diagnostic testing resources (i.e. polysomnography) in the Yukon, Northwest Territories, Nunavut, Saskatchewan, New Brunswick, Prince Edward Island, and Newfoundland and Labrador. Furthermore, the majority of practitioners are affiliated with the respirology department of their respective institutions, followed by psychiatry and general paediatrics. No paediatric sleep medicine practitioners directly affiliated with a developmental paediatrics department were identified in this survey (Katz et al., 2014).

xii Seeing patients more individually and investigating the clinical and genetic phenotype is the main theme of our time. This trend has been enabled by the advances in genetics (e.g. genome sequencing) and leads to the customization of healthcare. Medical decision-making and practices are tailored according to the individual patient.

xiii Similarly, the case vignette of the manuscript presented in section 2.2.1. demonstrates the dimension of chronic and early onset WED/RLS-related insomnia, which negatively impacted developmental coordination, sensory processing, and mental health, causing anxiety and mimicked ADHD behaviours. By observing the patient, her mother and father enhanced the impact of our clinical therapeutic employment concept. Home-videosomnography-recordings not only enabled us
to assess and understand the dimension of the falling asleep and sleep maintenance problems, but also provided us with insight into the patient’s burden of suffering as demonstrated by her distressed facial expressions, and her help-seeking interactions with her parents. Fascinatingly, the home-videosomnography-recordings also revealed information on the familial (and genetic) nature of the patient’s and her parents’ WED/RLS presentations. Further, home-videosomnography prompted us to understand the impact of trivial phenomena, e.g., scratching, which is likely not mentioned in most sleep studies and has never been investigated in a systematic way in the sleep of children, but is associated with the sensory profile of children with WED/RLS (Ipsirolgul, Beyzaei, et al., 2015).

xv The Peter Wall Exploratory Workshop in 2012 was another protected academic space I had created with my mentors and enabled a brainstorming discussion. The outcome of the meeting became a plea for transdisciplinary thinking as well as active communication and interaction with patient families. For this purpose, the attendees agreed to create the Children’s Sleep Network (http://www.sleep-phenotype.pwias.ubc.ca/).

xv Recently, an eight-year-old child with autism and severe anxiety who sat under tremendous tension began visibly to relax when he saw us testing his mother and, in follow up, he even felt comfortable to be examined.

xvi The following is a quotation from the initial assessment report of a male adolescent patient with sleep difficulties, diagnosed with ADHD and a neurobehavioural disorder, who experienced a particularly significant neurological adverse drugs reaction:

After being started on Adderall® by his paediatrician [...] [patient] had an adverse reaction (shivering, twitching). He was hospitalized at [hospital], and was presumed to have Tourette syndrome. However, to our understanding, he has never had a formal psychiatric evaluation for this diagnosis.
Later on, in a private discussion with a physician who was a paediatrician and a pharmacologist, I realized that the patient had experienced akathisia and was fitting to all descriptions of akathisia—a clinical presentation that most clinicians are not familiar with. Please note that my missing clinical understanding was compensated for by the clinical description that I provided, which was subsequently picked up by my more senior colleague who could support me in adjusting my understanding.

The following quotation is from a follow up report of a four-year-old boy who was diagnosed with autism spectrum disorder at age two and a half, and a history of hypothyroidism. The patient was seen at the sleep clinic for his long-term difficulties with falling asleep and sleep maintenance. The patient was prescribed a neuropathic pain medication to address his WED/RLS-related discomfort and his mother described the experience:

Ms. [mother] reports that she did not notice any significant difference on the first night (100 mg). On the second night (100 mg), [patient] woke up in the middle of the night and was “pretty hyper;” Ms. [mother] gave him Gravol® and it took him another 1.5 hrs to fall asleep. On the 3rd night (200 mg), [patient] was able to fall asleep in 20 minutes and did not wake during the night. On the 4th night (200 mg), Ms. [mother] reports that [patient] was hyper for an hour before bed; however, “once he was asleep, he stayed asleep.” Ms. [mother] describes from that from the 5th night and onwards, [patient] was extremely hyperactive (e.g. flipping around the bed), uncontrollable, and “appeared to be possessed” (e.g. “laughing in a hysterical and evil way”; “not like himself”). Ms. [mother] elaborates that “all of the calming activities and bedtime procedure that he was so great at from the past few months of training, needed to be tossed out during these nights as it took everything in my power (and in one of his therapist’s power who did one bedtime routine) to keep him just on the bed.” However, Ms. [mother] reports that once [patient] was able to fall asleep with the gabapentin, he did sleep through
the night. The gabapentin was stopped on the 2nd night of the 300 mg dose and Ms. [mother] continued to give [patient] Gravol® or Benadryl® during his night awakenings.

With a notion similar to that of ADHD, we analyzed common diagnostic questionnaires: BASC-2 Behaviour Assessment System (2004), Vanderbilt ADHD Diagnostic Rating Scale (1998), SNAP-IV Teacher and Parent Rating Scale (2003), ADHD Rating Scale IV (1998) and the revised version of the Conner's Rating Scale (1997). We investigated the degree to which they ask about (i) sleep problems, and (ii) for symptoms associated with sleep disorders, which have to be investigated further. None of these ADHD assessment tools screens systematically for primary or secondary sleep problems, which is actually a recommendation of the American Academy of Pediatrics when making an appropriate ADHD diagnosis (American Academy of Pediatrics, 2011). Tools published after 2000, i.e. BASC-2 and SNAP-IV, have some inconsistent questions that explore excessive daytime sleepiness and sleep disordered breathing and possible WED/RLS; however, tools published before 2000 ask fewer questions about sleep. It is not clear which questionnaire are used when and the inconsistencies trouble also my colleagues from psychology, e.g. the WED/RLS characteristic symptom of climbing, also included in the Diagnostic and Statistical Manual of Mental Disorders – IV, is asked in most questionnaires but not all (American Psychiatric Association, 1994).

In order to understand the background of the current dimension of ADHD medication prescriptions, Ms. Ellis also interviewed Dr. Margaret Weiss, a clinical researcher and professor of psychiatry at the University of British Columbia: “The rate of use of medication still does not meet the prevalence of the disorder. So statistically, we’re definitely not over-treating (it)” (Ellis, 2012). Furthermore, the article reported that “[i]n terms of the rise of treatment for ADHD, that’s totally appropriate given the recognition that it exists in boys and girls, that it exists in multiple sub-types and that it exists in adults. In other words, when we see those people who are suffering and very
much impaired and we have a treatment that's available to reduce that suffering, it's very appropriate that those people are getting the care they deserve” (Ellis, 2012).

In 2014, I participated in a research call by the Doctors of British Columbia (previously BC Medical Association) for testing novel concepts of sub-specialized service delivery. We failed at the first letter of intent level, despite being a strong team with strong supporters and focusing on the problem of iatrogenic harm. In the rejection letter, the main concern was that we might create a new program, which will cost British Columbian health authorities additional funding; most interestingly, no methodological or organizational concerns were raised. This understanding solicits the question: are budgetary considerations a better argument and do they justify, at the administrative level, continuation of iatrogenic harm? As making such a claim could be considered ridiculous, one may argue that it is not fair to review committee decisions, which have been made out of political calculations in a non-academic setting, and the defence of the decision-makers is not possible. On the other hand, between the announcement and the rejection letter, there was such a contextual controversy, which cannot be excused as political decision-making for the reason of fairness, but has to be investigated further in this discourse.

The names of the authors read as a who is who of British Columbian clinical (not administrative) decision makers: Dr. Whitfield was the Director of the Neonatal Follow-Up Program, the program evaluating neonatal outcomes; Dr. Chessex the Division Head of Neonatology. Dr. Miller is an experienced chronic care management researcher and Dr. Armstrong was the previous Department Head (only Dr. Recsky was a young researcher at that time). Dr. Armstrong enabled the trial starting the development of a service for patients with neurodevelopmental conditions suffering under sleep problems.

According to the literature, it takes 17 years on average until medical evidence is implemented into practice (Balas & Boren, 2000). After reviewing a couple of the recent recommendations of the
American Academy of Pediatrics (Bull, 2011; Marcus et al., 2012), I have proposed to review the services currently offered in British Columbia, and the degree to which they are meeting the state-of-the-art criteria recommended by the Canadian Sleep Society for the last three years [https://css-scs.ca].


The CanMeds framework, a synthesis of the scope of the different professional responsibilities of physicians, provides also an ethical framework, which supported my personal decision-making in overcoming the current situation (Royal College of Physicians and Surgeons of Canada, 2014). Since 2013, I have invited an ethicist and public health researcher into the discussion about novel observations and current state-of-the-art, and started a public discussion with parent groups and parent organizations offering service to patients with neurodevelopmental conditions. This way the information and the emotional burden of the observations were shared and the protected discussion space finding solutions were created within a collaborative research network.

Worst case scenario, with the connotation I have used and justified with my case report series and/or grenzsituationen (limit situations) as Jasper (1980) uses in his text ‘What is Philosophy’ sound pathetic, but are situations we all experience, situations which challenge us and create questions about are doing are the starting point for philosophical thinking.
In the European paediatrics context, the word *holistic* was introduced by advocates propagating *alternative medicine* and was reviewed critically by advocates of modern medicine. Over the time this connotation has apparently changed. This change from a ‘no’-word to a different and more positive connation became clear when the previous Head of the Department of Pediatrics at the University of Tuebingen, Germany presented the request of student representatives for a more comprehensive curriculum and made a plea for more “holistic medicine,” opposing highly sub-specialized academic paediatrics (Niethammer, 2007).
References


American College of Physicians. (2013). *Understanding the Patient-centred Medical Home: Get the Facts on the PCMH Care Model and Make an Informed Decision for your Practice and Patients*.


Ferini-Strambi, L. (2012). Does idiopathic REM sleep behaviour disorder (iRBD) really exist? What are the potential markers of neurodegeneration in iRBD? *Sleep Medicine, 12*(Suppl. 2), 129-135. doi: 10.1016/j.sleep.2011.10.010


Ipsiroglu, O. S., Jan, J. E., & Freeman, R. D. (2009). *Proposal for the Paediatric Sleep Medicine Program at BCCH/SHHCC and Development of Paediatric Sleep Medicine Services in British Columbia.* Report to the Department of Pediatrics, BC Children's Hospital, Vancouver, BC.


Ipsiroglu, O. S., Loock, C., & Carleton, B. (2014). *Poly-pharmacy in Children and Youth with FASDs and/or Prenatal Substance Exposure.* Poster session presented at the 22nd Annual Meeting of the German Sleep Society, Cologne, Germany.


189


193


Veer, D., Chan, R., Loock, S., Matthew, S., & Ipsiroglu, O. S. (2011). Applicability of sleep screening studies in the home or hotel setting: quality improvement research. Poster session presented at the 88th Annual Meeting of the Canadian Paediatric Society in Quebec City, QE.


Appendix A: Patient Assessment Reports

A.1. Typical Sleep Consultation Report

This is a typical sleep consultation report of a patient with Down syndrome from 2007. Note that at this time, I had not yet started to implement therapeutic emplotment in the clinical assessments.

SUNNY HILL HEALTH CENTRE FOR CHILDREN
3644 Slocan Street
Vancouver, BC V5M 3E8
Tel: (604) 453-8300 Fax: (604) 453-8301

CHILD DEVELOPMENT AND BEHAVIOUR PROGRAM

Pediatric Assessment of Sleep

NAME: [Redacted]
CHART: [Redacted]
DOB: January 7th, 2002
DATE OF CONSULTATION: June 4th, 07
DATE OF REPORT: June 5th, 07

Dr. [Redacted] referred [Redacted] to me regarding her sleep issues. Yesterday, I had the pleasure to see [Redacted], a delightful young girl, who was accompanied by her mother [Redacted].

History/Assessment: [Redacted] is 5 5/12 month old, has Trisomy 21 and increasing sleep disturbances for the last 1 to 1.5 years. There are some hints of sleep disordered breathing (SDB) over night: [Redacted] is a mouth breather, shows an orofacial hypotonia (no sore throat), but doesn't snore loudly or all the time (only when she is ill), no excessive sweating. However, she sleeps in prone position with buttocks up, sometimes waking up with gasping, and [Redacted] has also witnessed apnea. In summary the last three observations are quite strong symptoms of sleep disordered breathing due to hypopnea or obstructive apnea, and suggest surgical procedure.

There is an additional issue: [Redacted] likes napping, which may be indicative for non restorative sleep, but the [Redacted] family does not like [Redacted] to nap, as it is hard to wake her up, and bedtime gets postponed to late night hours. However, Ms [Redacted] observed that on days when [Redacted] is allowed to nap, she is less grumpy and distracted after the nap, and more collaborative.

Based on our discussion I would suggest finding out whether “power naps” which will not last long (~ 10-20 min, on the coach, in the garden, etc.) will help to improve her need for resting. I can imagine [Redacted]’s concentration to increase and her learning skills to develop more adequately, if she gets the possibility to nap.

Follow up/ Recommendations:

1. A sleep study can be recommended, but clinical symptoms are quite convincing, and I wonder whether we should lose time with waiting for a sleep study. This decision should be made by [Redacted] ENT BCCH.
2. In case of continuation of sleep disturbances I would be happy to see [REDACTED] any time.

Thanks for your referral.

Yours sincerely,

_______________________

Osman Ipsiroglu, MD
Clinical Associate Professor

CC:
- Dr. Fred Kozak, ENT
- Dr. Grace Yu, Community Pediatrician
- Dr. David Wensley, Respirology, Sleep Lab
- Dr. James E. Jan, Sleep Research/EEG BCCH
A.2. Sleep/Wake Behaviour Assessment Report

This is an in-depth report from 2015 using the concept of therapeutic emplotment. It is, in fact, a follow up assessment report of the patient assessed in the Typical Sleep Consultation Report from 2007 (Appendix A; Section A1.)

SUNNY HILL HEALTH CENTRE FOR CHILDREN
3644 Slocan Street
Vancouver, BC V5M 3E8
Tel: (604) 453-8300 Fax: (604) 453-8301

CHILD DEVELOPMENT AND BEHAVIOUR PROGRAM

Paediatric Assessment of Sleep

Confidential

OUTPATIENT CLINIC NOTE

MRN/Encounter:
Date of Service:
RE:
MRN:
PHN:
DOB: 7 Jan 2002

DIAGNOSES
1. Trisomy 21.
2. Recurrent AOM, now OME, myringotomy tubes in place, followed by Dr. [redacted]
3. Prior sleep-disordered breathing seen by Dr. Ipsiroglu, adenoidectomy April 2003.
4. ADHD.
5. Mild intellectual disability based on early psycho-educational assessment.

Dear Dr. [redacted];
Dear Mr. and Ms. Tudhope:

Thank you so much for your re-referral of [redacted], a now 13-year-old girl seen previously with sleep-disordered breathing and re-referred for chronic nighttime awakenings. She was accompanied today by her mother, Ms. [redacted]. Given the complexity of the presentation, we have completed a comprehensive assessment; this is a summary of our understanding.

MEDICAL HISTORY
As you are aware, [redacted] is a now 13-year-old girl who lives in Vancouver with her parents. Previous diagnoses are listed above. [redacted] was seen previously by the Sleep Clinic at the age of 5-1/2 years and was found to have sleep-disordered breathing with likely obstructive apnea. She has been followed by ENT and
had an adenoidectomy in 2013, which apparently was quite helpful. ___ is quite athletic, with good gross motor skills, and enjoys basketball and swimming.

From a developmental standpoint, ___ is functioning well within her abilities. She is now in Grade 7 with a modified program. The year had a rocky start as ___ struggled to meet the demands of course work and new routines; however parents and teachers made significant changes and things are now going well. At school ___ is working on self-regulation and problem-solving skills in the classroom setting, reading with the Grade 2 class (although she independently reads books above this level), and in maths she is working on adding, simple multiplication, and time concepts. She will be transitioning to the Exploration program at the high school next year. ___ has a history of ADHD diagnosed in 2012. Without medication, ___ requires significant redirection throughout the daily tasks, routines, and academic tasks. On the medication, she is much calmer and additionally more still. Ms. ___ also told us that when sleep-deprived or tired, ___ will be more distractible and more fidgety than usual.

**SLEEP ASSESSMENT/CURRENT SLEEP SITUATION**

Ms. ___ describes a long-standing history of challenges with disrupted sleep, specifically sleep maintenance and morning sleepiness. She is additionally concerned about some characteristic movement patterns; ___ is reported to be a very restless sleeper, sitting up, 'folding' and continuing sleeping. These behaviours were also observed in the formal sleep study. ___’s nighttime awakenings have gone on for years, and significantly impact the quality of life of both ___ and other household members, and the family has had to take a number of steps to address these behaviours. Often the parents’ sleep is sacrificed in order to manage ___’s insomnia related challenges.

1. **Bedtime/routine/regularity:** ___’s bedtime routine starts at 8:30 p.m. with teeth brushing, face washing, and pajamas. ___ is shortly after in bed and will read for 20-30 minutes prior to falling asleep. Typically, ___ is able to fall asleep between 15 and 45 minutes after the lights are turned out. On average, she is asleep between 9:15 and 10 p.m. Occasionally, she does have difficulty going to sleep but this is generally infrequent and happens on average twice a month. If Adderall® is given at a later time than usual, this will lead to prolonged sleep onset.

2. **Sleep maintenance/behaviour/parasomnias:** Without fail, wakes up at 3 in the morning. On the extremely rare occasions where she has slept through the night, it is in the setting of a much later bedtime. When she wakes up, ___ will often get out of bed and previously would go downstairs to play video games, where her parents would find her later in the morning, asleep on the couch. She was also starting to make food and use the microwave during the nighttime hours. ___ was exceedingly tired on the days following these nights awake, and parents were concerned about safety as she was unsupervised in her use of the kitchen and appliances. They have initiated door locking at night in an effort to reduce night time wanderings and ensure ___’s safety. If she wakes up and needs to use the washroom or wants a glass of water, parents will come and escort her there and subsequently back to bed. Sleep onset following these episodes is not an issue. ___ is quite active in her sleep. She tosses, turns, and frequently will sit up. After sitting up, she will often fold over and they will find her folded in half, asleep, in the morning. If bed-sharing, ___ will often kick.

3. **Sleep-disordered breathing:** ___ previously had significant evidence of sleep-disordered breathing, sleeping in a prone position, waking up gasping, and witnessed apneas. This had improved following adenoidectomy back in 2013; however, Ms. ___ did have concerns about this during the visit today. They had recently been evaluated by a sleep study in the medical day unit at BC Children's Hospital, but according to this, oxygen saturations remained stable and apnoea/hypopnea index was 1.7 and within the norm. Ms. ___ tells us that this was not a typical night’s sleep due to delayed sleep onset in the new environment and the need to wake up earlier to be out of the medical day unit in the morning. There is occasional noisy breathing at night and abrupt sitting upright, which could be consistent with sleep disordered breathing. However, there is no noted apnea, gasping, or nighttime sweating. Interestingly her arousal index was 12.3, which means that approx. 12 events fragmented her sleep per hour. Possibly due to that, in the first
part of the night did not experience any REM sleep, but did experience prolonged light/deep sleep. She also woke up approx. 11 times over the night.

4. **Excessive daytime behaviours:** wakes up independently at 7 a.m. The parents have noted a change in her waking behaviour over the course of the last year. On school days, she will not wake up independently at 7 and instead will require help from parents. On nights where she has been up and is still quite tired in the morning, will have to enter the room up to 3 times. will also report that she still wants to be sleeping. There is no clear history of daytime napping and no napping in the car that noticed. is seemingly tired on most days, and this does have an impact on her behaviour and her ability to focus.

**MEDICATION HISTORY**

is on the following current medications: Adderall® 20 mg q.a.m. between 7:00 and 7:30 am.

**Previous medications:** Melatonin taken for 3 months, from January to March, 2014, up to a dose of 9 mg by mouth at bedtime. There was a period of approximately a week where this was felt to be helpful. However, after this it no longer appeared to have any effect and so was subsequently discontinued.

There been no adverse side effects from the Adderall®. Initially there was some reduction in appetite but this resolved and is no longer a concern. It is apparently quite easy to note when the Adderall® wears off, between 3:00 and 4:00 p.m., and the parents do not feel that it impacts sleep onset on a regular basis when taken in the morning. It does, however, delay sleep onset if the Adderall® is taken later, around 12:00 or 1:00 p.m. also notes that the sleep problems experienced by predated the initiation of Adderall®, and they feel that these are unrelated.

**FAMILY HISTORY**

’s dad, Mr. , is a light sleeper. He has no problems with sleep onset, but does have difficulties with sleep maintenance and sleep disordered breathing (obstructive sleep apnea) has been questioned. Mr. often has a daytime nap and will go to bed quite early, between 8:00 and 9:00 p.m., because of exhaustion; this is interpreted as stress-related. reports fewer difficulties with her sleep. There is a maternal grandmother with nighttime awakening difficulties, and a maternal aunt who requires sleeping aids. There is also a paternal aunt with sleep maintenance challenges who also uses sleeping pills. is 46 years old and is a kindergarten teacher in Abbotsford. Mr. is 49 and works as a mechanic in Maple Ridge. Outside of sleep challenges, his health history is significant for depression and hypothyroidism requiring Synthroid.

**PHYSICAL EXAMINATION**

Height 139 cm (<3rd %ile). Weight 45.3 kg (15th-50th %ile). Cardiovascular, respiratory, and abdominal examinations were normal. Cranial nerve examination, excluding olfaction, was normal in the office today. had impressive strength and was 5/5 in all upper and lower extremity groups. She had low peripheral tone. She had symmetric reflexes in the upper and lower limbs, and gait including toe, heel and tandem walking, was normal. There was no fidgeting observed in the office today and, in fact, was unusually still, which mom tells us is typical when she is taking the Adderall®. We note that she prefers “yoga-like poses” while sitting. On the SCIT, exhibited restlessess and some large movements, however had very few small fidgeting movements. had no fidgeting or unusual movements during the SCIT.

**INTERPRETATION AND HYPOTHESIS**

Our clinical findings and observations suggest that chronically experiences non-restorative sleep, and is chronically sleep deprived as a result of:

1. **Behavioural sleep interruptions** with regularly timed nighttime awakenings with getting out of bed.
2. **Likely familial Willis Ekbom disease** (restless legs). ’s history of being quite athletic and her significant strength in the office today, as well as the unusual sitting postures that we observed, are in keeping with Willis Ekbom disease. Her history of tossing and turning at night is also consistent with spontaneous limb movements in sleep (as confirmed by the sleep study). The Adderall® medication that is
active during the day certainly will suppress any fidgeting type movements that we may otherwise see, but we note that the medication is not active during the nighttime hours, and \textit{Ms. Tudhope}'s frequent complaints of sitting upright or moving may be consistent with arousals caused by the spontaneous limb movements which lead to disrupted sleep. We were not able to see any signs of Willis Ekbom disease in Ms. \textit{Tudhope} today; however it is quite possible that Mr. \textit{Tudhope} has similar symptoms and an informal SCIT will be done with him at home. \textit{WED is characterized by a discomfort of the feet, legs, hands and/or other body parts, which is relieved by movements. The urge to move increases in periods of rest and towards the evenings, and seems to be aggravated mainly upon falling asleep. Iron deficiency, among other causes, can cause or worsen WED presentation.} The athletic history and yoga-like movement patterns are very much in keeping with this diagnosis, and are typical self-soothing approaches to WED-related discomfort.

3. Sleep-disordered breathing: Sleep disordered breathing is a significant risk factor for poor quality sleep. While \textit{Megan} no longer has complaints of snoring, as she did prior to adenoidectomy, a review of the sleep study report carried out recently does indicate a substantial number of what appeared to be obstructing episodes during the night. These were not apneas but were rather hypopneas considered to be within the normal range. While these may not clearly meet criteria for a significant apnea finding, they are not insubstantial and may be more aggravated when she experiences better REM sleep at night, and contribute to \textit{Megan}'s clinical presentation. Of note, a significant number of spontaneous awakenings were documented on the sleep study, all together contributing to non-restorative sleep.

In summary, we strongly believe that some of \textit{Megan}'s daytime behaviour seen in the absence of Adderall® is aggravated by chronic sleep deprivation due to likely Willis Ekbom disease and sleep interruptions. A WED like presentation and ongoing mild sleep-disordered breathing may also be contributing.

RECOMMENDATIONS FOR CONSIDERATION AND FURTHER INVESTIGATIONS
1. Blood work: We have requisitioned CBC, ferritin, CRP, electrolytes, extended electrolytes, TSH T4, vitamin D and vitamin B12. This will be helpful to provide an initial assessment currently of \textit{Megan}'s iron status.
2. Videosomnography and formal sleep study: \textit{Megan} has just undergone a formal sleep study at BC Children's Hospital. We have reviewed the results of that testing today. Given the frequent arousals with unclear etiology, it may be worthwhile in the future to go ahead with videosomnography which is a home-based sleep assessment video, to provide further clinical information.

NON-PHARMACOLOGICAL/PHARMACOLOGICAL THERAPEUTIC RECOMMENDATIONS
1. Sleep health/sleep health measures: We recommend going over the attached sleep health recommendations and identifying applicable/achievable goals (i.e. reducing screen time, increasing outdoor activities in order to tire \textit{Megan} out).
2. Daylight or artificial light: We recommend increasing \textit{Megan}'s exposure to daylight or artificial light during the day and reducing light as much as possible during the night to help regulate melatonin secretion. Recognizing that \textit{Megan} is afraid of the dark, transitioning gradually to lower wattage light bulbs and a dimmer night light would be achievable goals.
3. Medications: (a) We have advised \textit{Megan} to start iron supplementation, 30 mg of elemental iron b.i.d. at breakfast and lunch. We will discuss duration of medication administration with the family following the results of blood work. (b) In addition, low dosage melatonin (e.g. 2 mg long acting) could be used to initiate sleep at earlier hours and increase the current amount of sleep \textit{Megan}.

COMMUNITY SUPPORT FOR IMPLEMENTATION
1. Goal attainment scaling: We would suggest that the family develop a list of ranked goals and concerns regarding \textit{Megan}'s sleep; see attached goal attainment scaling form. This will help to evaluate the effects of the various medications in a personalized manner, and to monitor \textit{Megan}'s therapeutic plan.
2. Family health: \textit{Megan} has symptoms of disrupted sleep. Given that Willis Ekbom disorder is familial, we have asked the family to do a SCIT in the home and to report back to us with the results. If this is positive, it would be reasonable to obtain blood work to look for iron deficiency in Mr. \textit{Tudhope} and to supplement if required.

In summary, \textit{Megan} is a lovely young woman with a longstanding history of sleep problems due to behavioural interruptions, likely familial Willis Ekbom disease, with a possible component of sleep-disordered breathing. We will start with our initial blood work and iron supplementation, and follow with...
the family from there. It was a pleasure to see [redacted] once again. We will follow up with them after receiving the results of the blood work.
A.3. Abbreviated Immediate Report

Therapeutic emplotment is an important endeavour for capturing and understanding the patient’s narrative; however, it is also a time consuming process and leads to lengthy reports. For this reason, I developed an immediate abbreviated report concept. This is the complementary Abbreviated Immediate Report from 2015 for the patient assessed in the Sleep/Wake Behaviour Assessment Report (Appendix A; Section A.2.).

<table>
<thead>
<tr>
<th>ASSESSMENT OF SLEEP/WAKE BEHAVIOURS</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Immediate Abbreviated Report</td>
<td></td>
</tr>
<tr>
<td>Team Dr. Ipsioglu</td>
<td></td>
</tr>
<tr>
<td>Sleep/Wake Behaviours Clinic located at SHHCC</td>
<td></td>
</tr>
<tr>
<td>3644 Slocan Street, Vancouver, BC V5M 3E8</td>
<td></td>
</tr>
<tr>
<td>Phone: (604) 453-830</td>
<td></td>
</tr>
<tr>
<td>Fax: (604) 453-8338</td>
<td></td>
</tr>
<tr>
<td>Email: <a href="mailto:sleepassessments@phsa.ca">sleepassessments@phsa.ca</a></td>
<td></td>
</tr>
<tr>
<td>DATE: February 5, 2015</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Attention Deficit Hyperactivity Disorder</th>
<th>Autism Spectrum Disorder</th>
<th>Down syndrome</th>
<th>Other neurodevelopmental condition Specify:</th>
</tr>
</thead>
<tbody>
<tr>
<td>CHILD’S NAME: (Gender: M F )</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

| CHILD’S CURRENT AND/OR WORKING DIAGNOSIS:                                                                 |       |
| Trisomy 21; ADHD; chronic otitis media with effusion (tympanostomy tubes in place); mild intellectual disability; chronic insomnia (sleep maintenance) & secondary behavioural insomnia most probably triggered by Willis Ekbom disease (caused by iron deficiency) and presenting as stereotyped episodes of rhythmic motor activity during sleep; status post adenoidectomy & formal sleep study; excessive daytime sleepiness and hyperactive like behaviours. |       |

| PHN:                                                   |       |
| MRN:                                                   |       |

| PRESENTING PROBLEMS:                                   |       |
| SLEEP/WAKE BEHAVIOUR ASSESSMENT                        | INTERPRETATIONS:                                                                 |
| Bedtime:                                               | has chronic sleep deprivation, in addition to difficulties with sleep maintenance that are likely multifactorial and may be the result of: |
| - Routine starts at 8:30 pm with teeth brushing, face wash and PJs; | (1) Not getting enough amount of sleep (~7-9h with interruptions); (2) Behavioral sleep interruptions: getting up from bed with night time awakenings, most probably triggered by non-restorative/light and restless sleep (see sleep study results); |
| - Reads from approx. 9 pm to 9:30 pm;                  | (3) Possible iron deficiency contributing to limb movements in sleep, presenting as restless/non-restorative sleep and causing (4) Sleep-related-movement disorder, and |
| - Falls asleep within 15 - 45 minutes of reading;       | (5) Possible Willis Ekbom Disease (Restless Legs Syndrome). Significant Sleep Disordered Breathing (SDB) has been excluded by overnight polysomnography; however results show non-restorative ‘restless’ sleep, which has to be investigated further. |
| - Requires substantial night light due to fear, not required if room sharing; | |

| Excessive Daytime Behaviours: (/Sleepiness):            |       |
| - Prior to Adderall®:                                  |       |
| > Needed frequent redirection to complete routines;     |       |
| > Fidgeting and inattention in classroom setting;      |       |
| - No daytime napping, no car napping;                  |       |
| - Significant “Excessive Daytime Sleepiness & hyperactive-like behaviours”. |       |
Awakenings:
- Wakes once/night at 0300h, ongoing for years;
- If allowed will go downstairs, make food, and play video games;
- Currently door locked at night; [ ] will knock for drink/bathroom;
  > This has improved morning wakefulness, fewer nighttime disruptions;
- Described as a very restless sleeper, sitting up, ‘folding’ and falling asleep (observed also in the formal sleep study).

Routines:
- Previously would wake spontaneously, since Sept. has needed up to 3 ‘wake ups’ when tired (after being up at night);
- Takes Adderall® shortly after waking (7:30 am).

Sleep Disordered Breathing:
- Occasional kicking movements;
- No gasps/witnessed apnea; snoring only with colds; in formal sleep study: 11 hypopnea (12-21s); 77 spontaneous arousals, indicative of non-restorative sleep (overall arousal index 12.3 per hour);
- Frequently sits upright then falls back to sleep in ‘folded’ position.

RECOMMENDATIONS:

Further Investigations:
(1) Review of formal sleep study (done);
(2) Blood work to check electrolytes (including calcium and magnesium) and for iron deficiency (requisition provided);
(3) Perform the SCIT on other family members (especially brother) as discussed in the visit;
(4) After formal sleep study & blood work results review we will determine the need for the home-based sleep assessment video.

Therapeutic Recommendations:
(1) Sleep Health Measures (see attached form); increase amount of sleep (up to 10-11 hours);
(2) Medications: Iron Supplementation (suggested initial dose: 5 mg/kg elemental iron twice a day at breakfast and lunch);
(3) Increase exercise frequency for deeper night time sleep. We will follow-up with you once the investigations are completed and the formal sleep study has been reviewed, and will review the results of your home SCIT tests at this time.

COMMENTS:

Physical examination: Ht 139 cm (<3rd %ile), Wt 45.3 kg (15-50%ile); Cardio/Resp/Abdo - normal; Neuro - cranial nerves normal, 5/5 strength UE and LE, low peripheral tone, reflexes symmetric UE and LE, gait (incl toe, heel, tandem) normal.

Observations: No fidgeting observed, unusually still. Prefers ‘yoga-like’ poses while sitting. Informal SCIT: [ ] displayed some restlessness with large movements but had few small fidgeting movements; impressed as very quiet; (mother): no fidgeting or unusual movements during SCIT.

FULL REPORT TO FOLLOW

CC: PAEDIATRICIAN/FAMILY DOCTOR: [ ]

OTHER INVOLVED PROFESSIONALS: SHHCC/BCCH Chart

Osman Ipsioglu, MD (FRCPC), PhD, Clinical Associate Professor, Paediatrics, UBC.
A.4. Outreach Sleep/Wake Behaviour Assessment Report

This is an example of an adaptation of our typical Sleep/Wake Behaviour Assessment Report (Appendix A; Section A.2.) for an outreach activity in 2014, demonstrating the complex clinical presentation of typical patients of the Sleep/Wake Behaviour Clinic and Research Lab at BC Children’s Hospital.

SUNNY HILL HEALTH CENTRE FOR CHILDREN
3644 Slocan Street
Vancouver, BC V5M 3E8
Tel: (604) 453-8300 Fax: (604) 453-8301

CHILD DEVELOPMENT AND BEHAVIOUR PROGRAM

Paediatric Assessment of Sleep – Kamloops Outreach
Collaboration with Insight Support Services &
TRU Respiratory Therapy Sleep Clinic, Kamloops

Confidential

NAME: Mallory Bagri
DOB: March 1, 2000
PHN: 9858 208 259
DATE OF CONSULTATION: March 3, 2014
FAMILY CONFERENCE: March 6, 2014
CONTACT: engebretson@shaw.ca

DIAGNOSES: Moderate Developmental Disability; Cerebral Palsy; Chronic Medical Health; Attention Deficit Hyperactivity Disorder; Oppositional Defiant Disorder; Severe Sensory Disorder; Neurobehaviour Disorder; Emotional Dysregulation (with agitation & aggression); Enuresis

PRESENTING MAIN SLEEP/WAKE BEHAVIOUR SYMPTOMS: Excessive Daytime Tiredness due to Sleep Disordered Breathing (Obstructive Sleep Apnoea probably due to upper airway narrowing; R/o Enuresis due to OSA) and Insomnia caused by familial Willis Ekbom Disease with Periodic Limb Movements in Sleep

Dear Dr. [Redacted];
Dear Dr. [Redacted];
Dear Ms. [Redacted];

Thank you, Dr. [Redacted], for referring [Redacted] to the collaborative clinical service and teaching project in Kamloops of the Sleep/Wake Behaviour Clinic of BC Children’s Hospital (University of British Columbia; located at Sunny Hill Health Centre for Children), Insight Support Services and the Sleep Clinic of the Respiratory Therapy Program at Thompson Rivers University.
This is the consultation report of Mallory Bagri who was collaboratively assessed on March 3, 2014; also present at the assessment was her mother, Ms. Gail Bagri. This report consists of 3 interconnected steps: (1) summary of sleep/wake behaviours; (2) our interpretations; and (3) recommendations for parents and results of the urgent family conference (on March 6, 2014) with Ms. Bagri and Mallory’s behavioural therapist Ms. Julie Chambers, for your consideration.

General: Mallory Bagri is a 14-year old girl living in Kamloops, BC with her parents; she is a single child. Mallory experiences significant difficulties with falling asleep, night awakenings, waking up in the morning, and excessive daytime tiredness. Ms. Bagri reports that Mallory’s difficulties with falling asleep, night awakenings, waking up in the morning, snoring, sweating during the night, and restlessness began at birth. Currently, she continues to mouth-breathe and wet her bed. The sleep problems have affected both Mallory and her parents, and Ms. Bagri feels that if Mallory’s sleep problems were resolved the quality of life of both Mallory and her parents would significantly improve.

Medications: Current medications: Abilify® (apiriprazole, 2.5mg during the day), ReVia® (naltrexone; 25mg in the morning and at 1500hr for the last 3 weeks), Largactil® (Chlorpromazine; as needed for extreme aggression), Intuniv® (Guanfacine, just, recently started). Mallory is also taking liquid iron supplements. Past medications: Ativan® (lorazepam), Biphetin® (methylphenidate extended release, caused mania and made her “extremely wild”), Catapress® (clonidine), Depakene® (valproic acid), Desyrel® (trazodone, used for 4yrs for sleep), Epival® (valproic acid), Prozac® (fluoxetine), Risperdal® (risperidone), Seroquel®/Seroquel XR® (quetiapine). Ms. Bagri noted that Mallory reacts poorly and/or is allergic to most medications and reported that “none of [the medications] worked”; the medications seemed to cause an opposite effect than intended.

Mallory was previously prescribed a contraceptive at the age of 13 because her menses was very strong (however, it had to be stopped shortly after because it resulted in persistent menstrual bleeding). Currently, Mallory’s menses is still strong now lasting 6 days, which is a normal time range.

Bedtime: Ms. Bagri describes Mallory as having severe difficulty going to bed and falling asleep every night as she is very agitated and unable to calm down. Ms. Bagri reports that Mallory refuses to go to bed as she becomes very anxious and will do anything and everything to avoid bed time, and sometimes even breaking down into rages and showing aggressive behaviours. As a consequence, Mallory needs a parent with her to fall asleep every night. It can take Mallory anywhere between 15min-3hrs (sometimes up to 5hrs) to fall asleep after she is put to bed. Ms. Bagri reports that Mallory’s problem of going to bed or falling asleep has a severely negative impact on herself as well.

(Excessive Daytime) Sleepiness/Behaviours: Ms. Bagri reports that Mallory is extremely tired and edgy at school. Nevertheless, Mallory will not take naps during the day. Mallory is described as being significantly overtired every night, which also has a significant effect on her mother’s wellbeing as well: Mallory becomes manic and hyperactive before bedtime, 3-4 times per week during which time she targets her mother in a physical manner and tries to hurt her. These episodes are very dangerous and frightening for Ms. Bagri.

(Nightime) Awakenings: Mallory wakes up almost every night (sometimes 2-3 times per night). Ms. Bagri describes that Mallory’s awakenings during the night are significant to her but that there have also been rare, severe periods during which Mallory woke up and could not fall back asleep again for five hours. However, during the last weeks Mallory’s awakenings during the night have improved in comparison to before, although they are still significant and cause significant problems for her parents. Mallory’s parents can hear her talking in her sleep every night and sometimes
shouting out "mom" or "Maggie" (their dog). No night terrors, teeth grinding or pain interfering with sleep are reported.

Sleep Disordered Breathing: Shows several pathologic symptoms: she snores every night, breathes through her mouth, wets the bed, sweats profusely, and tosses and turns constantly. She sweats to the point that her pyjamas are soaked, and is so restless that she has bruises on her legs from banging into the walls and falling on the floor. Describes sleep disordered breathing in general as moderate, but mentions that has a persistent dry mouth every night, which results in coughing.

Routines, Regularity, Timing and Sleep Logs: Aims to start preparing for bed between 7:30pm and 8:30pm. It takes at least till 10pm to fall asleep every night. Believes that does not get enough sleep in general and shows a very irregular sleeping pattern ranging anywhere from 2-10 hours. Sometimes when is very agitated and aggressive before going to bed, will give chlorpromazine to her which helps her to fall asleep. During the two week sleep log, showed irregular sleeping pattern as well. Sometimes, she got up very early without falling asleep again anytime later. Missed three days of school during those two weeks of sleep log.

Functional Diagnoses: Describes the following functional diagnosis that fit for difficulties with achievement, attention, executive functioning, memory, gross motor skills, social communication, adaptive behaviour, cognition, language, fine motor skills and sensory problems. Has been diagnosed with the following mental health diagnosis: Psychotic Disorders, Generalized Anxiety Disorder, Mood Disorder, Attention Deficit Disorder, Sleep Disorder, Reactive Attachment Disorder, Oppositional Defiant Disorder, Attention Deficit Hyperactivity Disorder, moderate developmental disability, Cerebral Palsy, Neurobehaviour disorder, severe Sensory Processing Abnormalities, and emotional dysregulation.

Family Ecology and Goals: ’s family used several resources to improve ’s sleep: community organizations, other family members and friends, respite care, and medication. Notes that communication between family members is hard pressed but they do their best to work together. Nevertheless, ’s mother reports that when spends the night with her grandparents her behaviours are worse because she stays up longer (they give her sweet treats before she goes to bed). ’s major stress sources include difficulties related to ’s disability, and she notes that they don’t have much social support. Goals for and her family include improved sleep quality for everyone and less awakenings/more solid sleep throughout the night.

Physical Exam: Weight: 62kg (75-90 percentiles); Height: 163cm (50-75 percentiles); BMI: 23.3 (75-90 percentiles). Significant retrognathia and over-jet; Mallampati Score: 3-4; brief neurological exams reveals strength +++; no further clinical assessment in order not to increase the patients’ significant distress. Impressed as an anxious and very distressed patient.

Interpretation: We strongly believe that some of ’s daytime challenges and co-morbid psychiatric disorders, e.g. anxiety, are aggravated by her chronic sleep deprivation, due to non-restorative sleep (signified by her waking up tired and being very “edgy” and aggressive over the day) and that is chronically sleep deprived. Shows significant signs of Sleep Disordered Breathing (snoring, mouth breathing, enuresis, diaphoresis or excessive sweating, excessive daytime sleepiness, hyperactive like behaviours and aggressive behaviours and restless sleep with Periodic Limb Movements in sleep secondary to SDB and WED), which is a risk factor for poor quality sleep. Along with SDB she has significant signs of familial Willis Ekbom Disease (familial as describes Mr. to have restless legs syndrome with characteristic movement patterns). ’s descriptions that is a very restless sleeper that kicks while sleeping fit very well with WED with Periodic Limb Movements in Sleep (PLMs). WED is
characterized by discomfort (up to pain) of feet, legs, hands, and/or other body parts, which is relieved by movements. In majority of the cases WED is accompanied by PLMs. The urge to move increases in periods of rest and towards the evenings, and seems to be aggravated mainly upon falling asleep. Iron deficiency, among other causes, can cause or worsen WED presentation.

Recommendations for Consideration. Further investigations:

1. **Video-somnography**: In order to determine the current baseline for Mallory’s sleep fragmentation, and optimize our clinical understanding, we recommend completing an HBSA-video for 2 to 3 nights. *Our team will provide this as an urgent step for optimizing our clinical understanding and being able to support Dr. [Redacted] in her clinical decision making process. The videosomnography will be conducted ASAP.*

2. **Formal Sleep Study**: To determine the dimension of Mallory’s sleep disordered breathing she requires a formal sleep study. At the family conference we discussed how the concept of a sleep study in the hospital setting can be introduced to [Redacted] and agreed that Ms. [Redacted], behavioural therapist, will work and ‘prepare’ [Redacted] for supporting Ms. [Redacted] activities. As soon as [Redacted] seems to be ready for this clinical investigation a lab study will be offered; *we have initiated the referral.*

3. **ENT / Dentist / Orthodontist Assessment**: We recommend a comprehensive ENT (query adenoidectomy/tonsillectomy), Dentist (query jaw positioning appliances) / Orthodontist (query optimizing airway management) assessments for working out the conventional versus surgical upper airway management options. As an experienced dentist working with jaw positioning appliances you may consider a referral to Dr. Edmund Liem (Burnaby).

4. **Blood work**: We recommend for Mallory to obtain a blood work to test for iron deficiency, as there is a known association between iron deficiency and WED (test for iron, TIBC, ferritin, transferrin, transferrin saturation, CRP, haematology panel, TSH, T4 Free, calcium, and magnesium).

Therapeutic recommendations:

5. **Medications:**
   a. Should the blood work show an iron deficiency – the American Academy of Sleep Medicine recommends that all symptomatic children/adolescents/adults with ferritin levels below 50 mcg/l should be supplemented with iron, **Suggested iron supplementation**: Usual iron replacement dosing of 3-6mg/kg/day of elemental iron to be taken twice a day, at breakfast and lunch, in order to decrease the possibility of gastrointestinal irritation (*Weight 62 kg x 4mg = 248 mg; suggested initial dosage*). *We recommend giving [Redacted] a total of 250 mgs of elemental iron (Fe) supplementation, which would be 125 mg twice a day.* It will be helpful to take the iron supplement with orange juice (not milk) in order to increase its absorption.

   b. We suggest starting low-dose gabapentin, a neuropathic pain medication, to help with the discomfort caused by WED and to help improve Mallory’s difficulties with sleep maintenance. Suggested gabapentin dosage: 200 mg for three nights at dinner time to enable parents to observe the effects of the medication on [Redacted]; if well tolerated (less tension, less agitation) increase to 300 mg for one week and then decide whether a higher dosage 350 or 400 mg maybe necessary. [Redacted] may also benefit from a low dose (e.g. 100 mg) mornings and possibly noon times.
c. To help optimize Mallory’s breathing (reduce any allergic inflammation), we recommend Nasonex® Nasal Spray (Mometasone furoate monohydrate). The spray is to be administered to the nostril aiming to the lateral side with one spray in each nostril every night for 3 weeks. Should the Nasonex® be helpful, we recommend that she continues with the spray for 3 months.

6. Given the behavioural Adverse Drug Reactions Mallory has presented in the past to several medications (e.g. Biphentin® (methylphenidate extended release) caused mania and made her “extremely wild”), we have initiated a referral to the Division of Translational Therapeutics at BCCH for pharmacological advise and further investigations.

Community Support for Implementation:

7. Goal Attainment Scaling: We would suggest Mr. and Ms. Bagri in collaboration with Ms. Julie Chambers to develop a list of ranked goals and concerns (i.e. regarding Mallory’s sleep and medication); see attached Goal Attainment Scaling form. This will help to evaluate the effects of the various medications in a personalized manner (to ‘titrate’ the dosage) and monitor the therapeutic plan for Mallory – if optimizing sleep reduces Mallory’s challenging and disruptive behaviours further medication changes will be discussed. The Division of Translational Therapeutics at BCCH is the address for reviewing pharmaceutical management.

8. Community Involvement: As initially stated we had an urgent family conference on March 6th and discussed with Ms. Bagri and Ms. Chambers how to prepare Mallory for the next assessments, reduce her anxiety and navigate through the medical health care system.

In summary, Mallory is a lovely young girl with a very complex medical history, including chronic and ongoing sleep problems due to significant Sleep Disordered Breathing and Willis Ekbom Disease both associated with PLMs. We strongly believe that the chronic and ongoing sleep deprivation amplifies many of the co-morbidities Mallory experiences. Therefore, for us, addressing the sleep problems in a structured way appears to be worthwhile. Thank you for involving us in the care of this patient. It was a pleasure to meet Mallory and her mother, Ms. Bagri.

Sincerely,

[Signatures]

Osman Ipsiroglu, MD (FRCPC), PhD
Clinical Associate Professor
Department of Paediatrics, Faculty of Medicine, UBC
Adjunct Professor, Faculty of Science, TRU

In collaboration with,

Les Matthews, RRT(A), MA
Associate Professor of Respiratory Therapy
Faculty of Science, TRU

CC:
Mr. & Mrs. parents
Dr. Denise Chapple, Paediatrician
Dr. Sheik Hosenbocus, Psychiatrist
Dr. Susanna Amalia De Wet, GP
Ms. Julie Chambers, Insight, Kamloops
Ms. Carla Monson, RT
Further community-based team members via Mrs.
Dr. Division of Translational Therapeutics
A.5. Immediate Abbreviated Report – Medication History

This is an Immediate Abbreviated Report, from 2015 similar to Appendix A; Section A.3, which shows the complexity of the medication history typically seen at the Sleep/Wake Behaviour Clinic and Research Lab at BC Children’s Hospital. Note that the family started to medicate the patient with alcohol.

---

**ASSESSMENT OF SLEEP/WAKE BEHAVIOURS**

**Immediate Abbreviated Report**

**Team Dr. Ipsirolgu**
Sleep/Wake Behaviours Clinic located at SHHCC
3644 Slocan Street, Vancouver, BC V5M 3E8

**DATE: July 2, 2015**

**CHILD’S NAME:**

- **Gender:** M F

**BIRTHDATE:**

June 28, 1996

**CHILD’S CURRENT AND/OR WORKING DIAGNOSIS:**

- Migraines, fainting (ages 10-12), Anxiety, self-reported OCD-like behaviours.
- Diagnosed 2014 by Dr. [Redacted] ADHD - Inattentive Type, Sleep Disorder NOS.
- Diagnosed 2015 by Dr. [Redacted] ADHD - Combined Type, Major Depressive Disorder, Generalized Anxiety Disorder, Math Learning Disability. Working hypothesis: Significant WED/RLS, causing insomnia and triggering co-morbidities & CRSD

**MEDICATIONS/THERAPIES:**

- **Current:** Fluvoxamine; Quetiapine (started 1 month ago) causes 2-hour period of “deliberating tiredness”, improves sleep onset but not maintenance, increased appetite but no weight gain yet; Zopiclone (started 1 year ago; mod. effective for sleep onset). Recently family agreed on self-medication with alcohol at bedtime.
- **Previous:** OTC medications (Gravol, Benadryl, melatonin) ineffective for sleep in childhood; Fast-acting Ritalin (trialed in childhood) caused anxiety; Ativan (trialed 3 years ago); Fluoxetine (trialed 3 years ago, caused severe depression and nosebleeds); Concerta (increased depression); Vyvanse caused initial euphoria but “crippling depression” when wearing off, stopped after 2 weeks.

**PRESENTING PROBLEMS:**

**SLEEP/WAKE BEHAVIOUR ASSESSMENT**

**INTERPRETATIONS:**

**Bedtime:**

- Severe difficulty falling asleep;
- Goes to bed around 4-5am (takes 2-3 hours to fall asleep if bedtime is earlier).
- Developed Circadian Sleep Rhythm Disorder (CRSD) not suitable for school schedule (home schooled).

Chronic Familial Willis Ekboth disease [WED/Restless Legs Syndrome]: [Redacted]'s history of insomnia from an early age and significant sensory processing issues / urge to move and associated day- and nighttime restlessness (including positive SCIT), suggest that Willis Ekboth disease has disturbed her sleep since an early age.

It is familial as Ms. [Redacted] reports significant symptoms (+ previous diagnosis).

Our understanding is that WED/RLS is currently not the primary
**Excessive Daytime Behaviours: (/Sleepiness):**
- Wakes around 3:00-4:00pm;
- Often feels "un-rested" and tired in the morning;
- Does not nap or fall asleep once awake during the day.

Concern, but has contributed to [insert name]'s insomnia, circadian sleep rhythm disorder and triggered the co-morbidities. The challenge to the diagnosis is that [insert name] presents as a sensitive personality and apparent tendency to self-blame - causing her presentation to match her diagnosis.

Possible Dysautonomia as suggested by the history of fainting, migraines, high heart rate (vegetative symptoms), and pale skin.

**Awakenings:**
- Light sleeper (sensitive to sounds);
- Frequent nighttime awakenings;
- Under tension during sleep (wakes stiff/sore);
- Restless: "wiggly" when co-sleeping in childhood, currently moderately restless (bed is sometimes messed in the morning).

**RECOMMENDATIONS:**

**Further Investigations:**
(1) Review of formal sleep study (done);
(2) Blood work to check electrolytes (including calcium and magnesium) and for iron deficiency (requisition provided);
(3) Perform the SCIT on other family members (especially brother) as discussed in the visit;
(4) After formal sleep study & blood work results review we will determine the need for the home-based sleep assessment video.

**Therapeutic Recommendations:**
(1) Sleep Health Measures (see attached form); increase amount of sleep (up to 10-11 hours);
(2) Medications: Iron Supplementation (suggested initial dose: 5 mg/kg elemental iron twice a day at breakfast and lunch);
(3) Increase exercise frequency for deeper night time sleep.

We will follow-up with you once the investigations are completed and the formal sleep study has been reviewed, and will review the results.

**Awakenings:**
- Light sleeper (sensitive to sounds);
- Frequent nighttime awakenings;
- Under tension during sleep (wakes stiff/sore);
- Restless: "wiggly" when co-sleeping in childhood, currently moderately restless (bed is sometimes messed in the morning).

**Routines:**
- Bedtime and morning wake time varies significantly;
- Nearly inverted sleep rhythm: CRSD!

**Sleep Disordered Breathing:**
- No snoring;
- Sometimes dry mouth when waking (likely Zopiclone side-effect);

**COMMENTS:**
Sleep History: Colicky baby, falling asleep difficulties in infancy (slept in a swing/hammock), nighttime awakenings and need for co-sleeping started at age 2.5, late sleep onset (1 am) started at age 8, mental health difficulties worst at age 18 (1 year ago). Physical Exam: heart rate 84 (slightly high), teeth impressions on tongue, overbite. Abbr. neurologic exam clear. Positive SCIT (reports "tingly sensation" in calves relieved by movement). Family History: Mother reports history of mild insomnia + "night owl" pattern, currently 4-5 hours sleep per night, reports preference for sleeping in recliner with feet raised, takes trazadone. Sensory Processing: likes light skin pressure/tickling, nail-biting, sensitive hearing, reports internal "tense" feeling and difficulty relaxing, high pain threshold.

**FULL REPORT TO FOLLOW**

**CC:** PAEDIATRICIAN/FAMILY DOCTOR: [insert name] (referring paediatrician)

**OTHER INVOLVED PROFESSIONALS:** Family will distribute report as needed (GP)

Osman Ipsioglu, MD (FRCPC), PhD, Clinical Associate Professor, Paediatrics, UBC.