SUPPORTING DECISION-MAKING IN WHOLE GENOME/EXOME SEQUENCING: PARENTS’ PERSPECTIVES

by

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Abstract

Whole genome sequencing/exome sequencing (WGS/ES) technology is becoming more affordable and accessible, and will become more frequently used in various clinical settings, including for diagnosing rare childhood diseases. However, its use means that parents face decisions that could uncover life-altering information, unrelated to their child’s illness that may also have personal and ethical implications for their families. The purpose of this study is to explore and describe parents’ perceptions of their decisional needs when deciding on WGS/ES for their child.

The qualitative methodological approach known as Interpretive Description, the concept of shared decision-making and the Ottawa Decision Support Framework were used to inform and guide this study. Parents of children who had previously undergone WGS/ES informed consent were invited to participate in a focus group or individual in-person or telephone interviews. Parents had children with a range of undiagnosed conditions suspected to be genetic in origin. 15 parents were interviewed and transcriptions were analyzed concurrently and iteratively. Repeat interviews were conducted with 5 of the parents to confirm, challenge or expand on the developing conceptualizations.

Participants felt that their decision to proceed with WGS/ES for their child was easy. However, they expressed many unresolved decisional needs including: a lack of knowledge about certain topics that became relevant and important to them later, unmet expectations, and a need for more support and resources. Participants also acknowledged that the high volume of information and urgency of their circumstances may have caused them to be less receptive (or even unreceptive) to information during their WGS/ES decision-making (DM) process. Additionally, participants had ongoing informational and psychosocial needs beyond the single clinical encounter where their WGS/ES DM occurred. The content and amount of information that participants considered to be important varied.

Prior to the widespread use of clinical WGS/ES, parents’ perspectives about their decisional needs should be considered in order to implement parent-tailored education, counselling, decision support and informed consent processes.
Preface

The researcher (KCL) designed the entire study with review and feedback from the thesis supervisory committee (Dr. Bernie Garrett (BG), Dr. Maura MacPhee (MM), Dr. Jan M. Friedman (JMF) and Patricia Birch (PB)) and other consultants (Shelin Adam (SA), Dr. Anne Townsend (AT)). KCL recruited all the participants and conducted eleven individual interviews and five repeat interviews. An experienced moderator (AT) and KCL both facilitated the focus group. KCL transcribed the focus group session, five of the interview transcripts, checked all transcripts for accuracy and analyzed all of the data.

This research study was done in collaboration with the Friedman Lab, Department of Medical Genetics, University of British Columbia. KCL conducted the entire study and wrote the thesis from Chapter 1 to 5. The thesis committee reviewed and provided feedback on KCL’s work after each chapter. This research was approved by the University of British Columbia Children’s and Women’s Research Ethics Board, certificate number CW13-0005/H12-03121.

An e-poster resulting from this work was presented online at http://issuu.com/inspirenetbc/docs/li_vs2a/0.

The researcher certifies that she is the sole author of this thesis and that no part of this thesis has been published or submitted for publication. KCL certifies that, to the best of her knowledge, her thesis does not incorporate, without acknowledgement, any materials previously submitted for a degree or a diploma in any institution of higher education. KCL certifies that her thesis does not contain any material previously published or written by another person except where due reference is made in the text. KCL declares that this is a true copy of her thesis.
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List of Abbreviations

ACMG: American College of Medical Genetics and Genomics
DM: Decision-making
DNA: Deoxyribonucleic acid
ES: Exome Sequencing
HCPs: Health Care Professionals
IFs: Incidental Findings
ODSF: Ottawa Decision Support Framework
SDM: Shared Decision Making
VUS: Variants of Unknown Significance
WGS: Whole Genome Sequencing
Definition of Terms (Glossary)

*Bolded and italicized* words in the body of the thesis indicate a matching entry and definition in the Glossary.

**Closed communication:** In Hall et al.’s (2012) study, closed communication meant that parents avoided talking about the child with a disability. This type of communication was associated with situations when parents were highly stressed (Hall et al., 2012).

**Coles Notes:** In Canada, Coles Notes are guides that help students study literature or other materials in a condensed manner. The saying, “Coles notes”, has become an expression for a simplified version of instructions, descriptions, or a complex book.

**Decision aids:** Decision aids are “evidence-based tools to prepare people to participate in making specific and deliberate health care choices among options in ways they prefer” (O’Conner, 2006, p.3).

**Decision coaching:** Decision coaching is a type of support that a trained facilitator provides to a person facing a decision. This facilitator is someone who is supportive but neutral about the decision. Decision coaching can be provided through varying methods (in-person, telephone, online) and can be used alone or coupled with decision aids. Decision coaching includes the following strategies: a) clarifying decision and monitoring needs; b) facilitating access to evidence-based information, verifying understanding, clarifying values, building skills in deliberation, communication, and accessing support; and c) monitoring and facilitating progress in decision making and decision quality (O’Conner, 2006).

**Decisional conflict:** Decisional conflict is a person’s state of uncertainty about the course of action to take, when choice amongst options involves risk, loss, regret and challenge to personal life values (O’Conner, 2006).
**Decisional needs:** Decisional needs are one of the three key elements in the Ottawa Decision Support Framework (ODSF; O’Connor, Stacey & Jacobsen, 2011). Decisional needs are the entities that influence individuals’ decision-making, which include the type, timing, and stage of a decision, the presence or absence of decisional conflict (i.e., uncertainty about the course of action and related to feeling uninformed), one’s knowledge and expectations, one’s values, one’s support and resources, one’s personal and clinical characteristics (O’Conner, 2006). Decisional needs should be met or resolved, otherwise there will be adverse effects on the quality of the decision(s) (O’Connor, Stacey & Jacobsen, 2011).

**Decision support:** Decision support is another key element in the Ottawa Decision Support Framework (ODSF; O’Connor, Stacey & Jacobsen, 2011). Decision support can be provided in the form of clinical counselling, patient decision aids or coaching, and is personalized to the person’s decisional needs (O’Connor, Stacey & Jacobsen, 2011). Providing decision support involves clarifying the decision, the person's needs and values, providing facts and probabilities, guiding in deliberation and communication, and monitoring/facilitating progress in decision-making (O’Connor, Stacey & Jacobsen, 2011).

**Decision quality or high-quality decision:** Decision quality is another key element in the Ottawa Decision Support Framework (ODSF; O’Connor, Stacey & Jacobsen, 2011), and it is the extent to which the informed patient or participant has chosen the option that is most congruent with his/her perceived values for benefits, harms and scientific uncertainties (O’Connor, 2006; O’Connor, Stacey & Jacobsen, 2011). To make high-quality decisions means that the decisions were informed by the best available evidence and based on the person’s values (O’Connor, Stacey & Jacobsen, 2011).
**Decision support needs:** In this study, decision support needs are defined as entities that help individuals gain relevant information and clarify their values about the decision they are trying to make (Jackson, Cheater, & Reid, 2008). Decision support needs are influenced by the type, timing, and stage of a decision, the presence or absence of decisional conflict (i.e., uncertainty about the course of action), one’s knowledge and expectations, one’s values, one’s support and resources, one’s personal and clinical characteristics (O’Conner, 2006).

**Diagnostic odyssey:** In an attempt to find the cause of a condition, people undergo many repeated diagnostic tests for different conditions (Friedman et al., 2006).

**Exome sequencing (ES):** ES is the sequencing of an individual’s exome (the protein-coding portion of all the genes), or about 1% of a person’s entire genetic information (Biesecker & Green, 2014). Most of the known genetic variants that are clinically relevant are located in the exome (Wright, Middleton & Burton, 2013).

**Incidental findings:** Incidental findings are unanticipated or secondary findings that are unrelated to the condition or indication for which WGS/ES was performed (ACMG, 2012).

**Mendelian cause:** A mutation in a single gene can cause a genetic condition that is inherited according to Gregor Mendel's laws of inheritance.

**Parents:** In this study, parents refer to adults who have a child with a suspected genetic condition, and who: have considered WGS/ES for their child, had children who are candidates for (or have been offered) WGS/ES, or have already consented to WGS/ES for their child. All parent(s) and children included in the study have provided consent (or assent, for minors) to obtain WGS/ES for research or clinical purposes.

**Patient agency:** Patient agency refers to a patient’s perceived ability to be active and capable in managing his/her own health (Street et al., 2009).
**Therapeutic alliances:** Therapeutic alliances are the interrelationships among the patient and other parties (HCPs, friends, family or caregivers) that positively affect patient health outcomes (e.g., emotional well-being, satisfaction with decisions) (Street et al., 2009). Mutual trust among all parties, coordinated and continuous health care, and the patient’s perception of feeling respected and cared for are some indicators of a strong therapeutic alliance (Street et al., 2009).

**Whole genome sequencing (WGS):** WGS is the sequencing of an individual’s entire genome (Conley et al., 2012), although, for technical reasons, only 90% of the genome is evaluated (Biesecker, 2012).

**Variants of unknown significance (VUS):** VUS are genetic variants that currently have unknown clinical importance or meaning.
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Chapter 1: Introduction

Major advances in genomic technology have enabled the transition from single-gene diagnostic tests for specific genetic disorders to broader examinations of genetic material by the clinical genetics community (McGuire, Cho, McGuire, & Caulfield, 2007). This broader examination of genetic material includes technologies such as whole genome sequencing (WGS) and exome sequencing (ES). WGS targets the entire genome and provides information on almost all of a person’s genetic information (Biesecker & Green, 2014). ES examines the exome (i.e., the protein coding portions of genes) and provides information about 1% of a person’s DNA (Biesecker & Green, 2014). WGS and ES are being used to diagnose rare or unrecognizable conditions that are suspected to be genetic in origin (Biesecker & Green, 2014).

In 2001, the estimated cost per genome test was $95,263,072; in only thirteen years (2014), costs have fallen to $4920 per whole genome test (National Human Genome Research Institute (NHGRI), 2014). More recently, the company Illumina has made the thousand dollar genome possible (Sheridan, 2014), enabling some companies and university genome centres to sequence whole genomes for a thousand dollars each. The costs of WGS and ES are already comparable to some clinically available tests (Biesecker, 2012), and both technologies are used in clinical care (Biesecker & Green, 2014).

WGS/ES increases the extent of genetic testing, from a small number (one to a few) of genes – those suspected as the cause of a particular condition – to all of the genes at once (Ormond et al., 2010). As WGS/ES offers a vast amount of information, such testing presents both opportunities and challenges for health care professionals (HCPs) and the public. WGS/ES has been shown to be an effective alternative to gene specific or gene-panel tests in discovering the genetic basis of disease (Bainbridge et al., 2012; Bamshad et al., 2012; De Ligt et al., 2012;
Need et al., 2012; Worthey et al., 2011). Unfortunately, WGS/ES is also complex, often difficult to interpret, and can pose serious ethical, social and practical challenges (Townsend et al., 2012). Health care professionals who offer WGS/ES may be challenged as they discuss the benefits and potential adverse implications of genomic testing with patients (Clayton, 2003). WGS/ES results are often sought for patients who are children, so it is frequently parents who have to make the decision on WGS/ES. Families will increasingly require accurate, sufficient, and meaningful information about the nature and implications of WGS/ES in order to make an informed decision about whether or not to proceed with testing (Ormond et al., 2010).

Clinical literature on the Canadian public’s perspectives on the decision-making process and whether to proceed with WGS/ES testing is limited. Specifically, we have little understanding of parents’ perceptions of the tests, and their decisional needs when making a choice for their child. The purpose of this study is to examine parents’ perspectives on their decisional needs prior to deciding whether or not their children will receive WGS/ES testing. Understanding their viewpoints will enable HCPs to improve the education, counselling, informed consent, and other processes for parents considering such testing for their child.

1.1 Background to the Problem

Single-gene sequencing is used to analyze genes suspected of causing a particular undiagnosed disorder (American College of Medical Genetics and Genomics (ACMG), 2012). Single-gene sequencing is a powerful technique for diagnosing patients with rare heritable conditions, but it has limitations (Wright, Middleton & Burton, 2013). A HCP has to select one gene to test for – from the roughly 20,000 genes in the human genome – on the basis of clinical suspicion (Wright, Middleton & Burton, 2013), and there exist disorders for which mutations of many hundreds of different genes can produce a very similar clinical picture. There is
considerable interest in offering more robust technologies (such as WGS/ES) on a clinical basis, because testing single genes sequentially can be slow, expensive, labour intensive, and often unsuccessful (ACMG, 2012; Wright, Middleton & Burton, 2013).

The major advantage of WGS/ES is that it can sequence nearly all the genes in a person’s genome through just one test (ACMG, 2012), greatly reducing the time and expense of diagnosing a genetic disease when a precise genetic diagnosis is not apparent (Ayuso et al., 2013). In clinical practice, making a precise genetic diagnosis of a disease provides the patient an immediate benefit, especially for rare diseases (Ayuso et al., 2013). In Canada, WGS/ES is not yet the standard of care in clinical practice, and is predominantly used in the research context (Kleiderman et al., 2013). Some clinicians and researchers have focused their attention on rare disorders in Canadian children through a consortium called Finding of Rare Diseases in Canada (FORGE) (Beaulieu et al., 2014). Through WGS/ES, children with undiagnosed conditions may have a better chance of receiving a definite diagnosis, and more efficient care (Ayuso et al., 2013; Welch et al., 2011); HCPs can also provide more accurate counselling, prognosis, and care management with the test results (Regier, Friedman & Marra, 2010). For example, the cause of intellectual disability is unknown in one-third to one-half of all affected individuals (Regier, Friedman & Marra, 2010). Children with intellectual disabilities often endure a “diagnostic odyssey” of multiple diagnostic tests in an attempt to find the cause (Friedman et al., 2006), but ES can provide a specific genetic diagnosis in at least 25% of these patients (Yang et al., 2013).

While WGS/ES can provide families with the cause of their child’s health condition, it can also reveal incidental findings (IFs). IFs are unanticipated findings that are completely unrelated to the problem for which the test was performed (Ormond et al., 2010). WGS/ES can reveal unsuspected genetic variants that cause, or predispose a patient to conditions ranging from
trivial to those much more severe than what led the patient to seek medical consultation. In some cases, effective interventions are available to prevent the onset of these conditions, or ameliorate their symptoms. Other times, nothing can be done except to prepare the patient psychologically, and to help him/her plan their future accordingly.

In addition, the functions of more than half of our approximately 20,000 genes are still unknown; in many cases of WGS/ES, genetic \textit{variants of unknown significance (VUS)} are also found (ACMG, 2012). VUS are genetic variants that currently have unknown clinical importance or meaning. As the number of WGS/ES tests performed increases, so will the number of patients with uninterpretable findings and IFs (Wolf et al., 2008; Van Ness, 2008). Thus, although WGS/ES can offer significant clinical and health benefits, handling and disclosing IFs will continue to be a challenge for HCPs (Townsend et al., 2012).

If a family decides to proceed with WGS/ES for their child, the test can reveal significant IFs related to the child’s current health status and future health risks, as well as those of the parents, should they also be tested (Ayuso et al., 2013). The parents and their affected child are often tested together; this allows for comparisons between the two sets of genetic material, which can aid in analysis (Ku et al., 2013). More knowledge of genetic information can negatively or positively affect individuals’ and families’ life decisions or coping strategies (Ormond et al., 2010). For example, WGS/ES could reveal that a parent is at risk for an untreatable disease; there is also the possibility of receiving a false negative or false positive result. Other information that WGS/ES can reveal includes risks for psychiatric disorders or behavioural traits (Ormond et al., 2010), ancestry & family relationships (e.g., non-paternity, covert adoption), and normal traits (e.g., intelligence). Furthermore, because WGS/ES will often indicate that a person has an above-average risk for some diseases, or may have children with particular genetic conditions, every
person choosing to do WGS/ES could potentially face consequences from finding out those risks (Ormond et al., 2010). The ramifications may include alteration of people’s reproductive choices, increased difficulty obtaining insurance or employment, and the experience of stigma (Ormond et al., 2010). Thus, the decision to proceed (or not) with WGS/ES should integrate many considerations, including: its impact on families, potential discovery of non-paternity results, the identification of treatable and non-treatable findings, the possibility of false positive or false negative results, implications for reproductive choices, effect on insurance or employment, and the potential for stigma (Bick & Dimmock, 2011; Ormond et al., 2010). The numerous potential implications of WGS/ES may lengthen and complicate the decision-making process, and present challenges for HCPs as well as for families considering such testing (Ormond et al., 2010). American and European recommendations (Green et al., 2013; Van El et al., 2013) and a Canadian proposal (Zawati et al., 2013) have helped outline some of the genetic HCPs’ responsibilities, but more input is needed from patients, parents and public groups.

Parents of children offered WGS/ES testing have had their perspectives on the current, informed consent processes brought to light, even though these perspectives are situated in specific clinical or research settings in the United States (Tabor et al., 2012; Levenseller et al., 2013). These parents expressed specific concerns with the decision-making process, including the complexity of WGS/ES, the potential risks, (Levenseller et al., 2013), and the lengthy informed consent process (Tabor et al., 2012). Much of the recent literature focuses primarily on the public, research participants’, or parents’ perspectives about the return of results or IFs (Appelbaum et al., 2013; Bennette et al., 2013; Bollinger, Scott, Dvoskin & Kaufman, 2012; Daack-Hirsch, Driessnack, Hanish, Johnson, Shah, Simon & Williams, 2013; Facio et al., 2013; Facio, Brooks, Loewenstein, Green, Biesecker, & Biesecker, 2011; Harris et al., 2012; Murphy
et al. 2008; Sapp, Dong, Stark, Ivey, Hooker, Biesecker & Biesecker, 2013; Shahmirzadi et al., 2013; Townsend, Adam, Birch, Lohn, Rousseau & Friedman, 2012; Wright, Lewis, Fisher, Hooker, Emanuel, Biesecker & Biesecker, 2013), and not specifically about what support parents think they need in order to make an informed decision about WGS/ES.

1.2 Problem Statement

WGS/ES technology is getting more affordable and accessible, and will become part of routine healthcare (Bick & Dimmock, 2011). Given that WGS/ES offers a wealth of information that has numerous personal, social, and ethical implications (Ayuso et al., 2013), parents deciding for their children may need more support during the decision-making process in order to make informed, high quality choices. These parents face decisions that could uncover life-altering information, and understanding their perspectives on the decision-making process seems essential. Little is known about the support framework required by parents, including the content of information, the timing of its delivery, and methods of patient education. Exploring their decisional needs will help inform current decision-making processes, and support the development of better decision support tools.

There exists inadequate work on exploring Canadian parents’ perspectives in either clinical or research settings. Canada’s legal principles, publically funded health care system, and focus on patient/family centered care (British Columbia Ministry of Health, 2013) may create a different context compared to the environment in the United States. This study offers an exploration of the parents’ perspectives, situated in a Canadian research and clinical context.

1.3 Purpose

The purpose of this study is to explore and describe parents’ perceptions of their decisional needs when deciding on WGS/ES for their child.
1.4 Research Questions

In this study, the central question to be answered is: What are the perceptions of parents whose children have experienced genetic testing, regarding their decisional needs related to undertaking WGS/ES for their children?

In addition to this, the researcher seeks to explore the following sub-questions:

I. What information do parents consider important to their decision-making on WGS/ES for a child with suspected genetic disease?

II. How do personal (e.g., values, beliefs, knowledge, expectations, resources), social (e.g., relationships, support), and contextual factors (e.g., circumstances, type and timing of decisions) impact parents’ decision-making process?

III. When making a decision about WGS/ES, what type of support would parents find helpful?

1.5 Definition of Terms

Bolded and italicized words in the body of the thesis indicate a matching entry and definition in the Glossary.
Chapter 2: Literature Review

2.1 Trends in Canadian Health Care: “Patients as Partners”

In British Columbia (BC), key health care goals include providing quality clinical services, and enhancing patient and HCP experiences, while sustaining an accessible and publically funded health care system (Province of British Columbia, 2013). Some activities that reflect the work being done in BC are patients and families assuming the role of partners in their care, patients and HCPs receiving key information to make informed decisions, and the increasing use of available decision support to help manage patient populations (British Columbia Ministry of Health, 2007). The current trends in Canadian health care are building a strong foundation of health – with patients and families at the centre of care (British Columbia Ministry of Health, 2013) – and shared decision-making (SDM) (Légaré, Stacey & Forest, 2007). SDM is a collaborative approach that engages patients and HCPs to make health decisions together; it is fundamental to patient-centered care, informed consent (Jull, Stacey, Giles, & Boyer, 2012), and quality and safety in health care (Godolphin, 2009). SDM decreases the power asymmetry between HCPs and patients by increasing the patients’ knowledge, and sense of autonomy and control over decisions that affect their health and well-being (Charles, Gafni & Whelan, 1997). To help inform decision-making, SDM advocates for a two-way information exchange between the HCP and patient (Charles, Gafni & Whelan, 1997). This information exchange factors in information relevant to the decision, the HCP’s expertise, research evidence, and the patients’ wider values system and goals, beliefs, and existing knowledge (Charles, Gafni & Whelan, 1999).
2.2 Shared Decision Making and WGS/ES

Given the complexity of WGS/ES, the wealth of genetic information it produces, and its potential to change lives, parents of children considering WGS/ES should collaboratively deliberate with HCPs in order to make high quality decisions. **High quality decisions** indicate that the parent’s knowledge, perceptions of the possible outcomes and associated probabilities, and final decision are all aligned with their values (Légaré, Stacey & Forest, 2007; Volk, & Llewellyn-Thomas, 2012). The detailed examination of the human genome may lead to more complex decision-making and consent processes for both parents and HCPs (Netzer, Klein, Kohlhase, Kubisch, 2009; Ormond et al., 2010). Parents and HCPs need to thoroughly discuss all relevant benefits, risks, and implications of WGS/ES testing, and to consider what other information or support may be necessary to enable parents to make informed decisions that are right for them and their families (Biesecker et al., 2012; Ormond et al., 2010).

In contrast to some other HCPs, genetic counsellors and clinical geneticists traditionally adopt a non-directive approach in counselling their patients (Elwyn, Gray & Clarke, 2000). To be non-directive means to provide accurate information, and to help patients arrive at the best decision without steering them towards any particular choice (Elwyn, Gray & Clarke, 2000). However, there have been arguments for and against non-directiveness; support for more flexible approaches to genetic counselling – based on patients’ and families’ needs, values, circumstances, and desired counselling outcomes – is needed (Weil, Ormond, Peters, Peters, Biesecker & LeRoy, 2006). SDM has been suggested as a complementary approach within genetic counselling (Elwyn, Gray & Clarke, 2000), or as an alternative to non-directive practice models. It integrates both medical and psychosocial goals, improves the consent processes, and supports patient autonomy (Weil et al., 2006).
2.3 Health Care Professionals’ (HCPs) Perspectives

Much of the literature has focused on HCPs’ decision-making, and their perspectives on returning genetic research results or incidental findings (IFs) to research participants (Angrist, 2011; Abdul-Karim et al., 2013; Bookman et al., 2006; Fullerton et al., 2013; Evans & Rothchild, 2012; 2013; Klitzman et al., 2013), parents (Evans, 2013; Lohn et al., 2013; Zawati et al., 2013), or individuals (Berg, Khoury & Evans, 2011; Yu, Jamal, Tabor, Bamshad, 2013). The challenges with IFs include their quantity, answering whether or not they should be disclosed, and if yes, selecting which subset to reveal. These challenges result from the goals of respecting patient autonomy over their genetic information, beneficence (maximizing the potential benefits of personal or clinical genetic information), and non-maleficence (minimizing the potential harms of unwanted genetic information, or the harms resulting from not receiving important genetic information). To provide context around the ongoing challenges with IFs, the researcher will briefly discuss HCPs’ perceptions about them in both research and clinical settings.

2.3.1 Health Care Professionals’ (HCPs) Perspectives: Return of Results and Incidental Findings

In the research setting, there are additional complexities to deciding whether or not to return IFs to participants (Klitzman et al., 2013). Researchers have struggled with uncertainty about which IFs to return, and how to make these decisions (Klitzman et al., 2013). They often face conflicting duties, having to balance information quality (e.g., findings have to be scientifically valid), adherence to rules (e.g., to stay within the scope of the study protocol and informed consent) and perceived participant welfare (Meacham, Starks, Burke & Edwards, 2010). Much of the literature discusses whether or not research results should be returned, and if
so, which types (Abdul-Karim et al., 2013; Angrist, 2011; Caulfield et al., 2008; Evans & Rothchild, 2012; Fabsitz et al., 2010; Fullerton et al., 2012; Klitzman et al., 2013; Knoppers, Joly, Simard & Durocher, 2006; Meacham et al., 2010; Wolf et al., 2012). Even if the purpose of some genomic research is not to provide results, there may be IFs that could reveal a person’s genetic disposition for a particular condition, an increased susceptibility to a disease, or misattributed paternity (Meacham et al., 2010). Therefore, the larger discussion is motivated by ethical issues surrounding beneficence, nonmaleficence, respect for participant autonomy, and whether or not it is the researcher’s duty to warn participants about genomics findings that could be important or relevant to them. Other challenges in the research setting include the ability of researchers to address arising clinical issues, but these will not be discussed as they fall outside this study’s scope.

In the clinical setting, one focus has been the attempted development of expert consensus on the different categories of genetic variants, and which results should be disclosed to patients (Berg, Khoury & Evans, 2011; Green et al., 2012; Berg et al., 2013; Green et al., 2013). A 2013 American College of Medical Genetics and Genomics (ACMG) statement recommended the evaluation and reporting of a minimum of 56 disease-associated genes in any patient undergoing WGS/ES, regardless of the test’s indications (Green et al., 2013). This list of genes must be evaluated, and incidental variants reported irrespective of patient age or choice (Green et al., 2013). These ACMG recommendations are controversial, and some (Burke et al., 2013; Townsend, Adam, Birch & Friedman, 2013) argue that they challenge patient autonomy, respect for patient preferences, the concept of shared decision making, and the best interests and future autonomy of the children who are tested. Others (Evans, 2013) favour returning some of these IFs to families, and argue that the new recommendations arise from a different context. Evans
(2013) argues that the context has shifted from knowing about a familial risk and deliberately testing the child, to one of *incidentally* discovering a mutation in a child that reveals a high risk for a preventable, adult-onset disorder and can also impact the health of that child’s parents. He further argues that although there exists the risk of harm from violating a child’s autonomy, there are significant benefits to both the parents (e.g., smaller chance of disease and death) and the child (e.g., would prefer that their parent(s) not develop a life-threatening disease) if the IFs are disclosed to the family (Evans, 2013). Despite the contrasting views about the recommendations, the ACMG has recently released an update, allowing patients to "opt out" of having these results returned (ACMG, 2014).

Another suggestion is to provide “qualified disclosure”, which means that IFs should only be withheld under special circumstances (Bredenoord, Kroes, Cuppen, Parker & van Delden, 2011; Christenhusz, Devriendt & Dierickx, 2013). While there is debate, the consensus is to disclose – upon request – IFs with confirmed clinical utility, and that have the possibility of treatment or prevention (Berg, Khoury & Evans, 2011; Cho, 2008; Caufield et al., 2008; Christenhusz, Devriendt & Dierickx, 2013; Kaye et al., 2010; Lohn et al., 2013). Still, some genetic HCPs have suggested that the disclosure of IFs be done with caution, especially in specific contexts such as WGS and genetic testing in minors (Christenhusz, Devriendt & Dierickx, 2013). Some reasons to be cautious include the need to respect the right not to know, the autonomy of the patient, and the fact that IFs have implications – both positive and negative – for other family members (Christenhusz, Devriendt & Dierickx, 2013).

In their proposal for Canada, Zawati et al. (2013) suggested that “the decision-making concerning WGS/ES with children should be guided by their best interests, including timely medical benefit to the child during childhood” (p. 2). It has also been suggested that clinically
significant conditions that are actionable during childhood be reported to parents, whereas adult-onset genetic conditions should not be (Zawati et al., 2013). These recommendations are a valuable starting point in developing Canadian guidelines; however, the parents’ perspectives were not considered in the development of these recommendations.

On the contrary, Yu and colleagues (2013) have argued for shifting the focus from the return of results to the management of results. They propose a self-guided management approach that enables people to select among results in a convenient, value-based, and personalized context (Yu et al., 2013). The researchers suggest that the results should be seen as a source of information, and as part of an ongoing process of receiving and translating results over time (Yu et al., 2013). In this approach, patients play a greater role, and have more autonomy over their results. The responsibilities of initiating care, decision-making, and managing an individual’s health are shared between the HCP and the patient. This may be a suitable approach for parents deciding on WGS/ES for their child. However, it focuses primarily on HCPs’ suggestions about the management of results, and not on the parents’ views on their decisional needs prior to testing.

2.3.2 Health Care Professionals’ (HCPs) Perspectives: Parents’ Information and Decisional Needs

Few studies have explored parents’ information and decisional needs before they make decisions on WGS/ES. In their systematic review on WGS informed consent, Ayuso et al. (2013) found a general consensus on the contents of informed consent for WGS in the clinical setting. They proposed that the components of the WGS informed consent should include: the scope, a description specifying the type of information to be obtained, the expected benefits and risks, the existence of alternative options (if any), the voluntary nature of the test, the possibility of refusal,
future uses of the data, and confidentiality of the outcomes (Ayuso et al., 2013, p. 3-4). However, this consensus was based on input from genetic experts, scientific societies, and ethical boards, and there is no indication that either patient or parental input was considered.

The decision-making process often includes a combination of education, genetic counselling and informed consent. Each of these components are different but are often conflated as one process in the literature. Informed consent is often viewed as a medico-legal requirement, however, some experts argue that informed consent is a process, and not a one-time event (Lidz, Appelbaum & Meisel, 1988) or a simple signature on a legal form (Annas, 2001). Genetic counselling focuses on the patient's and family's needs and not only on informed consent. Genetic counsellors provide information on the nature, inheritance, and implications of genetic disorders, help patients and families understand and adapt to the medical, psychological, and familial implications of how genetics contributes to disease, and help them make informed medical and personal decisions (Canadian Association of Genetic Counsellors, 2014).

Bunnik and colleagues (2013) have proposed a tiered-layered-staged model for informed consent, which may suit the volume and complexity of information produced by WGS/ES. This model of informed consent is divided into tiers (or categories) of traits and diseases, distinguishes between different layers of information, and is seen as a process that provides time between counselling and decision-making (Bunnik, Janssens & Schermer, 2013). This model of the informed consent process may be a good starting point to help support parents deciding on WGS/ES. However, the proposed model was intended for consumers of personal genome testing in a commercial and online context. There is also limited evidence of consumer input in its development.
Recommendations based on the views of genetic experts or scientific societies may not accurately reflect parents’ information and decision support needs. Research has shown discrepancies between genetic HCPs’ and the public/parents’ perspectives on the amount of time available for informed consent (Appelbaum et al., 2013), the process and content of informed consent, preferences for the return of IFs (Levenseller et al., 2013), and the patients’ choices relating to the disclosure of IFs (Townsend et al., 2013). Parental groups have emphasized responsibility for their own health and decision-making, patient choice, and autonomy relating to WGS/ES, whereas genetics professionals have asserted the limitation of data for analysis to avoid confusing patients and causing unnecessary anxiety (Townsend et al., 2013). Parents have also questioned the HCPs’ concept of what is “clinically relevant” (i.e., what results are considered “serious” and “relevant”), and argued that the disclosure of IFs should not be based solely on clinical relevance (Townsend et al., 2013).

The evidence and recommendations generated by various HCPs, researchers, and experts form a good foundation for guiding further discussions as WGS/ES evolves. However, research on parental perspectives on their decisional needs remain limited. Given that the parents’ decisions on WGS/ES may significantly impact their families, it is crucial to assess their views and decisional needs.

2.4 Parents’ Perspectives

For the purposes of this study, parents refer to parents of children with a suspected genetic condition, and who: have considered WGS/ES for their child, had children who are candidates for (or have been offered) WGS/ES, or have already consented to WGS/ES for their child.
Research on parental perspectives to date has focused primarily on their views on the informed consent process for IFs (Appelbaum et al., 2013; Rigter, van Aart, Elting, Waisfisz, Cornel & Henneman, 2013), and their preferences for the return of IFs (Daack-Hirsch et al., 2013; Kleiderman et al., 2013; Levenseller et al., 2013; Rigter et al., 2013; Sapp et al., 2013; Shahmirzadi et al., 2013; Townsend et al., 2013).

Researchers have also examined the public’s perspective on the return of results (Bollinger, Scott, Dvoskin & Kaufman, 2012; Murphy et al., 2008) and, more specifically, IFs (Townsend et al., 2013). Others have explored individual patients’ perspectives on the return of their results (Wright et al., 2013; Facio et al., 2013) and IFs (Bennette et al., 2013). However, for the purposes of this study, the researcher will only discuss parental perspectives on the return of results and IFs.

### 2.4.1 Parents’ Perspectives: Return of Results and Incidental Findings

Parents have expressed a strong desire to learn about genetic variants that relate to the problem for which WGS/ES was indicated (i.e., primary results) (Sapp et al., 2013; Tabor et al., 2012). They have also expressed a desire to know that IFs are possible, to be given the choice on whether or not they want to be told about them (Townsend et al., 2013), and if so, how they want to be told about IFs (Daack-Hirsch et al., 2013, Tabor et al., 2012).

Most parents have asked or opted for IFs to be disclosed to them (Daack-Hirsch et al., 2013; Kleiderman et al., 2013; Shahmirzadi et al., 2013). Parents have reported that they prefer to receive all the results from clinical WES (Levenseller et al., 2013), or all the types of results (e.g., primary results and IFs about treatable or untreatable conditions, or carrier status) (Sapp et al., 2013). They have asked to be given choices over which types of results to receive (Levenseller et al., 2013; Tabor et al., 2012), and have focused on results that could be
personally useful to them (Daack-Hirsch et al., 2013; Levenseller et al., 2013). Following the return of IFs, parents expect that they would need additional support, such as counsellors, psychologists, family friends, support groups, or the medical community (Kleiderman et al., 2013). It is apparent that many parents want knowledge of at least some IFs, and control over which results they receive, and in what manner.

On the other hand, attitudes toward receiving IFs have not been uniform. Parents have indicated ambivalence over whether or not they want to receive IFs, and uncertainty concerning what kind of information might be returned, and how their lives may be impacted (Tabor et al., 2012). In addition, parents have varied expectations for ES, and assign different, personal weightings to both the potential benefits and harms of genomic testing and the discovery of IFs (Rigter et al., 2013).

Some parents felt unsure that the benefits of a genetic diagnosis outweighed the risks of IFs, while others were certain (Rigter et al., 2013). Parents have also expressed reservations about the receipt of results relating to their child’s carrier status, untreatable adult-onset conditions (Kleiderman et al., 2013; Levenseller et al., 2013; Sapp et al., 2013), or early onset conditions (Shahmirzadi et al., 2013).

Although many parents want to know the results and IFs, other parents are concerned and ambivalent. This variation in parental preferences indicates that there can be no single standard approach to the return of IFs. It seems necessary to further explore parents’ perspectives in order to inform dynamic and flexible approaches to the WGS/ES decision-making process.
2.4.2 Parents’ Perspectives: Information and Decisional Needs

2.4.2.1 Information Needs

There is little research on the content parents perceive should be discussed pre-testing. In the genomic research setting, parents have endorsed the following for inclusion in the informed consent discussions for IFs: risks, benefits, impact on family, return of results in the event of a death or incapacity, and terms of re-contact (Appelbaum et al., 2013). The “return of results in the event of a death or incapacity” indicates that participants should be told how IFs will be handled if he/she dies, or becomes incapable of making decisions, before findings are available (Appelbaum et al., 2013). Terms of re-contact relates to the fact that as more genetic variants and IFs are discovered, the need to re-contact participants about the new discoveries may arise. Parents endorsed the potential for being re-contacted about such discoveries to be mentioned during consent (Appelbaum et al., 2013).

Parents and genetic HCPs agree that certain information should be discussed pre-testing to ensure that parents can make informed decisions. This information includes: the primary reason for WGS/ES, the potential for IFs, variants of unknown significance, the associated potential for anxiety and uncertainty, the scientific uncertainty of IFs, and the probabilities for conditions that may develop (Townsend et al., 2013). Parents have also expressed concerns over potential risks related to WGS/ES, such as stigmatization, discrimination in insurance and employment, loss of privacy, and possible restrictions on future reproductive decision making for the child (Levenseller et al., 2013). Discussing this content pre-testing appears to be important to parents.
On the contrary, parents have little to no interest in genetic concepts, genomic variation, and WES limitations (Levenseller et al., 2013). Parents’ perceptions of their informational needs pre-testing are varied, and the literature on the subject is limited.

2.4.2.2 Decisional Needs

Parents agree that the pre-test discussion is essential to informed decision-making, and to avoid the surprise of IFs (Townsend et al., 2013). Prior to testing, parents have emphasized the need for education, and a consent process that prepares them for the possibility of an IF, and that gives them a choice to learn about different types of IFs (Daack-Hirsch et al., 2013).

Parents have placed importance on family participation in the decision-making process (Tabor et al., 2012). There is also evidence that family members involved in the decision-making prefer for the informed consent process to be spread out over several, shorter discussions (Tabor et al., 2012). In addition, parents have reported a preference for information to be delivered face-to-face, instead of the telephone or live streaming video (Tabor et al., 2012).

Parents likely have varied opinions on the amount of time they find most supportive to their decision-making during the consent process. Appelbaum et al. (2013) found discrepancies between genomic researchers’ and participants’ views on the reasonable amount of time for informed consent for IFs. Most genomics researchers thought less than thirty minutes was a reasonable amount of time for informed consent, whereas research participants – which included parents who were sequenced as part of the testing for their children’s condition – thought that the consent could be five to ten minutes, two hours, or ‘however much time it takes’ (Appelbaum et al., 2013).

In the context of WGS/ES, there is little evidence on what parents perceive as supportive to their decision-making.
2.4.3 Parents Perspectives: Challenges and Concerns with WGS/ES Decision-Making

Parents have voiced concerns with the decision-making process, including the lengthy informed consent process (Tabor et al., 2012), the complexity of the information provided to them (Rigter et al., 2013), the complexity of WGS/ES compared to other routine diagnostic tests, the lack of time for discussion, being distracted by their child, and their lack of careful consideration for the possibility of IFs (Levenseller et al., 2013). Despite a general enthusiasm to receive IFs pertinent to their children, parents admit that the decisions could be difficult (Kleiderman et al., 2013). Due the complexity of the information provided to them, parents may inaccurately interpret the implications of IFs in ES (Rigter et al., 2013). For example, in Rigter et al.’s study, one parent did not understand that ES could also reveal genetic information relevant to them as parents (Rigter et al., 2013). Furthermore, some parents may not understand what they are consenting to, or why (Rigter et al., 2013).

2.4.4 Parents’ Perspectives: Gaps

Much of the research regarding parental perspectives has focused mainly on their views, preferences, and decision-making on the return of IFs (Daack-Hirsch et al., 2013; Kleiderman et al., 2013; Levenseller et al., 2013; Sapp et al., 2013; Shahmirzadi et al., 2013; Townsend et al., 2013). The return of IFs is likely a large part of decision-making discussions. However, there is limited exploration of what other elements and support parents perceive as being the most important for their educational preparation, pre-test discussion, and informed consent process. No study to date has specifically examined parents’ pre-testing decisional needs with respect to
genome-wide sequencing. The primary studies the researcher reviewed are presented in Table 2.1 below.
<table>
<thead>
<tr>
<th>Author(s)</th>
<th>Study Aims</th>
<th>Method</th>
<th>Context</th>
<th>Description &amp; Number of Participants</th>
<th>Comments</th>
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</table>
| Appelbaum et al., 2013 | -Explore genomic researchers’ and study participants’ views of the informed consent process and their preferences for how it should be conducted | Mixed methods approach Online survey and semi-structured interviews | New York City, USA. **Research WES** | **Survey**: genetic researchers, n = 245 **Interviews**: n=28 genomic researchers, n=20 research participants (n=18 **parents** sequenced as part of an assessment of their children’s disorders) | -Limited representation of participants’ views (e.g., more HCP opinions, a lot of endorsements from participants)  
-Focused more on content of informed consent, not other factors that may affect their decision making  
-Lacking views about the processes of informed consent                                                                  |
| Daack-Hirsch et al., 2013 | -Explore public viewpoints on IFs in genome-based testing in clinical and research situations | Focus groups and interviews                                               | Midwestern state, USA. **Hypothetical vignettes if participants were to get tested.** | **N=63**  
9 **Focus groups**: public members – African Americans, clergy, elders, Hispanics, parents with children with hearing impairment, rural community, support groups, young middle aged adults  
9 **Interviews**: **Parents** of children who had chromosomal microarray analysis (n=7), people unable to attend focus group | -Hypothetical scenarios  
-Mostly a mixed group of people from the public, only 7 parents  
-Primarily focused on what IFs are, people’s desire to know or not want to know about IFs, and how to manage IF information |
| Kleiderman et al., 2013 | -Explore parental perceptions and experiences regarding the return of genomic incidental research findings in children with rare diseases | Focus groups and open-ended individual telephone interviews | Montreal & Ottawa, CANADA **Hypothetical vignettes about genomic disorders discovered incidentally** | **N=15**  
2 **focus groups**, 9 **interviews**: **Parents** of children affected by various rare diseases | -Hypothetical scenarios  
-Primarily focused on decision-making for IFs in the research setting  
-Canadian context, similar parent population |

Table 2.1 Primary studies on parental perspectives
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<tr>
<th>Author(s)</th>
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<th>Method</th>
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<th>Description &amp; Number of Participants</th>
<th>Comments</th>
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<tbody>
<tr>
<td>Levenseller et al., 2013</td>
<td>-Assess professionals’, parents’, adolescents’ views on implementing WES in the pediatric clinical setting: -Their understanding of the risks and benefits of WES, preferences for informed consent discussion, process for return of results, decision-making role of the pediatric patient</td>
<td>Focus Groups</td>
<td>Philadelphia &amp; Baltimore, USA. Hypothetical scenarios to assess opinions and intent</td>
<td>3 Focus groups: 1 Professional, 1 Parents, 1 Adolescents -2 parent focus groups n= 20 -only included parents with children who may be offered sequencing</td>
<td>-Hypothetical scenarios -Compared and contrasts the views of professionals, parents and adolescents -Discovers a few barriers and challenges to the process of informed consent and return of results</td>
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<tr>
<td>Sapp et al., 2013</td>
<td>-Characterize the values and beliefs that shaped parents’ preferences for learning their child’s sequencing results -Assess how parents conceptualize genomic data and factors that contribute to their decision making</td>
<td>Semi-structured interviews</td>
<td>Maryland, USA Research WES</td>
<td>13 families – 12 fathers, 13 mothers -Participants were recruited from an exome sequencing study at the National Institutes of Health -Each family had at least one affected child</td>
<td>-Discovered the following values and beliefs: parental responsibility, need to know/preference for knowledge, control, faith, altruism -Focus on parents’ decision-making about the return of primary and different categories of secondary variant results (IFs) -Does not examine parents’ decision support needs</td>
</tr>
<tr>
<td>Shahmirzadi et al., 2013</td>
<td>-Provide preliminary data regarding preferences and impact of secondary findings results disclosure based on the first 200 families</td>
<td>Retrospective analysis of patient responses</td>
<td>USA and Canada – not specific Clinical diagnostic ES</td>
<td>N=200 -All patients were affected with a genetic disorder n=162 Children (&lt;18 years old) -Parent or guardian for children n=38 Adult patients</td>
<td>-Patients (187/200) chose to receive IFs for one or more available categories -Focus on the return of IFs -Parents’ and adult patients’ views are clumped together -Doesn’t look at the processes of parents’ decision or their decision support needs</td>
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Table 2.1 Primary studies on parental perspectives (continued)
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<tr>
<th>Author(s)</th>
<th>Study Aims</th>
<th>Method</th>
<th>Context</th>
<th>Description &amp; Number of Participants</th>
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<tbody>
<tr>
<td>Tabor et al., 2012</td>
<td>-Explore participants’ experiences with the informed consent process, their perceptions of the risk associated with sharing of WGS data and their preferences for return of results</td>
<td>Interviews</td>
<td>USA and “not USA” = not specific</td>
<td>Research WGS-2 families with Miller syndrome</td>
<td>-Reveals parental experiences and opinions about the consent process, their motivations for participation and expectations of the study, and perspectives on return of results</td>
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<tr>
<td>Townsend et al., 2012</td>
<td>-Explore issues concerning IF in clinical settings -Explore and compare parent, public, and professional perspectives of IF and their implications for clinical practice</td>
<td>Focus groups</td>
<td>Vancouver CANADA Mostly hypothetical views about IFs in clinical WGS</td>
<td>3 focus groups: n=10 genetic health professionals; n=10 general public (non-genetics health professionals, unemployed, homemakers); n=8 (5 mothers, 3 fathers) parents whose children have experienced genetic testing*</td>
<td>-Primarily focused on people’s views about IFs *Unclear what type of testing “genetic testing” included</td>
</tr>
<tr>
<td>Rigter et al., 2013</td>
<td>-Explore the first experiences with and the needs for informed consent procedure in diagnostic ES</td>
<td>Semi-structured interviews and observation</td>
<td>Amsterdam, Netherlands Diagnostic ES</td>
<td>Interviews: 11 professional experts, written response: 1 professional 3 parent cases: observation counseling of parent, patient (child) and HCP interaction -2 testing of children, 1 testing of fetuses</td>
<td>-Mostly professionals’ views, limited parental perspective -Focused on decision-making processes, consent for IFs (e.g., opt out options, advisory board’s role) -Points out some challenges with their current informed consent processes</td>
</tr>
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</table>

Table 2.1 Primary studies on parental perspectives (continued)
A few studies have involved parents who have actually made WGS/ES decisions for their child (Appelbaum et al., 2013; Rigter et al., 2013; Sapp et al., 2013; Shahmirzadi et al., 2013; Tabor et al., 2012). Other studies are based on hypothetical scenarios, where its participants have not undertaken WGS/ES (Daack-Hirsch et al., 2013; Kleiderman et al., 2013; Levenseller et al., 2013). In these studies, the participants include only individuals with children who may be offered ES (Levenseller et al., 2013), or parents were asked to report on their predicted reactions to hypothetical situations or vignettes (Daack-Hirsch et al., 2013; Kleiderman et al., 2013). The use of hypothetical scenarios may not accurately reflect parental decision-making in reality.

Other studies have broadly focused on parents with children who have undergone other kinds of genome-wide testing (Daack-Hirsch et al., 2013; Townsend et al., 2013), and not on parents deciding on WGS/ES for their child.

Few studies have been situated in Canada (Kleiderman et al., 2013; Townsend et al., 2013), whilst many other studies were based in specific research or clinical contexts in the United States (Appelbaum et al., 2013; Daack-Hirsch et al., 2013; Levenseller et al., 2013; Sapp et al., 2013; Shahmirzadi et al., 2013; Tabor et al., 2012) or in the Netherlands (Rigter et al., 2013). As Canada has a public health care system, and WGS/ES is still used primarily in the research setting, there may be important contextual differences in what parents perceive as important. Furthermore, the resources and support available in research and clinical settings for both genetic HCPs and parents may also differ.

2.5 Genomics and Implications for Nursing

Experts foresee that advances in genomics, such as the growing research discoveries and new technologies of WGS/ES, will translate into new clinical approaches (Biesecker & Green, 2014). It is imperative for practicing nurses to be aware of and involved with these genomic
advances now (Calzone, Jenkins, Nicol, Skirton, Feero & Green, 2013). Without knowledge of the genomic technologies, the rationales for their use, and the possible ramifications that can result from genetic diagnosis or treatment, nurses will be unable to provide quality care (Loescher & Merkle 2005). Nurses have a legal and professional duty of care to their patients (CRNBC, 2014; Young 2009). Duty of care requires nurses to, amongst other things, perform processes and procedures with reasonable care and skill, and to inform patients of risks related to procedures and interventions (Young, 2009). Thus, the responsible translation of genomics into health care requires all clinical professions – including nursing – to continually educate their practicing workforce in genomics (Calzone et al., 2013).

In addition, nurses are well situated to incorporate genetic and genomic information across the lifespan, and all aspects, of the healthcare system (Calzone, Cashion, Feetham, Jenkins, Prows, Williams & Wung, 2010). The nursing profession has an ongoing commitment to being patient-focused by striving to understand their patients’ and families’ perceived needs, priorities, and expectations for health care, and reorganizing services around the patients’ and families’ needs (Lutz & Bowers, 2000). Not only do nurses have an intimate knowledge of patients’ and families’ perspectives, but they are also skilled in communications, building partnerships, and understanding the biologic underpinnings of health and disease (Calzone et al., 2010) as well as clinical processes and procedures. Furthermore, nurses are among the most trusted HCPs (Gallup, 2014), make unique contributions, and complement the work of other HCPs to improve the health of patients and families (Calzone et al., 2010). In some settings, nurses already play a key infrastructure role in supporting the delivery of services that use genomic information (Calzone et al., 2013). For example, nurses are often involved in obtaining
bio specimen consent, recruiting vulnerable and minority populations, and disclosing research results (Badzek, Henaghan, Turner & Monsen, 2013).

Nurses have social and professional responsibilities to understand the complexities that may arise for individuals and families, to serve as patient advocates (Hamilton, 2009), to be knowledgeable about genomics in order to assist patients and families in becoming better informed decision-makers of genomic-based health care, and to ensure fairness amidst the rapidly developing technologies (Badzek et al., 2013). Nurses can impact policies on the use of genomic information through raising related ethical, legal, and social concerns (Badzek et al., 2013; Calzone et al., 2013). A primary goal of nursing research into clinical genetics and genomics is to enhance the quality of health care for patients and families (Calzone et al., 2010). I aim to explore the potential for nurses to assist in this by exploring parental perspectives prior to the widespread implementation of clinical WGS/ES.
Chapter 3: Method

This study was designed to address the gaps in knowledge on parents’ decisional needs prior to WGS/ES for their child.

3.1 Research Design

Considering the nature of the research problem and the limited literature on parents’ perspectives, the researcher chose a qualitative approach. The objective of qualitative research is to generate knowledge about human phenomena for which depth and contextual understanding would be useful (Thorne, 2008). Qualitative inquiry tends to be inductive, exploratory, flexible and holistic in nature (Polit & Beck, 2012; Thorne, 2008). This type of research typically focuses on patterns and themes within individual human experience, on subjective knowledge, and illustrates experiential reality (Thorne, 2008). Parents may have unique perceptions and priorities and respond differently to their daily experiences. The researcher aimed to obtain knowledge from the perspective where participants describe and identify their own personal realities and viewpoints as they experienced them (Polit & Beck, 2012).

Many qualitative methods exist, deriving from various disciplines, including sociology, cultural anthropology, and philosophy. Each approach tends to reflect the different philosophical values, assumptions, methodological standards and objectives inherent in the particular discipline from which it arises (Thorne, Reimer Kirkham, & MacDonald-Emes, 1997; Polit & Beck, 2012). Grounded in nursing’s epistemological foundations and systematic reasoning, KCL chose interpretive description (Thorne et al., 1997) because it is a pragmatic, inductive analytic approach that enables the understanding of clinical phenomena and produces knowledge that is relevant for practice (Thorne et al., 1997; Thorne, Kirkham Reimer & O'Flynn-Magee, 2004).
The foundations of nursing knowledge, which assume that human health and illness experiences are made up of complex interactions between both psychosocial and biological phenomena, informed the methods in this study (Thorne et al., 1997). Interpretive description does not prescribe a single way to conduct a study but rather serves as a guide for using various data collection and analytic strategies that are congruent with the logic and intent of the study (Thorne, 2008). The logic of one’s disciplinary orientation can also justify application of specific methods and procedures outside of their traditional context (Thorne, 2008). The researcher’s orientation is in nursing, which is a professional discipline that applies knowledge to resolve human health and illness concerns (Thorne, 2008), provides safe, compassionate, competent and ethical care, promotes and respects informed decision-making, and serves in the public’s interest (Canadian Nurses Association, 2008, 2014).

Interpretive description is an approach that allows the researcher to examine and more fully understand parents’ perspectives; it appreciates the constructed, complex, and contextual nature of a person’s health experience while allowing for the construction of shared realities (Thorne et al., 1997). Interpretive description allowed the researcher to characterize and explain experiences from the parents’ perspectives, which will assist in constructing knowledge for practical clinical application and care, and form the basis for future research (Thorne et al., 1997).

3.2 Theoretical Frameworks about Health-Related Decision-Making

Decision-making (DM) is the process of choosing between two or more options, which may include deciding to do nothing (O’Connor, Stacey & Jacobsen, 2011). Most individuals want to decide on the options that produce the most desired outcomes; however, many health care decisions do not have an obvious best choice (O’Connor, Stacey & Jacobsen, 2011).
There are a range of health-related DM theories and models, such as DM in the transtheoretical model of behavior change (Prochaska, 2008; Prochaska, & Velicer, 1997), and the theory of reasoned action (Fishbein & Ajzen, 1975). The transtheoretical model of behavior change postulates that people make decisions through six stages of change (precontemplation, contemplation, preparation, action, maintenance, and termination) as they weigh the pros and cons of changing their health-related behaviours (Prochaska, 2008; Prochaska & Velicer, 1997). The theory of reasoned action (Fishbein & Ajzen, 1975) suggests that self-efficacy, attitudes toward behaviours and subjective norms (i.e., how one thinks others perceive him/her) influence behavioural intentions, which in turn predict actual behaviours. These theories have increased our understanding of how individuals’ make decisions about, change or adhere to specific health behaviours (e.g., compliance with medical recommendations). However, these theories are prescriptive, as they assume how people should make decisions. These theories may not operate well in the context of parents making a decision about WGS/ES for their child, for which there is no “right” or “wrong” approach.

The decisional conflict model (Janis & Mann, 1976, 1977) is another DM theory, which postulates that people often make decisions under stress and theorizes what people do during decision-making. This model assumes that stress affects the DM process and describes five patterns of coping (unconflicted adherence, unconflicted change, defensive avoidance, hyper vigilance, and vigilance), suggesting that vigilance is the most adaptable coping strategy. With each coping pattern, the decision-maker is suggested to have different predecisional behaviour characteristics such as the thorough investigation of objectives and alternatives, careful evaluation of consequences of current and new policies, thorough search for information, unbiased assimilation of new information, reevaluation of consequences, or planning for
implementation and contingencies (Janis & Mann, 1976). Although the amount of stress and different coping patterns are important to consider during DM about WGS/ES for a child, there are other dimensions (e.g., informational and educational needs, types of resources and support) that may be necessary to improve the DM process in the clinical setting.

3.2.1 Shared Decision Making

SDM is a collaborative approach where HCPs and patients use the best available evidence and make health decisions together (Elwyn, Laitner, et al., 2010). This approach promotes patient autonomy and engagement through a two-way exchange of relevant information and available options; this facilitates patients to express their values, reflect on their preferences and make decisions that are right for them (Elwyn, Laitner et al., 2010; Elwyn et al., 2012). SDM engages patients’ expertise in understanding their personal circumstances and weighing the value they attach to the benefits, risks, and scientific uncertainties of the options provided to them (O’Connor, Stacey & Jacobsen, 2011).

As described in Chapter 2, parents want to play a greater role in health-related decisions and emphasize their autonomy and right to choose. Thus, SDM may be a suitable approach to guide parents when they are deciding on WGS/ES for their child.

3.2.2 The Ottawa Decision Support Framework

Grounded in SDM, the Ottawa Decision Support Framework (ODSF) is a framework that guides patients in making social or health-related decisions (Ottawa Hospital Research Institute, 2012). The ODSF has three key elements, decisional needs, decision quality, and decision support (O’Conner, 2006).
Figure 3.1 The Ottawa decision support framework (O’Conner, 2006).

The ODSF holds that a person’s decisional needs (e.g., knowledge and expectations) will affect decision quality (e.g., informed decisions that are aligned with patient’s values), which then affects the individual’s actions, health outcomes, emotions and appropriate use of health services (Ottawa Hospital Research Institute, 2012). Decision quality improves when patients’ with unmet decisional needs have the opportunity to clarify their values, are provided with relevant information, and are offered value-based choices (O’Conner, 2006). Individuals who make better decisions experience less decision regret and conflict and may take actions that lead to more positive health outcomes (O’Conner, 2006). On the other hand, unmet decisional needs
have undesirable effects on the quality of the decision, thereby negatively affecting actions, health outcomes, emotions, and appropriate use and costs of health services (O’Connor, Stacey & Jacobsen, 2011). Decision support using tools like clinical counselling or decision coaching can help address unmet decisional needs, thereby improving decision quality (O’Connor, Stacey & Jacobsen, 2011).

The ODSF uses the following three-steps:

1) Assess client and practitioner determinants of decisions to identify decision support needs
2) Provide decision support tailored to client needs
3) Evaluate the decision-making process and outcomes

(Ottawa Hospital Research Institute, 2012).

For the purposes and scope of this study, KCL focused on step 1, where she only explored parents’ decisional needs (Figure 1). For example, KCL explored parents’ perceptions of their knowledge requirements (i.e., what knowledge parents assess as important). This information is needed to enable HCPs to recognize what information the parents of a child for whom WGS/ES is being considered may prioritize in the clinical setting.

The ODSF was originally designed to guide patients’ decision-making, however, the researcher extended its use to parents of children undertaking WGS/ES. In this study’s context, WGS/ES is for a patient who is a child, so it is frequently parents who have to make the decision about WGS/ES.

The ODSF and SDM are more suitable theoretical frameworks for the context of this study. Janis and Mann’s (1976) decisional conflict theory is one of the theories that inform the ODSF (O’Conner, 2006); therefore people’s stress and coping patterns during decision-making
are also considered. With SDM and the ODSF guiding KCL’s exploration of parents’ perspectives, she was able to explore parents’ decisional needs in a more holistic manner and subsequently help inform decision support interventions for the clinical setting. This exploration included entities that influence individuals’ decision-making, such as the type, stage and timing of the decision, as well as knowledge, expectations, support, resources, values, and beliefs (O’Conner, 2006). SDM, the ODSF and a review of the literature guided the development of a topic guide (Appendix G) for the focus groups and semi-structured interviews (Data Collection).

### 3.3 Ethical Considerations

Prior to the recruitment of participants, approval was obtained from the University of British Columbia Children’s and Women’s Research Ethics Board (REB). Parents were invited to participate using a REB-approved letter of invitation. During recruitment, KCL also explained the details and purpose of the study and emphasized the voluntary and confidential nature of the study. Participants were also informed about confidentiality and privacy implications, the study’s risks and benefits and their right to withdraw from the study at any time. Participants were informed and aware that there was no direct benefit to them from participating in this study. Participants provided consent prior to the first interview or focus group.

Interviews and focus groups took place at a convenient, accessible and neutral location, such as a room in a local community centre or a mutually agreed-upon location. Focus groups and interviews were audiotaped and transcribed. Audiotapes, field notes, consent forms, and participants’ demographic information were stored in a locked cabinet within a locked area. Electronic transcripts were stored on a password-protected computer on a secure network in a locked area.
Every effort was made to protect anonymity. Participants, their children, and other mentioned names were deleted in the transcripts. Pseudonyms and generic descriptions (i.e., daughter, husband, doctor, or nurse) were used in place of the names during transcription and analysis of the data. Only KCL had access to the match between the pseudonyms and actual names. Identifiable information, such as names will not be used in publications. Digital audiotapes will be kept for 5 years and all files erased and formatted after this period. All paper files will be dated, including anonymized paper transcripts, which will be kept for a minimum of 5 years prior to shredding.

A summary of the research findings will be provided to the participants and publications will be provided to APOGEE-Net/CanGène Test Network (the study’s funding source).

3.4 Recruitment and Sampling

3.4.1 Convenience Sampling

At the inception of the study, there were a limited number of parents with children who have undergone clinical WGS/ES in British Columbia; therefore, KCL recruited parents through non-probability convenience sampling. At the British Columbia Children and Women’s Department of Medical Genetics, KCL had access to potential participants who had given consent to participate in genetic research and expressed an interest in being contacted for future studies. KCL also collaborated with genetic physicians and researchers who had lists of parents or research participants who had participated in previous genetic research studies and expressed an interest in being contacted for future studies. KCL recruited participants from April 2013 to June 2013. The selection criteria included: 1) At least one parent fluent in English and 2) parents who had already consented to research or clinical WGS/ES for at least one child in Canada. The intention was to recruit parents who had already gone through the DM process for WGS/ES.
The researcher invited twenty-four parents from twenty-four families via email and/or telephone. The researcher explained the purpose, benefits and risks, and the commitment parents would expect from the study. If the parents expressed interest and agreement to participate, they were emailed or mailed: an explanatory cover letter, a consent form for a focus group or interview, a one-page information sheet about WGS/ES, and video links to information about WGS/ES (Appendix A-E).

3.4.2 Theoretical Sampling

Once initial data analysis has been performed, interpretive description (Thorne et al., 1997) recommends targeted theoretical sampling, which is a non-probability approach in which the researcher purposefully selects participants who will provide the most useful information about a phenomenon. The aim of this sampling strategy is to obtain maximum variation on the themes that initially emerge from the inductive analysis (Thorne et al., 1997).

As KCL’s insights about patterns and themes began to emerge in the initial phases of data collection and analysis, she went back to ask targeted questions to participants she had previously interviewed. These repeat interviews served to confirm, expand and clarify the researcher’s understandings from the initial analysis (Thorne, 2008).

3.5 Data Collection

Interpretive description suggests that the careful application of a range of data sources can add considerable strength to the value of the data (Thorne et al., 1997). In this study, KCL used focus groups, individual semi-structured interviews with repeat interviews. Four other members of the research team (PB, BG, SA, and AT) reviewed and made suggestions for improvements of the topic guide, and KCL revised it accordingly before use with study participants.
Interpretive description, the qualitative approach used for this study, holds that research participants may provide qualitative responses with infinite variability (Thorne et al., 2004; Throne, 2008). Thus, KCL did not use data saturation as the only criterion to stop the process of data collection. KCL continued to collect data until she was satisfied that all of the major themes that related to the phenomena of decision-making and the study’s context had been identified. She also reached a point during data collection in which participants were not raising new issues or themes. KCL also encountered practical limitations with regards to the number of participants available for interview. KCL acknowledged that more conceptualizations may exist outside the scope of the study.

3.5.1 Focus Groups

KCL used focus groups to take advantage of the group engagement and process (Thorne, 2008). Focus groups capitalize on the fact that participants react to what is being said by other participants, leading to a deeper level of discussion or expression of opinion (Polit & Beck, 2012).

In June 2013, KCL conducted one focus group (n=4) at a local community centre. She was unsuccessful in recruiting enough participants for more than one focus group.

Prior to attending the focus group, KCL instructed the participants to read a one-page summary (Appendix D) about WGS/ES and watch two video links (Appendix E). The two videos are both publically available on the Internet, and are titled “Sequence Me” (Aulakh, 2010) and “Whole Genome Sequencing and You” (Sanderson, 2012). These educational materials were intended to help participants gain a basic understanding of WGS/ES.

After KCL obtained written consent (Appendix B) and demographic information (Appendix F) from each participant, the focus group was initiated. The video “Sequence Me”
(Aulakh, 2010) was shown because some parents reported that they did not watch the video. KCL and AT moderated the focus group, which was audio-recorded and took ninety minutes to complete. Discussions were initiated and prompted by the topic guide (Appendix G) and then participants brought up topics that were important to them. The topic guide included questions about types of information, resources, support and contextual, personal or social factors that influence decision-making. KCL also took fields notes, recording participants’ responses and non-verbal behaviours and reactions.

3.5.2 Semi-Structured Interviews

KCL also obtained data through eleven individual semi-structured interviews. KCL conducted all the interviews and each interview was audio-recorded. The interviews ranged from thirty to sixty minutes in length, and were conducted over a three-month period between May to July 2013.

These interviews took place at a convenient time and location chosen by the participant. Five interviews took place at the participant’s home, and six interviews were conducted over the telephone. Four of the eleven interviews took place with the participants’ child present and interviews were paused as necessary to allow participants to care for their children. KCL received written consent (Appendix C) and demographic information (Appendix F) from the participants in-person or via mail or email. Prior to attending the interview, KCL instructed the participants to read a one-page summary (Appendix D) about WGS/ES and watch two video links (Appendix E). All participants reported that they had watched one or both of the videos prior to the interview. The same topic guide (Appendix G) that was used for the focus group was also used to stimulate and promote discussion for the interviews. Participants were prompted to expand on topics that were important to them.
As the first few interviews progressed, questions shifted from a broad perspective to questions that searched for similarities, differences or other specific attributes of participants’ responses.

3.5.3 Repeat Semi-Structured Interviews

Repeat interviews serve to enhance the rigor of the study by ensuring that the conceptualizations identified were firmly grounded in the data and represented participants’ shared perceptions (Thorne et al., 1997). These repeat interviews were used to reinforce, challenge, refine, provide insight, or expand conceptualizations in the developing analysis (Thorne et al., 1997; Thorne et al., 2004). In order to broaden the conceptual relationships, I also searched for alternative, exceptional, contrary cases or themes (Thorne et al., 2004).

I read, re-read, compared and reflected upon the initial interview transcripts. Tentative conceptualizations from the initial interviews were discussed with the research team (BG, JMF, PB, SA), and these discussions assisted in generating new or clarifying questions for the repeat interviews. After initial conceptualizations were formed, another member of the research team (PB) reviewed them to ensure that they were logical and congruent with participants’ responses, and to explore any further unique, contrary or unclear conceptualizations. After discussion with PB, KCL created questions for the repeat interviews. The questions for each repeat interview varied and were influenced by the concepts that participants brought up in their initial interviews. The questions were intended to search for further similarities, differences, or an elaboration about specific attributes of the participant’s perceptions.

KCL chose five participants from the focus group and interviews who were situated in unique contexts or expressed unique perspectives or themes that needed clarification to re-interview. Participants situated in unique contexts meant that they had less common
circumstances compared to the majority of the participants. For example, these participants included a parent who adopted her children and a father with a healthy child. Repeat interviews ranged from fifteen to thirty minutes and were conducted between October 2013 and January 2014.

3.5.4 Field Notes

Field notes were written directly after the focus group and each interview as well as during data analysis. Field notes were made according to personal insights, reactions, decisions, and study complications, if any. I recorded methodological, theoretical/analytic, descriptive/observational and personal field notes during data collection and data analysis (Polit & Beck, 2012). KCL used reflective journaling to guide the research and document reactivity (Thorne et al., 1997). Reactivity takes into account the influences that occur between the researcher and the participants and helps to interpret and account for bias within the study (Thorne et al., 1997).

3.6 Data Analysis

In the interpretive description approach, data analysis is concurrent with data collection (Thorne et al., 1997; Thorne, 2008). This means that data collection and analysis inform each other iteratively and shape the direction and evolution of the study as new possibilities are considered (Thorne et al., 2004).

Interpretive description involves the use of inductive analytic techniques that encourage repeated immersion in the data prior to coding (Thorne et al., 1997). These procedures allowed KCL to support synthesis, theorizing and re-contextualizing of the data, leading to more meaningful and deeper interpretations (Thorne et al., 1997). KCL re-examined the data repeatedly to become re-immersed, and obtained a better understanding of each participant’s
story, asked questions about their statements, such as: What is this? What is going on? What does this stand for? What else is like this? What is this different from (Thorne et al., 1997)?

Interpretive description also suggests constant comparative analysis within and between data sources to generate emerging themes (Thorne, 2008). This means that the data from one source are also compared with data from other sources (e.g., other interviews). KCL conducted this process simultaneously with data collection and continued until all the sources had been compared with one another. This process allowed for the discovery of similarities, dissimilarities, and the meaning of the phenomena being studied (Polit & Beck, 2012).

The focus group and interviews were transcribed verbatim. KCL re-read and checked all the transcripts for accuracy, deleted all identifiable names, and replaced them with generic descriptions or pseudonyms. KCL listened, read and re-read the entire data set, each individual data source in context and compared data sources against each other. These procedures helped to inform the discussions KCL had with participants as she continued to collect data. Given that the data were voluminous and complex, following the initial immersive reading, a significant amount of time was given to labelling sections of the transcripts with tentative conceptualization notes. After the focus group and all the interviews were completed, and KCL had reviewed and considered all the data in detail, she identified and coded key words, phrases, concepts, themes and connections that were salient and reoccurred within the data.

Data analysis was facilitated by using NVivo 10 (QSR International, 2013), which helped to organize and visualize the data and provided an audit trail for the inductive analysis.

3.7 Rigor

There is ongoing debate about the techniques that ensure credibility and rigor in qualitative research and whether or not there is a single set of criteria that should be used
(Sandelowski 1986; 1993; Thorne, 2008). Thorne and colleagues (2004) argue that the credibility of qualitative research findings derive primarily from the way researchers present and contextualize their analytic decisions. Rather than a rigid adherence to a set of rules and procedures, rigor is more about the researcher’s faithfulness to the essence of qualitative work, which involves artfulness, versatility, and sensitivity to meaning and context (Sandelowski, 1993).

KCL used the rigor-enhancing strategies described by Thorne and colleagues (1997; 2004; 2008). For example, interpretive description assumes that eliminating biases in qualitative inquiry is naïve; instead, it is crucial for researchers to be explicit and account for the influence of their own biases on the research findings (Thorne et al., 1997, p.175). KCL recognized that she came into this study with pre-existing personal beliefs and assumptions and that these may have influenced processes during the study, such as her dialogue with the participants, data analysis, and the resultant research findings. These personal assumptions and her understandings of the phenomena were consciously challenged, revised and refined through the use of reflective journaling. Through this reflective process, KCL acknowledged her personal biases and biases within her nursing perspective and recognized how these biases may have affected the decisions and actions she undertook. For example, KCL noted her thoughts, assumptions and reactions about the research problem before the onset of the study, as well as before, during and after data collection and analysis. She recorded these notes regularly from the beginning to the end of the study and in the context of all data collection sessions. Maintaining a reflective journal helped to guide and document the reactive processes on interpreting or countering bias within the research process (Thorne et al., 1997).
A second strategy to enhance the rigor of the study involved conducting repeat interviews. These interviews allowed KCL to challenge and refine the initial conceptualizations. Repeat interviews are described in detail in section 3.5.3.

A third strategy to enhance rigor involved maintaining a decision audit trail. The ability of another researcher or reader to retrace the development of my conceptualizations, to follow the analytic reasoning process and to judge the degree to which the data analysis was grounded within the data are all important aspects of a rigorous study (Thorne et al., 1997). For these reasons, KCL maintained a decision trail from the beginning to the end of the study. This decision trail included paper and electronic documents, Excel spreadsheets and an NVivo project file that traced or described entities such as: when and how participants were selected, timeframes for recruitment and data collection, the nature of the data collection sessions, the data analysis process, and why particular participants were chosen for repeat interviews. NVivo 10 facilitated the provision of an audit trail for KCL’s inductive analysis.

The researcher’s reflections and decision log are available from the researcher.
Chapter 4: Presentation and Discussion of the Findings

In this chapter, KCL presents the findings and discussion. She describes the participant demographics and then the findings through four overarching themes. Each overarching theme includes a number of components. Afterwards, KCL discusses the possible meaning and explanations of the findings.

4.1 The Participants

From the twenty-four parents invited, fifteen parents (N=15) consented to participate and took part in at least one interview or focus group. Five parents were interested but did not participate due to scheduling conflicts, one parent consented but dropped out of the study due to urgent personal circumstances, one parent declined to participate and two parents did not respond. The demographics of the fifteen participants were collected during April 2013 to July 2013 and are shown in Table 4.1. The participants resided in a range of locations in British Columbia and Alberta. Four parents went through the DM process for clinical ES and eleven parents, for research ES. The participants had children with a range of rare, serious, congenital or childhood-onset conditions suspected or known to have a Mendelian cause.
<table>
<thead>
<tr>
<th>Demographic</th>
<th>Number of Participants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>12</td>
</tr>
<tr>
<td>Male</td>
<td>3</td>
</tr>
<tr>
<td>Age (years)</td>
<td></td>
</tr>
<tr>
<td>20-29</td>
<td>2</td>
</tr>
<tr>
<td>30-39</td>
<td>4</td>
</tr>
<tr>
<td>40-49</td>
<td>9</td>
</tr>
<tr>
<td>Highest Level of Education Completed</td>
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</tr>
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</tr>
<tr>
<td>Postsecondary Trade /Vocational School</td>
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</tr>
<tr>
<td>University/College</td>
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<tr>
<td>Graduate/Doctorate</td>
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<tr>
<td>Marital Status</td>
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</tr>
<tr>
<td>Common Law</td>
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</tr>
<tr>
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</tr>
<tr>
<td>Separated</td>
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</tr>
<tr>
<td>Employment</td>
<td></td>
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</tr>
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</tr>
<tr>
<td>Homemaker</td>
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</tr>
<tr>
<td>Unemployed</td>
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</tr>
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<td>Total Household Income</td>
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<td>Less than $10,000</td>
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<tr>
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Table 4.1 Participants’ demographics
### Demographic

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</tr>
</thead>
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<td>2</td>
</tr>
<tr>
<td>Outside Vancouver but &lt;15km from Vancouver</td>
<td>1</td>
</tr>
<tr>
<td>&gt;15km but &lt;30km from Vancouver</td>
<td>2</td>
</tr>
<tr>
<td>&gt;30km from Vancouver but within British Columbia</td>
<td>8</td>
</tr>
<tr>
<td>Outside of British Columbia (in Alberta)</td>
<td>2</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Number of Children</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
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<td>2</td>
</tr>
<tr>
<td>2</td>
<td>5</td>
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<tr>
<td>3</td>
<td>7</td>
</tr>
<tr>
<td>4</td>
<td>1</td>
</tr>
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</table>

<table>
<thead>
<tr>
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</tr>
</thead>
<tbody>
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<td>1</td>
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<tr>
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</table>

<table>
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<tr>
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<td>10</td>
</tr>
<tr>
<td>No response</td>
<td>2</td>
</tr>
</tbody>
</table>

4.1 Participants’ demographics (continued)  

4.2 Findings

The four overarching themes included: 1) context 2) informational needs, 3) psychosocial needs, and 4) supporting strategies. Figure 4.1 illustrates how the four themes relate to one another. A participant’s decision-making is embedded in his or her context. This means that a participant’s informational and psychosocial needs and supportive strategies vary depending on their individual context. Implementing strategies that meet participants’ informational and psychosocial needs help to support participants’ decision-making about WGS/ES.
Figure 4.1 Four overarching themes

4.3 Context

Figure 4.2 is a block tree-map diagram that illustrates the number of items coded within the theme of context. The size of each box represents the number of coding references. The colour of each box represents the number of coding references on a spectrum from lowest to highest (red-yellow-blue). Red boxes have a lower number of coding references and blue boxes have a higher number of coding references.
Figure 4.2 Block tree-map diagram: context
The overall data highlighted the context-dependent nature of DM for WGS/ES. The diverse circumstances and other elements (e.g., personality, values, and beliefs) that participants’ brought to the DM process changed the context of decision-making. Many participants believed that each of them differed in regards to their personalities, reactions, the amount of prior knowledge and the amount and type of information they needed.

Well, I’d get to know [other parents], I wouldn’t tell them to do it [or] don’t do it. That would depend on who they were, and how they would deal with the information. If they didn’t want to know, I wouldn’t suggest to them that they do it. Although I would tell them my own story, you’re doing it for the kids, their future, not for curiosity…. But I wouldn’t’ push it down their throat, because some people...there’s certain things that some people just can’t deal with at that time…. [Sharon]

….[Parents] just want to get it done, right. Again, having information is powerful for people but a lot of people are just very desperate at that stage. It will depend on circumstances I’m sure. Like for my daughter it was easier because it wasn’t a life threatening thing. [Felix]

Well I think every family and every patient is totally different. And from what I can see at the hospital, many families are not interested in details, say medical details, they only want specific answers. [Gloria] I think that under our circumstances we would have done it, we did it anyways because the circumstances, I don’t know if I would just randomly do genetic testing…. I think if there’s extenuating circumstances, …. Then it’s a personal decision…. [Aida]

A lot of people can think their way through things; a lot of people cry their way through a lot of things. [They can] spend a lot of time crying.... Not everyone can sit down [and say] “that's where I want to go; this is what we've got [to] do.” [Patrick].

For some people it's really important to know all the facts…. For other people, it's not. [Cheryl]

Most participants (n=12) described their circumstances as having children who were sometimes acutely ill, requiring medical attention or hospitalization, or undergoing a diagnostic odyssey for several months or years. Two participants did not comment about their children’s circumstances and one participant described his child as “fairly healthy.”

<table>
<thead>
<tr>
<th>Circumstances described by the Participants</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acutely ill, frequent medical attention or hospitalization, or undergoing a diagnostic odyssey</td>
<td>12</td>
</tr>
<tr>
<td>“Fairly healthy”</td>
<td>1</td>
</tr>
<tr>
<td>No comments on their child’s current health/circumstance</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 4.2 Children’s circumstances
Despite the participants’ diverse personalities, values and circumstances, commonalities emerged from the data. Participants commonly spoke about their experiences prior to WGS/ES and motives to proceed with ES. Many (n=12) also felt that their own decision was a “no brainer.” These commonalities will be discussed in the next sub-section.

4.3.1 Hospital Experiences Prior to Exome Sequencing

Participants spoke about the range of both positive and negative experiences prior to being offered WGS/ES. These included feeling supported and optimistic, while also feeling uncertain, exhausted, frustrated and stressed.

4.3.1.1 Positive Experiences

Participants described some of their positive experiences in the hospital:

Everyone treats you, I felt for us, we were being treated, it was extreme professionalism. They didn't hide anything from us. They told us, it could either be downhill, uphill, they told us it could sideways or it could be good. Tons of support from everybody in that hospital, every person in the, in that whole wing there, like I mean just treated you like an equal. [Patrick]

For another participant, the positive hospital experiences and information she received about WGS/ES may have influenced her decision to proceed with WGS/ES:

We spent lots and lots and lots of time in the hospital so I have met other families that went through genome sequencing to get diagnosis for their children. There’s two families in particular that I know had genome sequencing where they found gene variants in their children that helped diagnose, so that’s kind of something that I already knew prior to it being brought up with [my son], and, back then I didn’t really read into it a lot or know much about it, but, things that I had heard about it prior to the physicians approaching me about it were, like, positive. So, I guess that’s kind of like a personal influence, right? If I had heard maybe bad things about it from families, maybe I would have asked more questions, got more information, but I already had like a pre-conceived judgment about it in a positive way. [Candace]

4.3.1.2 Negative Experiences

Participants described some of the unhelpful or negative experiences with the health care system and/or HCPs as they endured their diagnostic odyssey.
Nobody ever offered to come and help and because doctors didn’t believe me there was anything wrong [with my daughter]…. They told me I couldn’t get nursing support…. Nobody that had the power to do anything wanted to help…. We were all constantly taking [my daughter] to the hospital, and the nurses [were] always were on my side, they were always wanting to help, but their hands are tied, there’s nothing they can do. If the doctor says take your baby home, there’s nothing wrong with her, the nurse can’t do anything right? So the people that had the power to help didn’t want to help. [Claire]

I was fighting for the care discussion meeting about [my daughter’s] case. But the surprise was the care meeting was not to discuss her case, it was about [her] discharge. And when I start confronting the doctor about why now, when she is worse, why now when [no] doctor [has checked on] her for more than 2 months? We did not have… any rounds, even rounds! …. And [my daughter’s] condition was…at that point, when I start[ed] to… almost fight with everybody at the hospital… because [my daughter’s] condition was going down and at that point the doctors in charge wanted to make the discharge, when her condition was down! Falling down! [Gloria]

Regardless of their previous negative experiences, participants felt thankful that WGS/ES was available to find potential health solutions for their children.

…. But now that we know [the diagnosis], thanks to the genetic testing…it’s just, changed our lives, it’s changed her life and I know it’s going to make a huge difference in the years to come. [Claire]

…. But I don’t know another word for say[ing] how helpful it was… the genetics work. And the options they provide[d] me about the genetic test. How much thankful I am right now, and how much helpful [it was]…. Because they [made] the difference about [my daughter’s] health and her treatment. [Gloria]

4.3.2 Motivations for Exome Sequencing

Most participants (n=11) participated in research WGS/ES and few participants (n=4) participated in clinical WGS/ES. The different purposes or outcomes of research or clinical WGS/ES were not apparent to participants. Participants commonly described multiple motives for proceeding with WGS/ES. Regardless of whether they consented to clinical or research ES, most participants’ primary motivation for proceeding with ES was to find a diagnosis or answers to explain their child’s condition. Other motivations included: 1) to prepare and plan for their children’s futures and 2) to help research and others, 3) to follow the doctor’s recommendations and for 4) personal benefits. Many participants mentioned more than one motive.
Motivations for Exome Sequencing | Number of Participants (N=15)
---|---
Find a Diagnosis | 13
Prepare for the Children’s Futures | 9
Help Research and Others | 7
Follow the Doctor’s Recommendations | 4
Personal benefits | 3

Table 4.3 Participants’ motives to proceed with WGS/ES.

4.3.2.1 A Diagnosis

Participants wanted a diagnosis or answers to explain their child’s condition. Many focused on the possibility that the WGS/ES results might inform the treatment or management of their child’s current condition.

[The most important information was] finding out what specifically [was] the problem with [my child]….I mean it was a struggle when she was born trying to convince doctors that there was something wrong with her, nobody believed me, until we saw [my genetics doctor]. [Claire]

The information I was looking for always was… if possible, [a] diagnosis for her. [Gloria]

I think that the most [important information] is that you find results… [about] what’s causing your child’s illness, and finding the right medication to treat it. [Sydney]

4.3.2.2 Preparing for the Children’s Futures

Participants hoped that WGS/ES would provide information to help them prepare for their children’s futures. Participants described their intentions to prepare for the children’s futures, as well as their children’s future reproductive choices.

I’m hoping to get a bit of answers on their condition. On what their future looks like and… what we could do for them to… make sure that they stay healthy. [Tracy]

Let’s say she gets better, will there be other possibilities that she could have children with this kind of syndrome too? …If there is a possibility that this syndrome can also be inherited by her generation….and the next generation. [Francis]

I would like to give [my] children an opportunity to know what the genetic basis is so that they can make decisions on their own reproductive choices, as they go forward in life, and…even better, I would like to give them an opportunity to treat or prevent the condition from occurring…. [Sharon]
…The information is very important for me, my husband and my [unaffected] daughter because if there is something that is really important, I need to pass that information on to my [unaffected] daughter, because my [sons] don’t understand what’s going on…. ‘Cause [my daughter] may continue and get married and decide to have children and so on. [Margaret]

But my concern [is] definitely for my kids’ futures. Not that I want to know what’s good and what’s negative, but what’s the chance of their kids having these conditions, and things like that, right. [Sandy]

### 4.3.2.3 Helping Research and Others

Participants wanted to help advance research for current and future generations.

The first thing was yes [I’m] going [to] do it. I wanted to… I'm sure it's going to help another child. When it comes out, it could help another child. [Cheryl]

…. Another thing is that if we are doing some test, somebody will approach me, “well we’re doing this test, but we’re doing this study” I’m always supportive for these kinds of things so… I’m that type of person which, hey if we can help, why not? …and it’s for a good cause, good study. [Kirsten]

If we’re going to have to experience something or go through something in life, informing other people of it could help them. So if it’s going to benefit other people, by all means…I would rather be open if I could help other people [Claire].

…. We were more than willing to help this team do their research and maybe they’ll help other people. [Sandy]

### 4.3.2.4 Follow the Doctor’s Recommendations

Some participants highly trusted their doctors’ and wanted to follow the doctors’ recommendations about WGS/ES. These participants did not need much information to make their decisions.

There wasn’t much discussion, and like I said, I’m very trusting of his physician so I really just…what they say goes….If it’s recommended by the doctor then, I just go with it….The decision was definitely made by me secondarily, I would say. [Candace]

Originally I just did it just to help out this doctor because he’s done a lot for us, and not really knowing what I’m getting in to. [Sandy]

The doctor just said we wanted to do gene testing to help with [my child’s] diagnosis. So I had no contact with anybody, she just said sign this, and I trust the doctor, she’s been working with my kids for many years, so I didn't really question it. [Tracy]
4.3.2.5 Personal Benefits

Two participants were curious and wanted to find other information about their families.

If we can find out something is going to happen [to my family] then maybe somehow we can be proactive and try to do things to make it less harmful…. I thought we would find out why we have so many autoimmune conditions. That was a big thing that I wanted to know…why. [Sandy]

It gives [our family] some information….And it’s kind of curiosity as well….Just to know more about it, and what do you get out of it, I’m just curious what [results] you get out of it as well. That was the reason to do it. [Cheryl]

4.3.3 A “No Brainer” Decision

In this study’s context, most participants (n=12) felt that their decision to proceed with WGS/ES was a “no brainer,” – a decision that required little or no thought. All participants felt that it was an easy decision to be made. The ease of the decision did not differ between participants who partook in research or clinical ES, or between participants who were in urgent (i.e., an acutely ill child) or non-urgent circumstances.

If somebody offers you that kind of help, you're not going to say no…. No brainer, that's the term. [Patrick]

It was a fairly simple decision…. So [the researchers] just suggested it. We didn’t really have that much involvement in the decision making process other than saying “yes, go ahead and do it.” [Felix]
Oh it was a no brainer! I didn’t even have to think, it was like, you bet, sign us up, as soon as she asked me if I’d be interested, I was a yes. [Claire]

In particular for my son it was… an immediate decision for me because he is kind of an enigma so it was important to find out as much as possible about him…. I really didn’t know a lot and I just made the decision to do it right away…. I didn’t really contemplate my decision. [Candace]

One parent described the informed consent for WGS/ES as “desperate consent.” This suggested that parents’ decision-making required little thought because they were desperate to do anything to find out what was wrong with their child.

I think for a lot of parents at this stage where they need to get genetic testing on their kids, something has really gone wrong, and they’ll just do whatever they need to do to try get any answers they can get. A lot of desperation falls at that point….Again, having information is powerful for people but a lot of people are just very desperate at that stage….the medical side, they can’t actually get informed consent but it’s just desperate consent. [Felix]
4.4 Informational Needs

A second overarching theme was participants’ informational needs. Figure 4.3 illustrates the different sub-themes within informational needs, and the number of coding references. The colour spectrum from lowest to highest is from red to yellow to green. Red boxes have a lower number of coding references, and green boxes have a higher number of coding references.
Figure 4.3 Block tree-map diagram: informational needs
Participants focused their attention on the potential results or outcomes (i.e., a diagnosis, clear treatment for the child) of WGS/ES compared to any other information that was provided by HCPs.

[My son has] got multiple issues, so whatever happened with this [consent], it's like so what? I'm not saying I don't care, I'm like get to the point, you know, what caused it, that's where we were coming from. [Mona]

Generally for me, there’s not that much I need to know except that it’s going to provide some answers [for my daughter]. [Felix]

I wasn’t really concerned about how it was done or where things were going, I just kind of was like… sign me up if there was a possibility of finding out more about [my son]. [Candace]

I know with my daughter’s condition it’s more important to find out as soon as possible what’s causing these [symptoms] instead of jumping through a bunch of hoops and finding information, it’s like, do the test first then ask questions later. [Sydney]

Despite this concern with outcomes, participants also perceived that more detailed relevant information about WGS/ES needed to be provided to them at some point in time.

I think for myself, you need to have a basic understanding of what the DNA analysis or the genome analysis will give you. Just so you know what the test is generally going to do. Like that level of explaining of what could come out of this, a well-defined informed consent process. [Felix]

I think, just the basics for me was enough, like, this is what we will be doing… this is how we will be collecting… is it a tissue sample, is it a blood sample, that sort of information. Just kind of like a general idea of the process and a general idea of what the outcome may or may not be, that was enough for me. [Candace]

I think for most parents going through what we’re going through I think just you know, sort of just a little bit of an overview so that they at least… have a basic understanding of what it looks like, or what we’re agreeing to. [Aida]

Well what I would have liked, like even in our rushed situation is that once we decided and we went to the hospital to get the blood and everything and sent it off and all the paperwork and all that kind of thing, that we would’ve got maybe a pamphlet and then a link…. I understand with the rush, and then send it off, do all that stuff, but actually get information rather than waiting a whole year to get the results and getting the information. [Sydney]

Subsequent categories of informational needs included: participants perceived priorities for WGS/ES decision-making, volume of information, lack of knowledge and open disclosure:

“put it on the table.”
4.4.1 Participants’ Perceived Priorities for ES Decision-Making

Participants spoke about topics that they considered as important as they made a decision to proceed with WGS/ES.

4.4.1.1 Participants’ Top Priorities

The topics that seemed most important to participants are summarized in Table 4.4. The benefits, risks, realistic time frames and what to expect from the results were the most frequently mentioned topics and participants introduced these topics on their own.

<table>
<thead>
<tr>
<th>Topic</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td>Benefits</td>
<td>0</td>
</tr>
<tr>
<td>Risks</td>
<td>2</td>
</tr>
<tr>
<td>Realistic Time Frames</td>
<td>0</td>
</tr>
<tr>
<td>Expectations for the Results</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 4.4 Participants’ top priorities

a. Benefits

Participants referred to benefits as the advantages and best-case scenarios that could come from doing WGS/ES for their child. The benefits that participants hoped to receive included: a diagnosis, a better treatment plan, finding out more about the family’s genetics and disease risk, benefiting research and other children and helping to plan and prepare for their family’s future. These benefits aligned with participants’ motivation (Table 4.3) to proceed with WGS/ES. Participants often listed more than one of these benefits.
### Perceived Benefit

<table>
<thead>
<tr>
<th>Diagnosis/Root Cause</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Help better the child’s treatment/management plan</td>
<td>13</td>
</tr>
<tr>
<td>Planning for the future</td>
<td>9</td>
</tr>
<tr>
<td>Help research and others</td>
<td>7</td>
</tr>
<tr>
<td>Learn more about the family genetics/risk for disease</td>
<td>3</td>
</tr>
</tbody>
</table>

Table 4.5 Participants’ perceived benefits about WGS/ES

### Risks

Participants referred to risks as the risk of discovering incidental findings. Knowing the type of physical risks that WGS/ES posed to their children was important to participants. Participants also perceived risks as worse-case scenarios, such as discovering results that indicated that the parent or child could die from a health condition soon. Participants listed more than one of these risks.

<table>
<thead>
<tr>
<th>Perceived Risk</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possibility of Incidental Findings (e.g., results impact on child, other unaffected children, parents, disease risk, carrier status, non-paternity)</td>
<td>15</td>
</tr>
<tr>
<td>Worse-case Scenarios (e.g., parent or child might die soon, finding out about untreatable conditions about the parent or child)</td>
<td>4</td>
</tr>
<tr>
<td>Type of physical risks (e.g., invasiveness and frequency, blood draw)</td>
<td>6</td>
</tr>
<tr>
<td>No concerns about risks</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 4.6 Participants’ perceived risks about WGS/ES

Two parents said they were not concerned about risks. One parent stated “we were already in worse situation possible,” and the other parent said “I [already] lost [my son] many many times over the years.” On the contrary, one parent later stated that the “risk” of discovering incidental findings was actually not a risk:
I mean and as far as the risks, that’s to me…that’s not a risk finding out…I mean if we find out there’s something else wrong with one of us, that’s even a bonus! As far as I’m concerned (laughs) you know…to find out there’s something wrong with someone else in the family that you had no idea, I think that would be a bonus to get the heads up before something is so severe that you can’t treat it. [Claire]

c. **Time Frames**

Most (n =12) participants placed importance on receiving realistic projections about when they would receive results, and if there were going to be delays. Participants felt appreciative when they received realistic time frames. When participants did not receive realistic time frames or updates about delays, they felt anxious or disappointed. In addition, participants wanted to understand why the time frames were so long.

For us, maybe just time. It took a long time to get the results, a long (emphasis) time. Like I don't know what long time is in medical terms, but as a parent, waiting or an individual waiting for an answer for something [sigh]…. There's a big gap where we didn't get a phone call saying “yeah it's taking a little longer” whatever, whatever…. You know another 4 weeks went by or 3 weeks went by and we got a call saying “okay well we didn't find anything and it's going take another x amount of weeks”. And then it kind of did it again…? Again, …I'm not trying to…not be respectful of the time. It's kind of the opposite, if they would've said it would've took a year, then we would've just closed our eyes or ears for a year. [Patrick]

….For concerned parents, “soon” means nothing…like don't even use that word "soon." “We'll have some answers soon...soon.” Like don't make empty…it's like dating someone, don't make any promises, empty promises…. We're not kids here…we're adults right…. Look us in the eye and say…“This is going to take 6 to 12 months.” [Mona]

It’s been driving my wife nuts about just having to keep waiting and waiting or having no results back in a timely manner…. I guess the biggest thing is just realistic timelines, say “this is… the testing will start now, based on current loads, we expect to have answers at this point.” Or at least say “we will call you at this time, and tell you what’s going on.” I get that everyone is busy and people are working hard, but if we can find someone in the office to be able to just… calling every two weeks or month or whatever, “we’re at this stage with your paperwork, and this is where we stand.” People just get desperate if they’re waiting 2-3 months of no word, …they start panicking right? [Felix]

I wanted to know timelines, how long it would take, and it takes a very long time. [Aida]

For me…. And I think for other people too, it would be very important to actually [know] beforehand, like “ok, we’re drawing your blood, it’s going to go to [another country], and it could take up to…a year.” [Sydney]

d. **Expectations for the Results**

All participants (N=15) prioritized knowing what exactly they could discover or expect from the results. These included topics such as what type and how much information they would receive, what the results could mean and the fact that there could be no results. Participants had
expectations about the benefits from the primary results, as summarized in Table 4.5. Most participants hoped that the primary results would lead to beneficial outcomes such as a diagnosis, better treatment and planning for the future.

I think it’s just important to know what you’re getting yourself into like what the outcome will be….Yeah, for me that was the most important….The results, yeah, like what they could be what they could mean. [Candace]

[The] essential information is confirming if we were right about her condition…. Was [it] from some genetic syndrome or mutation? And what can we expect [from the results]…. If we find something, what can we expect, progress or no progress or anything. [Gloria]

4.4.1.2 Participants’ Agreed Upon Priorities

Most participants agreed that the following topics were important to discuss: process and procedures, possibility of incidental findings, basic genetics and genomics and implications for the immediate family. Only some participants agreed that the following topics were important to discuss: privacy and confidentiality, limitations and insurance implications. The frequencies of these topics are presented in Table 4.7.
<table>
<thead>
<tr>
<th>Topic</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td>Process and Procedures</td>
<td>1</td>
</tr>
<tr>
<td>Possibility of Incidental Findings</td>
<td>0</td>
</tr>
<tr>
<td>Basic Genetics/Genomics</td>
<td>4 Initially not important</td>
</tr>
<tr>
<td>Implications for the immediate family</td>
<td>0</td>
</tr>
<tr>
<td>Insurance Implications</td>
<td>0</td>
</tr>
<tr>
<td>Limitations:</td>
<td>0</td>
</tr>
<tr>
<td>-No guarantee for meaningful results or solutions</td>
<td>1</td>
</tr>
<tr>
<td>Privacy and Confidentiality</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 4.7 Participants’ agreed upon priorities

e. Process and Procedures

Most participants perceived that the procedure for ES was a simple blood or saliva test. Across participants, there were inconsistencies about how much detail they wanted about each stage of the WGS/ES process. There was, however, a consensus that a general overview and basic understanding of the process was adequate to make a decision about WGS/ES. Almost all (n=14) participants wanted a general idea of the collection procedure including: if there was going to be any physical harm to the child, what was being taken from which child or parent(s) (e.g., blood, saliva or tissue sample) and how long participants should realistically expect to wait for results. One participant didn’t prioritize knowing the processes and procedures.
Many participants (n=9) had further questions and wanted more details. This included: how WGS/ES was done, what researchers/HCPs were looking for, where samples were sent and where ES was being done (e.g., which country, lab or company), how often were the tests needed to be done and what happened to samples after WGS/ES was completed (e.g., discarded or stored).

.... What are you going to do with it after this procedure is done? Where is your blood going to go? …are they going to keep it for a period of time? Or are they going to… once they get the sequencing are they just going to throw it away or donate it or…? [Kristen]

Oh yeah of course, it is important [to know who is looking at it]… which company is looking at it and what [are] they going to do, if they are going to destroy it or not, or if they are going to keep it as a sample. [Cheryl]

[I want to know] just the procedure of how it is being done. Am I right, you just get from the blood samples that we have [and] you can do the sequencing from those right? …. But the sequencing… is it only done in [a specific province]? [Francis]

Well, I knew that it was going to be… a blood test and then they went into some detail of how they do it and where they send it, those types of things, you know, the general aspects of the test of how it is done and where it is sent, and what they’re exactly looking for. I think that’s important for parents to know. [Aida]

f. **Incidental Findings**

<table>
<thead>
<tr>
<th>Topic</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td>Possibility of Incidental Findings</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 4.8 Participants’ agreed upon priorities: possibility of incidental findings

Aside from the primary results, all participants (N=15) spoke about incidental findings that could “pop up” as “surprises,” “scary stuff,” or “other things [that] may come back.” Participants agreed or placed importance on knowing the possibility of incidental findings being discovered with WGS/ES. Most participants (n=8) prioritized their personal choices or assumed that they would be given choices on which findings they wished to receive or not receive.
...part of the informed consent process needs to be that, “we’re going to do this”, let’s see why she [has a condition] but we might find out... [that] she might have some other challenge. People should be informed that... to be aware that the testing may be give you more than what you were looking for... To me it’s just a matter of how much information you want or do you think you can handle pretty much. And I am sure I assume somewhere along the process, you can choose not (emphasis) to know the results of the DNA tests. If there isn't, there should be an option. [Felix]

...if you will put it in the consent that, is this all you want? Then... that information would be given to you, but if you want...well do you want surprises? (laughs) Then it will be given to you right? [Kirsten]

[Unanticipated findings]...that, you should be aware of, or you should know about it before you do any test.... Because there is always a chance they can find something else if they do certain kinds of tests.... [Cheryl]

I don’t know if this is possible but would say... people should have a choice, here's the main goal of the investigation, they know what that is, they check yes I want to know the results. But they get the option of, no I don’t’ want to know anything else, or, I want to be asked before I’m told. [Sharon]
I think it’s important to also explain that... you maybe be looking for something, but that outcome may not be what you’re looking for. [Aida]

It would be nice for either a website or somewhere where we could have like a checklist or something that maybe what you want to check for. Like, what do you want to find out, and then you can pick whatever you want. [Sydney]

g. Basic Knowledge about Genetics and Genomics

<table>
<thead>
<tr>
<th>Topic</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td>Basic Genetics/Genomics</td>
<td>4 Initially not important</td>
</tr>
</tbody>
</table>

Table 4.9 Participants’ agreed upon priorities: basic genetics and genomics

The amount and need for basic genetics and genomics knowledge varied among the participants, from "not necessary," and “I knew a lot already,” to “I would have liked a little bit more information.” Most (n=10) participants only wanted a “basic understanding”, an “overview,” “Coles notes version”, a “synopsis”, a “bird's eye view.” Some participants (n=6) claimed that they had prior basic knowledge of genetics, genomics and sequencing, but still prioritized the importance of educating parents that are making decisions about WGS/ES in the future.
I really just wanted to know the basic underlying …the basic stuff. I had a small grasp. They sort of enlightened me a little bit more, but I think every parent is going to be a little bit different in that, I think it’s good to give a little bit of a synopsis. [Aida]

My husband, he knows all this… genetic process. So it wasn’t something new for us. [Margaret]

I knew a lot of this stuff already, the DNA, I mean I know all that. Yes, but yes, I think I did need to have that basic idea. [Sandy]

Some participants (n=4) reported that basic knowledge about genetics and genomics was initially not important when they were deciding to proceed with WGS/ES. However, it became more important when they were closer to receiving the results.

Initially, no. I think once it was getting closer to finding out results and then once we found out there was a result of the testing then it was more…like okay I need to understand what’s going on now (laughs). Because… we found this gene that has a mutation and then all of a sudden I was having information thrown at me about variants in my genes, variants in the father’s genes, and how it affected [my son’s] outcome, so then, it became important to learn about how…the DNA works, and how they look at the DNA to come to those conclusions… so that’s probably the point where I started to learn about it, ask questions. I think I asked a ton of questions. But initially, it was not… I wasn’t too concerned about all the specifics of it. [Candace]

\[h. Implications for the Immediate Family\]

<table>
<thead>
<tr>
<th>Topic</th>
<th>Not Important</th>
<th>Agreed on importance (agreed with other participants or asked by the researcher)</th>
<th>Important (Participants brought up)</th>
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</tr>
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<tbody>
<tr>
<td>Implications for the immediate family</td>
<td>0</td>
<td>9</td>
<td>5</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 4.10 Participants’ agreed upon priorities: implications for the immediate family

Almost all participants agreed (n=9) or placed importance (n=5) on being informed about the implications of WGS/ES for the immediate family. These implications included discovering incidental findings for the parents themselves or the other unaffected children in the family.

[I want to know]…basically what else we are at risk for…in the future of my kids or myself. And which of course, I mean if you think about it, from a non-medical point of view, it is kind of scary (laughs) isn’t it? [Sandy]

If some more information that would affect the family, then we’d like to know those. [Francis]
…if there is something for my kids which is important to know for the health of... the kids, future kids or whatever. For me, it's normal to tell them... then when they are old enough and let them know about it.... But for other parents, it's... maybe it's not so important.... I think it's important to inform the parents... how it could be, how it could affect them, and ... because... some don't realize... don't realize it, or don't know about it, that there are different kind of outcomes. [Cheryl]

I would have to ask…. Any ramifications of the results to me or…? [Felix]

Out of the nine participants that agreed on the importance of telling families about the implications of WGS/ES on the immediate family, four of them were unaware of the implications of WGS/ES on the family or it did not cross their minds.

…But now looking back there was actually one, like, nothing major, but there was an incidental finding with my genes as a part of the whole sequencing for my son that was kind of, it just popped up, it was kind of a surprise, and, so yeah, it would have been good to know beforehand that that sort of stuff could have popped up. [Candace]

… like I didn’t actually know how big this test could be or how important this is, like how much stuff you can find out about with your genes. [Sydney]

My [other] son was also tested, my second boy. And I actually haven't heard back anything negative….cause he is so young, I am not thinking of "oh he's going to get married and have kids" at this point…. To be honest with you, we never even thought about what we're going to find, like never even asked… never crossed our minds to ask, are we going to have good news or bad news. [Mona]

Mine either, yeah. Their plate is so full already, what's another one? I don't know, I couldn't imagine that there's going to be anymore, like I don't know, I never asked. [Tracy]

Only one participant did not mention this topic. Interestingly, participants lacked awareness or did not initiate discussion about implications of WGS/ES beyond the immediate family.
i. Insurance Implications

<table>
<thead>
<tr>
<th>Topic</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td>Insurance Implications</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 4.11 Participants’ agreed upon priorities: insurance implications

Insurance implications included topics such as nondisclosure of participants’ results to insurance companies, and how to proceed with future life insurance claims or new purchases of life insurance. Only one participant brought up the importance of informing parents of insurance implications, while six participants agreed that it was important to inform parents of insurance implications, if any. Most participants (n=8) did not mention this topic. It was apparent that most participants did not realize that there could be insurance implications.

well don’t forget to [mention it], because there’s a lot of people that… have insurance and then, it will affect their claim, whatsoever. If something happened during the study or whatever, or if you get a disease during the study then you will make a claim and they will refuse you… and you don’t know right? [Kirsten]

Yes [mention it], so I would know what to do if I ever had to fill in that question or not, ‘cause you know, insurance companies, if you don’t fill it in absolutely 100% truthful, they’re going to… if you need to… claim something, they will find whatever reason they can to not pay out, so yes I’d like to be as truthful as possible (laughs). [Claire]

I think it would be a problem if [the insurance companies] know that you’re cancer prone then maybe your insurance premiums will be higher….I think it should be put on the table so that you know that it could affect… if it will, Uh Oh! Then we have… (laughs) yeah, that’s a good point, yeah insurance is… it’s quite expensive already right now, and if it will affect it, then I think we’ll have second thoughts. [Francis]

I didn't know…I didn't know [insurance companies] have questions like that… implications, I mean if we have to let them know we would, but I mean right now my husband and I already have life insurance, and I don’t think… I think it’s locked. [Sandy]

…I don’t really have any report or results, if an insurance company would ask me for that, I wouldn’t know where to begin because I never received one myself….We never got life insurance, but we’re very interested in it and we’re actually thinking about getting one… [Sydney]
### Limitations

<table>
<thead>
<tr>
<th>Topic</th>
<th>Not Important</th>
<th>Agreed on importance (agreed with other participants or asked by the researcher)</th>
<th>Important (Participants brought up)</th>
<th>No Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Limitations:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>- No guarantee for meaningful results or solutions</td>
<td>0</td>
<td>4</td>
<td>4</td>
<td>7</td>
</tr>
<tr>
<td>- False positive/negatives</td>
<td>1</td>
<td>4</td>
<td>0</td>
<td>10</td>
</tr>
</tbody>
</table>

**Table 4.12 Participants’ agreed upon priorities: limitations**

Limitations included comments reflecting the possibility of false positives and false negatives, and considerations that there were no guarantees that WGS/ES would produce meaningful results or answers. Four participants agreed that the possibility of false positives and negatives should be discussed with parents before proceeding with WGS/ES.

It’s… I mean it’s the same with anything in life. You roll the dice sometimes right. But again, it’s part of the informed consent process people need to be made aware of that they maybe errors or there may be false positives. [Felix]

Yeah, definitely. Obviously… I think obviously that would be important, so… I think that one’s pretty black and white. Nobody wants a false result of a test. [Candace]

Six participants agreed or prioritized knowing that there were no guarantees that WGS/ES would produce results or answers for their children.

Every conversation, every phone call, it was repeated that, we're not going to fix my son, but this is for future kids. [Mona]

I was explained that there could never be a solution… They were pretty clear that it would take some time, and… they might come up with some information or they might come out with nothing. [Tracy]

Even if she has a certain condition, sometimes there’s nothing that shows up, and I understand that and it’s a frustration. And it’s a part of informed consent that would be important, that they’re told you know that this may result in no information, no useful information. [Felix]
…like I think it was made pretty clear before the testing that it was possible that we didn’t find out any information at all…. So, I was aware that we might not find out anything from it but the hope was that we would. [Candace]

Before the test [the doctor] did tell us…. Everything… what the test… may show, may not show, “you may go through it and not get results…” so those were all put on the table. [Margaret]

…like how… how far can [we] go and… there are things that… will not be answered or unanswered. [Kirsten]

k. Privacy and Confidentiality

<table>
<thead>
<tr>
<th>Topic</th>
<th>Not Important</th>
<th>Agreed on importance (agreed with other participants or asked by the researcher)</th>
<th>Important (Participants brought up)</th>
<th>No Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Privacy and Confidentiality</td>
<td>2</td>
<td>3</td>
<td>2</td>
<td>8</td>
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</tbody>
</table>

Table 4.13 Participants’ agreed upon priorities: privacy and confidentiality

Some participants (n=5) agreed or prioritized the privacy and confidentiality of their families’ identities associated with their results. Participants did not express concerns about privacy and confidentiality and some even mentioned their confidence and trust with current privacy safeguards. Many participants (n=8) did mention the topic of privacy or confidentiality.

…it’s a part of the medical world…. People that are usually involved in research or the health care world are very private people in general so it’s never been a concern to me where his samples go, where his information goes, I just sign. [Candace]

I’m pretty confident in current privacy, that there’s… almost over-protection of private information. [Sharon]

Some participants (n=4) brought up potential concerns related to privacy and confidentiality, which included: the importance of not openly sharing the families’ identities and results with unauthorized people or companies and the eventual fate of the results.

…it was [a] study [that] was just used for the research in [a local hospital], so if there’s other companies coming in or they would ask us to have a look at it, maybe [I] would change [my mind]… it depends on the company who want[s] to have a look at it…. I mean you don’t want to spread it out everywhere actually because you never know what people [are] going to do with it [Cheryl]
…what I want is privacy. It’s not like “ok I need the DNA of this such and such person” and there you go [snaps finger], you have access to it…. I want privacy. [Kirsten]

I think for sequencing there is more delicate information that you can get, right. So having this delicate information in the hands of other people could be dangerous too. So it’s really good that you have the right consent and you inform the people that these are all the possibilities. And that it will be kept confidential…and things like that. [Francis]

…what was going to eventually happen to the information I think would be important to any parent… I understand that getting the information out to the scientific community in… an anonymous way will help find the answer…. But maybe emphasizing that the children would not be identified, that the family would not be identified…. As long as the data was anonymized in terms of their names, that there would be no leak through to say insurance providers or something like that. [Sharon]

4.4.1.3 Less Common Topics

Participants also discussed other topics. However, these topics were discussed less frequently and were inconsistent across participants. Furthermore, some of these discussions were introduced by the researcher or other participants. The less common topics included: financial costs to the family, ownership and access to the results and data, discussion of more options, new scientific discoveries and implications of the results outside the immediate family. These less common topics are summarized in Table 4.14.
<table>
<thead>
<tr>
<th>Topic and Description</th>
<th>Number of Participants (N=15)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Not Important</td>
</tr>
<tr>
<td><strong>Financial costs to the family:</strong> Participants wanted to know if WGS/ES would cost anything for their families and if they had to financially prepare for it.</td>
<td>0</td>
</tr>
<tr>
<td><strong>Ownership and access to the results and data:</strong> Participants wanted to know who owned or would have access to their genetic data and results. For example, they wanted to know the data belonged to the research institute and would not be withheld from them, leaked to external companies (e.g., insurance, pharmaceutical) or to the court of law.</td>
<td>0</td>
</tr>
<tr>
<td><strong>Discussion of more options:</strong> Other than being told by genetic HCPs to not have more children or to go ahead and proceed with WGS/ES; two participants wanted to know and discuss more options to help mitigate or solve their current situation (e.g. undiagnosed child, child with a rare condition).</td>
<td>0</td>
</tr>
<tr>
<td><strong>New scientific discoveries:</strong> Participants agreed that it was important to know the possibility of new scientific discoveries from their WGS/ES results as research progressed over time.</td>
<td>1</td>
</tr>
<tr>
<td><strong>Implications of the results outside the immediate family:</strong> Two participants agreed that they would like to know that WGS/ES results could involve or affect members beyond their immediate family.</td>
<td>0</td>
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</tbody>
</table>

*Table 4.14 Participants’ less common priorities*
4.4.2 Volume of Information

Other than discussing specific topics that were important to them, participants also described the volume of information they received. Each participant received varying amounts of information (i.e., educational information and/or informed consent) from different HCPs. Most participants (n=13) felt that they received “enough” information.

I know that my husband and I didn’t feel nervous or [that] we didn’t get what was going on, or that we needed more information…. We felt… that we were doing the right thing. And that this was going to be something that we were going to do… [and] we didn’t feel slighted in any way. [Aida]

I’m an easy going person, and I think what they did tell us was…very informative, probably more than I even would have thought to ask you know, so I was impressed…. But… I wouldn’t ask for more than what was done and… the information I was given was perfect. [Claire]

Two participants felt they received “too much” information.

It was too much information. It’s like one thing leads to the other…and to the other…it’s like too much to remember …it’s a big thing….Especially with a lot of things going on with my son…and you know…you’re trying to absorb things…little by little [Kirsten]

It's just too much information…there was too much. Just…take the blood work already…. Just do it… don't need to like read through hundred pages just for one blood sample…. It was just a big ordeal for a little blood sample. [Mona]

Even though most participants received “enough” information, they still spoke about the large volumes of information given to them prior to WGS/ES. Many participants (n=10) thought that large volumes of information given at one point in time as not helpful.

There was a lot of paperwork. I remember signing and they gave me stacks of papers, and… I don’t know, I didn’t really have time to go through, read each and every document....I don’t… you know it’s kind of like, you ask a baker to bake you a cake, you don’t need to know all the little steps they take in the meantime because if you don’t know how to do it anyways, what’s the point of being told? The outcome is the more important part, that’s where I’m curious you know. [Claire]

I guess if I had asked… the questions in the beginning, then, it wouldn’t have been just a flood of information and flood of knowledge all at one time. I would have kind of known a little bit along the way. [Candace]

…you don’t want to overwhelm somebody…. I know some people love lots and lots of information, but I know sometimes you actually can get too confused with too much. [Sydney]

It's got to be pointed and direct. I'd say, you can read 25 pages on what this may result in, but the big thing is… you may find out some really scary stuff… some real basic things that people know that it's not just a simple blood test. [Felix]
4.4.3 Lack of Knowledge

Despite the fact that most participants (n=13) reported receiving “enough” information during their DM for WGS/ES, several participants (n=8) later realized that they had personal knowledge gaps. Some of these knowledge gaps included: the extent/scope of WGS/ES, where their samples were sent to, time frames and when to expect results back, incidental findings, possible insurance implications and consideration of the results affecting relatives beyond the immediate family.

Well, actually, it’s the first time that I came to hear about this genome sequencing so I really don’t know anything about it. [Francis]

Originally I just did it just to help out this doctor because he’s done a lot for us, and not really knowing what I’m getting in to…. But definitely I would have liked a little bit more information. But regardless I think we still would have gone ahead with it. [Sandy]

With my situation… I didn’t even know where [our blood samples] were going…. It was never mentioned that it was going to [another country] and it could take up to a year, or anything like that. So honestly I thought I was just like one of those other blood tests that they can run, you know? I didn’t really know at all, anything about this, until afterwards (laughs). [Sydney]

No, I didn’t really think about it. Thinking about it now, obviously you know if it’s genetic, then outside the immediate family is going to get involved too, but I didn’t put any thought into that at all. [Felix]

Participants who spoke most about “lack of knowledge,” lived greater than 30 kilometers from Vancouver, or outside of British Columbia (Figure 4.4). Participants who completed high school spoke most about “lack of knowledge.” However, participants who completed college or university also spoke about “lack of knowledge” (Figure 4.5).
Figure 4.4 Lack of knowledge matched against participants’ residence
Figure 4.5 Lack of knowledge matched against participants’ education
4.4.4 Open Disclosure: “On the Table”

Some participants (n=4) used the phrase “on the table.” This phrase suggested the importance of transparency: HCPs openly disclosing all relevant information and informing participants of the potential of both good and bad news.

They have multiple diagnoses, so they're battling so much right now, like why not get it all on the table? [Tracy]

We were explained that it may be something that we never find or it may be something that we can find. Can or can't do something with…totally laid it out on the table. For us it was awesome as far as the layout on what they were doing and why they were doing it. [Patrick]

4.5 Psychosocial Needs

The third overarching identified theme was participants’ psychosocial needs. Participants commonly spoke about the relational and psychosocial aspects of their DM process. This included their relationships with the HCP and their well-being as parents. Figure 4.6 illustrates the different sub-themes within psychosocial needs, and the number of coding reference. The colour spectrum from lowest to highest is from blue to yellow to red. Blue boxes have a lower number of coding references, and red boxes have a higher number of coding references.
Figure 4.6 Block tree-map diagram: psychosocial needs
4.5.1 Relationship with Health Care Professionals (HCPs)

Most participants (n=13) described the importance of the HCP-parent relationship during DM about WGS/ES. Two participants lacked a relationship with their HCPs and wished they could have communicated their fears, and had “somebody with a personal touch” or “access at a personal level”. One participant did not comment on the parent-HCP relationship and another participant said that there “wasn’t much of a relationship.”

<table>
<thead>
<tr>
<th>Psychosocial Need: Relationship with HCP</th>
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</tr>
</thead>
<tbody>
<tr>
<td>Importance of the HCP-parent relationship</td>
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</tr>
<tr>
<td>Parents lacking a relationship with the HCP</td>
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</tr>
<tr>
<td>No comments or relationship not present</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 4.15 Number of participants who placed importance on the HCP-parent relationship

Participants prioritized different attributes that existed within their HCP-parent relationship. They most frequently spoke about certain attributes, which are listed in Table 4.16. Although less frequent, participants also described other attributes in the HCP-parent relationship including: connection, openness, transparency, comforting and intuitive, supportive, “like family”, integrity, fondness, respectful, listener and caring.

<table>
<thead>
<tr>
<th>Attribute in parent-HCP relationship</th>
<th>Number of Participants (N=15)</th>
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<tbody>
<tr>
<td>Trust</td>
<td>7</td>
</tr>
<tr>
<td>Continuity, ongoing relationship</td>
<td>7</td>
</tr>
<tr>
<td>Knowledgeable and informative HCP</td>
<td>10</td>
</tr>
<tr>
<td>Effective, Clear Communication from the HCP</td>
<td>6</td>
</tr>
<tr>
<td>Collaborative</td>
<td>4</td>
</tr>
</tbody>
</table>

Table 4.16 Participants’ most frequently stated aspects of the HCP-parent relationship
A HCP-parent relationship with these attributes helped participants feel at ease with the DM process.

To every parent that has a special needs… that personal…. [My doctor] knows my cell number, she knows how to text me. Her secretary talks to me… like there's almost like a relationship… like we want to figure it out. We're going to do it together. [Mona]

I really like [my genetics doctor] and I think she’s very nice to talk to… she’s very informative… she has a great aura about her… she’s soothing, she seems to have like a soothing… aura about her, I don’t’ know how else to put that… so it’s comforting. You know I always felt like I was in good hands, or that [my daughter] was in good hands. [Claire]

[My doctor and I] have a close connection. I was glad she was there, and I knew that if she supported it, it was a good choice, and made it very easy for me…. I really just put my… I have a very strong relationship with my [doctor]…. And, you know, a lot of [other parents], some of them want… needed more information and maybe it’s ‘cause they don’t have the relationship with their doctors. [Tracy]

So myself… our family and my son, we had already established our relationship with his team of physicians and primarily the physicians that were interested in his genes. We had established a really good relationship and built a foundation of trust so, any type of communication we had about the genome sequencing was really just suitable at the time….. I have a lot of trust in [my son’s primary physicians], so they were definitely my 100%, my main influence on deciding to go ahead with it. [Candace]

My husband didn’t feel like we needed to sort of explore more. We felt comfortable, and remember we’ve also been working with our [doctor] and our nurse for over a year, and we see them weekly… more than weekly…. We have a very good connection with them…. [Our doctor and nurse have] been working so hard with us and supporting us and they’ve been incredible… they’ve become almost like family to us, because well, we see them more than family really, and they see us at our most vulnerable and intimate times really. [Aida]

4.5.2 Parents’ Well-Being

Many participants (n=10) felt that their own well-being was affected or compromised due to their current circumstances. Participants described feelings of fear, anxiety, uncertainty, frustration and feeling out of control.

I did a lot of reading, which [long pause]… it then scared me a bit. But then I went back to [my doctor] and… we sat down and we talked… and I was able to ask a million questions, and she was able to… set my mind at ease. [Tracy]

Just… the frustration of not knowing…. My daughter… has challenges… and it all adds up to the ongoing conditional challenge plus having no control over when the information comes [from the HCP]. So basically you’re out of control, and when you don’t have control over the situation, it’s just frustrating. [Felix]

I was very anxious to be honest with you, I was extremely… ‘cause like I mentioned, the doctor told me it would take three months, so the first few months I was very nervous every time I saw the hospitals’ number on my phone calling (laughs). I was always very nervous to… (laughs) hear the doctor calling me about the results, and so that [was] a good little while, and then I was still nervous up to six months. [Sydney]
I mean I have no regrets from doing [WGS/ES] and the only thing that… like when it comes up when I’m talking, it’s like, God, do I really want to know [the results]. That’s definitely something that’s eating me. [Sandy]

I don’t know what’s going on actually, it’s like “oh okay...” maybe they can use [our families’ DNA samples], or maybe they are not interested in it. I just have no clue what happened to the samples. Which is… which is sad actually. [Cheryl]

Two participants reported feelings of distress and vulnerability when other people had challenged them on the basis of having an undiagnosed child who was frequently ill or about their decisions to proceed with particular medical treatments or genetic testing.

There was… one person… related to me…. she thought I was purposely making [my child] sick... basically accusing me of [making my] child sick…. It took, a scientific diagnosis, to get that person off my back, and apologize... that was probably one thing that…. I mean yeah it hurts to have somebody accuse you of something so horrible…. (crying) [Claire]

There’s a lot of ethical dilemmas that come with a lot of the decisions we’ve made. I mean people have even challenged us when… people found out our daughter did chemo, well, people challenged us on that…. You sort of, you learn…who you can sort of discuss things with at that time, because you’re so vulnerable…. Sometimes people get very you know opinionated on things about chemo and genetic testing and things like that…. With genetic testing...you know, challenging on the fact of people get into the “you’re making the perfect person” conversation. [Aida]

Participants valued the instances when HCPs recognized and considered their well-being as parents in a vulnerable and stressful situation.

Parents are... are people, and I think we’ve been really blessed with having a really good team that treats us really kindly, professionally, and treats us with integrity, I think that’s really important for doctors to always remember that, you know, that we’re… especially in this situation with oncology and life threatening diseases that we really are vulnerable, and we’re in a very serious crisis situation…. I know that with studies and everything it’s interesting and exciting and all those things, but to remember that behind the scenes, that someone is really suffering and vulnerable, and I think that’s a really important component to always remember…. It’s always good to check in with parents to see where they’re at also. [Aida]

Something [that] I liked from [my genetic doctors] was that they were thinking …. everything about the baby and also caring for [me]… “Are you ready?… [you’re] not [in] too much stress? [Do] you have too much stress here?” …I like[d] that they were worried about me. [Gloria]

[HCPs can support us]… afterwards… just a matter of keeping contact. Obviously [the HCPs] don’t have time to handhold everybody. But to this stage, if you’re doing genetic testing usually it means something’s pretty serious. [Felix]
4.6 Supporting Strategies

Participants made suggestions or agreed with other participants about strategies that would be supportive during the DM process for WGS/ES. They felt that these could be helpful for parents undergoing similar experiences. These strategies are summarized in Table 4.17. Figure 4.7 illustrates the different sub-themes within supportive strategies, and the number of coding references. The colour spectrum from lowest to highest is from green to yellow to red. Green boxes have a lower number of coding references, and red boxes have a higher number of coding references.
Figure 4.7 Block tree-map diagram: suggested strategies
<table>
<thead>
<tr>
<th><strong>Supportive Strategy</strong></th>
<th><strong>Description</strong></th>
<th><strong>Number of Participants (N=15)</strong></th>
</tr>
</thead>
</table>
| Brief and Understandable Summaries | - A “basic understanding,” “big pointers,” “Cole notes,” versions of the educational material and the informed consent process  
- Laymen’s terms, avoid medical jargon, at the parents’ level of understanding | 11 |
| Varying Media and Approaches | - Tailor to different communication needs, personal preferences and learning styles  
- Examples: face-to-face discussions, telephone, email, teleconferencing, web-conferencing, mail, pamphlets, frequently-asked questions, video, online resources (web-links, videos, media) | 10 |
| Layers of Information: | - Receive adequate and relevant information to make an informed decision about WGS/ES, but also receive additional layers of information through varying methods as required:  
1) Educational material and/or the informed consent ahead of the informed consent discussion  
2) Take-home material available before and/or after the decision  
3) Help direct parents towards credible resources (e.g., other people, websites, books, videos) or receive additional information directly from credible sources (e.g., the HCP)  
4) Knowledge of and access to available HCPs to answer additional questions before, during or after their decision. Accessibility can be through varying media. | 11 |

Table 4.17 Participants’ suggested strategies
4.6.1 Brief and Understandable Summaries

Many participants – especially those who had acutely ill children – said that high volumes of information were not helpful. Participants preferred brief and understandable summaries of the educational material and the informed consent process. Participants wanted a “basic understanding,” “straightforward and to the point,” “abbreviated,” “birds’ eye view,” “big pointers,” “outline,” “Cole notes,” “synopsis”, or “general idea” of the information and what to expect from WGS/ES.

Maybe an abbreviated option of what the product and what it will result in. You're doing this testing, and your options when this comes out are you know "you can do something, you can't do anything” …. Most of our information… starts out with the bottom line up front. Sort of drop one paragraph of "this is really what you're about to read", and then it goes into detail as required…. And so basically they just gave me the bottom line up front. this is the worse things that can happen, this is the advantages that could come out of it and it worked really well. Sort of an abbreviated version of the best case scenario…worst case scenario. [Felix]

I think it's good to give a little bit of a synopsis…. Most people want the Cole notes version. I mean you will find people obviously who want everything, and that’s different…. I think for most parents going through what we’re going through…. Just a little bit of an overview so that… at least parents… have a basic understanding of what it looks like… of what we’re agreeing to. [Aida]

…like an outline is great, somebody telling you, “ok we’re going to do this blood, but you have to get it drawn, you have to go to the hospital, it’s going to get to [another country], it might take up to a year”…kind of just the big pointers about it…to tell the family so they kind of know what to expect and then get the test done and then get information while you’re waiting, I think that would be definitely good. [Sydney]

Participants valued explanations that were in laymen’s terms or pitched at their level of understanding.

I think the best thing would be a summary…an easy summary about this. So it's understandable for everybody and easy to understand. [Cheryl]

…there was no word too big that they would've used. They would've said it and the guy looked at me and he'd say, “do you get it or not?” And If I didn't get it, he would say it again a different way. [Patrick]

…these words are new to me… like I’m an ordinary Jane, you know so… this is new to my vocabulary…. Because I’m just an ordinary mom…. One thing I wanted to emphasize is that [the doctor] leveled with me… he explained to me things step by step… the simplest way that he did… so I would be able to understand things. So that’s very important…. This is not our daily language (laughs). [Kirsten]
It’s good to sort of maybe say it in layman’s terms for people who aren’t in the medical professional field…. I’m not trying to say that parents aren’t intelligent, but if they’re not in the medical field then it is important to give them the layman’s terms as opposed to the big medical spew. I think it’s important to lay it out and supply it in a sense so that parents can understand. [Aida]

…as long as it’s not too medical, you know what I mean, with all the medical terms. But if there is something online that would help me decide, I would have…. As long as it’s in more understandable layman’s terms, I definitely would be interested. [Sandy]

Well the most important support in this process is the explanation about everything… because even if I am reading or studying by myself… of course… [there] are a lot of technical words… [and] process[es] that I cannot understand. [Gloria]

4.6.2 Approaches to Information Delivery

Participants acknowledged that every person has different communication needs and personal preferences for obtaining information.

I think a lot of it depends on how you are wired, myself and a lot of people are very much goal orientated, like cold information…. There has to be some way to cater it to different individuals and find different ways of approaching it, cause there's no way you get two people that want stuff the same way. If it was myself and my wife, again, we have very different ways of approaching things. [Felix]

It’s all just been kind of… go with the flow… and that works really well for us… with our doctor… the way we work together. Maybe for… other patients and physicians it would be different. Physicians all have their different personalities of how they like to give information and patients have different ideas of how they like to receive it. [Candace]

I’m more of a visual person so yes, I like a pamphlet or a DVD or… online. I know for my husband he likes face-to-face and then he’s good… he asks all the questions right away and he’s very into the scientific aspect of it and once he understands it he’s good… whereas I need a little bit more. Just different people right, different styles of learning. [Aida]

Participants (n=10) suggested varying approaches and media for receiving educational materials to support the DM and informed consent processes. These included: face-to-face discussions, telephone, email, teleconferencing, web-conferencing, mail, pamphlets, frequently asked questions summaries, DVDs, and online resources such as web-links to additional internet resources, videos and media.

I think maybe just something like [a video], very straightforward and to the point. I mean I wouldn’t sit and watch a 2 hour thing right? [Sandy]

Less optimal but possible is a video conference, something where you can actually see the person’s body language and… certainly some people live in very remote areas. [Sharon]
I think definitely the information to get was good on the paper, but I’m a visual person too, so I think those [video] links…that was really really good….I think with the videos and having the sheets in front of you I think that’s a pretty good explanation. [Sydney]

Most participants (n=12) had a preference for a face-to-face discussion regarding the initial informed consent process, the return of results, receiving “bad news” or any situation that required multiple questions or explanations.

I would say the best… the most engaged you're going to get... like a real meeting is going to be the best one. For something important for getting consented on something as big as this…. It’s a decision that has emotional ramifications as well, and I think we listen better when we’re talking. Like I say, more engagement…. You’re sitting right there, you’re kind of just forced to really deal with it for 10 minutes, or half an hour whatever it is, I think it’s better. [Sharon]

…let's have a dialogue face-to-face, because my son was having [multiple] issues…sometimes it's too much information on the Internet. I think [another parent] said it…have the dialogue and direct them to the Internet…onto the site. But…we have hearts, we are people and we are probably old school still…maybe the new generation will want no face-to-face contact (laughs) [Mona]

I think with this kind of thing…because this is…sensitive… it has to be face-to-face….It has to be explained very well like it was to us…. That face-to-face definitely needs to happen….Because like what I said, it’s a sensitive thing. [Kirsten]

We prefer face-to-face. Even though we were going back and forth with emails as in appointments… You know, we preferred face-to-face…. Because we had questions back and forth. So we preferred face-to-face [Margaret]

Of course face-to-face is best but… is not always possible because we don’t live in the same city. But I would prefer face-to-face…. If I was going to get some news, like some bad news I would definitely want it face-to-face. [Sandy]

4.6.3 Layers of Information

Participants favoured receiving layers of information rather than receiving a large volume of information at one point in time. Participants wanted to receive adequate and relevant information to make an informed decision, and then receive additional layers of information as required. Each participant had different perceptions of what was “adequate” or “relevant” information and desired different amounts and types of information. Receiving layers of information included: receiving information in advance, receiving take-home material, receiving information from credible sources, and access to a HCP for additional questions.
4.6.3.1 Receiving Information in Advance

Receiving information in advance included receiving educational material and/or the informed consent forms ahead of the informed consent discussion so that participants had time to review and absorb the information given to them.

…educational material should be provided in advance of the consent because rarely can something sit and absorb a lot of the information and then reasonably be able to provide consent for something. So there should be a period to read and review the materials before the consent is signed. [Sharon]

I think what makes the most sense to me, is you have the information prior to doing the consent, you’ll have a more informed consent…. I think the information should be… [sent] via email or have a link or if snail mail works… if you have time then do that way as well. I don’t think a meeting before the consent meeting is going to help, but at least then people who want to read the information have the opportunity. [Felix]

4.6.3.2 Take-Home Material

Participants suggested that take-home material be made available to review at a different time.

…I always think it’s good to have a walk-away, like something to give parents like a pamphlet or a video and the reason I say that is because, generally when you’re being taught something… you’re listening but maybe once you’ve left you’re like “oh, jeez I forgot to ask that” well if you have a pamphlet or, a Q and A type DVD, you can always put that on and be like “Oh, okay, I get it, oh right, that’s what they were talking about” I think it’s always… I prefer that, I like a handout or a DVD. [Aida]

Like things that would explain…to me stuff…. Getting me to sign papers…that would be nice… in person. But then to support that…[that] could be online… could be a hard copy for me to take-home or web-based. [Kirsten]

Participants with more urgent circumstances or for those who were more concerned about timeliness suggested take-home material or additional information after their decision.

Well what I would have liked…even in our rushed situation, is that once we decided and we went to the hospital to get the blood and everything and sent it off and all the paperwork and all that kind of thing, that we would’ve got maybe a pamphlet and then a link….That would’ve been perfect…. I understand with the rush, and then send it off, do all that stuff, but actually get information rather than waiting a whole year to get the results and getting the information. [Sydney]

…I think it’s a matter of timeliness, then I’d do the consent and take the stuff home later. Because people are going to consent anyway, I can’t imagine anyone not consenting. [Felix]
4.6.3.3 Credible Sources

Participants also valued being directed to credible resources or receiving additional information from credible sources such as a HCP.

In the office, no one is going to read it, no one is going to be paying attention (small laugh). They’ll look at it, but no one is going to be processing any of it, I’m pretty confident. Except if they take it home with them, then they can get information later. You know, most people will be jumping on the Internet these days just Googling whatever they can get their hands on. So it’s better to have some information to compare it against, so they don’t just freak out with bad information. [Felix]

...a lot more people would be at ease if they would actually get information strictly from an actual doctor or a professional, somebody that deals with it, and links to the pages to read, rather than you trying to Google it, search for it (laughs) and then who knows what site you end up on…so I definitely think that actual families would feel a lot better about having someone personally send them an e-mail with all the links, and then… or videos and whatnot (small laugh). [Sydney]

…the internet is flooded with misinformation on many levels, so I personally like to keep that simple, if there’s you know, webpages that the [health] professional says “you know what this is a really good site, this is medical information, this person’s in charge of this site, or you can go to children’s website and you’ll find this” that’s different, than just ‘Googling’ and so that I don’t just use Google for my information. [Aida]

Some participants had avid supporters of their decision to proceed with WGS/ES and one participant suggested that credible take-home information or resources about WGS/ES be available for parents so they can also be prepared to educate and engage other people involved in the child’s life.

When I told [our family] “oh yeah we’re doing this testing, genome sequencing” it wasn’t like “oh, I know what that is!” They were like “what is that?” (laughs) So, you’re kind of trying to explain something that at that time I didn’t really know what it was (laughs) really about, really…. and so [my family members] want pamphlets, they want…e-mails…or books. It’s definitely helpful to have things like that, not just for the people who are going through it, but like the immediate families…that are very interested in it and they want to share this with you, or you know support you, but they don’t understand, and you don’t understand, you don’t know how to explain to them, so…it’s kind of like a chain reaction. If you have the proper information, even links and stuff like that, then they can literally just forward it to grandparents, or mothers, or fathers, and then sisters…brothers, and then it’s more clear. [Sydney]

4.6.3.4 Accessibility and Availability of HCPs

Participants emphasized the accessibility and availability of a HCP to answer their additional questions during and after their decision had been made. Access to a HCP could be in-person, over the phone or via email.
…I always go straight to the source. Super open with the doctors… I can phone any time (emphasis) and ask any questions, send an email at 2am and get an answer the next morning. So… I’m more for getting it from the horse’s mouth… like I usually just bombard them with questions. [Candace]

I like the fact that I can email [my doctor] for the one or two questions…. I would rather be able to ask questions right away than, you know, sit with it in my mind. [Tracy]

…I think just… making sure that the parents know that if they do have any more questions, even after the information that they can contact whoever is presenting the information for more questions. [Aida]

…you could always look at the information or look at videos but something might pop in your mind and so you want to just have somebody that you can call or have access to ask questions, on a personal level. [Sydney]

4.7 Discussion

The findings indicated that participants felt that their decision to proceed with WGS/ES was not a difficult one. The potential benefits of WGS/ES appeared to outweigh the potential risks, such as learning about IFs.

On the other hand, the findings also revealed that participants had some unmet decisional needs. These included a lack of knowledge about specific topics that later became relevant to them and, in some instances, the delivery of too much information at one time. Participants had both informational and psychosocial needs, as well as decision support needs at different points in time.

4.8 A “No Brainer” Decision

Given most of the participants’ circumstances, it was not surprising that proceeding with WGS/ES was perceived as a “no brainer” decision. Most participants’ motivation to proceed with WGS/ES was to find a diagnosis for their child. Other studies have found that a majority of parents value a diagnosis (Makela, Birch, Friedman & Marra, 2009) and that parents are unlikely to pass up the opportunity to find a diagnosis for their child (Rosenthal, Biesecker & Biesecker, 2001). One participant described the informed consent for WGS/ES as “desperate consent” and explained that most parents would do anything to try and find answers for their child. Similarly,
Levenseller et al. (2013) found that parents recognized that they were likely to accept the offer of ES immediately in order to find the cause of the child’s condition or prognosis, without carefully considering the limitations and the possibility of incidental findings.

For some participants in this study, a possible factor that contributed to the ease of their decision was their established relationship and high trust in their HCPs. Some participants (n=4) based their decision primarily on their trust in their physician(s) and following the physician’s recommendations. When those participants reflected back to the informed consent period, they expressed that they “didn’t know a lot” or “there wasn’t much discussion” or they did “not really [know] what [they were] getting into.” Evidence suggests that parents who highly trust their HCPs are less likely to take an active role in the decision-making process, and they are more likely to rely on the HCP to make the decisions (Thorne & Robinson, 1989; Kirschbaum & Knafl, 1996). Most participants were living with the circumstances of having an ill child and managing competing life priorities, perhaps making them more reliant on HCP decision-making.

Given that there is a high level of uncertainty in what results will be obtained from WGS/ES and how these results will affect individuals and families, genetic HCPs need to be cautious about “overselling” the benefits of WGS/ES to parents (Brunham & Hayden 2012; Pinxten & Howard, 2014). HCPs must be mindful of the potential for their undue influence or persuasion on parents (Singhal, Oberle, Burgess & Huber-Okrainec, 2002) and make decisions in a shared manner. This may be especially important because participants’ motives to proceed with WGS/ES mirrored their perceived benefits of WGS/ES (Table 4.3 and 4.5).

4.9 Informational Needs: Participants’ Perceived Priorities

Prior to making a decision about WGS/ES, participants prioritized receiving specific information. There were both similarities and differences in the participants’ informational needs
compared to other investigations of informational needs prior to WGS/ES. Other investigations of informational needs prior to WGS/ES have come from professionals’ perspectives (Ayuso et al., 2013) or both professionals’ and parents’ viewpoints (Appelbaum et al., 2013; Levenseller et al., 2013; Townsend et al., 2012).

There were topics that participants prioritized that had not been mentioned in previous studies related to parents’ informational needs prior to WGS/ES. Participants also had knowledge gaps about certain topics and had varied informational needs over time.

4.9.1 Similarities

Compared to other studies, the similarities in this study’s participants’ informational needs included: the expected benefits and risks (Appelbaum et al., 2013; Ayuso et al., 2013), expectation of the results (e.g., the scope of WGS/ES and a description specifying the type of information to be obtained) (Ayuso et al., 2013), the potential for IFs (Townsend et al., 2012), impact on the family (Appelbaum et al., 2013) and possible issues with insurance (Levenseller et al., 2013). These topics may be the key priorities for parents considering WGS/ES for their child.

4.9.2 Differences

Compared to other studies, the participants in this study infrequently discussed, did not speak about or did not seem concerned about the following topics: the existence of alternative options if any, the voluntary nature of the test, the possibility of refusal, future use of the data, and confidentiality of the outcomes (Ayuso et al., 2013), return of results in the event of a death or incapacity, being re-contacted about new discoveries (Appelbaum et al., 2013), variants of unknown significance, the scientific uncertainty of IFs (Townsend et al., 2012), stigmatization and discrimination in employment, loss of privacy, and possible restrictions on future reproductive decision-making for the child (Levenseller et al., 2013). These topics may be
important to participants, but they may have not had knowledge about these topics or had not realized they were important or relevant. On the other hand, participants may consider these topics as “extra” information that they already knew about, didn’t need or would like to learn about later.

Many participants thought that basic genetics and genomics and the limitations of WGS/ES were important topics to know. In contrast, Levenseller et al. (2013) found that parents had little interest or no concerns about genetic concepts, genomic variation and ES limitations (Levenseller et al., 2013). This difference may be because the participants in this study went through the DM process for WGS/ES and later realized that these topics were important and relevant to them. Whereas in Levenseller et al.’s (2013) study, participants had not undergone WGS/ES and were presented with hypothetical scenarios to assess their opinions and intentions related to WGS/ES.

Two other topics that participants’ prioritized were realistic time frames and process and procedures of WGS/ES; these have not been mentioned as important in other studies of parents’ informational needs prior to WGS/ES.

4.9.3 Realistic Time Frames

Participants emphasized realistic time frames. This is important because participants reported feelings of anxiety or frustration when they did not receive realistic time frames or updates about time delays. The desire to know realistic time frames may be related to participants’ desire for control over their child’s health and well-being, to reduce participants’ frustration or anxiety related to waiting and the unknown, and a mismatch between HCP and parent expectations.
4.9.4 Process and Procedures

Participants agreed that knowing the basics of the process and procedures was important. However, many participants wanted more details at the time of the DM or after the decision had been made. Thus, it is important to make information about the process and procedures available to parents at some point during the DM process.

4.9.5 Variability in Information Priorities

The participants desired varying amounts of detail about each topic. Some only wanted basic information while others wanted more specific and detailed information. Some participants also realized that they were not aware of certain topics and wished they had known more about them. Whether each of the prioritized topics was actually critical to participants’ during decision-making is unclear because some participants proceeded with testing even if they only had limited knowledge about ES during decision-making.

Participants may also have different informational and decision support needs over time, or during times of stress and urgency compared to their day-to-day circumstances. For example, during the decision-making and informed consent process, participants were often focused on the benefits of WGS/ES such as possibility of a diagnosis for their child, but they later realized that they lacked information that was relevant to them (e.g., about incidental findings pertinent to themselves or another family member). Furthermore, the information that participants agreed would be important to know may not have been necessary during participants’ actual decision-making. The information may have only become important because the researcher or another parent had introduced the topic. In addition, the specific content may have actually been discussed during the decision-making process but participants may have had poor recall of the information.
These findings suggest the importance of tailoring the content and detail of the information based on the relevance to parents as well as their individual needs, values, priorities, prior knowledge and readiness to learn more information. Other researchers have also emphasized the importance of tailoring the information to a person’s needs (Durand, Stiel, Boivin, Elwyn, 2010; Jackson, Cheater & Reid, 2008) and specifically in the context of informed consent in genetic testing (Ormond et al., 2007). Tailoring information to the patient has been shown to improve recall, raise patients’ and caregivers’ satisfaction with the information and decrease anxiety (McPherson, Higginson & Hearn, 2001).

4.10 Informational Needs: Lack of Knowledge and Volume of Information

4.10.1 Lack of Knowledge

Some participants’ lacked knowledge about key topics (e.g., what they consented to, incidental findings) and some reported little knowledge about WGS/ES when they decided to proceed with WGS/ES. This lack of knowledge may be attributable to numerous factors such as the urgency of participants’ circumstances, a lack of time during the informed consent process, the complexity of the information presented, participants’ high trust in the HCPs, participants’ poor recall of information or participants feeling overwhelmed with too much information.

Despite the fact that participants felt that they received adequate or too much information, some participants realized that they lacked information they would like to have had. Similar variation in information needs has been reported in other patient populations and types of decision-making, such as women making decisions about amniocentesis (Durand, Stiel, Boivin & Elwyn, 2010). Regardless of the amount of information they received, participants still consented to WGS/ES in the interest of their child, a decision that they considered to be a “no brainer”. However, this should not take away the importance of properly informing them about
the nature and possible outcomes of proceeding with WGS/ES (Dondorp, Sikkema-Raddatz, de Die-Smulders & deWert, 2012). From an ethical standpoint, providing this information to parents respects their role as representatives of the best interest of their child (Dondorp et al., 2012). Furthermore, participants preferred to have information available or access to more information as it became more relevant to them. For example, one participant stated that learning about basic genetics (e.g., DNA, how sequencing works) was initially not important. However, when an incidental finding came up, she realized it became more important to learn and understand basic genetic knowledge in order to understand more about incidental findings. Participants realized that they lacked knowledge about certain topics that were later relevant and important to them. These findings suggest that not all participants’ informational needs were met prior to the decision to proceed with ES and thereafter.

Even though the participants did not have a difficult time making their decision to consent to WGS/ES, their reported knowledge gaps indicated that they may have had incomplete or incorrect understandings of matters relevant to the decision to proceed with ES. Rigter and colleagues (2013) found that some parents may interpret the implications of IFs in ES inaccurately or may not understand what they are consenting to or why. Since there is uncertainty in what results will be obtained from WGS/ES and how these results will impact families, participants’ knowledge gaps may be problematic when parents receive primary results and incidental findings.

4.10.2 Volume of Information

Participants spoke about the large volumes of information presented to them during the informed consent process for WGS/ES. Some participants also admitted that they did not read the consent form. Research and clinical observations have shown that effective informed
consent, even without WGS/ES, can often be challenging (Lidz, Appelbaum & Meisel, 1988; Pinxten & Howard, 2014). Lidz and colleagues (1988) suggest that informed consent can often be an “empty ritual,” as patients are presented with complicated information that is difficult to understand and that does not help them to make a good decision.

Additional challenges specific to the consent process for WGS/ES include the time needed for genetic counselling, education and discussion and the extent and complexity of information regarding WGS/ES (Bick & Dimmock, 2011; Levenseller et al., 2013; Ormond et al., 2010). It is too extensive to discuss all of the possible outcomes from WGS/ES prior to testing, and HCPs do not have time to provide information about all these possibilities (Dondorp et al., 2012). Furthermore, parents can only be expected to comprehend, absorb and reflect upon a certain amount of information before “information overload” occurs, which further challenges well-informed and meaningful decision-making (Dondorp et al., 2012). Thus, there is a need to use and examine approaches that avoid making parents feel overwhelmed with too much information, while at the same time, ensuring they are well informed.

In the genetic screening context, Elias and Annas (1994) proposed “generic consent.” This involved providing patients with information about categories of general outcomes and options instead of providing very specific information about each of an extensive range of possible outcomes (Elias & Annas, 1994). However, generic consent might not provide adequate information for some parents, and some may feel uncomfortable asking for additional information from the HCP (Ormond, Iris, Banuvar, Minogue, Annas, Elias, 2007). Furthermore, patients had contrasting views about generic consent (Ormond et al., 2007), which, therefore, needs further study in the context of parents deciding for their children about WGS/ES.
Appelbaum et al. (2013) suggested “staged consent” as one way to improve current informed consent practices. For example, the possibility of IFs could initially be discussed with the participant; then when and if IFs are found, additional information relevant to the particular findings would be provided to facilitate informed decision-making about return of the results (Appelbaum et al., 2013). One participant in the current study proposed a similar idea, “layers of consent,” in an effort to differentiate the primary reason for investigation from incidental findings. The participant suggested that parents undergo a first layer of consent and receive information related to WGS/ES and the primary reason for investigation. The participant suggested that incidental findings be treated differently – just being mentioned as a possibility during the first discussion and considered in more detail in second layer of consent. This concept of layers of consent had some parallels with Bunnik, Janssens & Schermer’s (2013) tiered-layered-staged model for informed consent, and Yu and colleagues’ (2013) self-guided management approach for the return of results (see Chapter 2).

Layers of information and consent may address some of the challenges of informed consent described earlier and may be a suitable approach for parents considering WGS/ES. This, therefore, deserves further research.

4.11 Psychosocial Needs

At the beginning of the study, the researcher imagined that the challenge would be to define the body of information that was essential during parents’ decision-making for ES. However, the findings suggested that participants’ psychosocial needs were also important. Participants described factors that contributed to their psychosocial needs, such as having a good HCP-parent relationship and HCPs’ consideration of parents’ well-being. A good HCP-parent relationship was most often characterized by attributes summarized in Table 4.16
In this study’s context, the parent is considered the “patient” in the HCP-patient relationship because the parents interacted with the HCPs and made the decisions for their children. The importance of the HCP-patient relationship and its’ associated positive effects on health outcomes is not new (Kaplan, Greenfield & Ware, 1989). Psychosocial health can be viewed as the degree to which a person has more positive beliefs and feelings (e.g., psychological well-being, self-efficacy), and fewer negative beliefs and feelings (e.g., worry, anxiety, fear) (Street, Makoul, Arora, & Epstein, 2009). Aspects of the HCP-parent relationship such as trust and the provision of emotional support have been found to lead to more positive working relationships and less *decisional conflict* (Stewart, Pyke-Grimm & Kelly, 2005). Other aspects of the HCP-patient relationship such as communication can directly influence positive psychosocial outcomes (Street et al., 2009). Effective communication can contribute to increased patient knowledge and shared understanding, enhanced *therapeutic alliances*, enhanced emotional self-management, higher quality decisions and enhanced *patient agency* (e.g., self-efficacy, empowerment) (Street et al., 2009).

Building a good relationship (Elwyn et al., 2012) and the context of the entire clinical encounter can facilitate shared decision-making (Matthias, Salyers & Frankel, 2013). For the participants in this study, continuity in the HCP-parent relationship was important. Participants perceived that they had ongoing informational and psychosocial needs, rather than only during the in-person or telephone meeting where DM about WGS/ES occurred. Matthias and colleagues (2013) found that SDM is complex and continues beyond the one clinical encounter where DM occurred. Thus, it is important for HCPs to be mindful of the aspects of the relationship that have the potential to impact parents’ psychosocial needs and their DM (Matthias, Salyers & Frankel; 2013; Roter, 2000; Stewart, Pyke-Grimm & Kelly, 2005).
The other important factor that contributed to participants’ psychosocial needs was HCPs’ consideration of participants’ well-being. Some participants wanted not only information from their HCPs but also to be able to turn to them when feeling particularly vulnerable or uncertain. This is not surprising because the participants experienced a range of both positive and negative emotions as they endured a diagnostic odyssey. In other studies, parents with children living without a diagnosis have similarly described their journey as an “emotional rollercoaster” filled with a range of emotions (Lewis, Skirton & Jones, 2010).

Hall and colleagues (2012) studied stress in parents of children with conditions that have confirmed or suspected genetic causes and varying degrees of intellectual disability. They found that parents who were stressed had a maladjustment experience. This experience included family vulnerability (e.g., pile up of stress, stressors of having a child with a disability, being ostracized by others), closed communication (i.e., avoiding communication about the child), negative appraisals of their children’s situation and lack of support (Hall et al., 2012). Thus, it is especially important for HCPs to assess parental stress among this parent population (Hall et al., 2012). The stories of two of the participants about the misconceptions that other people had about their child without a diagnosis, or their child undergoing genetic testing, illustrate another important concern. Parents may be challenged by other people about their decisions to proceed with WGS/ES, a technology that is still novel. The study’s findings showed that participants’ psychosocial needs may be as important as their informational needs.

4.12 Supporting Strategies

Participants suggested strategies that could be helpful during decision-making. These were summarized in Table 4.17.
4.12.1 Brief and Understandable Summaries

Participants suggested brief and understandable summaries of the necessary information and informed consent process. Some of the participants’ reasons for brevity included: a lack of time, a sense of urgency given circumstances and avoiding information overload. Kleiderman et al. (2013) also found that information presented in a concise and understandable manner was important to parents of children who were affected by rare diseases. Information that is tailored to the individual’s reading level has been important for other patient populations (Jackson, Cheater & Reid, 2008; McKibbin et al., 2014). This could be a relatively simple strategy to implement in current clinical settings.

4.12.2 Approaches to Information Delivery

Participants suggested varying approaches and media, as it would better suit different parent’s communication needs and personal preferences for receiving and learning information. This resembles other studies that have found parents’ desire for information presented in multiple ways or in multiple formats to account for individual differences (Durand, Stiel, Boivin, Elwyn, 2010; Jackson, Cheater & Reid, 2008; Townsend et al., 2012).

In addition to the face-to-face discussion with a HCP, the availability of a variety of written, audio and visual information was considered to be most helpful by participants. These findings are also similar to other studies that have found that written text or videos alone were not considered helpful (Jackson, Cheater & Reid, 2008), the importance of visual elements of relevant information (Durand, Stiel, Boivin, Elwyn, 2010) and an interest for interactive videos or websites in addition to print or audio materials (McKibbin et al., 2014).

Evidence has shown that a combination of oral and well-designed written information (e.g., pamphlets) can help improve patients’ health knowledge and recall, especially if the
information is personalised (McPherson, Higginson & Hearn, 2001). Other resources – such as patient-oriented interactive websites – may also help improve knowledge and have been shown to have beneficial effects on people’s comfort in posing questions to physicians, confidence with decisions, emotional health, social support, and their participation in health care (Gustafson et al., 2002). These varying strategies need further study in the context of WGS/ES and this parent population.

4.12.3 Layers of Information

Participants suggested layers of information over time, which included receiving information in advance, take-home material, credible information sources, and accessibility and availability of HCPs. The suggestion of layers of information may be related to participants’ desire to avoid information overload. Many participants acknowledged that they may experience information overload, or need time to absorb, comprehend and reflect on the information and think of additional questions. The process of providing information at different points in time can help avoid information overload (Jackson, Cheater & Reid, 2008).

Other studies have found that adult patients and parents find it helpful when information is provided before an appointment with the HCP (McKibbin et al., 2014), as it helps prepare them for discussion and prepare questions to ask the HCP (Jackson, Cheater & Reid, 2008). Appelbaum et al. (2013) made similar suggestions, suggesting that standard teaching materials such as videos and computer-based programs be sent to participants in advance as an adjunct to the in-person informed consent processes in genomic research.

In studies that involved adults making health decisions about their child or about their own genetic results, participants also suggested take-home information (Durand, Stiel, Boivin,
Elwyn, 2010; McKibbin et al., 2014) and the desire for HCPs to direct parents towards credible sources (Jackson, Cheater & Reid, 2008).

Participants also emphasized the importance of the availability and accessibility of HCPs to answer their questions during the decision-making process and thereafter. Thus, it is important for HCPs to create opportunities for parents to ask questions as many parents do not immediately know what questions to ask or may feel intimidated to ask questions (Jackson, Cheater & Reid, 2008).

4.13 Challenges and Opportunities for Tailored Layers of Information

4.13.1 Challenges

If there is only one informed consent process and genetic counselling session that encompasses both the primary reason for investigation and the disclosure of incidental findings, it may be problematic to provide layers of information after parents have already made their decision to proceed with WGS/ES. If participants want to review information more carefully at a later time, they may change their minds about WGS/ES and want to withdraw their consent. Withdrawing their consent is possible, but if the WGS/ES processing is already underway, then the results might go into the patient’s chart. Patients and families may not want these results recorded in their medical chart. Therefore, layers of information may be more effective if given together with the earlier suggestion of layers of consent, as opposed to giving parents layers of information in isolation.

Each participant had different perceptions of what they considered “adequate” and “relevant” information, and each desired different amounts or layers of information. Similarly, Ormond and colleagues (2007) found that patients’ “personal information style” impacted the type of information they desired about genetic carrier testing. For example, some patients sought
out as much information as possible, while other patients waited for more information based on what was relevant for them (Ormond et al., 2007). Thus, it is important for HCPs to assess parents’ individual values, priorities and informational needs and tailor related information accordingly (Durand, Stiel, Boivin, Elwyn, 2010; Jackson, Cheater & Reid, 2008; Ormond et al., 2007). This will likely be a challenging task given the insufficient capacity of genetic HCPs, geographically unbalanced access to genetic services and genetic HCPs (Canadian Institute of Child Health, 2014), and the substantial time required for effective genetic counselling (Johansen Taber, Dickinson & Wilson, 2014). For example, in the province of British Columbia, there is approximately one genetic counsellor and less than one medical geneticist per 100,000 people (Canadian Institute of Child Health, 2014).

4.13.2 Opportunities

Decision support interventions could help address the insufficient human resources, help parents clarify what exactly is important to them, instigate discussion about their values and any personal issues and provide information tailored to their needs. These decision support interventions can include a variety of methods including a service (e.g., decision coaching), a system (e.g., an interactive decision aid), or products such as pamphlets, books, videos or websites (Elwyn, Frosch et al., 2010). Decision aids are interventions that typically provide support for people who may be facing difficult decisions (Elwyn, Frosch et al., 2010). Decision aids can help support parents to participate with their HCPs to make deliberative and tailored decisions among available options (O’Conner, 2001). When the idea of a web-based decision aid for WGS/ES was introduced by the researcher, there was an interest among participants for the use of such web-based tools. Participants did not consider web-based tools as a replacement for face-to-face discussions but as an adjunct to the face-to-face discussions with HCPs. A web-
based decision aid could also serve parents that are geographically farther away. Interestingly, three participants who lived far away from the primary location for genetic services seemed to have the most critical knowledge gaps, including what exactly they consented to, the scope of WGS/ES and what to expect from the results, if any.

The strategies that participants suggested resemble the different categories of decision support interventions; those that can be used during the face-to-face clinical encounter or those that are used outside the clinical encounter (Elwyn, Frosch et al., 2010). Face-to-face interventions facilitate SDM by acting as prompts and props, usually making options visible and organizing information according to elements that patients find relevant (Elwyn, Frosch et al., 2010). Decision support interventions outside the clinical encounter can be used after an initial consult with the HCP with advice to return for further discussion, or ahead of the time so patients can arrive at their appointment better informed and prepared for greater involvement in the decision-making (Elwyn, Frosch et al., 2010). Implementing these different categories of decision support interventions may be a way to help mitigate the limited number of genetic HCPs, information overload, prevent critical gaps in knowledge about WGS/ES, tailor information that is most relevant and important and help support parents to make deliberate decisions that are right for them and their families.

Genetic HCPs have also suggested the delivery of web-based information or e-counselling tools (Birch, 2014; Dimmock 2012, Roche 2012). Evidence suggests that the use of interactive e-learning and decisional support e-tools may be comparable or better than face-to-face counselling in promoting knowledge acquisition and may be effective in reducing decisional conflict (Birch, 2014). However, further research is needed to effectively design such decision
support tools and explore whether or not they are feasible, effective and sustainable in the context of parents making a decision about WGS/ES for their child.

Another possible way to support effective genetic counselling and the limited number of genetic HCPs is a team-based approach that involves non-genetic HCPs (Johansen Taber, Dickinson & Wilson, 2014). Other HCPs – such as family doctors, oncologists or nurses – could play a role to help to mitigate the lack of human resources within the genetic specialty. For example, among some existing decision support tools, nurses are the HCPs that typically present and discuss information with the family, guide them to find resources, help explain any issues and support the family’s deliberation process (Elwyn, Frosch et al., 2010). It may also be valuable to families to have the HCP that they are most comfortable with to offer WGS/ES, or to have the HCP work in collaboration with genetic HCPs. Comfort and trust was evident in the study participants’ relationships with non-genetic HCPs. However, this means that non-genetic HCPs would need to be knowledgeable and able to answer or re-direct families’ inquires about WGS/ES. Johansen Taber and colleagues (2014) suggest increased education and training of non-genetic HCPs and more opportunities for collaboration between different specialties and HCPs. As WGS/ES becomes more frequently used in many clinical settings, there will be a growing need for HCPs that are specifically trained (Johansen Taber, Dickinson & Wilson, 2014) to implement strategies that facilitate parent-tailored education, counselling, decision support and informed consent processes.
Chapter 5: Conclusion

Despite the abundant research that emphasizes education, counselling, and consent processes for WGS/ES, this is one of the few studies that focuses on parents’ needs during the DM process for WGS/ES. This study sheds light on parental perceptions and views regarding the types and quantity of information they received, and the education and overall decision support they valued most when considering WGS/ES for their child.

Participants felt that their decision to proceed with WGS/ES was a “no brainer”. However, they expressed unresolved decisional needs including: a lack of knowledge about certain topics that became relevant and important to them later, unmet expectations (e.g., realistic time frames) and a need for more support and resources. Participants also acknowledged that the high volume of information and urgency of their circumstances may have caused them to be less receptive – or even unreceptive – to information during their WGS/ES DM process. Additionally, participants had ongoing informational and psychosocial needs beyond the single clinical encounter where their WGS/ES DM occurred. These unresolved needs may adversely affect decision quality, which could affect the parents’ actions and emotions, and the patient’s (i.e., the child’s) long-term health and appropriate use of health services (Jacobsen, O’Conner & Stacey, 2013; O’Conner, 2006). It is hence essential for HCPs to re-examine current settings and processes that may contribute to creating unmet decisional needs, and to execute strategies that address these unmet requirements.

Contextual (e.g., circumstances), personal (e.g., prior knowledge, learning preferences), and social factors (e.g., relationships) influenced the participants’ DM processes in different ways. Contextual factors, such as the circumstance of having an ill child, can make the decision to proceed with WGS/ES a “no brainer”. Personal factors – such as being presented with
information in a manner incompatible with one’s learning style – may increase the difficulty of communication and understanding between HCPs and parents. This factor could have contributed to the participants’ lack of knowledge on certain topics. Social factors, such as a “good” HCP-parent relationship, may ease the DM process. HCPs need to be mindful of these contextual, personal, and social factors, because they can both positively and negatively impact the DM process.

Prior to making their WGS/ES decision, participants prioritized knowing the following information: the benefits and risks, realistic time frames, and expectations for the results. However, the content and amount of information that participants considered to be important varied; there is hence a need to tailor education and counselling to each person’s assessed informational needs.

Participants suggested strategies that would support their DM. These include brief and understandable summaries, varied approaches to information delivery, and layers of information. Layers of information include methods such as: receiving information in advance, take-home material, credible sources, and accessibility to knowledgeable HCPs. These strategies should be considered to help improve education, counselling, decision-making, and the informed consent process for parents considering WGS/ES for their child.

5.1 Limitations

There are some limitations to this study. The participants were primarily female Caucasians, university- or college-educated, and had higher than average household incomes. Also, only participants who consented to WGS/ES were included. Parents who do not consent to WGS/ES, and those from more diverse demographics, should be included in future studies in case they have different perspectives on parental decisional needs.
There were also limitations related to use of a focus group, in which some participants may feel uncomfortable expressing their views or openly participating in the group discussion (Polit & Beck, 2012). The dynamics of the focus group session may have facilitated a dominant group culture that impeded individual expression (Polit & Beck, 2012). To mitigate these limitations, and to enhance the credibility and authenticity of the developed concepts, the researcher also conducted eleven individual and five repeat interviews. Concepts from the single focus group session also emerged in the individual interviews.

In addition, participants were part of different WGS/ES research trials or clinical investigations, so the types of WGS/ES experienced varied. There was also variability in the HCPs that participants encountered. No direct observations of the participants’ decision-making and informed consent process were made, so the information that the HCPs actually conveyed to each participant – and their interactions – were not accessible. Finally, participants may have poor or inaccurate recall of their experiences with the information, decision-making, and informed consent processes. However, despite these issues, common concepts still emerged from the data.

5.2 Future Research Recommendations

As some participants did not read the consent form, or had limited knowledge about WGS/ES prior to consenting, it is unclear if their decisions will result in future problems. Additionally, it is unknown how the parents’ views about their decisional needs will change over time, and what their reactions may become when their primary results and/or incidental findings are returned; perhaps their perceptions depend on the results received (i.e., negative or positive impact on the family). As WGS/ES is used more frequently in various clinical settings,
longitudinal studies are needed to examine the personal, emotional, social, and economic impact they have on parents over time.

The participants’ suggestions about supportive strategies should be empirically tested in the research and/or clinical settings to assess their effectiveness on parents’ knowledge acquisition, comprehension, positive psychosocial outcomes, and ability to make a quality decision about WGS/ES for their child. For example, the design, feasibility, effectiveness, and sustainability of providing layers of information alongside layers of consent should be clinically studied. There is a need to empirically test this and other strategies to avoid overwhelming people with too much information, while simultaneously providing them with sufficient education to ensure meaningful and well-informed decisions. Some example strategies designed or suggested by HCPs and researchers include: different informed consent or return of results models (such as generic consent) (Ormond, 2007), staged consent (Appelbaum et al., 2013), a self-guided management approach for WGS/ES results (Yu et al., 2013), and a tiered-layered-staged informed consent model (Bunnik et al., 2013). Testing these strategies, and the subsequent soliciting of the participants’ (i.e., parents’) perceptions of each approach’s utility (i.e., how helpful it is in facilitating their understanding and ability to make a meaningful and well-informed decision), would be useful. In addition, specific further research is targeted to inform the design of decision support interventions (e.g., decision aids) that integrate both the HCPs’ and participants’ perspectives, and to explore whether such interventions are feasible, cost-effective, and sustainable over time.
5.3 Implications for Nursing and Nursing Education

This study provides some initial insights on how HCPs can better support parents making a WGS/ES decision for their child. The aim of this study is to enhance the quality of health care for patients and families, which is one of the goals of nursing research in clinical genetics and genomics (Calzone et al., 2010). Nursing – through its emphasis on patient advocacy (Hamilton, 2009), health promotion, caring, understanding individuals and their relationships with patients and families (Calzone et al., 2013) – brings a valuable perspective to the application of WGS/ES. Nurses are well positioned at the forefront of patient care, and can help incorporate genetic and genomic information into different aspects of the healthcare system (Calzone et al., 2010). For example, pediatric or neonatal nurses that take regular care of an infant or child could identify possible genetic issues, and refer the patient and their family to genetic HCPs. Nurse case managers – who often have longstanding relationships with, and a more holistic understanding of, the patient and family – could work with genetic HCPs, and support the family through a shared decision-making process. Nurses could also help parents clarify their values and expectations, and guide them to relevant resources. Furthermore, the nurse may be the HCP that the patient or family is most comfortable with during the DM process. There are hence opportunities for nurses to become more involved in supporting patients and families who are considering WGS/ES.

Due to the advances in genetic knowledge and technology, nurse clinicians, educators, researchers, and administrators have new responsibilities (Badzek, Turner, & Jenkins, 2008). As WGS/ES becomes more prominent in many clinical settings, the need for nursing to continue educating their practicing workforce in genomics, and to be prepared to respond to patients’ WGS/ES inquires, only grows (Calzone et al., 2012; Calzone et al., 2013). In the United States,
experts have made efforts to integrate genomic knowledge and competencies into nursing education and training (Calzone, Jenkins, Rust, 2007; Jenkins & Calzone, 2007; Tonkin et al., 2011). However, many nurses are still unprepared to respond to patients’ genetic testing inquiries (Thompson & Brooks, 2011), and general nursing competency in genomics remain limited (Calzone et al., 2012).

In Canada, there is consensus about the importance of genetic nursing roles, but there are several barriers to the development of genetic nursing practice (Bottorff et al., 2005a, 2005b). Amongst other challenges, the most significant barrier is the inadequate attention to genetics and genomics in nursing curricula at all levels (Bottorff et al., 2005a, 2005b). Efforts to address nursing competency in genomics are furthest along in the United States and the United Kingdom (Williams, et al., 2011).

Genetics and genomics will become increasingly important in nursing across all areas of nursing practice (Bottorff et al., 2005a). Genomic technologies have far-reaching applications in health promotion, disease prevention, diagnostics, and treatment strategies (Thompson & Brooks, 2011). Therefore, continued efforts to educate nurses on genomics are necessary.
References


Angrist, M. (2011). You never call, you never write: why return of “omic” results to research participants is both a good idea and a moral imperative. Personalized medicine, 8(6), 651–657. doi:10.2217/pme.11.62


Appendices

Appendix A  Explanatory Cover Letter

THE UNIVERSITY OF BRITISH COLUMBIA

[Address Information]

DATE

ADDRESS

Dear MR/MRS ____________,

Thank you for your interest in our study.

I have attached the following documents for your review:
1) Consent Form – For a Focus group
2) Consent Form – For an Interview
3) Information Sheet – Whole Genome & Exome Sequencing
4) Demographic Information

Please review these documents before the focus group/Interview date. Please do not hesitate to contact me if you have questions about any of these documents or the study. We will go over the consent form together in person.

I also emailed you two video links titled “Whole Genome Sequencing and You” and “Sequence Me.” I highly recommend for you to watch these informative and engaging videos, especially closer to the date of the focus group or interview. I look forward to meeting you.

Please feel free to contact me if you have further questions.

Sincerely,

[Contact Information]

Contact information and identifiers have been blacked out.
Appendix B  Focus Group Consent Form

CONSENT FORM

Study Title       Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

Principal Investigator     Identifiers have been removed.

Student-Investigator

Co-Investigators/Student Supervisors

Funding Agency
This project is supported by APOGEE-Net, a research and knowledge network on genetic health services and policy, and funded by the Canadian Institutes of Health Research.

Study Purpose
The purpose of this study is to explore and describe parents’ decision support and informational needs when deciding on whole genome or whole exome sequencing (WGS/WES) for their child. This research is also being conducted for a Master of Science in Nursing thesis.
WGS/WES (sequencing of all or part of our cells’ genetic materials) is a method used in research studies to find genetic changes. This new technology identifies the causes of many genetic conditions, but potentially also many unexpected or unknown changes in your genetic material. WGS/WES offers a wealth of information, which can complicate the decision-making process for patients, parents and health care providers.

Each family requires accurate and relevant information to make informed decisions about undergoing WGS/WES testing. Prior to choosing WGS/WES, it is important that individuals and their families are part of the decision-making process, and fully understand genomic testing and its implications. Our goal is to define the body of information that parents view as important for informed decision-making. We will carry out focus groups amongst parents with children who have experienced WGS/WES. Then, we will invite some parents back for individual interviews to confirm the research findings.

**Your Participation is Voluntary**
Your participation is entirely voluntary, so it is up to you to decide whether or not to take part in this study. Before you decide, it is important for you to understand what the research involves. This consent form will tell you about the study, why the research is being done, what will happen during the study and the possible benefits, risks and discomforts. If you wish to participate, you will be asked to sign this form. If you do decide to take part in this study, you are still free to withdraw at any time and without giving any reasons for your decision.

**Research Procedures**
We will obtain information through focus groups, which are groups of people gathered together to participate in a guided discussion. Focus groups allow you to hear from people with similar or different views. We invite you to share your thoughts and opinions on what is important to you when making a decision about WGS/WES. ________ will guide the group, but the topics discussed will depend on what the group of participants finds important. There are no wrong questions or topics. We may discuss ideas such as personal concerns, impacts on society, and feelings and attitudes towards genomic testing. There will be some questions posed to get people started talking.

We will hold focus groups for individuals or family members with personal WGS/WES experience. Each group will contain 5 to 8 participants. Each focus group will last around 1 ½ hours. The groups will meet in a location and on a date that works best for the majority of the members. Possible locations include the BC Children’s Hospital, UBC, or a local community centre. We will let you know the location and date as soon as we have a group of interested participants. The facility will be comfortable, and light refreshments will be provided. You may also be invited to attend an interview to confirm the research findings.
**Remuneration**
Participants will be offered $20 in appreciation for their participation.

**Confidentiality and Privacy**
The focus groups will be audio taped on a digital recorder so that an exact transcription of the discussion is available for analysis. To protect your identity, this transcription will not use names, but generic labels (e.g. “person 1”, “person 2,” “person 3”) that cannot be used to identify the speaker.

The audio recording, transcriptions, and the consent forms will be kept in locked filing cabinets in the Friedman Lab, which is a secure area that only ______________________ will have access to. All digital tapes will be deleted five years after the focus group takes place.

We will remind all focus group participants to use discretion in the disclosure of personal information, and to respect the privacy of others in the room by avoiding the discussion of specific personal content outside of the focus group. However, please realize that some participants may decide to talk about the discussion afterwards with people outside the focus group.

Your confidentiality will be respected. However, research records and health or other source records identifying you may be inspected in the presence of the Investigator or his or her designate by representatives of Health Canada and the UBC Children’s and Women’s Research Ethics Board for the purpose of monitoring the research. No information or records that disclose your identity will be published without your consent, nor will any information or records that disclose your identity be removed or released without your consent unless required by law.

**Risks**
Depending on the topics discussed, there is the potential for psychological stress during the focus groups. Please understand that you are free to talk about only those topics you wish to discuss. Part of the job of the facilitator is to closely monitor the discussions to ensure it is not causing discomfort to any participants. You may leave or withdraw your participation at any time, but any information you have shared up to that point will be retained for data analysis.

**Benefits**
Although there are no immediate benefits, your participation in this study may be contributing to a greater understanding of patients’ and families perspectives, which enables health care providers to improve the existing counseling and consent processes. You may also experience some benefit from the opportunity to talk about your experiences.
Study Contacts
If you would like more information about this study, or if you are interested in participating, please contact _____________. Their contact details are:

Contact information and identifiers have been removed.

Contact for concerns about the rights of research subjects
If you have any concerns about your treatment or rights as a research subject, you may contact the Research Subject Information Line in the UBC Office of Research Services at 604-822-8598, or if long distance e-mail to RSIL@ors.ubc.ca, or telephone toll free: 1-877-822-8598.
Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

Consent

Your participation in this study is entirely voluntary, and you may refuse to participate or withdraw from the study at any time without jeopardy to your health care or relationship with anyone associated with the Friedman Lab, the Department of Medical Genetics, University of British Columbia, or BC Children’s and Women’s Hospital.

Your signature below indicates that you have received a signed and dated copy of this consent form for your own records.

Your signature indicates that you consent to participate in this study.

_________________________________          ____________________
Participant Signature                      Date

_________________________________
Printed Name of the Participant
Appendix C  Interview Consent Form

CONSENT FORM

Study Title  Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

Principal Investigator  Identifiers have been removed.

Student-Investigator

Co-Investigators/Student Supervisors

Funding Agency  This project is supported by APOGEE-Net, a research and knowledge network on genetic health services and policy, and funded by the Canadian Institutes of Health Research.

Study Purpose  The purpose of this study is to explore and describe parents’ decision support and informational needs when deciding on whole genome or whole exome sequencing (WGS/WES) for their child. This research is also being conducted for a Master of Science in Nursing thesis.
WGS/WES (sequencing of all or part of our cells’ genetic materials) is a method used in research studies to find genetic changes. WGS/WES offers a wealth of information, which can complicate the decision-making process for patients, parents and health care providers.

Each family requires accurate and relevant information to make informed decisions about undergoing WGS/WES testing. Prior to choosing WGS/WES, it is important that individuals and their families are part of the decision-making process, and fully understand genomic testing and its implications. Our goal is to define the body of information that parents view as important for informed decision-making. We will carry out focus groups and/or interviews amongst parents with children who have experienced and/or have been offered WGS/WES. Then, we will invite some parents back for individual interviews to confirm the research findings.

**Your Participation is Voluntary**

Your participation is entirely voluntary, so it is up to you to decide whether or not to take part in this study. Before you decide, it is important for you to understand what the research involves. This consent form will tell you about the study, why the research is being done, what will happen during the study and the possible benefits, risks and discomforts. If you wish to participate, you will be asked to sign this form. If you do decide to take part in this study, you are still free to withdraw at any time and without giving any reasons for your decision.

**Research Procedures**

We will obtain information through individual interviews. We invite you to share your thoughts and opinions on what is important to you when making a decision about WGS/WES. Depending on what works for you, ________ will conduct the individual interviews in-person, by phone or video-conference. There will be some questions posed to get the conversation started. There are no wrong questions or topics. We may discuss ideas such as personal values and concerns, impacts on family and society, and feelings and attitudes towards genomic testing.

Each interview will last about 1-1.5 hours. The interview will take place at a location and date that works best for you. Possible locations include the BC Children’s Hospital, UBC, a local community centre or at your home. ________ will let you know the location and date as soon as we have interested individual participants. You may also be invited to attend (in-person, phone or web) another interview to confirm the research findings.
**Remuneration**
Participants will be offered $20 in appreciation for their participation.

**Confidentiality and Privacy**
The interview will be audio taped on a digital recorder so that an exact transcription of the discussion is available for analysis. To protect your identity, this transcription will not use names, but generic labels (e.g., “person 1”, “person 2,” “person 3”) that cannot be used to identify the speaker.

The audio recording, transcriptions, and the consent forms will be kept in locked filing cabinets in the Friedman Lab, which is a secure area that only __________________________ have access to. All digital tapes will be deleted five years after the interview takes place.

We will remind all interview participants to use discretion in the disclosure of personal information. We will also ask individual participants to respect the privacy of other people by avoiding the discussion of specific names during the interview.

Your confidentiality will be respected. However, research records and health or other source records identifying you may be inspected in the presence of the Investigator or his or her designate by representatives of Health Canada and the UBC Children’s and Women’s Research Ethics Board for the purpose of monitoring the research. No information or records that disclose your identity will be published without your consent, nor will any information or records that disclose your identity be removed or released without your consent unless required by law.

**Risks**
Depending on the topics discussed, there is the potential for psychological stress during the interview. Please understand that you are free to talk about only those topics you wish to discuss. You may leave or withdraw your participation at any time, but any information you have shared up to that point will be retained for data analysis.

**Benefits**
Although there are no immediate benefits, your participation in this study may be contributing to a greater understanding of patients’ and families perspectives, which enables health care providers to improve the existing counseling and consent processes. You may also experience some benefit from the opportunity to talk about your experiences.
**Study Contacts**
If you would like more information about this study or if you are interested in participating, please contact _______ or _______ in the Friedman Lab. Their contact details are:

Contact information and identifiers have been removed.

**Contact for concerns about the rights of research subjects**
If you have any concerns about your treatment or rights as a research subject, you may contact the Research Subject Information Line in the UBC Office of Research Services at 604-822-8598, or if long distance e-mail to RSIL@ors.ubc.ca, or telephone toll free: 1-877-822-8598.
Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

Consent

Your participation in this study is entirely voluntary, and you may refuse to participate or withdraw from the study at any time without jeopardy to your health care or relationship with anyone associated with the Friedman Lab, the Department of Medical Genetics, University of British Columbia, or BC Children’s and Women’s Hospital.

Your signature below indicates that you have received a signed and dated copy of this consent form for your own records.

Your signature indicates that you consent to participate in this study.

_________________________          ____________________
Participant Signature          Date

_________________________
Printed Name of the Participant
Appendix D  Information Sheet

Information Sheet – Whole Genome Sequencing & Whole Exome Sequencing

Deoxyribonucleic acid (DNA) is located inside each of the trillions of cells that make up your body. DNA carries all the information your body needs to function and makes you who you are.

DNA is packaged into structures called chromosomes. Humans have two copies of all but the sex chromosomes and therefore, two copies of each gene. We inherit one copy of each chromosome from our mother and the other copy from our father.

DNA is made up of four chemical bases (letters) called Adenine, Thymine, Guanine, and Cytosine – A, T, G, C.

These letters are the building blocks of DNA. The human genome is made of about 3 billion of these letters. These letters are arranged into genes. Genes are important because they contain the instructions that cells need to make proteins. Proteins do most of the work in your body, by building and maintaining things such as your muscles and organs. A change in a gene can lead to a change in a protein, which could affect your body. For example, it can determine how your heart functions.
Your **genome** is your complete set of DNA, and the exact order of these letters is your **genome sequence**.

**Whole genome sequencing** is a technology that looks at **all** of your DNA. **Whole exome sequencing** looks at **part** of your DNA. Today, whole genome or whole exome sequencing are being used extensively in research studies to find the genetic cause for many diseases and disorders. As these methods become less costly, the use of whole genome or whole exome sequencing is likely to become routine in the near future.

**Whole genome or whole exome sequencing** offers a wealth of information that may or may not complicate the decision-making process for patients, parents and health care providers. One reason why decision making can be a complex matter is the chance of discovering incidental findings during the sequencing. **Incidental findings** are unexpected genetic changes in our DNA unrelated to the initial reason for testing. These unanticipated genetic changes may or may not influence future decisions and actions for you or your family.

Each family requires accurate and relevant information to make informed decisions about whether or not they want to undergo this type of testing. Currently, there is limited research about what information people need to make informed decisions about having whole genome or whole exome sequencing. **What is important information for you to know prior to making a decision? What else is important during your decision-making?** Our goal is to explore what parents view as important in their decision-making for their child’s genomic testing.
Appendix E  Video Links about WGS/ES

“Sequence Me” https://www.youtube.com/watch?v=Z0EaDpt6NXE (Aulakh, 2010).

“Whole Genome Sequencing and You” https://www.youtube.com/watch?v=IXamRS85hXU (Sanderson, 2012)
Appendix F Demographic Information

Demographic Information

Study Title: Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

We would be grateful if you would fill out this information that will allow us to describe our focus group attendees in general terms. Your responses are anonymous, and we will not be able to connect this information to you. Thank you.

Please circle your response:

Gender:
Male Female Other

Age: 20's 30's 40's 50's 60's 70+ prefer not to say

Education: Highest level of school completed
No schooling completed
Primary School
High School
Trade/Vocational
College/University
Graduate/Doctorate

Marital Status:
Single
Common Law
Married
Widowed
Divorced
Separated

What is your Occupation? ____________________________ (optional)

or

Student

Employed

Out of work and looking for work

Out of work but not currently looking for work

Homemaker

Retired

Other ____________

What is your total household income?

Less than $10,000

$10,000 to $29,999

$30,000 to $49,999

$50,000 to $69,999

$70,000 to $89,999

$90,000 to $99,999

$100,000 or more

How would you classify yourself?

Aboriginal

Black/African Canadian

Asian

Caucasian

Other ____________

Prefer not to say

Where do you currently live?

Within Vancouver

<15km from Vancouver

>15km from Vancouver

>30km from Vancouver

Outside of British Columbia

How many children do you have? _________________

Your child’s diagnosis (if applicable): _________________
Appendix G  Topic Guide

Study Title: Supporting Decision-Making in Whole Genome/Whole Exome Sequencing: Exploring Parents’ Perspectives

✓ INFORMATION SHEET
✓ “WGS & You,” “Sequence Me” VIDEOS
✓ Review and any Questions about CONSENT FORM
✓ Filled out DEMOGRAPHIC FORM

INTRODUCTION (10 minutes)

- Welcome and thank you
- Introduce first name and role
- Ground Rules: Respect and listen, one person speaks at a time, no right or wrong responses cell-phone, distraction etiquette
- Time: approximately 1-1.5 hours.
- Emphasize confidentiality, anonymity, de-identification
  o We encourage all participants to refrain from disclosing names
- Will be audio-taped and I’ll be taking notes
- Verify the Tape Recorder
- Format of interview – ask standard questions, might ask additional questions to clarify what you mean or to help you expand on your responses.

Remember, if you wish to stop at any point, we can stop the interview.

Purpose of the interview
- The purpose of today’s interview is to understand your perspectives and opinions about what you would need to support your decision making about Whole Genome/Whole Exome Sequencing (WGS/WES).
- Explain the format of the interview – will pose some questions, but you may respond or talk about anything that is important to you during the decision making process

Any Questions?
Introduction to Whole Genome/Whole Exome Sequencing (WGS/WES) (10 minutes)

CONTEXT
Presenting problem for sequencing?
Motive for sequencing?
For child? For family? Other Reasons?
More than one child affected?
Fears during decision making?

QUESTIONS ABOUT INFORMATIONAL/KNOWLEDGE NEEDS (20 minutes)

What is the most important information or knowledge before making a decision about whole genome/whole exome sequencing?

Probing Questions:

I. What type of information would you see as most important to help make a decision? – e.g., content/topic
II. What information do you think health care professionals should share with patients and families related to making a decision about genomic testing?
   I. In order to help yourself make a decision GS, what actions would you take? Or what information would you look for?
   II. Prior to making a decision, how important is it for you to have basic knowledge about genetics and genomic testing (like the brief summary at the beginning of this session)?
   III. What information would you like to know about the genomic testing process?
      a. E.g., how the tests are done, where and who analyzes the results, what happens to the results
IV. What do you consider as NOT helpful in supporting your decision?
V. What other information would you like to know?
   - If no response, list some examples from below
   - Would you think about that? Would you worry about that? Would you want to know more about that? Would you have questions about that?
   E.g., (Bick & Dimmock, 2011; Ormond et al., 2010).
      - Basic genetic knowledge (e.g., cells, genes, chromosomes, mutations) - similar to the brief explanation during the introduction
      - Patterns of inheritance
      - Types of DNA variants
         i. Pathogenic – has clinical consequences, has effect on the individual
         ii. Benign – no effect on the individual
         iii. Unknown significance – effect of the DNA change is unknown
      - Incidental findings, and impact on family (e.g., BRCA1 Mutation)
         i. Childhood onset versus adult onset
         ii. Medically actionable versus not medically actionable
      - False positives and false negatives
      - Scientific discoveries that result from test results
- Interaction of genes and environment
- **Information privacy**
- Non-paternity
- **Return of which results?**
- Genetic Information Nondiscrimination Act 2008 legislation (USA)
- Risks for common adult diseases such as diabetes, some cancers, heart disease, psychiatric conditions, behavioural traits
- **Implications for reproductive choices, children, relatives**
- **Implications for insurance, employment**
- Potential for stigma

VI. Do you think it is necessary to have a health professional available to offer their clinical knowledge and help with your decision making about GS? Why or why not?

**QUESTIONS ABOUT OTHER CONTEXTUAL, PERSONAL, SOCIAL FACTORS THAT MAY AFFECT THEIR DECISION MAKING**

VII. **What factors in your life do you think will affect your decision making around genomic testing?**
   E.g., knowing someone with a genetic condition, cost of testing…

**PROCESS OF COUNSELLING (probes)**
- Did you get counselled by a genetic counsellor?
- Who was at the counselling session?
- How did the consent process go?
- What do you think you really need to know?
- Who made the decision?
- What could be better?

**QUESTIONS ABOUT RESOURCES AND SUPPORT (20 minutes)**
What and/or who else do you think would help you make your decision?

Probing questions:
I. **Other sources:**
   - Individual decision
   - With partner
   - Discussion with family/friends
   - Health care professionals – e.g., family doctor, nurse practitioner
   - Anything else - e.g. spiritual? Personal values, beliefs, expectations

II. **What form should that information take? When would you want this information?**

III. **What are supports or hinders your ability to make a decision?**
What about online resources?
- E.g., Own research online – (factual)
- Google; (other peoples stories)
- Online support groups, discussion forums
- Webinars, podcasts
- Social media (Facebook, Twitter, Pinterest)
- web-based applications, other tools or applications designed to help you decide

In what medium (form) would you prefer to receive additional information and/or support?
- If participants need context: e.g., Some genetic counsellors spend hours of clinic time consulting with a patient/family
- E.g., face-to-face, telephone, online: email, forum, web-based application, seminars/webinars, podcasts, tools or apps designed to help you decide
- Where else might you get/search for information on: genomic testing/ how people decide?

End Questions (10-15 minutes)
I. Is there anything that anyone would like to add to any of the topics that we discussed today? Anything that you have not had a chance to say?
II. Is there anything that we did not talk about today that you would like to discuss?

SUMMARIZE

Is there anything else anyone would like to add?

Thank you
Remind: May be contacted in the future to confirm research findings (in consent)
Q-forms – name of payee and confirm mailing address