

**THE CHANGING MEANING OF GENE THERAPY:**

*Exploring the significance of curative genetic research in the narratives of families with  
Duchenne muscular dystrophy*

by

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## ABSTRACT

Gene therapy and stem cell therapy are symbols of a futuristic age in biomedicine. These experimental treatments have led to speculation that many presently untreatable diseases will soon be cured. However, there has been relatively little study of how this conjecture affects families experiencing a serious genetic illness. This study is based on interviews conducted with ten families in which a child has Duchenne muscular dystrophy (DMD) – a lethal childhood disorder. Parents were asked to relate their hopes and expectations for advanced genetic research, and its relevance in their everyday lives. Building on a tradition of scholarship in the social sciences that examines how individuals make sense of their experiences with illness through narrative, I show how the field of gene therapy research is implicated in the process of emplotting and telling a story about DMD and remodifying it as circumstances change. I point to some of the ways in which the significance of curative genetic research for parents varies over time. I illustrate how the field of genetics serves as a narrative device, taking on different meanings depending on its place in the story parents tell about their child's disease. I also show how parents learn about genetic research mainly by participating in socially constituted communities of practice, a process I liken to the concept of legitimate peripheral participation. This study contributes to a growing debate about whether lay-actors are sufficiently informed vis-à-vis the risks and benefits involved in experimental medical research, by showing how studies of "therapeutic misconception" can overlook the broader picture. Attention is drawn to the manner in which research is situated in parents' personal biographies and everyday lives, to the socially constituted ways in which parents come to acquire knowledge about and construct expectations for the field of genetics, and to the multiple places that it occupies in the stories they construct and tell about their child's chronic illness. Brief overviews of the historical trajectory of gene therapy research, and recent developments in curative medical research related to DMD are also provided.

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## INTRODUCTION

Genetic therapies are symbols of a futuristic age in biomedicine. Often described as “just around the corner”, these experimental treatments have generated immense speculation that many serious, presently untreatable disorders will soon be cured. There are perhaps no bigger stakeholders in the success of this research than the families experiencing lethal hereditary diseases such as cystic fibrosis, Huntington disease, and muscular dystrophy. But while the hopes that such families invest in genomic science are often obliquely acknowledged by researchers and clinicians, they are rarely documented or explicated in detail. One result is a curious gap in the cultural narratives that sustain scientific discovery; high-tech, cutting-edge genomic research is undertaken for the noble purpose of “curing the incurable” and “saving the sick”, but there is comparatively little discussion of the meanings and expectations attached to this science by the families for whom it is intended, or of the multiple ways in which it enters their everyday lives. The aim of this study was to investigate how gene therapy research is understood, interpreted and narrated by families experiencing a fatal genetic disease, who presumably have the most to gain (or lose) from the field of curative genomics.

Many of these families experienced the optimism of the past decade of bio-technical research, and have reason to be both awed and disappointed by its results. The Human Genome Project (HGP) generated an unprecedented quantity of information about the operation of the cell, and the aetiology of its dysfunction.<sup>1</sup> The widely-regarded success of the HGP, and the ambitious scientific agenda it has spawned, have led some commentators to refer to this period as a new “Genomic Era” of disease management and research (Guttmacher and Collins 2003).<sup>2</sup>

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<sup>1</sup> The Human Genome Project was an internationally coordinated research effort to catalogue, sequence and map the entire genetic structure of the human organism. It was completed in April 2003, and has been described as one of the greatest feats of scientific exploration in history. See <http://www.genome.gov/10001772>.

<sup>2</sup> Guttmacher and Collins (2002: 1512) define genomics as “the study not just of single genes, but of the functions and interactions of all the genes in the genome”. The word is used to draw a distinction with the more conventional

Against this backdrop of enthusiasm and accomplishment, however, is the reality that genetic therapies have yet to demonstrate substantial utility for treating human disease. Relatively few have gone beyond Phase I clinical trials, which seek only to evaluate the safety of novel treatments on a small number of patients.<sup>3</sup>

This study elicited narratives about hope and cure in the context of this new “Genomic Era” from western Canadian families experiencing a serious chronic illness. The objective was to explore how the current disconnect between the promise and reality of medical genetics acquires significance in everyday life. Parents of children with Duchenne muscular dystrophy (DMD), a progressive and fatal muscle-weakening condition that affects children, were interviewed about their perceptions of and expectations for gene-based therapeutic research. DMD has been a prominent disease in the field of gene and stem cell therapies, which have taken on an aura of “miracle technology” (Stockdale 1999). Experimental successes with potential treatments for the disease have been enthusiastically reported in both the popular and medical press (e.g. Bremmer-Bout, et al. 2004; Gregorevic, et al. 2004; Skuk, et al. 2004), leading some to speculate that a cure is tantalizingly close. Nevertheless, these prospective therapies still face significant obstacles before they can be widely tested on humans, let alone offered as treatments on a large scale (see Chamberlain 2002). For those coping with a genetic illness, gene therapy might therefore be compared to experimental treatments being tested for various kinds of cancer, for it is embedded and given meaning within a discourse of hopefulness (Delvecchio-Good 1991).

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term “genetics”, being “the study of single genes and their effects”. Gene therapy for DMD, the topic that concerns us here, falls somewhere in between.

<sup>3</sup> For example, 32 of 1076 (3%) of gene transfer trials conducted between 1989 (the first year for which data are available) and 2005, have gone beyond Phase I/II (Journal of Gene Medicine 2005). Information on currently registered clinical trials in the United States can also be located at <http://www.clinicaltrials.gov>. Also see this website for details about the phases into which the US Food and Drug Administration classifies clinical research.

This thesis adds an anthropologist's perspective to a growing debate questioning whether participants in clinical trials of genetic therapies are adequately informed about the considerable risks involved in the research, and the very low likelihood that the experimental therapy under study will benefit them personally (e.g. Arkin, et al. 2005; Henderson, et al. 2004; Kimmelman and Palmour 2005; King, et al. 2005). Much of this literature has approached this question by testing subjects for evidence of "therapeutic misconceptions" (Appelbaum, et al. 1982), which essentially amount to unrealistic expectations for research that are not consistent with the (often low) probability that it will bring direct improvement to one's health.<sup>4</sup> Widely seen as well-demonstrated in many types of clinical trials (e.g. Joffe, et al. 2001; Lidz, et al. 2004; Madsen, et al. 2002; Schaeffer, et al. 1996), therapeutic misconceptions are entwined with the hopes that patients have for curative medical science.

However, subjects' "inflated expectations" and "misconceptions" are usually explored only in the context of a clinical trial itself, and are often attributed either to a lack of knowledge on the part of lay-participants, or to the failure of protocols used for seeking informed consent (Sankar 2004). Reliance on this model leads us to overlook the way in which beliefs, expectations, values and decisions respecting genetic technology are framed by subjects' everyday lives – their family history with the disease, their personal biographies, and their clinical experiences, for example. Here, I look beyond the encounter(s) during which patient and researcher meet in the course of a clinical experiment, to ask how curative genetic research factors into the lives of patients and families in the everyday. How do people learn about it? How do they frame and construct their expectations for it? And how do they integrate it into the

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<sup>4</sup> A research subject is said to be "therapeutically misconceived" if she believes she is receiving regular medical treatment (and thus expects her health to improve), when in fact she is participating in experimental research. Research entails different risks and has different goals from treatment, and often there is an extremely low probability that one will benefit personally from participating in it. This is especially true in the case of Phase I research (the phase in which most types of gene therapy are currently tested). See Appelbaum, et. al (1982).

narratives through which they organize and communicate their experiences of chronic illness? When we ask these questions, I contend, we gain deeper insight into the multiplicity of meanings that genetic research acquires over time as families come to terms with, and attempt to make sense of, a serious chronic illness.<sup>5</sup>

This study draws upon and aims to contribute to a body of work by medical anthropologists examining lay-understandings of the new genetics and the way in which they often differ from those held by clinicians and researchers (e.g. Cox and McKellin 1999; Durant, et al. 1996; Richards 1996). It is also informed by a rich tradition of phenomenological research in the social sciences that examines how individuals make sense of and represent their experiences with illness through the process of narrative (e.g. Good 1990; Kleinman 1988; Mattingly 1994; Mattingly and Garro 2000). Within this theoretical framework, the subjective experience of illness is seen as consisting of a process of active engagement in “emplotting” a story about disease that is constantly being reworked and retold over time (Mattingly 1994). Parents use these stories to lend coherence, temporality and sense to the chaos and disruption imposed by disease. In a later section, I elaborate on the theory of narrative emplotment and its importance to the study of chronic illness.

Using data from in-depth interviews I conducted with parents of children with DMD, I argue that the field of advanced genetic research is implicated in this process of emplotting a narrative about DMD and remodifying it as circumstances change. I suggest that we can discern from parents’ narratives a basic narrative schema, or storyline, along which they proceed in coming to make sense of and establishing hopes for genetic research. As part of a broader

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<sup>5</sup> This study fits within a vast body of literature on chronic illness that takes the family as its unit of analysis (for reviews, see Shapiro 1983 and Ell 1996). It is now widely accepted that the experience of illness takes place within families rather than simply individuals, and is deeply influenced by the network of interpersonal relationships and by the exchange of knowledge and (usually) support that occurs within the family unit. Here, parents’ perspectives are taken to represent in some sense the perspective of “the family” due to ethical and practical constraints that prevented interviewing children and under-age siblings directly.



process of constructing a coherent personal biography that incorporates illness and reconciles its disruption, the significance of gene therapy varies over time. As parents begin to live successfully with their child's illness, there is a transition in how they perceive gene therapy that can be observed as they start to articulate narratives of hopefulness and healing from within the context of their everyday lives. Genetic research can thus usefully be viewed as a narrative device that is used for different ends, and that takes on different meanings depending on its place in the story parents tell about their child's disease. Further, I suggest that this process is one that is fundamentally social in nature, as parents learn about genetic science and interpret its significance mainly by participating in socially constituted communities of practice. Ultimately, this study draws our attention to the fluid and temporally variable nature of lay-actors' understandings of genetics, and to the ambivalence and contradiction that characterize their expectations for it. In the light of the rich complexity of parents' narratives about illness, the concept of therapeutic misconception appears rather limited.

This thesis is divided into several sections. The next section describes the design of the study. Thereafter, I give a brief overview of DMD, highlighting some of the features of the disease from a clinical perspective. I then explain the nature of gene therapy and offer a condensed description of some of the research that has led to speculation that a cure for this disease may be imminent. The thesis continues with a discussion of the theoretical framework that informed this study, in order to lay the groundwork for the analysis of parents' narratives that follows. Lastly, I conclude by raising some of the implications of the study as well as some of its limitations.

## STUDY DESIGN

This thesis is based on data from fieldnotes and transcripts of ten in-depth interviews I conducted with 14 parents of children with Duchenne muscular dystrophy (DMD) living throughout British Columbia, Canada. I met these families as part of a larger study carried out at a regional tertiary care centre examining parents' narrative descriptions of their child's chronic health condition and their experiences seeking health care (Miller, et al., in preparation).

Participants were recruited through a multidisciplinary care clinic, and via letter mailed out by a patient support organization.<sup>6</sup> An intermediary invited parents to participate if they had a child with DMD between the ages of 5 and 14. I employed these age restrictions in order to reach parents who had had a period of time in which to receive, adjust to, and reflect upon their child's diagnosis and prognosis, but who were still in the early to middle (i.e. not palliative) stages of the disease's progression. Consistent with qualitative research recruiting strategies, which aim to capture a broad view of the diversity of individuals' experiences, families from a variety of geographic locations across British Columbia, and from different cultural, educational, linguistic and class backgrounds were targeted.<sup>7</sup>

Interviews were conducted in the spring of 2005, and the majority took place in families' homes;<sup>8</sup> they lasted between 1.5 and 2.5 hours. The interview protocol contained two sections, and was open-ended and loosely structured. The first section asked parents to relate their experiences with the disease, as well as their attitudes toward, and hopes and expectations for advanced genetic research. The second section included questions about their experiences

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<sup>6</sup> The name of the care centre and the patient support organization are withheld to maintain anonymity. The study procedures, forms and interview protocol were approved by the University of British Columbia's Behavioural Research Ethics Board, and by the Research Ethics Committee of the hospital in which recruitment took place.

<sup>7</sup> See the conclusion of this thesis for more information on limitations due to the recruitment strategy used.

<sup>8</sup> One interview was conducted at a participant's workplace. Three were conducted at a local charitable organization where families from out-of-town reside while attending regularly scheduled clinic visits.

obtaining health care for their child, and sought their comments on continuity in care (Miller, et al. in preparation). Data for this thesis come primarily from the first portion of the interviews.

I attempted as much as possible to give interviewees the opportunity to guide the conversation to areas they deemed to be important and/or salient. This strategy is widely seen to enhance validity by allowing the interviewee to determine the timing, sequence, context and topical area within which the discussion takes place (Mishler 1991). Interviews and field observations were audio-taped, fully transcribed and analyzed manually using *Atlas-ti* for data management and support. Transcripts and notes were read iteratively during the interview process in order to identify themes for follow-up in later interviews. Data were coded thematically using an open-coding process to inductively capture themes emerging from parents' narratives, and to avoid imposing a preconceived analytic framework onto the data. Parents' narratives were considered holistically, and were systematically and repeatedly compared to search for similarities and differences. Themes identified by this process were then integrated into subsequent interviews in order to seek elaboration.

#### **DUCHENNE MUSCULAR DYSTROPHY: OVERVIEW OF THE DISEASE**

Duchenne muscular dystrophy is a neuromuscular illness that primarily affects male children. It is characterized by the progressive weakening of the skeletal muscles, which leads to loss of the ability to walk and perform some day to day tasks, and later to more serious complications. The illness is generally (but not always) diagnosed between the ages of 3 and 5, by which time parents start to notice that their child is slow in walking or "not keeping up with his friends". It can take some time before a parent's suspicions are acknowledged by a physician and confirmed by a clinical diagnosis (Marshall and Galasko 1995).

Most often, children require a wheelchair for mobility<sup>9</sup> by their pre-teens, and as the disease progresses they gradually need more assistance in the performance of everyday tasks such as using the toilet and bathing and, later, eating and breathing. A recent Dutch study calculated the median age of survival with the disease to be 19.4 years (95% CI 19.0-19.8 years), but some 23% of patients lived to the age of 23 and beyond (van Essen, et al. 1998). More recently, with improved nursing care and the use of assisted ventilation, some individuals are living into their thirties (Simonds, et al. 2000). DMD is relatively common among genetic diseases in Western countries, with an incidence of approximately 1 in every 3,500 live male births (Emery and Muntoni 2003).

The gene that causes the disease is inherited in an x-linked recessive fashion, meaning that mothers can be carriers of the gene but do not normally exhibit symptoms of the disease; DMD is usually passed from mothers only to their male children. Approximately one third of cases result from new genetic mutations (i.e. there is no family history of the illness) (Emery and Muntoni 2003). Currently there are very few treatment options available to children with DMD, much less a cure for the disease.

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<sup>9</sup> Here, I intentionally avoid the phrase “confined to a wheelchair”, or similar terminology that is more conventionally found in the medical literature (e.g. Wells and Wells 2002). Parents in this study pointed repeatedly to the fact that many day-to-day activities can still be carried out despite a reduced- or in- ability to walk normally. Several parents in this study discussed how for them, their child’s wheelchair has been “freeing” rather than “confining” since some activities are *easier* to carry out by using it as a mobility aid.

## **HISTORICAL CONTEXT: GENE THERAPY RESEARCH AND DMD<sup>10</sup>**

### **A Brief History of Gene Therapy**

Others have provided a more complete history of gene therapy than space permits here (e.g. Friedmann 1997; Martin 1999; Scollay 2001; Wolff and Lederberg 1994). The technique involves the introduction of new genetic material into the cells of an organism in order to either “improve” it, or to treat or cure disease. The idea for gene therapy first entered medical discourse in 1966, when Edward Tatum proposed that individuals suffering from a genetic disease could be treated by introducing a functional gene into their affected tissue (Tatum 1966). The objective is for this introduced gene to restore the lost function caused by the patient’s malfunctioning endogenous one. If the new gene can be integrated into the cells of the recipient and expressed, gene therapy offers the potential to treat or cure a host of chronic hereditary conditions for which there are presently few treatment options, including various muscular dystrophies, cystic fibrosis, sickle cell anaemia, thalassemia and many others.

Gene therapy research experienced a boom in the 1990s, especially following the success of French and colleagues in treating a child suffering from a severe immune disorder, reported in 1995 (Blaese, et al. 1995), and significant investment in the technology by the pharmaceutical sector. Researchers have sought to use variations of gene therapy to cure diseases as diverse as cancer (Yamamoto and Curiel 2005), heart disease (Jones and Koch 2005), and HIV/AIDS (Strayer, et al. 2005), with little tangible success to date. The field suffered setbacks in 1999 when an 18-year-old participant in a gene therapy clinical trial died as a result of the treatment (NIH Advisory Committee 2000), and again in 2001 when three children participating in a

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<sup>10</sup> Several commentators have argued that the term “gene therapy” is a misnomer, since very few genetic treatments have yet proven to be clinically therapeutic, and all are delivered under research protocols (e.g. Dubowitz 2004; Churchill, et al. 1998). The term “gene transfer” has been suggested instead. Nevertheless, the majority of parents in this study used the term “gene therapy”, reflecting the popularized conventional name assigned to this field of research. I see and use the terms “gene therapy” and “gene transfer” synonymously.

clinical trial in France contracted leukemia following the experiment (Hacein-Bey-Abina, et al. 2003). Today, gene therapy is regaining credibility as a potentially viable method for treating many diseases, including DMD; a new clinical trial for the disease is planned in 2006.<sup>11</sup>

### **Gene Therapies for DMD**

Research on possible genetic treatments for DMD emerged into the milieu of hype and hope that characterized the field of genetics in the late 1980s. Efforts have been focused on restoring the ability of muscle cells to produce a functional protein called dystrophin, which is essential for proper muscle function but lacking in those who have the disease. The two main strategies being pursued are myoblast cell transfer and gene transfer (Dubowitz 2004).

*Cell transfer* involves the direct injection of cells (either the patient's own cells genetically modified *ex vivo*, or those of a compatible donor) into tissue with the goal of achieving regeneration and repair. This experimental strategy is again in the limelight as a result of enthusiasm and controversy surrounding "stem cell research". Trials of cell transfer for DMD have yielded mixed results. The first successes occurred in the late 1980s and early 1990s on animal models (Partridge, et al. 1989), but there has been difficulty demonstrating similar results in humans (Karpati, et al. 1993). More recently, some success has been reported with this technique by researchers in Quebec, who injected three patients with donor myoblast cells and achieved dystrophin expression at levels approximately 10% of normal (Skuk, et al. 2004).

DMD *gene transfer* studies have generally taken place in the lab on animal models, and have attempted to re-establish dystrophin production in muscle tissue by either introducing a new, corrective gene into the cells, or by "masking" the existing genetic mutation and nullifying its deleterious effect.<sup>12</sup> Both techniques require the introduction of new genetic material, and

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<sup>11</sup> See <http://www.clinicaltrials.gov>, identifier no. NCT00159250.

<sup>12</sup> The latter technique involves the use of antisense oligonucleotide technology and aims to regulate gene expression.

much research has been focused on identifying and refining viruses to serve as vectors for carrying the modified strand of DNA. In the case of DMD, the large size of the dystrophin gene has posed a difficult challenge for researchers, since a vector large enough to deliver it is required (Chamberlain 2002).

“Successful” gene therapy experiments for muscular dystrophy have been widely reported and publicized, both in the popular media and via the newsletters and websites of muscular dystrophy charitable and support organizations.<sup>13</sup> Institutions such as the Muscular Dystrophy Association of America (MDA) and its international counterparts also regularly facilitate electronic and conference forums wherein patients and their families are able to pose questions to researchers about research developments. MDA and similar groups have a legitimate interest in promoting the results of gene and stem cell research because it encourages donations from the public and sustains the attention of government granting agencies. Nevertheless, on a number of occasions these organizations have also been criticised for heralding potential cures with unwarranted enthusiasm, thus inappropriately raising the hopes of patients and families (Dubowitz 2004). Several parents interviewed in this study referred to events following the Quebec study on myoblast transfer discussed above (the technique involving injections of healthy cells into muscle tissue):

There's that research they're doing in Quebec, where they're injecting the healthy muscle from a parent into the muscle of the child. Grafting muscle, and they were seeing some improvements but, guess what, one hundred shots in his hips and knees or something. Yeah, it's great that it worked. It was unfortunate that the [patient support organization] sent a letter to everybody, a press release promising results in the research. Everybody got all excited about it, and it was hugely exciting. It was like “we're almost there, we're almost there!” Then, you haven't heard anything since. The [clinicians] actually brought everybody back down to ground pretty quickly, and I think the [patient support organization] got a bit of a

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<sup>13</sup> These can be found by simply searching the internet for “muscular dystrophy” and proceeding to the “research” sections of any number of websites. See, as two examples of many, <http://www.jessesjourney.com> and <http://www.mdausa.org>.

slap on the wrist for sending that information out as early as they did, because it can be really disappointing. I don't want my kids...they experience enough loss on a daily basis with their bodies not doing what they want them to do, so I'm not going to start giving anyone false hopes.

As these comments suggest, and as others have shown (Dubowitz 2002; Stockdale 1999), hyped and exaggerated claims about the potential of advanced genetic research are anything but benign for families who have hopes invested in their success.

## **THEORETICAL FRAMEWORK**

This thesis invokes two theoretical paradigms that I elaborate here. These are 1) the cultural construction of illness narratives; and 2) the concepts of situated learning and legitimate peripheral participation.

### **Narratives of Illness**

There is now a well-established literature in anthropology examining how subjects attach significance to and create meaning out of experience by using the concepts and rhetorical tools of narrative and storytelling. From this theoretical perspective, situating ourselves within a narrative told as a story is seen as a key feature of the way in which individuals make their lives meaningful and create coherence out of disparate life events (Agar 1980; Becker 1997; Bruner 1986; Geertz 1973; Mattingly 1994; Ricoeur 1984). Producing and telling stories to others is seen as a way of giving "form to feeling" (Langer 1953) – of creating sense and conveying to others what it feels like to live in this world (Mattingly and Garro 2000: 11). In addition to being a fundamentally social act, assembling and telling a narrative story is also a process that occurs at a cognitive level. It entails the attribution of a beginning, middle and end, in relation to which meaning, moral, and purpose are established (Mattingly 1994). Telling stories allows us to order experience and communicate to researchers, clinicians and others what is significant in our lives



(Rosaldo 1986). For anthropologists, narratives offer a point of entry for exploring how experiences are made intelligible and represented.

The study of narrative has thus given impetus to a far-reaching body of work examining how “narrative mediates between an inner world of thought-feeling and an outer world of observable actions and affairs” (Mattingly and Garro 2000: 1). Scholars who use the concept to study illness and healing draw on diverse analytic strategies to examine the ways in which individuals create and convey the meaning of illness through storytelling.<sup>14</sup> Generally, however, the narrative tradition in illness research engages with the activities through which

healers, patients, and their kin construct and negotiate interpretations of their experiences and use those interpretive frames to guide future actions. There is an interest in the dramas which surround illness, in the temporal contexts in which it occurs, and in illness and healing as dynamic processes in which meaning is not a given but something actors struggle to discover (Mattingly and Garro 2000: 11).

This theoretical paradigm has been mobilized productively in work that examines the experience of both chronic illness (e.g. Garro 1992; Garro 1994; Good 1990; Good and Good 1994; Kleinman 1988; Landsman 2003; Mattingly 1998; Murphy, et al. 1988) and acute illness (e.g. Kirmayer 2000) in clinical and everyday settings. For example, anthropologists have explored the ways in which patients and clinicians work together (or, often, fail to work together) to co-construct narrative representations of illness that resonate with notions of self and create opportunities for stories of healing (e.g. Capps and Ochs 1995; Eisenberg 1981; Kirmayer 2000; Mattingly 1994).

The means through which individuals construct a narrative – by which they assemble life events into a coherent story – has been labelled by Mattingly (1994) and others as a process of

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<sup>14</sup> For reviews, see Hyden (1997); Mattingly and Garro (2000); Mishler (1995); and Somers (1994).

“emplotment”.<sup>15</sup> Though the term has been used in various ways, it is generally seen as the method by which one “narrativizes”, or orders experiences temporally, makes connections between them, and “creates a whole out of a succession of events” (Mattingly 1994: 812; see also Ricoeur 1980, 1984). When emplotting a story one invokes a process of “editing” (i.e. deciding what kind of information is significant or “worthy” of being included), and relies on culturally proscribed notions of what constitutes a story, to whom it should be told, and in what form it should be conveyed (Mattingly and Garro 2000; Shore 1995). Learning how to tell a story is “fundamentally a cultural matter...a constructive process and a learned skill” (Mattingly and Garro 2000: 25) (see also Bruner and Feldman 1996; Frank 1995). Narrative is thus a point of entry for examining the relationship between individual agency and cultural embeddedness (e.g. Good and Good 1994).

Stories vary too, depending on who is telling them and who is listening. As a dialogical (Frank 2005) and “relational act” (Linde 1993: 112-113) that implicates the audience in its production, emplotment also exposes the contingency of social interaction and communication, for it takes place in social time and place. Individuals emplot different stories in different contexts, choosing what to leave in and what to leave out depending on their relationship with the listener and their perception of her expectations. Moreover, stories are fluid and constantly being reworked to incorporate new experiences and perspectives (Price 1987). As researchers and clinicians, we are engaged with our subjects in the act of co-constructing a shared story. Its content is thus dependent on the particularities and context in which our interaction takes place. The relationship between the stories we hear and the lived reality of peoples’ lives is always an open question – a theme I return to in the conclusion of this thesis.

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<sup>15</sup> Mattingly (1994) traces the term “emplotment” to literary critic Northrop Frye (1957), who used it to describe four archetypal plot structures for representing experience: romance, tragedy, comedy and satire.

Narrative theory has impacted the way anthropologists view and talk about the experience of chronic illness in particular. Much of this research builds on work by Bury (1982) and others (e.g. Becker 1997; Frank 1995; Garro 1992; Williams 1984), who have argued influentially that the diagnosis of a serious disease is akin to a “biographical disruption” of one’s sense of self. The arrival of an unanticipated disease shatters hopes and plans for the future, and marks a “biographical shift from a normal trajectory through relatively predictable chronological steps, to one fundamentally abnormal and inwardly damaging” (Bury 1982: 171). It thus provokes a re-examination of a person’s hopes and expectations “in life”. Individuals and families are confronted with the task of reordering priorities, perspectives, and social relationships in accordance with the shortened time span imposed by disease (Becker 1997). Work on chronic illness has foregrounded this process of reconstitution of identity and self that takes place in the wake of being diagnosed with a serious illness. This process of “narrative reconstruction” (Williams 1984) occurs as an individual seeks to restore a sense of stability and order to her life, by reconstructing a personal biography that articulates a legitimate and meaningful place for illness within it. Over time, as illness narratives are reworked, retold, honed and smoothed, they are widely seen to acquire and create stability and coherence in one’s sense of self (e.g. Kirmayer 2000), and they constitute an important means by which individuals find a way to cope, and even thrive, with a debilitating disease.

This study was informed by and aims to contribute to this narrative tradition in medical anthropology. My analysis focuses on the stories parents told me about their everyday experiences with their child’s illness. I seek to unpack the way that gene therapy – or more specifically its potentiality – is implicated in this broader process of constructing a personal biography or narrative that incorporates DMD. But while narrative analysis offers endless possibilities for examining and theorizing the interface between culture, experience and text, I

am intrigued here by its usefulness as a means of getting at the messy details – the contradiction, multiplicity, anxiety and ambiguity – that characterize how illness is experienced as “present in a life” (Good and Good 1994: 841). Good and Good eloquently capture the essence of the perspective I adopt in this thesis:

Illness is grounded in human historicity, in the temporality of individuals and families and communities. It is present as potent memories and as desire. It embodies contradictions and multiplicity. As with aesthetic objects or complex narrative texts, illness cannot be represented all at once or from a single vantage. It is constituted, rather, as a “network of perspectives”... And illness, present in imagination and experience, is constituted with an openness to change and to healing (1994: 841, see also Iser 1978).

In the pages that follow, I show how the field of advanced genetic research, and specifically the *possibility* and the *potentiality* of gene-based therapies, are implicated in the process of emplotting a narrative about DMD and remodifying it as time progresses and life circumstances change. I argue that genetic research serves as a narrative device that is used for different ends, taking on different meanings depending on its place in the story parents tell about their child’s disease. As parents become acquainted with the day-to-day routines posed by DMD, and develop an ability to “live successfully with” their child’s illness, hopefulness and expectations for gene therapy are modified and resituated within a broad personal narrative through which their experience with the disease is organized.

### **Situated Learning and Legitimate Peripheral Participation**

In seeking to explore the socially constituted ways in which families learn about and construct expectations for genetic research, this study is also informed by a theoretical perspective on learning articulated first by Lave and Wenger (1991) in their work on “legitimate peripheral participation”. Drawing heavily on Bordieu’s (1977) work on social practice, symbolic capital and habitus, Lave and Wenger posit the processes of “learning, thinking and knowing [as] relations among people in activity in, with, and arising from the socially and

culturally structured world” (51). In other words, they view learning as an activity in which one acquires knowledge through social participation and via a process of becoming integrated within a wider “community of practice”. Lave and Wenger suggest that the activity of learning is best seen as acquiring mastery over time through a process that loosely resembles *apprenticeship* – by moving from the periphery of a community (as a newcomer or “apprentice”) to the “centre” as an experienced and knowledgeable “full participant”. Acquiring knowledge is seen as a dynamic process of social interaction, as one gains familiarity with the social traditions, practices and knowledge of a community or group.<sup>16</sup> This approach can be contrasted with didactic theories of knowledge acquisition that conceive learning as a linear process of information transfer between “expert” and “student” (see Uljens 1997).

These themes may sound familiar in the context of the topic that concerns us here, namely how families of children with DMD come to learn about, make sense of, and construct hopes and expectations for the field of genetic technology. As I mentioned in the Introduction, many studies that address this topic focus on the transfer of clinical trial-related information from investigators to subjects at the site where research takes place. Here, I argue that parents’ knowledge about genetic research is essentially cut from a much larger piece of cloth; it is acquired situationally over time through social participation in communities of practice.

I argue that parents of children with DMD can be usefully viewed as a geographically dispersed but still coherent community of practice that shares certain cultural features, defined in relation to their common experiences with the disease and its symptoms, their contact with health

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<sup>16</sup> I should note that, for the purposes of clarity and brevity, this description considerably simplifies a complex and eloquently articulated theory on learning contained in Lave and Wenger (1991). In particular, the authors assert that we should be wary of reducing “the end point of ... participation in a community of practice to a uniform or univocal ‘center’, or to a linear notion of skill acquisition” (36). The word “centre” is used cautiously here and only for the sake of clarity in presenting the theory to those readers who may not have encountered it elsewhere. Additionally, Lave and Wenger draw a number of important distinctions between theories of “apprenticeship” and their concept of legitimate peripheral participation. These fall beyond the scope of this thesis.

care services, and the shared knowledge that circulates among families via networks of friendship, support, and participation in various social arenas related to DMD. All of the parents in this study self-identified as members of a “DMD community” in some sense, membership that is constituted in diverse ways for each family, including through participation in charitable organizations, support groups, activities related to the sport of power soccer,<sup>17</sup> political advocacy on behalf of the disabled, or by communicating (often electronically) with other parents of children with this disease. For many, these networks were a key source from which knowledge about genetic research was acquired, and expectations for it established. This framing of community can also be seen in the vein of Simpson’s (2000) notion of “imagined genetic communities”. Following Benedict Anderson (1983), Simpson described the emergence of new forms of social bondedness that coalesce around the sense that individuals share a common genetic (and I would add, illness) identity. This perspective leads us to consider how knowledge and ideas about genetic potentiality are not simply held by bounded and disparate individuals, but rather “travel” through social connections among families embedded in a community of shared practices, experiences, and co-constructed identities.

### **DIFFERENT STORIES OF DMD**

The disease is one of the most interesting and at the same time most sad, of all those with which we have to deal: interesting on account of its peculiar features and mysterious nature; sad on account of our powerlessness to influence its course, except in a very slight degree...It is a disease of early life and of early growth. Manifesting itself commonly at the transition from infancy to childhood, it develops with the child’s development, grows with his growth—so that every increase in stature means an increase in weakness, and each year takes him a step further on the road to...an early and inevitable death. (Gowers 1879)

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<sup>17</sup> Power soccer is a competitive sport of growing popularity in the disabled community, and is essentially similar to the game of indoor soccer, only players are in power wheelchairs and maneuver an oversized ball.

Gowers' opening paragraph to his 1879 monograph summarizes a narrative representation of DMD that has a long history in biomedicine. This "clinically pessimistic" framing of the experience of chronic illness can be found in a vast clinical literature on the disease (Emery and Emery 1995; Roland 2000) and in charitable pamphlets describing muscular dystrophy using words like "progressive", "crippling", "devastating", "wasting" and "terminal".<sup>18</sup> Typically, the major points in this storyline include diagnosis before the age of 5, after parents and clinicians notice a child's slowness in walking and running during the first years of life. By age 13, the child is "confined" to a wheelchair, and death usually results from heart or lung failure by the late teens or early twenties. This representation of DMD has little variation and is written as a tragedy using the language of suffering; words like "coping", "challenge" and "struggle" against the "inevitable" progression of the disease suffuse this description of the disease. This perspective on chronic illness is likely to be familiar to the reader and is pervasive in popular accounts of what it is like to have a "serious illness".

The parents who participated in this study were at various places in the process of creating their own stories around their child's illness, and incorporating it into their everyday lives. In large part, this was due to differences in the time since their son was diagnosed, which ranged in this study from two to ten years. But in all cases, parents framed their experience with DMD in a way that contrasted sharply with the "clinically pessimistic" story of the disease told in the clinical literature and exemplified by Gowers' description of a prolonged and inexorable decline. This contrast was manifest in parents' frequent references to how DMD is for them a positive story in many ways, and certainly not a tragedy with a predictable end. Laura Brown, mother of 9-year-old Joseph, describes this perspective:<sup>19</sup>

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<sup>18</sup> See the websites [www.mdausa.org](http://www.mdausa.org); [www.jessesjourney.org](http://www.jessesjourney.org); or [www.duchennemd.org](http://www.duchennemd.org) for examples.

<sup>19</sup> All names are pseudonyms.

I think at first it really made me think about everybody's mortality. Which was a slap in the face. [chuckle] And then a gift. You know um...it's made me not live in the future. It's made me, live in the present. Which is a real gift. It's um, made me see some really, really good sides of the world that I didn't know were there...It made me realize who my friends were and, who they weren't. Like it's a really good filter. To, to, [chuckle] to, uh, life. It's a really good filter... You know our kids have a really rough road, but, really we see the better part of the world. And I really, I really believe that.

Grace Martin, mother of 9-year-old Aidan, had similar comments:

I think for those of us, we're the lucky ones. People think of us as unlucky because we have these terrible diseases, and our children, and our mothers and fathers or whatever but, actually, we're the lucky ones because, if we've learned our lessons, and it's touched us in that deep way, our lives are so much richer, and it's a life that isn't based on fear. We're more free in a sense? I don't know how to describe it. It's not religion, it's maybe spirituality, but it's not even that, it's just about being...They say "live for today", well, it's more than that. It's not "live for today", it's "live in the moment", cause you only have power in the moment, and the moment you start projecting into the past, "oh, he used to be able to walk" or the future, "oh, he might die when he's 18", I've lost it. You gotta muscle into every moment, right? So, that's kind of where the beauty is, is finding and living from that place and being centered in what is now.

These descriptions sum up a narrative construction of DMD held by all of the parents in this study – one that contests predominant popular and clinical discourses that present the disease as an unmitigated tragedy. They also show the richness with which the illness is experienced and narrated – as a constantly unfolding story that describes "lessons learned in life", and whose ending and morals are shifting and not yet finalized.

There was variability among participants in the severity and rate of progression of DMD among families.<sup>20</sup> Parents were also at diverse points in the process of reconstituting their personal biographies to incorporate and make sense of it. Differences between families, such as

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<sup>20</sup> For example, some children had progressed quickly to requiring a wheelchair for mobility and others were semi-mobile. Two children were still walking exclusively, and one required near-full-time care. The remaining children used a wheelchair full-time. Some parents had taken different strategies, such as administering corticosteroids to their children in an effort to maintain muscle function, or having surgery for heel cord release and/or spinal rod insertion (see Emery 2000 for more information). Clinical evaluations were not conducted in the course of the study and children's state of health was subjectively discerned from parents' descriptions.



their place of residence<sup>21</sup> (and thus their network of care providers), their different family structures<sup>22</sup> and work backgrounds<sup>23</sup> (i.e., their familiarity with the language of medicine and the institutional structure of care [Miller et al., in preparation]), as well as the uniqueness of each interview itself, resulted in variation in the stories and experiences parents chose to relate. Nevertheless, there were common elements to the narratives they told, and prominent themes within them, that emerged during comparative analysis of transcripts and fieldnotes. A general trajectory – or plot – of illness experience could be discerned, around which parents' descriptions varied to some degree idiosyncratically. I provide a synopsis of this basic narrative schema here.

None of the mothers in this study were aware of their carrier status<sup>24</sup> prior to their child's diagnosis, and thus the realization that their child had DMD came as a profound and devastating shock to families. Diagnosis was usually delivered by a general practitioner, pediatrician or in some instances, a neurologist, after a period of suspicion that "something was not right". After diagnosis (and in some cases, before) parents and child were referred to a specialist neuromuscular clinic in a large urban centre, where they learned more about the disease and obtained interdisciplinary care.<sup>25</sup> Nine of ten families had also met with a genetic counsellor relatively soon after their diagnosis to obtain genetic testing and to make decisions about whether (and how) to have more children. Most of the parents in this study had learned about the field of gene therapy either in the course of their own library and internet research on DMD soon after their child's diagnosis, or at these initial clinic and genetic counselling appointments. In general,

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<sup>21</sup> Five of the families in this study lived in an urban setting, and five in a rural setting.

<sup>22</sup> This study included seven two-parent households, one single-parent household, and one foster-parent household.

<sup>23</sup> This study did not ask specific details regarding socioeconomic status (such as household income etc.). Broadly, four of the families who participated in this study could be characterized as having a "working-class" background, four could be described as "middle-class", and two could be described as "wealthy" or "upper-class".

<sup>24</sup> In the case of DMD, carrier status refers to whether a mother carries a mutation on one of her chromosomes in the gene that "causes" DMD. Mothers who are carriers usually do not exhibit symptoms of the disease.

<sup>25</sup> Families typically travel to the urban centre (if necessary) for regular clinic visits every six months, unless there are pressing concerns, in which case visits would be more frequent. Day-to-day medical care is usually handled by a general practitioner or pediatrician, in consultation with the specialist clinic if necessary.

they recalled their clinicians providing a lay-summary of gene therapy and adding “the usual stuff” to the effect that “there is nothing available now, but things are hopeful and something might become available in the future.” After hearing this, all of my interviewees had eagerly and hopefully engaged in a process of inquiry and learning about gene therapy, in an effort to find out more about its prospects and availability. As I illustrate below, they described acquiring much of their knowledge about “what gene therapy is” through community involvement, personal research and social contacts, more so than from clinicians.<sup>26</sup>

By analyzing parents’ narratives systematically, and in particular by evaluating and comparing the stories told by parents of older children with those of younger children, I discerned a general trajectory along which parents progress vis-à-vis their perceptions and understanding of gene therapy, and its significance in their lives. This process is loosely structured, and entails transition from 1) an initial stage of learning about and making sense of the research; 2) coming to terms with its salience, establishing expectations and hopefulness for it, and learning to control and manage exposure to information about it; 3) confronting the lengthy timeline for research and the progressiveness of the disease; and 4) modifying and resituating hopefulness within everyday life. In the sections that follow, I elaborate each of these commonly mentioned themes. I also describe some of the variation around them, and the contradiction, ambiguity and uncertainty in which they are ensconced in parents’ talk.

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<sup>26</sup> Indeed clinicians were surprisingly absent from parents’ descriptions of learning about gene therapy. The reason for this is a topic for further research. Parents occasionally mentioned research news that they had heard to clinicians in order to seek their advice. Some cited a reluctance to ask busy clinicians what they perceived as “frivolous” questions about genetic research, and others perceived non-receptiveness on the part of some care providers to such inquiries. Still others felt that their clinicians would tell them about any new developments and that there was little need to ask. Further study might explore how clinicians approach and answer questions about genetic research from parents. For a discussion of similar themes in the context of cancer, see Delvecchio-Good (1991).

## **“GENE THERAPY” ACCORDING TO PARENTS: VARIATIONS IN MEANING**

Earlier, I provided a (vastly simplified) description of genetic research relating to DMD to give the reader a sense of the complex terrain that parents must navigate as they become familiar with their child’s disease and acquainted with the field of genetics. Many of my interviewees reported facing an enormous challenge in making sense of scientifically complex differences between the various therapeutic modalities. This process of learning “what to think of” gene therapy was a distinctive feature of their experiences with the disease, especially early on. As just described, all of the parents in this study reported becoming aware of the existence of current efforts to find a cure for DMD shortly after their child’s diagnosis. Typically gene therapy was mentioned and described briefly during clinic visits or genetic counselling sessions, where it was framed by clinicians as a “possibility down the line” and rarely explained in detail. The majority of parents in this study described learning most of what they know about gene therapy later, through community interaction and conducting their own research about their child’s illness.

A number of studies have suggested that the frameworks with which lay-persons approach and understand the field of genetics often differ significantly from those used by clinicians and research-scientists (e.g. Copley, et al. 1995; Durant, et al. 1996; Parsons and Atkinson 1992; Richards 1996). In a similar way, parents’ narratives from this study show distinct ways of framing and situating gene therapy in the context of existing treatments with which they are more familiar. One result was that, for parents, the very meaning of the term “gene therapy” often differed considerably from how it is used in the medical literature.

Consider this description:

Well I kind of lump...That’s what I think stem cell research would be, yeah...I didn’t even know that there was a difference like that. I, I, um, thought that when you were using stem cells that you were, inserting the genes into them. I don’t

know maybe I was wrong there, but yeah, we have, I would. ... That's what they were talking about at a conference. Yeah it was basically, you know attaching them to a virus and making them go to the muscles, you know, and they were saying that it would have to be, you know an intravenous therapy. Not an intramuscular therapy.

There were many similar instances of conflation in parents' talk of two treatment categories that are conceptualized in the scientific literature as separate. Thus, in parents' narratives references to "gene therapy" often indexed "stem cell research" as well.<sup>27</sup> This finding suggests that confusion or misunderstanding between clinician-researchers and lay-participants may be traceable to their respectively different understandings of what the term "gene therapy" signifies.

Parents' talk about gene therapy in this study also referenced perceived differences between it and other forms of research and treatment, such as pharmaceutical and surgical interventions, a finding that has also been noted elsewhere (Scully, et al. 2004). These differences were what gave the field of genetics its meaning, highlighting two key points. First, as Stockdale (1999) has noted, gene therapy is not easily assimilated to other models of therapeutic action, and this can pose considerable problems for lay-persons seeking to understand it. Second, they show how frameworks on the new genetics are constructed *in relation to* existing, more well-known treatments. For example, many parents described what gene therapy *is* by saying what it *isn't*. Two examples suffice here:

Henry Chung, father of 14-year-old Jacob:

Henry: Well you know, I got a leaflet, I got information from the Muscular Dystrophy Association of Canada, and it said that in Ottawa there is research about the treatment, and I believe that the treatment should be for the genetic, the genetic treatment, not like the regular, minor surgeries that Jacob had a month ago. But this is not for treatment, it's just a, a kind of you know, because he's growing, but his muscle is not growing, his muscle is weakening. So the doctor

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<sup>27</sup> Importantly, the term "gene transfer", introduced to remove the therapeutic connotation from the moniker "gene therapy" (Churchill, et al. 1998) had little meaning to parents in this study, who tended to recognize and use the term "gene therapy".

made the surgery to prevent the lower back from bending. But this is not that kind of surgery or treatment. [It is] “higher”, which means, kind of, you know, the basic genetic research and the genetic treatment.

Laura Brown, mother of 9-year-old Joseph:

Laura: Like, Deflazicort [a corticosteroid drug administered to some of the children in this study] [is] easy. You can say, okay, I don’t want to take this any more. And wean him off of it and it’s gone. Gene therapy may be a whole different story. I don’t know. No, like if they can say to me and said, “Laura you can take this tablet and in six months it’s out of his system.” [pause] That’s way different from, “you take this tablet and that’s it for the rest of your life.” You know? So there’s that too, right?

She elaborates later:

Laura: I think it’s the newest thing, I think it’s like chemo[therapy] was. You know what I mean? I think honestly when I think of gene therapy that’s what I equate it to...that’s where I put it in my brain, is sort of like it’s the chemo for ALS. It’s the chemo for muscular dystrophy or, or Alzheimer’s. That’s what I think of it as, so you know? And, I say chemo because I think there could be real detrimental parts to it too, right? Like you know what I mean? Like I don’t think it’s safe, I don’t think it’s a hundred percent safe. So that’s, and I don’t think of chemo as being a hundred percent safe but it’s better than the alternative. So, yeah, I don’t know, it would be nice if it were but I don’t think that we’re gonna get there quick enough for, well when I need it.

These descriptions shed light on parents’ efforts to assimilate and understand the concept of genetic therapy within existing frameworks of “conventional” biomedical treatment, and illustrate a process of meaning-making that takes place as they seek to make sense of what gene therapy entails. This process begins long before parents participate in any clinical trials, and it suggests that clinician-researchers and lay-actors are likely to draw on different frameworks to classify and give meaning to “gene therapy”.

#### **FALSE HOPES AND LEARNING TO CONTROL EXPOSURE TO THE DISCOURSE OF GENETICS**

For parents in this study, learning about DMD also entailed becoming familiar with prospective efforts to cure it. As they became acquainted with the disease and began to cope with their child’s diagnosis, many described a process of gradual learning about genetic research

through personal investigation and engagement with the “DMD community” described above. After hearing clinicians mention research, parents became curious and most families began to monitor developments in therapeutic research sporadically, either by consulting the internet, or tapping family, community and clinical networks for research news. As time progressed, eight of the ten families in this study became actively involved in activities related to research support, such as fundraising or volunteering for organizations that seek to enable and sponsor curative research. These parents monitored developments in research most closely, consulting the scientific literature and actively seeking information about new studies and clinical trials. Robert and Lisa Jones are one such couple who are involved with an organization that funds genetic and stem cell research relating to muscular dystrophy. They described following the developments in this field quite carefully. Their child Nathan was diagnosed in November three years ago and by the following January, they were actively engaged in fundraising for a cure:

CJC: Have you always been this closely involved in research?...

Robert: [I] just follow certain medical journals and stuff like that...I'm trying to keep up with that... It's, once you get over the shock of having a kid with a terminal illness and kind of dealing with that on the day-to-day. Um, we've been fortunate enough that he is not at the stage yet, where we have to start [worrying about] mobility and those other kinds of things. Like we're still cognizant of it. So after you get over the initial shock of that and dealing with that, then you know, getting involved with [name of organization] I think happened pretty quick?

Lisa: Oh yeah. It was, by January we were contacting them to see what's involved.

Robert: Yeah, so it was, it was pretty quick. You know, before we started getting into that, and of course their primary thing is raising funds for research. So they have the information on research available on their web site, to start. And then with the newsletters that are quarterly, so you get that kind of information. But, [that] mainly started [with] me looking around at least and taking a look. 'Cause you want to find out, is this gonna be possible in his lifetime? Or what kind of odds are against it? Kind of thing. And we looked at it and thought you know, this stem cell thing is positive. I liked what [name of organization] is doing that way.

For Robert and Lisa, this initial process of learning about the research field and assessing the likelihood that new treatments would arise was an important part of dealing with and making sense of Nathan's initial diagnosis. Assisting in the endeavour to "find a cure" for DMD provides them with an outlet for hope, into which they channel their energies and anxieties about their child's disease.

Mary and Richard Moore, parents of 12-year-old Ben, are closely involved with a large patient support organization. Information about genetic research comes to them via Mary's links as a volunteer. Here, she receives emails and phone calls on research topics and, like many of the families in this study, she attends conferences where she networks with other parents, clinicians and researchers:

Mary: We also have an annual meeting every year, and they try to do something in conjunction. When they do that, they bring in someone who is doing research on a certain type of muscular dystrophy. [Name of a prominent researcher] came and spoke to us, and she's in Toronto at Sick Kids [Hospital]. [Another prominent researcher], he spoke to us – a few of the bigwigs that have made major discoveries. So, they would come and give us an update, and, as a parent ... that's what the parents want to go and hear. That's the number one thing that we're interested in.

CJC: It sounds like you're fairly familiar...

Mary: Yeah. There's not a lot of changes. We hear the same thing, and the question comes up every time: "well, how close are they to a cure?" At [a Canadian university], there's a doctor that's quite involved, I don't know what his particular area is but, anyways, we all got excited and he said, "well listen, we're ten years from even some kind of a trial". So, it's nice to have it happening, but, as a parent, it's never fast enough.

Like most of the parents in this study, Mary and Richard also hear about research through their involvement in the muscular dystrophy community, including from contacts made through Ben's participation in a power soccer league. Genetic research is something that they are keenly interested in, but they are careful not to get "too excited" about new discoveries.

Nine of the ten families I interviewed felt that too much exposure to research, or over-excitement about its results, can be negative and harmful. These families told stories of adopting a reserved stance on news about therapeutic experiments, and described learning to approach pervasive public discourses about gene and stem cell therapies selectively, “in order not to go crazy”. Elizabeth Bailey’s comments were typical in this regard. She is the mother of 10-year-old Nicholas:

CJC: Can you tell me how relevant that is in your lives, that aspect of reading about genetic research and learning about it? Is it something you think about frequently?

Elizabeth: I find that every once in a while I’ll go and read and see if there’s anything new to read about, but I find also that I almost go into “Nicholas-muscular-dystrophy overload”, and if I get too many appointments or too many things happening with his disease at once, I find it emotionally overwhelming, and I have to step back. So when we feel we’re in a good place to go and research some more, then we’ll read about it. Other than that, we try and live one day at a time.

Learning to control one’s exposure to research news, and to manage one’s expectations, was a prominent theme in parents’ narratives. I suggest that it challenges constructions of parents of the terminally ill as having an uncritically cheerful attitude toward curative science. Rather, and more complexly, exposure to information about research was often carefully managed, and parents were usually immediately sceptical upon hearing news about “breakthroughs” in experimental studies. This trepid or hesitant approach was a learned one; parents adopted it after the ensuing disappointment of having had “hopes raised before”. Research news is perceived as capable of causing harm if exposure to it is not carefully controlled, because it confronts parents with the terminal nature of their child’s condition, raises the danger of “false hopes”, and makes salient their uncertainty about whether a cure will “come in time”.



One result is that these parents have learned to employ various strategies for selectively restricting their exposure to news about genetic research, including simply “not reading” during difficult periods, as Elizabeth Bailey described above. For Laura and Thomas Brown, whose 9-year-old son Joseph was diagnosed with DMD five years ago, the best way to limit exposure and still keep informed is to engage a family member in the task of “filtering” information. Laura’s mother, perceived to be strong and more capable of dealing with the difficult nature of some of this information, took up this role:

Laura: [She’s] the best, like my mom takes problems and she’s like, okay, yeah, here it is, let’s deal with it. And she deals with it [chuckle] you know.

Thomas: She’s not afraid to go on the internet and talk to people like, she does stuff that we might-

Laura: She’s amazing.

Thomas: that we might not be ready to do, right?

Laura: Yeah. I couldn’t do what she’s doing. I, I would dissolve [pause]. But um, my mom just is able to filter through everything and it’s whenever she hears anything interesting she’ll say, “hey Laura will you look at this?” and then I look at it.

This tendency to restrict or manage exposure to information about research highlights a feature of the chronic illness experience that emerges in the context of this new “Genomic Era”. As parents learn to navigate the terrain of their child’s illness, so too are they tasked with developing strategies to selectively filter pervasive discourses about potential new “genetic cures”. It also suggests an interesting hypothesis worthy of further investigation, namely that exaggerated or hyped claims about research findings may, in the aggregate and over time, cause some families to be *less* informed about current developments in genetic research. Dhillon and colleagues (2003) noted a similar phenomenon in their study of internet use among parents of children in a neonatal intensive care unit; parents were found to be less likely to access medical

information in light of “information overload” about their child’s disease, and the perceived unreliability of information found online.

Parents’ familiarity with the current status of genetic research changes over time, depending on their child’s disease stage and state of health, their attitudes toward the illness, and the simple vagaries of day-to-day life; this reflects the situational character of knowledge and learning about the disease (Lave and Wenger 1991). Lay-actors’ engagement with genetics is socially determined and best characterized as unstable, fluid, and developing within the nexus of community and family relationships, a finding also noted by Cox and McKellin (1999) in the context of predictive testing. John Davis captures all of these themes as he describes how, for him and his wife Michelle, interest in the field of genetics is presently limited and has declined over time, something others in the community do not realize:

CJC: Do you follow some of the research that’s going on?

John: I do, but I don’t really. I’ve gone through stages where I was on the computer constantly following the research but, at this point, there are so many other things that I need to be doing for my kids that are more important...I’m not going to hold my breath or have false hope on where this is going. The tougher thing is the information everyone else gives: the teachers, the TA [teaching assistant], the teacher down the street, or the next door neighbour who keeps forgetting the difference between muscular dystrophy and multiple sclerosis and cystic fibrosis, and he’s just sure if we do this. And people who are telling us, “you know, I’ve read this enzyme,” or “I’ve read if you give kids this, you get muscle, it’s going to come back.” I find that a lot more frustrating.<sup>28</sup>

The above narratives illustrate the tension between hope and scepticism that saturates parents’ descriptions of genetic research and the ways in which it enters their everyday lives. They also show the tension between hope and realism that underlies and structures how families manage their knowledge of, and their expectations for, the field of genetics.

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<sup>28</sup> Other parents related similar stories of unwanted advice about potential treatments, cures, and research “breakthroughs”. This advice was perceived as being given by others with good intention, but was generally not well received.

Taken together, these descriptions suggest that parents are engaged in a process of learning how to maintain a realistic perspective on the likelihood that new genetic treatments will become available, and how to avoid the quagmire of potentially harmful “false hopes”. This occurs as they become initiated into the experience of having a child with a chronic illness and start to gain familiarity with the disease. This process of situational learning through social engagement can productively be viewed in terms of Lave and Wenger’s (1991) concept of legitimate peripheral participation, in which parents move over time from the periphery (as “newcomers”), to being full participants in the socially constituted community of practices that surrounds the disease. As part of this process of initiation into the experience of chronic illness, they begin to acquire knowledge about the disease and about the field of curative genetic research. Parents are then faced with the challenge of developing strategies for managing the place of genetics in their everyday lives, and determining its role in the stories they construct about their child’s illness. Their learning about genetics can thus be viewed as a process of assimilating knowledge acquired through community participation into a broader illness narrative that is constantly unfolding and reworked over time.

**“IT WILL BE A LONG TIME, I UNDERSTAND THAT”: TENSION BETWEEN HOPING AND TIME**

As parents learn more about the field of genetics, they must assimilate its place within the story they emplot about their child’s disease. This raises questions about time and confronts parents with the need to reconcile the timeline for clinical progression of the disease with the (possibly much longer) timeline for genetic research. All of the parents in this study believe that advances in genetic technology will *eventually* bring a cure for DMD. However, this optimism is tempered and layered with ambivalence about *when* such a treatment might arrive. Hopefulness, seen by all parents as key to maintaining a positive outlook, must nevertheless be

kept realistic, and it abuts the slow pace of research progress. Patricia Smith describes her certainty that a genetic therapy for the disease will eventually be available, but she's not sure if it will be soon enough to benefit her 11-year-old son Kevin:

Patricia: Of course I hope for it. It will happen eventually for a lot of people. It will be a long time before, I understand that. I realize that, and I'm okay with that. Anything they can do, I hope they can do it. I know it's going to take some work. It's going to take a long time. If they're gonna do it, let them do it, more power to them. They've gotta do it, and I think they will in time. If they can beat cancer, we can take this down too...

CJC: Do you follow it closely?

Patricia: No. I don't really need to. I know it will happen. I would like to find out about it, even after I don't have Kevin anymore, it would be kind of neat to find out if they ever do. I know they will, it's just a question of time. For all of them. I think they will for anything. Wait and see.

The perceived inevitability of eventual success in the quest to find a cure for DMD is widespread among interviewees and notable in itself. It is a perspective that is also widely shared by researchers, clinicians, and those who produce material for lay-persons experiencing DMD, many of whom frame the disease as a problem for which a solution is forthcoming, "when we get it all sorted out".<sup>29</sup> However, though many parents use a similar language of inevitability, their narratives also reveal ambivalence about how to make sense of it in the context of their personal experience. Here, the framing of scientific discovery as "eventually inevitable" lies in tension with the perception that successful therapeutics are likely a long way off, and with the sceptical approach that most have adopted from having had "hopes raised before". Many reported that this stance was reinforced by clinicians who tended to encourage parents to "be realistic". The result is considerable anxiety and uncertainty, not about *whether* a cure will come, but *when*, and more specifically, if it will "arrive in time" to be of use for their child. One

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<sup>29</sup> See, as two examples among many, Emery (2000) and the website of the American Muscular Dystrophy Association (<http://www.mdausa.org>).

must not get one's "hopes up". Later in the interview, Patricia Smith's sense of optimism is more tempered:

Patricia: Right now, he's good. He's got everything going for him. It's amazing I don't have to do much for him, they pretty much do it all for me. What worries me about that is what happens when he is old .... What happens then?

CJC: You don't think there will be a cure by then?

Patricia: I don't think so.

CJC: What makes you think that you won't get it in time?

Patricia: I don't want to get my hopes up. I don't want to jump into something and then be disappointed. You gotta face reality. It could take years. It could take months. You just don't know. I just think that it might be too late for Kevin because I think he might be too old at that point. That's why I'm concerned. I'm not saying he won't be here, but I just think it will be too beyond him at that point. They'll look at him and say, "well, no, he might be too far along". There's no sense in that.

The themes of time and chronicity were salient components of parents' narratives and a sense that their child's disease is progressing tempers many parents' hopefulness that a new therapy is forthcoming. The fact that such a therapy is likely to be much more effective if administered when a patient is still young takes on greater significance as a child ages. Parents of older children thus referred to their concerns that potential therapies will have to restore function and repair muscle damage already caused by the disease, which may or may not be possible. Elizabeth Bailey's comments typify ambivalence about genetic therapies in the light of the disease's progression:

Elizabeth: I hope in his lifetime they'll find something to stop what's happening to his muscles. I think it's two stages. I think it has to stop what's happening to his body, and then I think they have to find a way to repair the damage that's already been done, so it's not just, "here's a cure, we're healthy again, and here we go". I think it's a two-long step kind of process. Other families will have to go through what we did, and maybe a 3-year-old being diagnosed today will have a better chance of beating it than Nicholas will. Who knows, it's hopeful. It sounds like they're making progress in a lot of areas, and they're just on the brink of cures for so many things...I hope, in his lifetime, that they do find something, but,

as I say, thinking about that question you're asking, we really don't look down the road.

For Mary and Richard Moore, the fact that their son Ben is already in a wheelchair complicates their expectations for new therapies:

Mary: Oh, we're always hopeful, we have to be. Unfortunately, now we have to look towards, because when you're first involved they say, if they find a cure, it can't repair the damage that's been done, it may stop it where it's at. So, of course, our hope was that they'd find it before he was in a wheelchair, and he went into a wheelchair at [age] 9, so he was fairly early. So then we decided, well, that's it, he's not going to get out of the wheelchair, but now, there's a possibility with stem cell, if they can find something, they might be able to repair or put in the cells that can regenerate and produce dystrophin, and we might be able to get something happening. You kind of just have to have the best you can at the moment and be hopeful.

Mary's narrative juxtaposes the disappointment and optimism about curative research that she and Richard have experienced as Ben's disease has advanced. Their hopes now rest with the potential of "stem cells", which are seen to have healing and regenerative potential. For Henry Chung, father of 14-year-old Jacob, optimism has waned over the years as prospective gene therapies have failed to materialize:

Henry: In the beginning, you know, I had really high expectations. When Jacob was diagnosed with the disease, the doctors told us that the medical technology was improving every day and every day, and so I really expected it. And they told me, maybe five years later there might be a higher treatment, and those kinds of things. But five years and five years, and double five years, and ten years after that, and there was nothing in particular – no good ones came up. And so I thought, "oh, it might be quite difficult, quite tough".

The narratives in this section capture the changing significance of gene therapy for parents as their child grows older. The (likely protracted) length of time before new therapies are developed becomes salient and must be reconciled with the reality of a child's progressing disease. Parents of older children in this study reported a decline in the relevance of genetic research in their everyday lives over time. Many began to stop actively seeking information about it, and a few reported simply "trusting the doctors" to mention any new developments:

The neurologist, we'll talk to her about it, but we don't ask about any research that's going on. We did in the beginning. We just don't anymore, we just make sure his needs are met...We trust that he's getting the best current care that he can get.

As I show below, this change in significance cannot be read simply as "giving up hope", since parents remain extremely *hopeful* about the prospects for genetic research, and take for granted the fact that a cure will eventually be found. Rather, I suggest that it indicates the embeddedness of hopes and expectations for gene therapy within the broader context of parents' self-perceived capacity to live successfully with the disease. As parents gain confidence in their abilities to cope with DMD, the significance of gene therapy and its place in the stories they tell about the illness changes. As I show in the next section, many parents look elsewhere for hopefulness as they begin to define and articulate their ability to live effectively with their child's illness in the absence of a genetic cure.

### **REDEFINITIONS OF HOPE: LIVING IN THE PRESENT**

The previous sections showed how, as parents gain experience with their child's disease and acquire knowledge about the status of experimental attempts to cure it, many have learned to manage hopes and expectations. As just described, they experience anxiety that stems from a sense that their child's disease is progressing in the absence of new therapeutics. Many told stories about how this experience has provoked a reconfiguring of "what hope is" – from "hoping for the future" to "hoping for the present". Michael Johnson's comments, quoted here in their entirety, capture this perspective well:

Michael: You know? Hope is an interesting thing, because hope is something that you can draw a line, in front of you and, put hope on the other side of the line. And say this is what I want to eventually achieve. Or hope can be something you live inside of. So, what we have found and this is, to me has been almost a revelation...Um, that if ...if, if I can continue...spending more time, with, Mathew, than probably a lot of people will spend with their children that will live

longer. What we are really doing is living *in* hope at that point. You know, because I do spend an awful lot of time at home, with the kids and stuff eh? So I am putting in more, like I am building up, time, as we have it...you know? And so, that perspective prevents me from looking, out, for hope too much. ...You still have that line, and there is hope on the other side of that line, as far as maybe a medical cure. But if you're living inside of a personal hope, that, you know, a quality of life kind of a hope, then, then you, you don't have a tendency to, to grasp quite as, vigorously sometimes. You know. And I shouldn't really lose that. I shouldn't really lose that sense, you know. It's still there, so my resolve has come to the point that, uh, if it is, if there is a cure in the cards, uh, for muscular dystrophy say or, or at least if they could stop the disease from progressing, wonderful. It's, it's just, it would be great. But I look at that as something that, and I would have that hope for my son, but I wouldn't want, at this point in time to push too hard to that hope and lose another one, which would be ah sort of our own sense of sanity. You know, and I try and keep that into perspective...If, there was a relatively significant medical advancement Mathew gets treatment, and he does better, great. But, that great, is, [pause] not [pause] as huge as [being able to say] "you know he's still with us and we had an awesome day in the park today."...The one that's here and now. The one that I can do something about today.

Michelle and John Davis narrate their current focus on living within and enjoying "the present moment" with their son Eric. Though they previously anticipated a cure and followed developments in genetic research quite closely, today they emplot a story of hopefulness from their everyday lives by focusing on their child's wholeness as a person, rather than on future possibilities for "physical healing":

Michelle: Physical healing would be great! It would be a great thing, but it is certainly secondary to making sure that my boy is a whole person.

John: I think wholeness is just living in the now. Today is important, not what's going to happen in five years with possibilities that there will be something [a gene-based cure] going on. We've learned you can't live for that, you live for what's important today, what's important tomorrow, next week, next month. That's kind of the all-encompassing importance. What happens next year, well, it would be great to go to [the southern USA] again, but, that's the plan, we don't worry too much about whether it's going to happen or not.

For many of the parents in this study, I suggest, the significance of gene therapy has changed over time as part of this process. Genetics is no longer simply a "source of hope" from which parents draw, as hopefulness becomes rooted in their day-to-day lives and their confidence in



living with the disease begins to crystallize. Gene therapy (or, more accurately, its lack), comes to serve as a symbol which many parents use to affirm their ability to live and cope with their child's illness:

Susan Taylor, mother of 9-year-old Justin: I'm not holding out for that [a gene-based cure] no, no, not at all. I think it would just take too much away from living in the here and now and enjoying who he is now and what we have now. Saying, "oh well, they're going to find something in five years and he'll walk again"... Well, if he is, and if they do find something to fix it in five years, then great! But, I'm not going to sit here now and wait for that five years and not enjoy that five years.

John Davis described the process of learning to live with the disease as one that leads to drawing hopefulness from different sources. The words "healing" and "cure" have come to mean different things over time:

I think that you always are holding out for a cure. You never would give up hope on that, but one of the things I think that I've learned to emphasize with my kids is that...we come from a very strong religious background. One day the youth group leader phoned me, she was really concerned because one of the studies that they were doing was on healing, and he said, "you know, I thought after that, it might just bring up some conversation, [about] why does God heal some people and not other people?" It did precipitate that conversation. So, what you end up doing is saying, "okay, well, what's healing?" Does it have to be physical healing? Is it emotional healing? Is God using you because He's going to heal a pain in somebody else, and what is more important? Would He be able to use you, if you weren't in a chair?

Redefinitions of "what hope is" and "what therapy is" were themes frequently narrated by parents in this study as milestones in a process of coming to terms with their child's diagnosis and learning about the status of genetic research on DMD. Comments such as these suggest that parents' anxieties about the prospects for a gene-based cure are resolved by focussing on what they can control: the use they make of each "present moment" and their child's holistic development "as a person". We end this section with the words of Grace Martin, whose description captures well this shift in an unfolding story about the disease, one that previously

involved “having hope for a future cure” but she now describes as “having faith in the present”.

Her comments stand well on their own:

Grace: You can't let it [curative genetic research] get to you, because it just might not ever evolve, and I think the longer you're on this journey, the more you let go of that kind of thing... There's different kinds of hope, and hope for a cure is just one. Then, you start focusing on other things cause you get tired of the wait.

CJC: What are the other kinds of hope?

Grace: Hope that your child can finish high school. You know, you start setting up other goals. Or, hope that he can see various countries, trying to, not be a “Mad Hatter” and trying to pack as much in, because that's just fear-based as well, but just ensuring that you try to make him proud of his accomplishments. For me, I think with Aidan is to try to get through high school. I'd love for him to go to college, [but] right now we're focusing on high school, getting him to grade 12, making sure he's well attended to, and is confident and has a good support network around him. And he wants to go to college, but right now, we have to be realistic, we don't know how long he'll be around, so you just have other little milestones that you lay out ahead of you, instead of the big milestone of the cure. It's too big, it's way too much... I think now it's more about faith than hope. Hope is hanging on to something. Faith is having faith that, no matter what happens, it will be okay... hope was hoping my kid will live to 21. Faith is, if he lives to 12, that will be okay because that's enough.

## **PRESENT HEALTH CARE AND THE POSSIBILITY OF FUTURE THERAPIES**

Parents' narratives of “the cutting edge” of curative biomedicine are rich and diverse, steeped in the emotional anguish of a fatal disease, and infused with ambiguity, tension, and contradiction. It should come as no surprise then, that (somewhat paradoxically) several interviewees, after referring to their efforts to keep their hopes reasonable and balanced, and reporting a shift in perspective away from “having hope for a future cure” to “having faith in the present” (described in the previous section), went on to discuss ways in which the possibility of a new genetic therapy plays a key role in decisions around health and family planning. Though their narratives often focused on their efforts to “be realistic”, it appears that some parents are reluctant to completely discount the possibility that an effective new therapy may be imminent,

and some described going to considerable lengths “to keep this door open”. Laura and Thomas Brown, for example, decided to have extra children after their son Joseph was diagnosed with DMD, partly in order to obtain and bank (at significant cost) umbilical cord blood, a rich source of stem cells that could one day be useful if a new therapy becomes viable:

Thomas: So we save all the stem cells from our other kids...What else are you gonna do?

Laura: Yeah, exactly. Yeah, and then you know, there’s [the question about] “oh, how many kids do we have to get before [chuckle] [you’ve got all of the different possible genetic combinations (haplotypes)]”?

Thomas: So we were gonna have four and then we got, like, fifty percent covered one half times, right? [chuckle]

Laura: Yeah. [laughing] Yeah.

CJC: So that was factoring into your decision? About how many kids to have?

Thomas: Sure.

CJC: ...And you were thinking that your second and third children would provide what?

Laura: Cells.

Others reported taking more aggressive measures to maintain their child’s mobility than they otherwise would have, acting in the hope that slowing the progression of the disease may improve their child’s eligibility for any clinical trials that could be forthcoming. For example, two couples discussed how potential future therapies were a factor in deciding on appropriate activity levels for their child:

Lisa Jones, mother of 5-year-old Nathan:

Lisa: Stem cell is definitely it. And I think they’re gonna find the cure some day I guess...And the sooner the better they say, as long as we can keep Nathan walking. The better off he is, ‘cause they stop the disease course, [but] they can’t reverse it. And, we keep him completely active. Like he’s gotta be probably more active than some normal boys out there. He does everything you could possibly want to do.

Laura and Thomas Brown, parents of 9-year-old Joseph:

Laura: Midor [an experimental drug]...they're saying that perhaps they have some, [pause] good medication on the rise and, basically ... Like if you're in a chair that's it, but if you're not there, it might be able to stop you from going there. And it was really promising what they were talking about. And uh that's my newest hope actually. Even -

Thomas: So they altered his, his latest field trip around the lake, 'cause he goes around the lake for...two and a half hours, and he over exerts his muscles. That's gonna be causing damage, right? So, so now we're more careful 'cause we're more hopeful from that latest thing...

Laura: Yeah, I think the longer we can preserve what we have, the, more of a chance there'll be something around when it happens, right...But I think that this may come before the stem cell.

One mother described how one of the main reasons for visiting the regional specialist clinic is to "be on the list" for, clinical trials of new therapies that may come along:

As I say...that's the only reason I really go to clinic, unless there was a problem with my child, going to clinic basically, is, our name is on record somewhere so that...in case there was a trial...Just to be in their books, on the records. Really, clinic does nothing for our family, unless there's a problem...Sometimes we go down there, and two or three of them [clinicians] aren't there or they have stand-in people, and that's all right, because they do the documentation. We know that we're in the books - they've got his weight, the height, so if they're looking for somebody [for a clinical trial], they can get that information and if we fit the bill then that's great. But, really, they all ask the same questions. [Our son] gets so tired of answering the same questions. They measure how much flexibility, it's on the records, so he has to go through it, but it doesn't really do anything for us. We see how he changes, and we adapt, and we get the school to adapt, and the OT [occupational therapist] to help us out, so, what they're telling us doesn't really help us at all. We never get the answer to the question, "how long have we got?" or, "look at our child, where is he compared to everybody else?" They can't tell, but we're on the records. They're watching, and if they need somebody to fix what's happening to him, well then that's great that we've gone, and we go every six months.

These narratives display parents' efforts to keep open the possibility that new therapies for DMD may soon become a reality. They show how parents have not yet drafted the ending to an unfolding story about their child's illness, and that the potentiality of genetic research is, for many, implicated in the process of maintaining a subjunctive stance toward DMD. Mattingly (1994), Good (1990) and others (e.g. Good and Good 1994) have developed the concept of

subjunctivity first introduced by Bruner, to show the way in which narratives about illness operate to maintain a focus on “human possibilities rather than settled certainties” (Bruner 1986: 26), or in other words, on the potential for a future in which multiple possibilities or endings exist. It appears that genetic research plays this role in parents’ narratives as well, functioning as a means by which parents construct themselves within a subjunctive and hopeful world, “one in which healing is an open possibility, even if miracles are necessary” (Good and Good 1994: 839).

## CONCLUSION

This thesis explored some of the ways that parents narrate the significance of gene therapy in their everyday lives. My focus was on how the meaning of gene therapy changes for parents as part of the broader process of reconstructing a personal biography that incorporates DMD. After describing briefly some of the clinical features of the disease and the types of genetic research seeking to cure it, I elaborated the theoretical frameworks of narrative and situated learning that informed my analysis. I suggested that we can discern from comparative analysis of parents’ narratives a basic narrative schema – a trajectory or plot – along which they proceed in coming to understand and establish expectations for genetic research.

In the beginning, parents struggle to make sense of genetic research and see it as a hopeful prospect. Through social engagement with the DMD community, and in a process that can be likened to Lave and Wenger’s concept of legitimate peripheral participation, parents learn about gene therapy, how to manage their exposure to the discourse of genetics, and to keep realistic their expectations for it. Later, they must reconcile the timeline for research with the timeline and progression of their child’s disease.

Here, a transition can be seen as parents begin to question whether gene therapy will “come in time”. While it may be facile to label this as a “loss of hope” for the technology, this hypothesis does not account for parents’ remaining extremely *hopeful* for genetic research, even as their child’s disease progresses in its absence. Rather, and more complexly, I showed how as parents begin to live successfully with their child’s illness, there is a shift in how they perceive gene therapy that is expressed in how they emplot and articulate narratives of hopefulness and healing from within the context of their everyday lives. As questions about time become salient, and as their confidence in living with the disease begins to crystallize, parents described looking elsewhere for hopefulness – to their day-to-day lives and to their use of “present moments”. For many of the parents in this study, the lack of a successful gene therapy has over time come to function essentially as a foil, against which they enunciate and affirm their ability to live and cope with their child’s illness. Genetic research can thus be usefully viewed as a narrative device that is used for different ends, and that takes on different meanings depending on its place in the story parents tell about their child’s disease. Throughout this thesis, I showed how this process of constructing significance for gene therapy is infused with uncertainty, ambiguity and contradiction. This was indicated in particular by parents’ efforts to maintain a subjunctive stance (Good and Good 1994) on the possibility for cure by “holding the open door” to clinical trials that may be forthcoming.

Further research might approach gene therapy, and parents’ expectations for it, from the perspective of narrative. In particular, there is a need to understand how genetic science is situated within a broader cultural discourse on hope that suffuses biomedical practice and research (Delvecchio-Good 1991). For example, additional lines of inquiry might explore how hopefulness for gene therapy is emplotted and influenced by clinicians, who likely manage disclosure of information about genetic cures according to culturally proscribed conventions of

clinical discourse and conduct (Delvecchio-Good 1991). Clinical conversations are thus likely to draw from myths or models of hope and genetics in interesting ways. In addition, there is potential for further exploration of how genetic research functions to maintain coherence and/or disrupt the stability of narratives told by chronically ill families. While there is a general consensus in the literature that the experience of chronic illness leads to refined, coherent, and progressively more stable narratives (e.g. Kirmayer 2000; Kleinman 1988), we might explore whether gene therapy, and the anxiety and uncertainty it provokes, in fact disrupts the attainment of stability and coherence in the personal biographies of the chronically ill – a hypothesis for which there is preliminary support in this body of data.

### **Limitations**

Like all research, the process of recruiting and selecting participants has a bearing on its conclusions. The likely accrual bias in this study is that the interviewees may be drawn from among those who believe that they have handled their child's illness successfully, while others who feel they are coping less effectively may be less willing to participate. This study did not reach families who do not access services at the clinic and patient support organization through which subjects were recruited (though the vast majority of families with DMD in British Columbia are believed to do so). In addition, there were challenges in overcoming linguistic and cultural barriers to recruitment. All but one of the participants were of European descent and spoke English as their first language. It is obviously possible that families with other ethnic backgrounds would perceive genetic research differently. The socioeconomic status and educational backgrounds of participants may not be representative of parents with children with DMD, but is likely to reflect the bias towards more educated participants found in other studies. If this is the case, however, one might expect greater interest in medical research among this cohort than among a representative sample. Further research should seek to incorporate more

diversity by employing a different method of reaching participants, or by conducting inquiry within different cultural milieux. Though the participants in this study were evenly distributed by gender (eight women, six men), future research might also explore differences between fathers' and mothers' perspectives on genetic therapies, and the cultural shaping of these perceptions in accordance with gender roles and divisions of household labour.

Qualitative research, dealing as it does with the messiness of human social interaction, necessarily raises questions about what participants choose to disclose and to withhold from an interviewer. The emplotment and communication of illness narratives is necessarily a dialogical act that implicates its audience (Linde 1993), and as a researcher I too, am engaged in the process of creating a story with an interviewee. The interview protocol used in this study also contained a section related to parents' perceptions of continuity in the clinical care their child receives (Miller, et al. in preparation), and so it is possible that parents had expectations about the kinds of themes that were important to include (or neglect) in the stories they told me. Though in all interviews I sought to clarify my background and role as an anthropologist without clinical credentials, it is likely that some interviewees viewed me in some sense as connected with their child's clinical care and thus avoided certain topics.

Finally, as a "positioned subject" (Rosaldo 1996 [1989]) I am an apprentice in a discipline that has particular expectations about what constitutes information "worth knowing". The act of ethnographic representation is itself a mode of storytelling that is influenced by my social position as a (male) anthropologist, my surrounding cultural world, and my status as a graduate student (Saris 1995). Though I hope to have provided a coherent and justifiable analysis of the complex and interwoven webs of stories that parents related to me, in some sense this thesis is a narrative itself – necessarily my own "story of stories".



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