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The lived experience of parents with children diagnosed with mucopolysaccharidosis waiting for enzyme replacement therapy

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THE LIVED EXPERIENCE OF PARENTS WITH CHILDREN DIAGNOSED WITH MUCOPOLYSACCHARIDOSIS WAITING FOR ENZYME REPLACEMENT THERAPY

by

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The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis waiting for Enzyme Replacement Therapy

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Abstract

Mucopolysaccharidosis (MPS) is a rare and life threatening disease, and is one form of a family of metabolic genetic diseases. MPS is classified as an orphan disease. There is no cure for MPS; however, supportive treatment through Enzyme Replacement Therapy (ERT) is available but is an orphan drug. Canada is the only member of the Organization for Economic Co-operation and Development (OECD) that does not have an Orphan Drug Policy. The purpose of this phenomenological descriptive study was to explore the lived experience of parents with children diagnosed with MPS and who had received ERT but were now waiting for ERT funding by the provinces/territories. Nine key informant interviews, using a semi-structured interview guide, were conducted and the following four key themes emerged: *A New Journey; A New Reality; A New Optimism* and *A Road to Nowhere*. This description is unique to parents in Canada as it describes their experiences of having to live out the policies set by the Federal and provincial/territorial governments.

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Dedication

This thesis is dedicated to the memory of my father, Gaetano Pasquale Maione, who taught me never to give up.

I miss you daddy. I love you with all my heart, Maria.

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CHAPTER 1: INTRODUCTION

Disease Background

Mucopolysaccharidosis (MPS) is a rare and life threatening disease, and is one form of a family of metabolic genetic diseases. There are seven types of MPS, each one representing a deficit of a particular enzyme. Over time, MPS affected individuals experience deterioration leading to a loss of function in one or several systems in the body resulting in mental and or physical disabilities or death (The Canadian Society for Mucopolysaccharide and Related Diseases Inc. [The Canadian MPS Society], n.d. [b]). There is no cure for MPS; however, supportive treatment through Enzyme Replacement Therapy (ERT) is available for three of the seven types of MPS (The Canadian MPS Society, n.d. [a]).

MPS is classified as an orphan disease. An *orphan disease* is a "disease, which has not been *adopted* by the pharmaceutical industry because it provides little financial incentive for the private sector to make and market new medications to treat or prevent it" (MedicineNet.com®). There is little financial incentive for the pharmaceutical industry because of the rarity of the disease. For example, the prevalence of an orphan disease is defined as less than 1 in 2,000 people (Canadian Organization for Rare Disorders (CORD), 2007[c]) making it difficult for pharmaceutical companies to recoup their initial financial investment. Drugs that are developed for MPs are classified as *orphan drugs* because the prevalence of MPS is difficult to estimate due to the shortage of human based studies and epidemiological data (Muenzer, 2004). One way to assist with the collection of this data is through a disease registry, because data on MPS would be collected to assess its' frequency and to document the natural disease process (Muenzer). At this time, Canada does not have an MPS disease registry.

Currently the empirical body of knowledge on MPS is focused on the physiological aspect of the disease such as clinical trials with ERT. These clinical trials have been ongoing for numerous years (Meunzer, 2004; Wraith, Clarke, Beck, Kolodny, Pastores, Muenzer et al., 2004), and ERT has shown significant improvement in MPS affected children such as being able to walk independently, improvement in stamina, decrease in oxygen use and an improvement in blood saturation values. However, to date there have not been any studies focusing on the psychological, emotional and social aspect of the child living with MPS and their family.

Statement of Study Purpose

The purpose of this qualitative phenomenological study was to explore and describe the lived experience of parents with children diagnosed with MPS waiting for federal drug approval and provincial/territorial drug funding for ERT. A study such as this will contribute to the body of knowledge about children living with chronic, life-threatening illnesses and will allow parents to tell their stories. When caring for MPS affected children and their parents, if health care professionals have a better understanding about the experiences encountered by parents waiting for ERT funding this understanding can guide the health care professional in developing interventions to effectively support these families. Exploring the lived experiences of parents as they wait for federal drug approval and provincial or territorial drug plan funding gave parents a voice in that they were able to share their unique experiences. Exploring these experiences could contribute to improvement in health practitioners and health care policy makers understanding of these parents' experiences.

CHAPTER 2: REVIEW OF LITERATURE AND POLICY CONTEXT

Introduction

Chronic illness is defined as a long-term or permanent disease that is either incurable or has residual features resulting in limitations and/or disruptions to daily living (physical, psychological and social functioning) requiring special assistance or adaptation (Coffey, 2006; Goble, 2004). MPS is one such chronic, albeit rare (the total population for orphan diseases account for less than 1 in 2,000 people), life threatening metabolic genetic disease that is caused by insufficient or deficient lysosomal enzymes (CORD), 2007). The loss of system function(s) results in cognitive and/or physical disabilities and, in severe or untreated cases, death (The Canadian MPS Society, n.d. [c]). There are seven types of MPS, each one representing a deficit of a particular lysosomal enzyme. There is no cure for MPS; however, treatment with ERT that involves ongoing/weekly therapeutic infusions is available for three of the seven types of MPS (The Canadian MPS Society, n.d. [d]).

The purpose of this literature review is to summarize the current literature and gaps about parents caring for children with a chronic illness or diagnosed with MPS and to describe the policy context regarding ERT in Canada.

The literature review was conducted using the *Cumulative Index to Nursing & Allied Health Literature* (CINAHL) and *MEDLINE* databases via *Ovid*. Key words used to facilitate the literature search included essences such as *waiting, parent coping, chronic illness, stress* and *special needs children*. The keywords and references sited on the retrieved literature served to further enhance search options and data retrieval.

There have been numerous studies of parents with chronically ill children. However, the number of research studies on the topic of MPS is few and are focused on drug trials.

There have been studies exploring the needs of children with chronic conditions but they are generic in nature, dealing with such chronic conditions as developmental disabilities, cancer and diabetes. Biomedical and technological advances are enabling chronically ill children to live longer and more independently, thereby, shifting the activities of daily living from health care institutions and health care professionals to parents (Baum, 2004; Brett, 2004; Hovey, 2003). The literature showed that the burden of care for the chronically ill child rests with their parents or designated guardians. When a parent cares for a chronically ill child at home, the parent is not only responsible for physical care, but they must become proficient in clinical assessment, clinical decision-making and coordination of their child's multidisciplinary care (Ray & Ritchie, 1993). Finding ways of managing problems/crises with the multiple demands of caring for their chronically ill child is difficult and pervades all aspects of family life (Ray & Ritchie). For many parents, these tasks are overwhelming and often beyond their financial, intellectual and psychological resources.

The literature search uncovered a substantial number of studies using both qualitative and quantitative designs. The majority of the studies to date have been situated within stress and coping models and therefore parents' experience of caring for children with chronic illness have been described from this particular perspective. The theoretical lenses of Lazarus and Folkman (1984), Folkman and Lazarus (1988a) and Folkman, Schaefer & Lazarus, (1979) have been used to elucidate a context in which to guide and interpret research findings including those of: Azar & Solomon (2001); Calderon & Greenberg (1999); Heaman (1995); and Ray & Ritchie (1993). The populations studied to date include children with diabetes mellitus (Azar & Solomon, 2001), hearing loss (Calderon & Greenberg, 1999), developmental disabilities (Heaman, 1995), and complex care requirements including tube

feedings, tracheostomy care, oxygen administration, peritoneal dialysis, apnea monitoring and central lines (Ray & Ritchie, 1993). Therefore the major findings from these studies centred on the parents' abilities or inabilities to *cope* with caring for their children.

Using the lens from Lazarus and Folkman (1984), authors have developed a conceptual definition of coping, which suggests coping is a behavioural and cognitive strategy that the individual brings to a stressful experience (Azar & Solomon, 2001). Folkman and Lazarus' (1988a) theory of stress and coping hypothesizes that stress is a developing reciprocal transaction in that the stressor is neither a reaction nor a response, but an assessment on the level of threat the individual perceives the situation has on their well-being (Azur & Solomon, 2001). In other words, the parent assesses their child's chronic illness and perceives the outcomes as either challenging or threatening or both (Azar & Solomon, 2001; Ray & Ritchie, 1993) and therefore is judged regarding their ability to cope with their child's condition. Since the researchers base their studies from the stress and coping models it results in the focus of their studies being at the individual level (i.e. coping ability of parent) and do not take the broader perspectives into account such as social, political or health system factors.

A study by Hovey (2005) used the Roy Adaptation Model (Roy & Andrews, 1999) to validate the conceptual framework for categorizing father's roles when parenting chronically ill children. The participants in this study included children with cancer, cystic fibrosis or juvenile rheumatoid arthritis (Hovey, 2005). The study confirmed Roy and Roberts' (1981) initial definition of adaptation, which stated that adaptation is the pattern of behaviour an individual exhibits during times of expected and unexpected change within their environment. These patterns of behaviour occur over time promoting integrity and are

exhibited through survival, growth, reproduction and competence. Again, this study looked at micro-level factors and because of the model used in the study did not examine broader system level factors that may influence individual's patterns of behaviours.

Although the studies examined diverse populations of children with chronic illnesses, there were common findings that emerged from the literature. The literature will be described below based on these common themes, which include: waiting; bureaucratic navigating; and parental gender differences.

There were a limited number of studies found on the theme of waiting. Only two studies had a paediatric focus as both described parents' experiences waiting for their children's surgeries (Miller, 2004; Stubblefield & Murray, 2002). Six studies looked at various adult patient populations such as those waiting for surgery (i.e., generic, transplant and cardiovascular), waiting for treatment (i.e., visiting the doctor and chemotherapy) and waiting for information on a loved one in an intensive care unit (Bournes & Mitchell, 2002; Brown, Sorrell, McClaren & Creswell, 2006; Krumwiede et al., 2004; Mitchell et al., 2005; Screeche-Powell & Owen, 2003; Sjoling, Agren, Olofsson, Hellzen & Asplund, 2004).

The first paediatric study by Stubblefield and Murray (2002) was a phenomenological study. The study looked at the phenomenon of parents waiting for their children to undergo lung transplantation and the experience of relocating to be closer to the transplant centre. This description of experiencing relocation was part of a larger study on "Waiting for Transplantation." Stubblefield and Murray interviewed six parents leading to one descriptive category (experiencing relocation) and six related themes (putting their lives on hold, experiencing diminished emotional support, establishing new sources of support, undergoing role change, worrying about money, and making the best of the situation). When parents in

this study described the phenomenon of undergoing relocation, they identified a key role for nurses. The authors note it is important for nurses to understand the family's perspective of relocation thereby aiding nurses to develop family-based plans of care to meet specific needs (Stubblefield & Murray).

The second paediatric study (Miller, 2004) used a cross – sectional method in which a questionnaire was mailed to parents to determine their concerns and attitudes about waiting for operative procedures and how it affected their children and families. The author stated that this was the first study of its kind on waiting. Attaining a response rate of 64% (57 surveys returned out of 89), 47% of the respondents felt that their child's physical health was deteriorating while waiting; 63% felt waiting was emotionally stressful for the child and 95% felt it to be emotionally stressful for the family; 37% of the respondents reported that their child's condition prevented them from participating in their usual activities; and 84% anticipated an improvement in the child's quality of life after the operation (Miller). In summary, Miller (2004) reported that families experienced stress and concern for their child's wellbeing waiting for operative procedures, however, the study did not provide descriptions of the families' experiences. Implications included the importance of developing paediatric sensitive wait time criteria to meet the issue of wait times.

The six adult studies were all qualitative in nature using semi-structured interviews to collect data (Bournes & Mitchell, 2002; Brown, et al., 2006; Krumwiede, et al., 2004; Mitchell, et al., 2005; Screeche-Powell & Owen, 2003; Sjoling, et al., 2004). Five of the studies were generic in nature as participants were patients waiting for either surgery or treatment that is well established (Bournes & Mitchell; Brown, et al.; Mitchell, et al.; Screeche-Powell & Owen; Sjoling, et al.).

Krumwiede et al. (2004) used a grounded theory methodology to learn how rural families understand and manage the neutropenic experience caused by chemotherapy and the family's waiting for chemotherapy to resume. The seven families that participated had family members with a variety of cancers who ranged in age from 30 to 76 years of age.. A key finding in this study was that health care providers who give consistent support and information were valued and trusted by these families. Therefore, nurses who are sensitive to the family's needs may enhance symptom management in cancer care for these families (Krumwiede et al.). Although this study involved adult patients, and not children it did explore the concept of waiting from the families' perspectives. Krumwiede et al. interviewed family members waiting for chemotherapy to resume while I interviewed parents waiting for their children to commence ERT.

Miller, Recsky and Armstrong (2004) are Canadian authors and offer a commentary on the needs of children with chronic conditions in an era of health care reform. The first point the authors make is that the presence of a chronic health condition during growth and development affects not only the child's medical status but also social and emotional development. Secondly, the chronic illness has significant implications for the entire family and thirdly that the epidemiology of children's diseases differ from adult onset chronic illnesses because of the period of growth and development. The article offers specific action for advocacy groups to capitalize on in order to bring their concerns to the forefront and into the public consciousness (Miller, Recsky and Armstrong).

Three studies (Ray, 2005; Ray & Ritchie, 1993; Freedman & Capobianco-Boyer, 2000) reported on children living with chronic illness and the supports needed for these children and their families. Ray, a Canadian investigator, reported on the challenges parents

of complex care needs children face (ventilatory support, motor disabilities, cognitive delays and enteral feedings) in locating and coordinating community-based resources and services for their children. One of the main findings identified in Ray's study was the fact that parents spend an inordinate amount of time navigating bureaucracy instead of caring for their children. In an earlier study by Ray & Ritchie (1993), the authors identify another issue, that of the parents becoming responsible for physical care-giving and thus they must become proficient in physical assessment, clinical decision making and coordinating care. Therefore, parents find that they are being challenged to find new ways of coping with not only having to care for their children's physical needs but also to advocate for their children's medical and social needs (Ray & Richie, 1993).

From an American perspective, Freedman and Capobianco-Boyer's (2000) qualitative study identified similar findings to Ray's (2005) study. Once again, one of the main themes identified was the complex service system parents need to navigate in order to obtain support for their children.

Numerous studies have explored, analyzed and compared the affects of chronic illness on mothers and fathers (Knafl & Zoeller, 2000; Goble, 2004; Heaman, 1995; Hovey, 2005). Knafl & Zoeller's participants included children with diabetes, asthma, and juvenile rheumatoid arthritis and they reported that mothers are prone to experience physical & psychological stress, guilt and sadness more frequently than fathers. Goble's sample included fathers with children diagnosed with cerebral palsy, autism and osteogenesis imperfecta. The foci of the fathers' experiences were: living with a limited social life, lack of intimacy between partners, the idea of being the primary caregiver to the healthy child and worrying about the future of the chronically ill/disabled child (Goble, 2004). Two of the studies

(Heaman; Knafl & Zoeller) compared the experiences of mothers and fathers. Knafl & Zoeller compared the perception of how the chronic illness impacted their family life and found that if there was a discrepancy in the couple's views, it was the mother that tended to have the more negative outlook. Heaman focused on the perceived stressors of parents and identified worrying about the child's future to be the greatest concern for both parents. Hovey found the coping strategies that were similar between fathers and mothers included problem solving and information gathering techniques such as: trying to figure out what to do; looking at options; getting information; and weighing choices.

There is a wide body of theoretical and empirical knowledge on parents caring for chronically ill children. However, the literature focuses on chronic illnesses such as diabetes, cystic fibrosis, cancer and developmental disabilities. These chronic illnesses do not require complex drug approval process for treatment, as MPS requires. No studies were found that focused on the psychological, emotional or social aspect of the MPS child and their family. Currently, the empirical and theoretical body of literature on MPS focuses solely on the physiological aspect of the disease such as clinical trials with ERT. While MPS families have much in common with families who also care for a chronically ill child, it is proposed that the MPS families face unique challenges not encountered by the others. Thus, this study will explore the lived experience of parents with children diagnosed with MPS and how the phenomenon of waiting for ERT funding affects them. Not only will this study add to nursing knowledge on the phenomenon of waiting but it will also identify the need for further study into and funding for to meet the health care needs of this specific population.

Policy Context

A literature review was conducted on health and drug policies that are in place at the federal and Ontario provincial government level. The purpose of this review was to explore how health and drug policies affect families with children diagnosed with orphan diseases. The first section will discuss how drugs are approved in Canada. The second section will explore what influences health policy issues using the framework of institutions, (with particular emphasis on the federal Special Access Programme and the Ontario Drug Benefit plans); ideas (Orphan Drug Policy and Catastrophic Drug Coverage); and interests (associations, advocacy groups).

How are drugs approved in Canada?

Approving drugs for treatment in Canada can be a very long and complicated process. The drug application must go through numerous federal/provincial/territorial (F/P/T) committees before the drug is available to the general population. The waiting for federal drug approval and subsequent provincial/territorial drug funding for patients that are on experimental drugs (clinical trials) can be a very distressing and emotional time.

The drug manufacturer must submit its new drug product to the *Health Products and Food Branch* (HPFB) at Health Canada, which evaluates the drug's safety and efficacy. This process can take up to 2 years (Ontario Ministry of Health and Long Term Care, [MOHLTC], n.d. [a]). Once Health Canada approves the product, a *Notice of Compliance* (NOC) and a *Drug Identification Number* (DIN) are issued. After the NOC has been granted the manufacturer must then apply to the Common Drug Review (CDR) to be considered for funding under the provincial governments' drug plans (MOHLTC [a]). The CDR is a process at the federal level and the CDR is responsible for reviewing and providing common

recommendations for new drugs "based on clinical and pharmacoeconomic reviews to participating federal, provincial and territorial (F/P/T) drug benefits in Canada" (MOHLTC [a]). The CDR is housed by the Canadian Agency for Drugs and Technologies in Health (CADTH) [(Canadian Coordinating Office for Health Technology Assessment (CCOHTA) prior to April 2006] and receives expert advice from the Canadian Expert Drug Advisory Committee (CEDAC). CEDAC then makes recommendations to the partnering jurisdictions (provinces/territories except for the province of Quebec) regarding coverage (MOHLTC [a]).

In Ontario, for new or brand name drugs, the manufacturer must then complete another application and submit it to the Ontario Drug Quality and Therapeutics Committee (DQTC) in order to be listed in the Ontario Drug Benefit (ODB) formulary/Comparative Drug Index (CDI) (MOHLTC, n.d. [a]). Factors, which may influence the DQTC, may include fiscal capacity, current formulary listings, the need for clinical criteria and the population served by the ODB programs (MOHLTC [a]). The DQTC then makes a recommendation to the MOHLTC and the drug is added to the ODB or is rejected for coverage (MOHLTC [a]). The ODB plans will be explored in detail in the following section under "Institutions."

What influences health policy issues?

Institutions, ideas and interests play a major part in health policy issues and decisions (Baranek, Deber & Williams, 2004). The *Institutions* segment discusses what government mechanisms are in place at the present time in Canada and the province of Ontario for patients with MPS diseases to access ERT. The *Ideas* section introduces two policy options; an orphan drug policy for Canada advocated by interest groups and a Catastrophic Drug Coverage plan put forth in *Standing Senate Committee on Social Affairs, Science and*

Technology (Kirby, 2002) and the *Commission on the Future of Health Care in Canada* (Romanow, 2002). The *Interests* segment introduces the major stakeholders in this health policy issue. These stakeholders include patient and family, advocacy groups and research and pharmaceutical industries.

Institutions

This section will examine the drug policies in force at the federal and the provincial (Ontario) government level and their effects on children with MPS.

In October 2005, the F/P/T health ministers met in Toronto and reviewed the issue of "expensive drugs for orphan diseases" (Health Canada, 2005). At this meeting the health ministers "…asked officials to take the necessary steps to proceed with time-limited-research programs, including clinical studies for patients meeting treatment guidelines…" (Health Canada). The health ministers agreed to this research on a risk-shared basis with the drug manufacturers (Health Canada). K. Wong from the provincial Health Minister's office (personal communication, December 6, 2005) stated that all ten provinces and the three territories, the federal government and the pharmaceutical industry representatives were in talks to come to some agreement over who "pays for what". She imparted that an announcement should and would be made in early 2006. As of April 2008 there has been no F/P/T agreement announced.

Federal Government.

At present the federal government has the *Special Access Programme – Drugs* (SAP). This program enables practitioners to request medications for their patients that are unavailable for sale in Canada. Accessibility is restricted to patients with life-threatening conditions on a compassionate or emergency basis when conventional therapies have failed, are unsuitable, or are unavailable (Health Canada, n.d.). Diseases treated under this program range from intractable depression, epilepsy, transplant rejection, haemophilia and other blood disorders, terminal cancer and Acquired Immune Deficiency Syndrome (AIDS). SAP can also be accessed in times of a communicable disease outbreak, providing access to nonmarketed drugs (Health Canada, n.d.).

The *Special Access Management System* (SAMS) is the database, which lists all the eligible drugs covered through SAP. This list changes continuously thus making it difficult to maintain a published database. The practitioner needs to complete a *Special Access Request* (SAR) *Form* and fax the form to SAP. Once a drug is approved through SAP the maximum quantity of prescribed drug authorized would be enough to last for a six-month duration (Health Canada, n.d.). This presents a dilemma, as the patient would need to reapply through SAP every six months.

Ontario Provincial Government.

In Ontario, the provincial government has the *Ontario Drug Benefit: Individual Clinical Review (ICR) Mechanism* to deal with non-listed drugs (MOHLTC, n.d. [b]). The ODB covers prescription drug products for the majority of illnesses. In the case of new drugs not covered by the ODB, an individual may request special coverage through ICR (MOHLTC, [b]). However, in order to request an ICR you must be eligible for drug coverage

under ODB. Ontario residents covered under the ODB include; people 65 years of age and older; residents of long-term care facilities and Homes for Special Care; people receiving Home Care professional services; social assistance recipients; and Trillium Drug Program recipients (MOHLTC [b]). According to the eligibility list, children with MPS do not fall into any of these categories.

The *Ontario Drug Benefit: Trillium Drug Program* (TDP) assists people who have high drug costs in proportion to their income (MOHLTC, n.d. [d]). Once an individual's application has been approved the TDP covers over 3,400 prescription drugs, over 400 limited-use drug products and some nutritional and diabetic testing products (MOHLTC, [d]). Individuals can apply to the TDP if their private insurance programs do not cover 100% of prescription costs; have a valid Ontario Health Insurance Plan (OHIP); or, are not eligible for the ODB program (MOHLTC, [d]). The TDP has a deductible based on income and family size. The program runs from August 1 to July 31 of the following year and the individual must pay for drugs up to the deductible level before the program starts to pay for drugs. However, even though an individual may be approved for the TDP, products that are non-ODB must be pre-approved by the ministry before costs can count towards the Trillium deductible (MOHLTC [d]). Therefore, even though a MPS family may be eligible for the TDP the family must still wait for the ERT drug to be approved by the ministry.

The *Ontario Drug Benefit: Special Drugs Program* (SDP) covers the full cost of particular outpatient drugs used in the treatment of specific conditions such as drugs for cystic fibrosis, thalassaemia, HIV and Gaucher's Disease (MOHLTC, n.d. [c]). The SDP covers the costs of drugs if you are a resident of Ontario with valid OHIP coverage; are a patient with one of the diseases specified under the SDP; meet the specified clinical criteria;

and/or are approved by a designated centre or physician for the specific drug (MOHLTC [c]). The Ministry designates special facilities (usually hospitals) to distribute these treatments and in some cases, special review committees have been established to ensure that clinical criteria have been met (MOHLTC [c]). If patients meet criteria for coverage under the SDP there are no costs to patients (MOHLTC [c]). At the present time ERT for MPS has not been classified as eligible under the SDP.

Ideas

This section discusses the two main ideas that would benefit orphan diseases in Canada, 1) an Orphan Drug Policy (ODP) and, 2) a Catastrophic Drug Coverage plan for Canada, put forth in the *Standing Senate Committee on Social Affairs, Science and Technology* (Kirby, 2002) and the *Commission on the Future of Health Care in Canada* (Romanow, 2002).

Orphan Drug Policy.

In 1997, the Drugs Directorate Renewal Project of Health Canada deliberated on a proposal for an ODP and concluded one was not needed (BIOTECanada, 2004). At the time, the panel concluded that Canadians had access to essential medicines used in treatment of orphan diseases through the existing drug programs. The report however, did mention the potential for financial hardship of patients without an ODP (BIOTECanada).

An ODP in Canada would be an incentive for drug development and would make it possible for small companies to assume drug development programs (BIOTECanada, 2004). Some features included in orphan drug policies from international jurisdictions include the following: 1) Market exclusivity of drug product ensuring a guaranteed revenue stream for biotechnology companies, thereby making companies attractive to investors; and 2) grants

for clinical trials. Governments also support clinical research by funding disease registries and patient networks; and 3) shorter review times of orphan drug products to prioritize these drugs within the regulatory framework. This review is usually completed through a separate review process (BIOTECanada, 2004).

Catastrophic Drug Coverage.

Kirby (2002) and Romanow (2002) both advocate a *Catastrophic Drug Coverage* plan. Romanow addresses two issues: 1) the importance of improving accessibility and ensuring there are no financial barriers to prescription drugs for Canadians; and 2) the continued quality, safety and cost-effectiveness of prescription drugs. Romanow outlines five steps that need to happen in order to address these two issues. These steps are: 1) a Catastrophic Drug Transfer: 2) a National Drug Agency; 3) a national formulary for prescription drugs; 4) a medication management program linked to primary health care; and 5) a patent review.

Kirby (2002) maintains that Canadians should not be denied prescriptions and treatment because of their inability to pay and that conditions for long-term sustainability of current prescription drug programs need to be created. This long-term sustainability plan would include existing provincial public and private supplementary insurance plans, therefore building on provincial insurance plans instead of replacing them. Kirby therefore, recommends a national Catastrophic Prescription Drug Insurance Plan and a National Drug Formulary.

Interests

Interests introduces the interest groups involved in emphasizing and converging public opinion on the issue of orphan drugs and an orphan drug policy for Canada.

Interest groups.

The Canadian MPS Society and CORD are two interest groups that are lobbying the federal and provincial governments for the implementation of an ODP. The mission of The Canadian MPS Society, which was founded in 1984, provides information and support to affected individuals and their families, and is active in helping to directly fund research into MPS related diseases (The Canadian MPS Society, n.d. [a]).

The mission statement on CORD's website states, "Through an educational and informational support network, CORD is committed to the enhancement of the lives of all persons affected by rare (orphan) disorders" (CORD, n.d. [a]). CORD is Canada's national voice for the more than 6,000 orphan diseases identified worldwide and links individuals and/or families together with the same orphan disease (CORD, [a]).

Biotechnical Associations and Pharmaceutical Industries.

BIOTECanada is an association representing the research and pharmaceutical industry in Canada. The BIOTECanada (n.d.) website states, "BIOTECanada is the national industry-funded association representing the broad spectrum of biotech constituents including emerging, established and related service companies in the health, agricultural, and industrial sectors." BIOTECanada supports an ODP for Canada and argues that an ODP in Canada would be an incentive for drug development and would make it possible for small companies to assume drug development programs (BIOTECanada, 2004) thereby making Canada much more competitive in the pharmaceutical industry worldwide (BIOTECanada). Some recommendations put forth by BIOTECanada include the following: 1) federal funding bodies to put aside monies to support orphan disease research. Monies would be used for research and development (R&D) and establishing patient registries; 2) federal tax credits

competitive with that of the US (50% on R&D costs); 3) guaranteed shorter review times to prioritize orphan products; 4) federal funding dedicated to orphan products as part of the proposed catastrophic drug funding; and 5) awarding 10-year market exclusivity for orphan products on par with the US and the EU. This incentive will ensure revenue streams for companies undertaking this commitment to orphan diseases (BIOTECanada).

The pharmaceutical industry has invested time and money into orphan diseases and orphan drug medications. One pharmaceutical company conducting clinical trials for MPS II is *Shire Pharmaceuticals Group plc* in the UK, while *Genzyme Corporation* is an American pharmaceutical company, which has developed ERT drugs for MPS I, Fabry and Gaucher diseases.

There have been and continue to be clinical trials for ERT in Canada, the United States (US), the European Union (EU), and Australia among others (CORD, n.d. [b]). Two enzyme replacement therapy drugs identified and trialed are Aldurazyme® (Laronidase – Type I MPS) and Elaprase™ (Idursulfase – Type II MPS). During clinical trials, significant improvement in physical condition has been documented in individuals receiving this enzyme replacement provision. The biggest deterrent for ongoing therapy is the cost. On average, it costs $300,000 to $350,000 per year to treat one individual with MPS (Wansbrough, 2007). Health Canada approved Aldurazyme® in 2004 and Elaprase™ in 2007. However, in July 2005 (Aldurazyme®) and in December 2007 (Elaprase™), the Common Drug Review (CDR) recommended the provinces not list these enzyme replacement drugs in their formularies, meaning the provinces do not need to fund these drugs (CADTH, 2007; CCOHTA, 2005).

The three main reasons for the negative recommendation for Aldurazyme® included, 1) the clinical significance of a 5.6% mean improvement in forced vital capacity and a non-statistically significant median change in the six-minute walk test were not demonstrated and no differences were noted in quality of life (measured by the Childhood Health Assessment Questionnaire [children] or the Health Assessment Questionnaire [adults]); 2) the majority of the patients develop antibodies against Aldurazyme®. The significance of these antibodies for benefit, harm and dose requirements is unknown, and 3) the cost of the medication. The average annual cost for a patient weighing 40 kilograms would be approximately $435,000 per year (CCOHTA, 2005). Taking these three reasons into consideration, plus the fact that the medication does not cross the blood brain barrier and is not cost effective, the recommendation to the provinces/territories was not to list the medication on their formularies (CCOHTA).

The three main reasons for the negative recommendation of Elaprase™ included, 1) the clinical significance of the drug's effects is not established as evidenced by an average improvement of less than 10% above baselines in distance walked during the six minute walk and did not "improve clinically relevant outcomes such as quality of life, pain, rates of hospitalization and resources required for home care support" (CADTH, 2007, p.1); 2) the medication unlikely crosses into the central nervous system, therefore, not improving neurological complications, and 3) the cost of the medication. A 6 mg vial of Elaprase™ costs $4,215. Therefore, the cost for treatment of a child weighing 35 kg is $657,000 per year (CADTH).

Dr. J. T. R. Clarke, a geneticist with a focus on the MPS population at The Hospital for Sick Children (Sick Kids) in Toronto, argues that Canada's process for reviewing drugs is

rigorous and duplicative (Clarke, 2006). The CDR came into being in 2003 and Dr. Clarke contends that the CDR is duplicating what was a provincial initiative. Dr. Clarke states that the CDR process was put in place to make sure that the public "...is getting its money's worth from its financial support for patients." According to Dr. Clarke the problem with the CDR process is that because the orphan disease population is so small, achieving a sufficient statistical power to prove an orphan drug's efficacy is almost impossible.

In conclusion, not much is written on the implications of the CDR process on families whose children are diagnosed with an orphan disease and are faced with policies that do not address the unique needs of orphan diseases. It is important to capture the broader context in which these families are living out the policies the federal and provincial/territorial governments have implemented. Studies have mainly focused on coping at the individual level and other than two studies that talked about parents having to organize their children's care (Ray, 2005) and the experience of parents waiting for surgery for their children (Miller, 2004) the studies reviewed did not utilize a framework (lens) that enabled them to elucidate the broader social and political factors impacting on the families coping.

The section on policy context described what health policies are in place currently and how they affect the families with children who are diagnosed with orphan diseases. Governments institute health policies with limited knowledge of how these policies will affect individuals once the policies are implemented. Exploring the lived experiences of parents as they wait for federal drug approval and provincial/territorial drug funding through the provincial drug plans will bring to the fore parents' experience.

CHAPTER 3: METHODOLOGICAL APPROACH

Introduction

This is a descriptive phenomenology study with Husserlian philosophical underpinnings as an approach. Caring for a patient's mind, body and spirit, is what makes nursing a holistic practice therefore, if caring for just a part of a patient is at odds with nursing practice, the study of humans should also be a holistic endeavour (Speziale & Carpenter, 2007). Originating in philosophy, phenomenology offers the opportunity to explore the lived experience of individuals and to describe their perceptual awareness (Ng & White, 2005). Phenomenology is also an appropriate method when there is limited or no research in an area of study (Donalek, 2004). As stated in Chapter 2, there are no published studies on MPS affected children and families experiencing living with MPS affected children and waiting for funding approval of ERT. A descriptive phenomenological study is also suited to exploring ill-defined concepts since descriptive phenomenological research focuses exploration on everyday occurrences and draws the essence from the phenomenon for others to experience (Brett, 2004).

I chose descriptive phenomenology as the framework for this study because having cared for children with MPS as a paediatric nurse in the past; I wanted to further explore the experiences of the parents. I attempted to describe a narrative of the collective experience of the participants in regard to waiting for federal drug approval and provincial/territorial drug funding approval for ERT, in comparison to the hermeneutic approach in which I would have searched for differences and uniqueness in each of the parents' contextualized lived experiences (Wojnar & Swanson, 2007).

For the purpose of this study, the term *parent*, *mother* or *father* will be used to represent parents.

Research Design

Husserlian Approach (German Phase)

Edmund Husserl is considered to be the founder of phenomenology as a philosophy and the descriptive (eidetic) approach to enquiry whereby he believed that consciousness was the condition of all human experience (Jasper, 1994; Wojnar & Swanson, 2007). Husserl focused on the meaning of lived experience, from a first person point of view and the belief that the meaning of the lived experience may be unravelled through one to one transaction between the investigator and the participant (Priest, 2002; Wojnar & Swanson). Husserl also suggested that you could not separate the phenomenon from the experience and the only way to understand the phenomenon was through description of the experience by the participant living through the phenomenon (Dinkel, 2005; Jasper, 1994). Husserl believed that since humans live their everyday lives without daily reflection, a scientific approach was needed to explore and describe essential components of a group of people's lived experience (Lopez & Willis, 2004). Husserl outlined three key components in phenomenological research: phenomenological reduction; intuiting; and a search for universal essences (Dinkel; Lopez & Willis). Essences are the basic elements of a common understanding of any phenomenon (Speziale & Carpenter, 2007).

Phenomenological Reduction.

Phenomenological reduction is a method used to describe phenomena as free as possible from conceptual presuppositions and cultural context to help make research findings more precise (Dowling, 2005; Giorgi, 1997). Phenomenological reduction commences with

the suspension of beliefs, assumptions and biases about the phenomenon under investigation, although this reduction may never be fully achieved because of the relationship individuals have with the world and their preconceived notions in everyday life (Lopez & Willis, 2004). In other words, we take things for granted in everyday life and we do not challenge reality unless something unusual happens. With phenomenology we are pushed to systematically and critically understand how a phenomenon has come into being and attempt to link things and events to causes and/or conditions (Giorgi).

Bracketing is a second component that occurs during phenomenological reduction (Giorgi, 1997). The investigator must identify preconceived ideas or notions about the phenomenon, and must bracket or isolate them so as to not influence the object of the study (Lopez & Willis, 2004). Giorgi writes, "…this does not mean that one empties oneself of all possible past knowledge", but he goes on to say that the investigator must put aside past knowledge so the phenomenon under study has a chance to present itself fully and a description can be written.

Intuiting.

The second step, intuiting is an accurate interpretation of what is meant in the description of the phenomenon under study. Intuiting requires that the investigator become immersed in the phenomenon under study and during this process the investigator begins to know about the phenomenon as the participants describe it (Pallikkathayil & Morgan, 1991). As the investigator becomes immersed in the data, there is a sense of what it might be like to "live in the participant's skin" and the investigator makes a concerted effort to understand what the participant lived through (Wojnar & Swanson, 2007). Balancing intuiting and bracketing is an all-consuming task for the researcher and a task that should respect insights

about the emerging evidence while simultaneously suppressing the investigator's premature assumptions (Wojnar & Swanson).

A search for Essences.

An essence gives common understanding to the phenomenon under investigation. Through phenomenological reduction the participant's lived experience is revealed, subsequently described and its essential structure uncovered (Priest, 2002; Salada & Adorno, 2002). Salada & Adorno write, "the core of phenomenology is the intentionality of consciousness, which is understood as the direction of consciousness towards understanding the world" (p. 283). Through consciousness, intentionality attributes meanings to objects (Salada & Adorno). As a result, without meanings it would be impossible to talk either about an object or an object's essence. As the researcher it is my task to describe and analyze the experiences of the parents and illuminate how they have given meaning to the phenomenon of "waiting for ERT drug funding," thus arriving at its' essence (Salada & Adorno).

Theoretical Underpinnings

Critical Social Theory guided this phenomenological study. Critical Social Theory is based on the assumption that reality is shaped by social, political, cultural, ethnic and gender based values (Speziale & Carpenter, 2007). Critical theory investigators believe that social reality is historically established and can be constructed and deconstructed by members of society. However, they contend that although people can change their social and economic situation, their ability to do so is ultimately delimited by social, cultural and political factors (Myers, 1997). Critical Social Theory, therefore, uses social critique as a means of exposing the oppressive and marginalizing nature of the status quo and, by focusing on the conflicts

and contradictions in society, attempts to eliminate the causes of alienation and domination (Myers, 1997).

Duchscher (2000) writes Critical Social Theorists believe that by relying solely on scientific objectivity as the only method of reconciling rational thought, invalidates human perceptions and experiences, therefore, Critical Social Theorists stress the importance of human subjectivity as important in knowledge generation. Critical Social Theory provides a voice for smaller often marginalized groups. As discussed in Chapter 1, MPS is an orphan disease with a small population of affected individuals. The literature review also revealed limitations as most of the studies examined the experiences of parents with children diagnosed with chronic illness more on a day-to-day basis than on a long-term basis. The key differences with MPS affected children compared to other children with life-threatening diseases is that these children are not "guaranteed" to get treatment so political, economic and social forces play a significant role in the child's treatments and outcomes. Duchscher explains, "Critical social theory epistemology maintains that knowledge as truth is socially constructed and that the facts are relevant only in the lived experiences of persons" (p. 454). Therefore, facilitating parents of MPS affected children to recount their experiences will help to elucidate their reality and will expose their experiences of marginalization. Thus, critical social theory was used to guide this study.

Research Question

The study was guided by the following research question: What is the lived experience of parent(s)/legal guardian(s) with children diagnosed with Mucopolysaccharidosis (MPS) waiting for federal drug approval and provincial/territorial drug funding for Enzyme Replacement Therapy (ERT) in Canada?

Data Collection Methods

Sampling/Selection of Participants

The Research Ethics Board (REB) at Ryerson University (REB #2006 – 127) approved the study. A copy of the Ryerson University REB approval letter was sent to the Canadian MPS Society. Approval to contact parents was obtained from the Canadian MPS Society board members.

The study used purposeful sampling. Purposeful sampling is the identification of a group of people who have or are experiencing the particular phenomena of study and are willing to share their experience (Speziale & Carpenter, 2007). This type of sampling was used because in phenomenological studies only participants who have experienced the phenomena under study can provide in-depth information or data that is relevant to the research question (Milne & Oberle, 2005).

Parents of children diagnosed with MPS and who were receiving ERT either through clinical trials, extension studies or compassionate treatment in Canada were identified by The Canadian MPS Society. I did not have access to the names of the parents identified by The Canadian MPS Society. The Canadian MPS Society mailed an introductory letter written by me out to parents on my behalf, asking if they would like to participate in the study. The introductory letter outlined the purpose and methodology of the study and full contact

information was supplied for the parents to contact me if they wanted to participate in the study (Please see Appendix A).

Inclusion Criteria.

1. Parents who cared for children diagnosed with MPS and were enrolled or had been enrolled in a clinical trial, an extension study, or a compassionate treatment protocol and had received ERT in Canada.

Exclusion Criteria.

1. Parents of children who had not participated in a clinical trial, an extension study or a compassionate treatment protocol for ERT.
2. Parents of children who had never received ERT.

Sample.

Nine interviews were conducted with parents from across Canada, all with children diagnosed with one of the seven types of MPS and receiving ERT. Within the parent dyad they selected which parent would participate in the study. There was only one instance in which both the mother and father of one child participated in the study but they were interviewed separately.

Consent Process

In the initial telephone conversation or e-mail I answered any preliminary questions and informed the parent that two copies of the consent would be mailed to them to read. The parent was then telephoned within two weeks and I then reviewed the consent with the parent, prepared them for the interview and answered any questions they had (Speziale & Carpenter, 2007). Parents were also informed that the interview would be audio taped and subsequently transcribed for analysis. The parents who agreed to participate then set up a

time and place for an interview. Both copies of the consent were mailed back to me if the interview was held via telephone and one copy of the consent was then returned to the parent with my signature. A signed informed consent was obtained at the start of the face-to-face interviews (please see Appendix B).

The interview was conducted on the telephone for parents living outside of the Greater Toronto Area (GTA). Telephone interviews were audio taped using a digital recorder (Olympus VN-3100PC) and a telephone-recording device (Olympus TP – 7). Parents from the GTA met at a mutually agreed setting such as my home or in an office at Ryerson University. The interview took place in a quiet place with visual and aural privacy where parents could discuss their experiences with ease and without the fear of being overheard.

Ethical Considerations

The consent form was reviewed and explained to each parent prior to the start of the interview. The consent form included the purpose of the study, how the data would be collected and analyzed, and potential benefits and risks for harm that may arise during the study. The parents were also informed that their participation was voluntary and that they could withdraw from the study at any time. They were informed that withdrawal from the study would not in any way jeopardize their child's ERT or nursing care. Parents were asked if they could be contacted again if further exploration of the phenomena was needed. Consent was also obtained from the parents for permission to use direct quotations from their narratives as this could potentially lead to a breach of confidentiality. One way to alleviate this breach of confidentiality was to assign a code to each participant (Speziale & Carpenter, 2007). In addition, when confidentiality is pledged to a research participant, it is understood

that any identifying information will not be accessible to anyone but those directly involved in the research and data analysis (Speziale & Carpenter).

When the consent was read and explained to the parent, confidentiality was addressed along with the strategies in place to maintain the confidentiality of parents. Examples of strategies to maintain confidentiality included the following (Speziale & Carpenter, 2007):

1. All audio recordings, transcribed narratives and findings were kept locked in a file cabinet in my home office.
2. All audio recordings, transcribed interviews and findings were password protected on my computer at my home office.
3. I omitted participants' names and a code was assigned to each parent when the description was written. Consent was also obtained from participants to use direct quotes from the transcripts in the final narrative of the study.
4. Names of parents and children were not used in any transcriptions of interviews.
5. My thesis committee members and I were the only people that had access to the raw data.
6. The audiotape recordings were only used to transcribe the parents' stories. Audiotapes will be stored for 2 years on my computer and will then be destroyed once the study has been completed and the thesis is defended.

Data Gathering

Semi-structured audio-recorded interviews were utilized for this study. This type of interview method consisted of open-ended questions based on the phenomena I wanted to explore. The advantage to utilizing open-ended questions was that they provided the opportunity for the parents and I to discuss particular topics in detail (Hancock, 2002).

Interview Guide

The interview started with a broad question asking parents what it was like to wait for federal drug approval and provincial/territorial drug funding for ERT (Please see Appendix C for Interview Guide). Interestingly, parents started telling their stories in chronological order often from the time the child was an infant and long before diagnosis. Therefore, the interviews took on a chronological structure with a history of the parents' experience. The next two questions explored the parents' experiences when they were told their child had MPS. These two questions were emotionally difficult for the parents to answer as they initially considered the diagnosis as the end to their child's future. These questions led to the fourth question of describing their experience of having their child included in a clinical trial and how they felt about the opportunity of participating in a trial knowing that ERT was an unknown entity. A follow-up question was asked regarding how ERT had impacted their life and that of their child. At the end of the first interview, I felt there needed to be closure for the parent, therefore a seventh question was added. I wanted to give the parents an opportunity to express their experience, opinion and disappointment by asking them to imagine they were speaking to their respective provincial/territorial Minister of Health.

Data Analysis

The Colaizzi (1978) method of data analysis was utilized for this study. Speziale and Carpenter (2007) summarize Colaizzi's (1978) method of analysis into a nine-step method. These steps include, 1) describe the phenomenon of interest; 2) collect participants' descriptions of the phenomenon; 3) reading and rereading the descriptions to gain an understanding of the participant's experience; 4) return the original transcripts to participants and extracting significant statements; 5) attempt to formulate meanings that discover and

illuminate meanings of each significant statement; 6) categorize into clusters of themes and validating with original text; 7) integrating the findings into an exhaustive description; 8) return the description to participants to validate the findings; and 9) incorporate new findings offered by the participants into the final description of the phenomenon.

As per Colaizzi's (1978) method found in Speziale & Carpenter (2007) the following occurred during data analysis. The phenomenon of parents' experience of waiting for ERT federal approval and provincial/territorial drug funding approval was identified by my interactions with the families while the children were enrolled in their respective clinical trials. The parents' descriptions of the phenomenon were then collected through individual interviews. Each parent was interviewed individually at a mutually agreed time and place. The interviews ranged from 35 to 65 minutes in length. I then transcribed the interviews verbatim. The transcribed interviews were returned to the parents and each parent had the opportunity to edit his/her transcribed interview. After all the interviews were returned to me and any requested changes were made, I read and reread the transcripts numerous times to become familiar with the contents of each transcript. With each subsequent reading of the transcripts, significant statements from the transcripts were extracted and placed into clusters (Dowling, 2005). The clusters were then analyzed and the formulization of meanings were uncovered and expressed as psychological and phenomenological essences (Dowling). The essences were validated with original text from all nine interviews. Once the essences were validated with text the findings were described and linked together into four themes to describe the phenomenon of the lived experience of parents with children diagnosed with MPS waiting for ERT approval and drug funding. After the description was written the findings were returned to the parents for validation and to compare to their own experience.

Only two parents acknowledged the opportunity to provide feedback. One parent did add a written passage (under the essence of guilt). The two parents agreed that the narrative was an authentic description of their experiences.

Rigour

There are a number of issues about rigour that must be taken into consideration when conducting phenomenological research. These issues include authenticity, i.e., the voice of the participant; credibility, i.e., reflecting how believable the research is; and integrity, displayed by reflexivity of the investigator (Milne & Oberle, 2005). In this study, there were several strategies that were utilized to attain and maintain rigour. These strategies are discussed below.

Authenticity

Transcription of audiotapes is an important factor that ensures authenticity in phenomenological research. I transcribed all the interviews. My thesis supervisor also listened to the tapes and read along with the transcript to check for accuracy. As reported in the *Data Analysis* section, after the description was written the findings were returned to the parents for validation/authenticity and to compare to their own experience. This process is referred to as "member checking" (Speziale & Carpenter, 2007).

Credibility

The credibility of qualitative studies stems in part from the experience of the investigator. When the *interview* is part of the process the investigator should have some experience in interview techniques (Tuckett, 2005). As a nurse, I have had significant experience conducting interviews. I conducted the first interview and then sought feedback

from my thesis supervisor regarding interviewing techniques. Once feedback was obtained from my thesis supervisor I continued with the rest of the interviews.

I used field notes to describe observations such as room setting, participant reaction and expressions such as body posture, facial expressions and eye contact as well as my own reactions to the discussion. Observations such as these served as corroboration for statements made by participants and assisted in facilitating the emergence of themes (Koch, 1994).

I had credibility as an investigator since I had cared for children with MPS throughout the last 5 years of my career as a paediatric nurse. Therefore I had some degree of understanding of the challenges these parents faced while their children were participating in their respective clinical trial. There was a sense of trust between the parents and myself partly because the parents knew I had an understanding of the experiences they had endured.

Integrity

Integrity refers to the reflexivity of the investigator with regard to potential sources of bias, as the investigator is also a participant (Milne & Oberle, 2005). During this study, I maintained integrity by actively reflecting on biases, assumptions, values and beliefs that working with these families for five years had instilled. An example of maintaining the integrity of the study was that I kept to the questions outlined in the Interview Guide and did not attempt to prompt the parents when they were relating their experiences. I wrote out my assumptions and biases prior to commencing the interviews and reflected on how I felt about these assumptions and biases with my thesis supervisor. Also, once the first interview was completed, and before the second interview, my thesis supervisor listened to the interview to monitor and critique my interviewing technique and ensure that I was not using leading questions or prompts.

The purpose of qualitative research is to provide a venue for the participant's voice; therefore, this step serves as a check that their story is indeed reflected. In this study, the transcripts were returned to the participants prior to data analysis to give them the opportunity to add or omit data from the transcripts. The transcripts were also returned to the parents at a later stage of data analysis to validate the description as an accurate description of their experience, as a form of "member check" (Milne & Oberle, 2005; Speziale & Carpenter, 2007).

Peer review is another method of assuring integrity in that colleagues ensure that the findings remain true to a qualitative description and the participant's meaning is presented in a clear picture (Milne & Oberle, 2005). My thesis committee served as the peer review for this study. The thesis supervisor received copies of each of the audio taped and transcribed interviews enabling her to check for accuracy of transcription and to become immersed in the parents' experiences. One-on-one meetings between my thesis supervisor and myself provided guidance with writing the descriptions. Periodic thesis committee meetings with the whole team were scheduled and during these meetings the committee members made sure that I adhered to the principles of phenomenology and the Colaizzi (1978) method of data analysis.

CHAPTER 4 – FINDINGS

Introduction

This chapter will describe the parent's experience of caring for their child with Mucopolysaccharidosis (MPS) waiting for federal drug approval and provincial/territorial drug funding of Enzyme Replacement Therapy (ERT).

Overall, thirteen essences emerged from the data. These thirteen essences were then categorized into four themes and included, 1) A New Journey, 2) A New Reality, 3) A New Optimism, and 4) A Road to Nowhere. Underpinning these themes is the passage of time in years. In the first theme "A New Journey" the parents describe striving to find a diagnosis while in the second theme "A New Reality" the parents are struggling to come to terms with the diagnosis of MPS. In the third theme "A New Hope" the majority of parents struggle with the decision of whether or not to place their children at risk with an unproven treatment (ERT), while others anxiously wait for the opportunity to start their child on ERT. Finally, in the fourth theme "A Road to Nowhere", the parents dispute the decision the provincial/territorial governments make regarding not funding ERT. Below is a conceptual framework of the themes and essences (Figure 1). A full-size, colour version of the conceptual framework is attached in the Appendices section (Please see Appendix D).

The sample included nine parents, two of which were a couple. Two of the parents interviewed had two children each that had been diagnosed with MPS. The nine parents interviewed represented 10 children with a diagnosis of MPS. Parents were given a code once the interviews were transcribed. These codes are found at the end of each participant quote sited in this chapter. For example, "P1" stands for "Parent 1." The ranking of the parent does not have any significance as the numbers were assigned randomly.

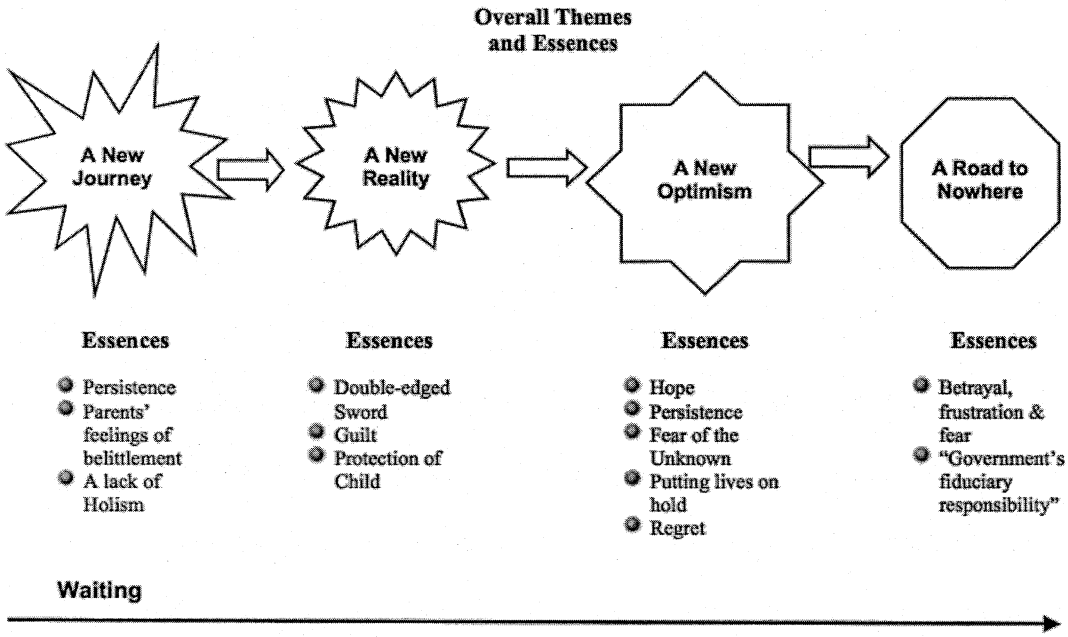


Figure 1. Overall Themes & Essences

Themes

A New Journey

The parents often began their stories talking about their entry into the health care system. The parents noticed that their child was "not right" and the journey began in trying to find a diagnosis for their child. The parents described the frustration of waiting for a diagnosis and through their stories the following essences emerged: persistence, parents' feelings of belittlement, and a lack of holism.

Persistence

Many parents were searching for a diagnosis as no one could definitely tell them what was wrong with their child. Parents were persistent and insisted on finding out what was

wrong with their child, taking any opportunity given to them to acquire a "second opinion." The following are passages from the parents' transcripts elucidating their experiences of trying to find out what was wrong with their children. The first parent describes how they were passed on from "doctor to doctor" trying to find a diagnosis for their child:

> "…when [X child] was four she started walking on her toes, so her doctor suspected that there was something wrong. We got sent to another doctor and from there we just got passed along from doctor to doctor." (P4)

In this second quotation, the parent described the hopelessness of going from specialist to specialist, but making no headway in finding a diagnosis for their child and the amount of time that was wasted:

> "So our family doctor did send us to a specialist and from that specialist we ended up at (a paediatric hospital) in neurology, I believe. We ended up wasting so much time because the neurologist asked to see [X child] the following year. So it ended up going a few years like that just doing nothing just investigating…" (P3)

The above two examples were powerful examples of what the parents experienced when searching for the elusive diagnosis. It was a new journey for the entire family going from doctor to doctor for second, third even fourth opinions. For some parents this search took up to four years.

Parents' feelings of belittlement

From the parent's perspective the period before diagnosis was one of upheaval and struggle, stress, tension, and fighting to be heard. At this point, the parents knew that their child was not healthy. They continued to take their child to the doctor and they described feeling "put off" by the doctors when they brought their child to the doctor. The symptoms

the children displayed were symptoms that any normal young child would exhibit. Examples of symptoms included; earaches, stumbling, and congestion. The parents were told that these symptoms are normal for the child at their particular point of development. One parent states,

> "But we kept being put off that it's an ear infection, it's this, it's that, right up until [X child] was diagnosed at about 18 months." (P1)

A parent recounted a similar situation:

> "There had been things leading up, but none of it had been out of the ordinary. Everything that had happened with [X child] was all very common childhood problems." (P2)

One parent's frustration is exemplified in this passage when describing their journey in finding a diagnosis. The doctors were reassuring her that her child was experiencing "normal" symptoms that all "normal" children of that age exhibited.

> "There were other things. Like a bellybutton hernia. But her doctor said "oh don't worry it's normal a lot of kids get this". Her hearing loss, her multiple hearing infections--don't worry it's normal. Everything that she had was normal for a child her age and I was told not to worry." (P4)

One parent was made to believe that their concerns were insignificant. These parents had been trying to secure a diagnosis for their older child and they (the parent and doctors) still did not know what was wrong with the older child. During one doctor's appointment, the parents brought up the fact to the doctor that their youngest child exhibited the same "claw like" hand contractures as their older child's, who had previously been diagnosed with MPS.

> "The thing that upset me the most, I have to say I was very upset when we brought Y (younger sibling) to [hospital A] just to sit in on X's (older sibling) appointment in

neurology. The doctor made light of [Y child]. Someone, another close person in our family noticed that [Y child's] hands were claw-like. And then we looked and we said "Oh my goodness, they are in fact, very similar to [X child's]." (They) were similar in so many ways. So this led us to, speak out at the appointment and say, "doctor, [Y child] seems to have the same hands as [X child]." What upset me was that he just made light of it and said that [Y child] is just imitating [X child]." (P3)

When the mother was recounting this episode I could sense she was very frustrated and distressed by the fact that the doctor had dismissed her concerns considering her older child had been diagnosed with MPS.

A lack of holism

Some parents voiced concerns that doctors did not have enough knowledge about the disease therefore, believing that the doctors were only able to treat the symptoms instead of treating the disease. In the following passage a mother recalls regret expressed by the doctor when the doctor reflects on how the child was diagnosed:

> "But when she finally got diagnosed and her paediatrician said, "you know I should have added it all up seeing the whole bigger picture instead of just looking at individual things." (P4)

One parent saw this as a medical education issue. The parent believed that this type of education needed to be provided at the beginning of a physician's training by putting more emphasis on genetic diseases. Her perspective on this issue is articulated in the following quote:

> "I think we'd have to have within our medical system, obligation to… to some sort of continuous education of our MDs about these types of diseases so that they can pick

them up. I know that most paediatricians will pick up this type of disease. But certainly even outside of the large cities children don't tend to go to paediatricians, they're just seeing their MDs. And time and time again, you will hear parents say well they just put it down to a cold or they just put it down to you know an earache, or bad hearing or the TV turned up. When in fact they get into almost their teens before they realize that it's been MPS." (P5)

The lack of knowledge about MPS overall was exemplified by how doctors had no real strategy for diagnosing the disease. The parents felt that they were passed on from doctor to doctor because there was a lack of understanding on the part of the doctors regarding MPS. This parent explains this common parental experience as follows:

"First, we had gone to seven doctors. They passed us along, so we started at a neurologist, we moved up to an orthopaedics and we went through a couple of orthopaedic doctors, and finally we ended up at [Dr. X], the head of the orthopaedics department. And, he suspected that it could be a storage disease. He was the first one to suspect something like that and he said that he would make an appointment with [Dr. Y]. And [Dr. Y] finally made the diagnosis." (P4)

One parent described how the doctors seemed to haphazardly investigate the symptoms their child exhibited for almost two years before making a diagnosis of MPS, again reinforcing the idea that doctors do not have enough knowledge about MPS.

"When [X child] was born, he was particularly large for his age…So at six weeks our midwives noticed that, sort of paired with a couple of other minor symptoms and thought that perhaps he had an overgrowth disorder so referred us to a paediatrician who tentatively diagnosed [X child] with an overgrowth disorder. So, we started

coming to [hospital A] when [X child] was just two months of age and seeing the genetics department for what was never confirmed as an overgrowth disorder. But gradually they weren't able to confirm it and they felt that his features were changing as he got older, but they couldn't really pinpoint what was unusual about him and developmentally he was doing great…we didn't think it was anything that urgent even if it was an overgrowth disorder…So we weren't really that concerned, we just had to do monitoring of symptoms. So, when he was 18 months of age we were at [hospital A] for a routine appointment…we brought up that he had a slightly rounded back. So they said, "well you know just to rule something out can you go and have a skeletal X-ray of his back and a urine sample done." So, we did those, and they wouldn't tell us what they were testing him for and we weren't really that concerned. So we got the phone call the next day to say come in right away to see [Dr. Y] that we realized that [X child] had a whole separate, a whole different type of disease altogether." (P6)

The passages quoted above gave a sense of parents wandering in the wilderness and a feeling of hopelessness, as expressed by this parent, "…none of the doctors could really help. They could only treat her symptoms. So basically it was watching her go downhill." (P4) However, as exemplified throughout this essence, a lack of holism, the doctors also appeared to be in the same wilderness in not being able to come to a definitive diagnosis. The issue of not being able to find a diagnosis for these children can be attributed partly to the fact that MPS is a very rare disease however, as pointed out by the parents, the issue was complicated because the doctors did not view the child as a whole (all the symptoms together), but rather tended to treat each symptom separately.

A New Reality

The parents continued their story by describing the realization of having their child diagnosed with a genetic, life-threatening disease. From the data the following essences were uncovered: a double – edged sword, guilt, and protection of the child.

Double – edged sword

Parents described a myriad of feelings such as devastation, shattered dreams as well as relief: a double – edged sword. The parents were finally given the diagnosis of MPS, and the majority expressed a feeling of devastation in learning that their child had MPS (a progressive, debilitating and terminal disease) and the fact that there is no cure for this disease. I described this is as a *double-edged sword* because there is also some relief experienced by the parents since they finally find out what disease their child has and so there was a definitive cause for their child's symptoms. After years of searching for a diagnosis, the disease finally had a name and the families now knew with what they were dealing.

The experiences associated with the essence of *double-edged sword* are demonstrated through the quote below from one of the fathers. He described how difficult it was at the beginning when they were told of the diagnosis but he also got to the point where he had to make decisions of how he and his wife were going to proceed with their lives and how they were going to deal with this disease.

> "Oh, how has it changed my life? Well, I think it's a kind of a double-edged sword. Obviously, there are all the very, very difficult things that come with it but it also makes you hyperaware of family. It makes you really appreciate why you're on this earth, makes you appreciate the important things in life like family and your kids and

your wife…And this disease for me, although it was difficult to take during those first couple of months, it made us realize that we couldn't just sit around and wait for our child to be ravaged by this disease. We were going to have to help him, so I guess it gave us some determination as well to fight this as a family." (P7)

The majority of the parents start with a feeling of relief that they finally have a diagnosis, but once they realize what the potential outcomes of the disease are, their feelings of relief quickly turn to feelings of devastation. This is described by one of the parents as follows:

"Well, in a way, it was a relief to finally find out that there is a reason for some of the symptoms that we've seen, discovering over the years because you're bounced from one doctor to another and no one can give you an answer and so it was a relief. But a shock too, to know that they have a genetic disease. I mean who would have thought…we knew that there were some strange things happening…but to understand that there was actually a reason for their, you know, how do I put this their posture to be a certain way and becoming more pronounced you know that was a relief and shock. I guess that's the best way to put it, because now we have to deal with the problem and then you go around it by saying "Well how do we deal with this genetic disease?" (P3)

During the interviews, the parents reflected on how their hopes and dreams for their children shattered upon hearing about the MPS diagnosis. One parent describes this common feeling of shattered hopes and dreams amongst families as follows:

"I don't think that there's much worse news a parent can hear. We spent weeks crying, trying to come to terms with what that meant, and what it meant for [X child].

Even now thinking about…we still think about the fact he's so gifted. I mean, I think he's gifted in terms of his brain and stuff, but because his language has been slow, it hasn't been as much of a focus as sports has been. He's always been so good at that we think about what could have been if he didn't have this. He could have been such a great athlete and who knows what else." (P2)

One parent also reflected that when their child was born, they had hopes and dreams for their child. When they were told that their child had a life threatening genetic disease they had to adjust their hopes and dreams for their child. The parent went on to reveal that being told about this disease after so many years of living with this child as "normal" and then being confronted with a sick child was especially confounding and hard to cope. They thought it would have been different if they had known of the disease right from birth.

"We were extremely devastated especially thinking he had MPS 1 for that week. That was just the worst week of our entire lives, mostly because different than other diseases you think… you have these two years or whatever, a year and a half to think that your child is healthy and normal and so you start to develop these dreams and goals and all of that, for the future for your child. And to have that taken away I think is that much harder because you're so far into their lives, whereas, if you had the diagnosis from birth it still would have been awful, but it would have been different. This was so extremely shocking because he seemed so perfect." (P6)

The parent explained that if they had known about the diagnosis right from birth instead of thinking they had a "healthy" child at birth, then they would not have formed significant dreams and goals for their child. Therefore, they believed these dreams and goals would not have been shattered in such a devastating way as they had experienced.

Guilt

Two of the mothers interviewed reflected on the essence of guilt. Guilt was expressed by these mothers in response to the question of how they felt when they were told their child had MPS. The first mother that spoke of guilt was a carrier of the MPS – 2 genes that were passed on to her son. She explained that in fact she felt no guilt because there was nothing she could have done to prevent her son from having MPS – 2 short of not getting pregnant. She also alluded to the fact that it is common that health professionals, in particular doctors have a way of making mothers feel guilty when children are born with congenital diseases. However she was not going to allow this to happen to her. She describes this as follows:

> "And the words are worse then any knife being stuck in your back. The question of feeling guilty…I never did, I asked him (the doctor) at the time what he meant for feeling guilty. He said most mothers do. I said well tell me what you mean about guilty. Why should I feel guilty? Have I done something? Do you know something about this disease? Did I do something that could have caused this, because it certainly wasn't known to me? That was a question that has stuck with me all my life and I've asked many doctors why he would ask that -- and one doctor said, simply because over time doctors have made mothers feel guilty for having these children." (P5)

When I asked this mother to read over the findings for authenticity, she supplemented the above passage with the following:

> "This question resulted in an underlying but permanent feeling of remorse, a conviction for unknown actions. This basic question brought on a lifetime nagging question: did the doctor know something I didn't, is he withholding something, is

there something that is common to all MPS mothers that he knows and not saying. This guilt keeps going and the lack of medical education or innocent question has condemned me to a guilty conscience that has the capacity to control my emotions and without the realization of my deed or sin – should I ask for forgiveness of those I offended – who." (P5)

This passage gives the sense that even though she believes there was nothing she could have done to prevent her child from being born with MPS, the question that was asked years ago by that first doctor has always stayed with her and unconsciously "condemned" her for her child's illness. This mother ties it back to the lack of medical education and knowledge since the questions commonly asked to mothers about their pregnancy often cause mothers to feel solely responsible for their child's poor diagnosis.

With regard to guilt, another mother pondered upon her decision to have two other children after having one child with MPS. This mother questioned if she had known that her first-born child had MPS whether or not she would have gone on to have her other two children. This mother described some feelings of guilt but also happiness that she had all her children, even though her second child is a gene carrier and her third child has a diagnosis of MPS.

"And then there was the guilt if we had been more observant perhaps as parents. But again, we never knew that this existed. We didn't think that this was a possibility. But yeah, the guilt of if we had noticed with [X child], from the beginning maybe should we have stopped having children perhaps. You ask yourself that question. Although our second child is a carrier, then again if we had caught it with [X child] and then maybe discovered that our second child was only a carrier, maybe we would have

stopped at two children. But again, then we wouldn't have our Y (youngest child). And she's our baby. So many feelings to deal with." (P3)

Listening to this mother, you can sense the conflict within her. The mother states she is happy to have all her children, but there is a bittersweet sense in that in order for her to have all her children she needs to reconcile with the fact that they have a genetic disease.

Protection of the child

The need for parents to protect their children was another prevalent concern. The parents worried about how others perceived their children so they tried to protect them from the outside world. The parents attempted to shield their child from the outside world and tried to have them grow up as happy and normal as possible, giving them the confidence of being the best that they can be. One parent even confessed that his child did not know that he was ill and his child believed that all children visit the hospital once a week as routine.

"He always assumed that every kid goes to the hospital once a week. We didn't tell him. There was no sense yet, no gain for him or us. So we just carried on. He just assumed every kid went through this." (P1)

The parents endeavoured to protect their children from the prying eyes of the public and aimed to make the child happy and encouraged him/ her to be the best that they can. This mother described her experience of protecting her child from people that looked at her child in derogatory ways and her mission of instilling confidence and self worth in her child.

"My focus was entirely on giving [X child] confidence…everybody looks at these children because they look differently, they're always subjected to stares, and they're always subjected to people making fun of them and the crass and crude remarks. Things like look at the midget and… So you are you trying to deal with a psyche and

a confidence that can be so fragile, simply because of what they are forced up against outside. So rather than go to an advocacy group I literally focused my time on making [X child] feel like he knew who he was; he was comfortable in his own skin and he liked who he was. And I felt if I could do that then the external forces would not affect him so much." (P5)

Another parent described the same feeling of protection and their attempt at trying to protect him from outsiders:

"You try to protect them from that and because this disease causes deformities…His eyes, he's always wearing thick glasses, and people hurt him. He's very sensitive, maybe because he's been cradled so much, I don't know. It's so unfair to a little guy. All he wants in life is his pets and his friends…you know he's like any other eleven year old. It's a little harder for him to move. His joints are seizing up; his eyes aren't there. A normal eleven year old can see, hear, and jump. Well [X child] and a lot of these children can't." (P1)

One mother talked about how the child himself felt about the disease. This child had started working and hated to bring attention to himself by having to miss work for appointments or treatment.

"Many times [he] worries about how this affects him at work. He says it's not nice for him to have to tell them at work that he has to come and have treatment done. He's almost somewhat embarrassed by it I guess or bringing attention to himself. He just wants to be like everyone else." (P3)

In conclusion, the parents described the upheaval and stress the family experienced with the confirmation of the diagnosis. The family had to deal with the diagnosis of a genetic

disease and the realization that their child's illness was terminal. However, over time the parents are introduced to a possible treatment that may alleviate some symptoms and stop the progression of the disease.

A New Optimism

Parents described the experience in which they were told of a new treatment – Enzyme Replacement Therapy (ERT) and that their child was eligible to participate in a clinical trial. One group described feeling new hope and optimism that maybe this treatment would help improve their child's quality of life and possibly extend their lives. The parents enrolled their child in a clinical trial but then had to ensure that they met the requirements of the clinical trial, which led to further upheaval in their lives. This upheaval included weekly travel to paediatric centres for ERT as some families had to travel by air to get to these centres. Some parents had to quit their jobs, as it was more prudent to move closer to the paediatric centre instead of travelling on a weekly basis. When this decision was made one family had to re-establish themselves by seeking new employment and a new home. Two of the mothers described how their sons (one in his twenties and the other in his late teens) had to disclose and negotiate with their employers that they would miss a day of work on a weekly basis for their infusions, thus making them feel "different."

The second group of parents' children were not eligible to partake in the clinical trials because they were either too young and had not yet been diagnosed with MPS when the clinical trials had started or there was no clinical trial for their type of MPS. This group of parents had to fight to get their children on ERT as compassionate cases. When parents described this "new optimism" the data revealed essences that included hope, persistence, fear of the unknown, putting their lives on hold, and regret.

Hope

The parents had been given a new hope for their child when they were told that their child could take part in a clinical trial for ERT. In the following passages, parents expressed their feelings when they were told their child was accepted into the clinical trial. The parents moved from a feeling of hopelessness to hope. The following parent expressed how she went from watching her child "go downhill" to the stabilization of the disease process:

> "None of the doctors could really help. They could only treat her symptoms. So basically, it was watching her go downhill. Then this drug came along…she didn't make great strides in improvement, but she has stabilized and I keep thinking that without this drug, her situation would be very grave right now. Her quality of life would be very poor. I mean she does have many problems day to day. She has quite a few problems even being on the drug. But, it's better than if she wasn't on the drug." (P4)

This parent reflected that with her child receiving ERT it had given her the opportunity to re-evaluate her hopes and dreams for her child.

> "And then having had this chance of getting him on ERT has changed that again because all of a sudden we have those things restored again. We can think about the future again." (P6)

Another parent associated his child being on ERT to winning the lottery. He explained that his son's diagnosis of MPS was unfathomable to begin with because of the rarity of the disease, but then to be able to participate in the only treatment in North America, it was like winning the lottery to him. It was clear that this parent chose the uncertainty of a

clinical trial (an unproven drug in humans) because the other choice (certain physical deterioration and a more imminent death) was unacceptable to him and his wife.

> "Ecstatic. It was like we won the lottery again. It's a one in I don't know to be diagnosed with MPS and then to find out that we actually got in on the only treatment in North America and it wasn't without risks but the other option was totally unacceptable. We weren't going to just sit back and let him just pass away. So it was huge. From that point on, we knew everything was a gamble, just with doctors and specialists and everything. But because he responded so favourably…you should have seen the doctors they were like they won the lottery when they realized how well he was responding to this." (P1)

Listening to the parents, the parents expressed feelings of renewed hope after a period of devastation and shattered dreams. They are able to reassert their hopes and dreams for their child with the opportunity of enrolling their child in a clinical trial.

Persistence

The group of parents whose children were not in the clinical trials because their children were too young or had not been diagnosed with MPS when the trials began talked about persistence of keeping their child's plight on their respective provincial/territorial government's agenda. Some parents decided to make their story public by parlaying their dilemma with the government through the media. One set of parents consciously had to decide how they were going to portray their child to the government. Were they going to present a sick child or a healthy child who without ERT would become sick and a burden on society? This dilemma is described in the following quote:

> "It was really frustrating. When we were getting nowhere with the government, we

had to look at how we were going to present this picture. Were we going to present a picture of a very sick child, that's not receiving treatment or were we going to paint a picture of a child that's extremely normal and bright and beautiful like my (child) is and talk about what's coming down the road for [X child]." (P7)

Another parent spoke of how they had to wait until ERT was approved in the United States before they could even approach their provincial government about obtaining access to ERT for their child.

"When we first were diagnosed, we heard there was treatment going to be available soon and so I looked into that. Until the FDA approved it, it was just a wait, because at that point it was not approvable so there was nothing we could do. We called [pharmaceutical], we called everybody basically to see if there was anything we could do to speed up the process but we pretty much learned that there wasn't. Once it got approved by the FDA, it was July 24th and from there…I know a lot of people have told me I've been too pushy, and done too much." (P2)

The same parent went on to describe that once ERT had been approved in the United States they started contacting their provincial government and lobbied for their child to receive the treatment in Canada. This parent was very honest that she was willing to utilize any means possible to get her child onto ERT.

"And I told him that I knew of other families that had gone to the media and had success getting ERT for their children after going to the media. I said I didn't want to do that but I said I was willing to go there because it's my kid's life we're talking about." (P2)

Anger seemed to play a part within the essence of persistence. One father described the opportunity he had to speak with his respective provincial minister of health and the anger he projected when he spoke of the government's decision to either fund the ERT and give his child a future or not fund ERT and destroy his child's future. The father recalls this encounter as follows:

> "And I just looked at him and said, "This isn't about politics, it's not people, it's about our son and he's the first and foremost on our minds right now." And he himself as the Minister of Health or as the representative for the (provincial/territorial) government for health care in this province/territory has the opportunity to make or break my child's future. He can either make the decision to fund treatment and my child will have a future or s/he can say no and he'd be without." (P7)

One mother did voice hesitancy on making her plight public. She explained her hesitancy in terms of seeing herself as one person against the entire system. This mother was the only parent that decided to sit back and wait for the government's decision. She implied that she put her faith in a higher being and waited for the government to make the right decision and fund ERT.

> "I just waited for them, as one of these cowardly parents. I just waited, I figure I'm just a small person, and there's not much that I could do, so I just waited for them to say yes or no and just prayed that they would come about soon for my children. I just figured I wouldn't have much say in the matter." (P3)

The parents had been shown and had witnessed a treatment for their children that improved their physical limitations. However, what differs in this situation is that for these parents unlike others, there is a treatment for their children but in some sense it is not accessible.

This situation is very different than what Canadian citizens are used to experiencing, because, for example if a child is diagnosed with diabetes or cancer, the child would have access to treatment without having to lobby government officials.

Fear of the unknown

Some parents were ecstatic that their child could participate in their respective clinical trial. However, it was not without soul searching. The parents knew ERT was unproven in humans and had been forewarned about possible adverse effects.

> "It was with very mixed feelings; one of elation that finally there was something that may help stem the progress of the disease and prolong his life. There was one of fear that this was basically an untested drug and was my son a guinea pig." (P5)

Another parent expressed the same sort of fear and hesitancy toward the clinical trial. She was happy that her children had been approached to take part in the clinical trial but she was so anxious and cautious in agreeing to her child's participation that she consulted a psychic.

> "Well I was very happy that [Dr. X] considered my children but also as a parent, you don't want to make the wrong decision for your children. They would be like guinea pigs in the trial obviously. And I think I even consulted a psychic to make sure that nothing would go wrong with my children. We were just concerned; you know "Will this work? Is it going to harm them?" But then, we said it's gotta be better than nothing in the end. How bad can it be? So we just thought we'd be brave and, go with it and try it out for their sake." (P3)

These parents had all taken a chance and a risk by enrolling their children in clinical trials, but as the following parent stated it was also done for altruistic reasons, i.e., for future MPS affected children.

> "I convinced my conscious, that someday everyone has to die…this is a good cause. Number 1, maybe we are successful in getting some relief from this medicine, so it's a direct benefit for us and second for the future we are contributing and taking a risk today for other kids." (P8)

Putting lives on hold

This essence described the upheaval of the family's everyday life. With their child accepted into a clinical trial the family had to ensure that they were able to attend the weekly infusions. The Canadian site for the MPS 1 clinical trial was located in Vancouver and the only North American clinical trial sites for MPS 2 were located in the United States of America (North Carolina, Houston, St. Louis, California). Families enrolled in these two clinical trials had to make the commitment of traveling each week to the centres for their children's infusion. The majority of families experienced upheaval during this initial period until the infusions could be moved to paediatric centres closer to their homes. This first parent describes his family's challenges at the beginning of the clinical trial until they finally came to a solution – moving closer to the hospital.

> "We travelled. We started out traveling back and forth at first, flying to [X city]. Once a week travel by car for five or six hours to [Y city], spend the night, get on a plane, spend two nights and three days in [X city] then back on a plane to [Y city], then drive back to (home town). So we moved to [Y city] and then from [Y city], we moved to [X city]… So what we did, because my wife had already been asked to

leave her job due to just a lack of being able to be there or when she was there needing to rush off to doctors. We decided that my wife would move to [X city] and I changed my employment so that I could be there. I was also a fulltime employee with the [X government] and we had to move to follow his treatment, which we did – that's neither here nor there." (P1)

Another mother talked about her shock of being told the clinical trial was being conducted in a city 3000 kilometres away and if she wanted her child to take part in the trial she would have to move to that city or fly out on a weekly basis. At first this mother decided against enrolling her child in the clinical trial until the doctor advised her that this trial was too valuable an opportunity to give up.

"He informed us that we would have to fly to [X city] on a weekly basis or move out there, which was a shock. I did not want to leave my family. (Child) didn't want to go. I wasn't going to leave (child's) sister behind. So I said no, in the beginning. I phoned and I said no we couldn't do this; it's just too much. I talked to [Dr. X] the head of the study out there. And he said that if we don't do it that it might be three to five years before she could be put on this drug before there is any kind of funding, any kind of approval for this drug, and that would probably be too long to wait. So that was the defining moment when I said yes, we are going to participate in this study and we were going to have to make it work somehow. We're just going to have to adapt and that's what we did." (P4)

This mother reflected on her experience of traveling with her child on a weekly basis. She pointed out that the challenges were not just about the costs and time involved but also related to the risks associated with traveling with a sick child.

"There was not only a time involved because it was a two-day event, sitting around airports, planes cancelled, the timing of it, that constant daily travel. It was horrendous. And then with [X child] having to have the pre-medication. We were traveling back the same night that he had the medication. But yes, I don't think anybody until they do it can understand the stress of that constant weekly travel. And then to come back with your body full of drugs and exhausted with all the pre-medication, that traveling back, exhausted." (P5)

Regret

Parents with older children expressed regret that treatment had come too late. Having received ERT, the parents witnessed an improvement in their child's physical wellbeing, but they also felt a great deal of regret that ERT had come too late for their child and the devastation the disease had harvested prior to ERT could not be "undone."

"And the only thing that I must say now that…I look at it and I find is… I'm human. I'm thinking, if only it had been 10 years ago…Unfortunately, [X child] is not getting the benefit of it all, but he's certainly getting the benefit in the major organs; the spleen and the liver. But there are other things that we've seen such as the joints etcetera; he's not getting the full benefit of it. So yes I was totally elated, a little bit disappointed that it had come too late, but better late than never." (P5)

A second parent echoes the same sentiments, however this mother also remarked that for a long period of time she felt that she was just watching her child die and not being able to do anything about it. With ERT this mother now felt that she was doing something to improve her child's quality of life:

"But as far as having an impact on our lives, I'm glad I'm doing something for my daughter, because for seven years there was nothing available. It hurt because I couldn't do anything for her. It's very frustrating, it's hard to watch your child go downhill, and there's nothing you can do for them. Now I feel like we're doing something and there's some help out there. I know it's not a cure. Probably the disease is still progressing but at a very slow rate. I know she'll have a longer lifespan due to this drug, which makes me very happy. Being on this drug has had a big impact on [X's child] life – physically and emotionally. I know she doesn't like coming to the hospital and getting IVs every week. This is very difficult for her, but in the end I think she's glad she has the drug, because it's giving her a better quality of life. And for me I know I'm doing something for my daughter." (P4)

A Road to Nowhere

"A Road to Nowhere" represented the disillusionment the parents felt and the beginning of another battle for the parents. Their children had been on ERT (ranging from 1 to 7 years) and there was now the realization that the provinces/territories would not necessarily cover funding for the different types of ERT even though the drugs had been approved by Health Canada. Parents described feelings of betrayal, frustration and fear, and the belief that it was the "Government's fiduciary responsibility" to provide funding for ERT.

Betrayal, frustration and fear

The parents described a feeling of betrayal, frustration and fear toward the federal/provincial/territorial (F/PT) governments. The parents had witnessed improvements in their children's condition while receiving the ERT. They had sacrificed time, work, and family life to make sure that their child could participate in the clinical trial and were now

faced with the threat that their child would not be guaranteed ongoing ERT. The following parent could not believe that the people he thought would oversee his son's treatment were now backtracking on their promises.

> "We've gotten through all of that…the transportation everything to get to and from these treatments and stuff, that once everything was approved was when we realized that not all governments and hospitals and pharmaceuticals…you need to get everything in writing. All the promises we were told that we would never have to worry about treatment disappeared in a blink of an eye. We were in for our treatment on a Friday and we were told not to…I can't even remember the date when we were told that unless we come up with some sort of funding then not to come back the following week." (P1)

Another parent expressed her frustration with the federal and provincial/territorial governments because there was a treatment available for her child and she knew it had improved her child's quality of life but all levels of government were not willing to pay for the treatment. She expressed her disbelief and frustration, as she believed that these government officials either had no compassion or understanding for what families had experienced.

> "The frustration of now getting something and knowing that it's not available or that we can't get the help where there is something that can help my son has resulted in perhaps a little bit more of rather than a hopeless frustration, but one of just sheer frustration of individuals that perhaps don't have the compassion and the understanding." (P5)

Another mother described the fear she lives with every day because she believes the provincial government will withhold ERT treatment from her child. She explained that parents have been waiting for a while for some form of F/P/T government policy on how ERT will be funded but nothing has been decided yet. She expressed that without an orphan drug policy she fears that ERT will be taken away and that she and her husband will not be able to plan for their family's future.

> "We have a fear that one day the doctor will tell us that he was not able to get [the drug] and the companies are not giving them. I'm afraid of that situation. Always it's in my mind, that maybe some day [this] will come. But so far, so long, it's OK I don't want to live with this fear, if government they really take [it away]… or they really decide to give." (P9)

Similarly, another parent's betrayal, frustration and fear are palpable in this passage. He was totally at a loss not knowing whom to turn to for help when he was told the clinical trial had ended and there was no funding to pay for his child's treatment the following week.

> "There was no warning it was a Friday afternoon when we finished his treatment, when one of (child's) doctors came in and he was very visibly upset and everything and that's when he told us that unless we came up with funding that we weren't allowed back the following week. At that point, right then and there it was like the whole world collapsed. We've already learned that this is a life-altering disease. It's a destiny for these children if they don't get their treatment – slow, painful and all you can do is sit and watch. Your whole world is going to come to an end even though you've helped develop it." (P1)

The parents also expressed a feeling of betrayal by the pharmaceutical companies. This betrayal stemmed in part from the parents' belief that they put their children at risk by having them participate in the clinical trials and in doing so the pharmaceuticals "owe" something to their children for assuming that risk. One parent in the following passage conveys this feeling of betrayal in the following passage:

> "[X mother] wrote a letter to (pharmaceutical), basically saying that [X child] did participate in the phase 3 study and that she risked a lot for them. Basically we put our life on hold for that study and that they owed us." (P4)

One father's feelings about the betrayal by the pharmaceutical company were evident. He felt that the pharmaceutical company turned their back on his family once the ERT was approved by Health Canada. The father believed that once the company had gotten federal approval for the drug that the company had obtained their goals and the children and families then became irrelevant to the company.

> "We started out in the double blind study, then the blind study, and then the regular study. Through all of this, we were dealing with the drug company and everybody that was involved to get our treatment. And since he was the youngest to be inducted into this, we thought we had a very bright future working together with these people. And as soon as the studies were over and everything was approved for manufacturing, distributing and whatever in Canada was the very first inkling that we got that they might withhold treatment. Through all of the studies and everything we had done we were told we wouldn't have to worry about it because we had participated in helping get this up and running…And in 15 minutes it went from a very bright future of finding a treatment that never even existed prior to [X child's] and other children's

involvement to ending up with… well in North America how many children are left and in an instant they're taking it away over a few measly dollars. It is very expensive. But this is something that shouldn't concern the children or the parents. This is something we've participated in." (P1)

The above statement summarizes how the parents felt about the whole situation, in that once the pharmaceutical companies had obtained Federal drug approval through Health Canada, their mission was accomplished and from the drug companies perspective the children and families then became irrelevant.

"Government's fiduciary responsibility"

The parents' betrayal, frustration and fear for both the F/P/T governments and the pharmaceutical companies is tangible but eventually the parents' anger and frustrations turned toward the F/P/T governments. It was their belief that the government should be caring for their citizens, especially children, and thus funding ERT. The majority of the parents stated that they never thought that something like this could happen in Canada and a country that prides itself on a universal health care system. They felt that the government was neglecting their children. The following passage highlights one parent's betrayal, anger and disillusionment with all levels of government. He believed all levels of government had abandoned him and his family and they had forsaken their "fiduciary responsibility" to him, his family and citizens in general.

"They're not looking out for their people. We've got two different First Nations governments, we've got a provincial government, we've got a territorial government and we've got a federal government and everyone of those people stand on a stack of bibles, and swear they're there to help you. They're here to make sure to look out for

you…but you're alone, you're there with your child, your son…your daughter. You're there alone and you know what we pay billions of dollars in taxes, and we go through all kinds of hoops to elect these people that we think they are going to look after us. Look out for our best interest and yet when it's all laid out on the table you're the only one there. You and your child…But we believed, we trusted, we were very naïve." (P1)

Another parent also frustrated with the government response, could not understand the "laggard behaviour" of the provincial government in committing funds to ERT. She stated that they have been waiting for two years and nothing has been done yet.

"It will be a hard life for [X child] without this enzyme and it's very hard for all of us to understand the behaviour of the provincial government by not…it has been 2 years and they keep on telling us that they are looking. Yes, something should be done, but it has not been done yet." (P9)

This same parent sounded cynical about whether or not, provincial/territorial government would ever "own up to their responsibility" and that the governments seemed to ignore what the families have experienced:

"Their behaviour shows that they will continue to do this for as many years as they can. They don't understand the feelings we have to go through, the agony of the family." (P9)

Another parent could not come to terms with what she considered to be the wayward logic the F/P/T governments have regarding ERT funding. The parent states the facts as she sees it: there was no treatment for MPS; her child received ERT and her child's health

improved; the "ruling type" are not funding ERT nor do they understand the pain that the families are experiencing.

> "In the initial stages, you were looking to see if there was any problem if there is anything. Now that he's had the drug and you see the results with him and the benefits that he is receiving from the drug. It rather exacerbates the frustration of not being able to get those of the ruling type and to be able to get the politicians to understand the pain that one's going through." (P5)

This same parent remarked that she could not understand how one entity at the federal level (Health Canada) could approve ERT and then have the provinces and territories make the decision to not fund ERT. She could not reconcile that the provincial/territorial governments would not fund ERT to treat the disease and prevent deterioration of these children, but the provincial/territorial governments were willing to pay for organ transplants, rehabilitation and any other treatment that would be needed when these children deteriorate.

> "I think I would first say that it makes no sense to me that we have one entity i.e., the federal government approve a drug and then say we approve something and then hand it over to another entity without any other recommendation and/or dictate for funding of that drug. Therefore, in the approval you're saying this will help these children, but it's up to you whether you want to decide to pay for it to help them. With regards to the (provincial/territorial) minister, I would say what rights has anyone to say that we will continue to let a child suffer and we will pay for the suffering and for all of the various surgeries. And, we will let a family sit and watch for each one of those surgeries. Watch the pain of those families and the suffering of each of those families." (P5)

Another parent plainly stated the facts as to why the provincial/territorial government would not fund ERT – it costs too much money and the return on investment is low:

> "The patient has to go through all those struggles and the people use them, especially in this one…this drug has been approved. The government is in deadlock talks. The government doesn't want to pay for 2 reasons. It's very expensive and the second reason, the patients are not many." (P8)

One parent spoke of the arrangement that was made between the family and their provincial government. The province/territory were threatening to stop ERT funding for their child but on the day the family was to go to court to challenge the decision the province/territory decided to fund the child for half the dose. During my study, this child was receiving 50% of what he was originally receiving during the original clinical trial funded by the pharmaceutical company.

> "Through the province/territory we are working on probably I think it's about 50% of what he was getting through the study. The way we accomplished that was our lawyers…the day we were going to court to get an injunction to keep the treatments going, was the day the province decided to cover 50% of it with the acknowledgement that they have to give us 30 days notice before they cut us off until there is an orphan drug policy or something like that in place. So we ended up, we're still running on that." (P1)

One parent described how she felt when she had to pick between her two children as to who would continue to receive ERT. The clinical trial had ended and the provincial/territorial government had decided to continue to provide treatment to the children but only for a limited time and to children who were severely affected. This mother described

how she and the doctor came to the decision. The mother talked about speaking to the doctor over the phone and discussing the situation. She explained how the doctor had called her at work and that they needed to make the decision as to which of her children would continue on with the ERT and which one would stop as there was no money to fund both of them. The mother explained her younger child was the more severely affected than her older child so the younger child was picked to carry on with treatment. The mother went on to say that it was a very difficult decision for her to make and that she was at the government's mercy as to when her older child would restart the ERT.

> "Well you know the biggest thing that struck me was the time I had to choose between my two children to receive the treatment. This was after our study period, I believe and we were receiving the treatment and at a certain point, there was no funding to provide treatment for both of my children. So, [Dr. X] as well as myself, had to choose which child would receive treatment. As you can imagine that was very difficult. But, it was only available for one and [Dr. X] and I both agreed on the same child. [X child] seems to be the worse off of the two children, but not to say [Y child] didn't need it. So that was a little bit difficult, to choose. For me as a parent it was difficult to say, "OK well [Y child] you can take a break", but knowing full well that it's because there's no funding that you can not continue with treatment. But eventually [Y child] got called back. Again we're at everyone's…whenever they're ready to put out some money for the funding. And not to say it's not a lot of money but we're at their…whenever they're ready." (P3)

Parents were also upset with the F/P/T governments, as they believed their children's rights were being compromised. These feelings are exemplified in the passage below as one

parent discussed the issue of fairness and how one province/territory varies from another. She could not come to terms with the fact that one province/territory could pay for ERT while another would not.

> "Well I don't feel it's fair. But in a way, I can understand since we have six children in [X province/territory] and the other [provinces/territories] only have one child. I know [X hospital] is funding the child out there. [X province] is such a rich province, why should our children be the ones not getting funding for the enzyme? I don't think the government is being fair in denying these children. And since it's approved by Health Canada I don't see why they're dragging their feet so much regarding funding." (P4)

Summary of Findings

In summary, the parents described a continuum of continual waiting. "A New Journey" described how the parents strived to find a diagnosis for their child. When facing "A New Reality" the parents are struggling to come to terms with the diagnosis of MPS and the certainty that their children will continue to deteriorate. During "A New Hope" the majority of parents tussle with the decision of putting their children at risk with an unproven treatment, while some parents wait for the opportunity to start their child on ERT. During "A Road to Nowhere," the parents dispute the decision the provincial/territorial governments make regarding no funding for ERT.

Looking at the four themes through the Critical Social Theory (CST) lens, there is a progression not only through time but also on the increasing awareness of the parents' sense of what is *right* for their children. The parents started the journey on their own, and asserted their needs by trying to find a diagnosis for their children. They sought answers by consulting

with different health care professionals and finally arrived at a diagnosis. When they couldn't find answers they made their questions heard by shedding light on their child's plight by whatever means necessary (i.e., seeking out multiple medical opinions; writing letters to government officials; going to the media). The parents' struggle and ultimate dispute with both the federal and provincial/territorial governments emphasizes the marginalization of MPS affected children and their parents.

CHAPTER 5 – DISCUSSION AND IMPLICATIONS

Introduction

The main purpose of this study was to describe the lived experience of parents/ caring for children diagnosed with Mucopolysaccharidosis (MPS) waiting for federal drug approval and provincial/territorial drug funding of Enzyme Replacement Therapy (ERT). The need for such a study arose because I became aware of the challenges parents of children diagnosed with MPS experienced when I was caring for the children during ERT clinical trials. I noticed the continual stress and worry the parents experienced regarding the fear that the provincial government would not fund ERT and an extensive literature search yielded no findings in the literature on this phenomenon. The parents, for the first time, had seen an improvement in their children's physical wellbeing since starting on ERT and believed that because of this improvement the government seeing this improvement would naturally agree to provincial drug funding.

The chapter begins with a discussion about the use of Critical Social Theory as a framework for this study and then is followed by a summary of the key findings as they relate to the existing literature. This is followed by a discussion on what orphan drug policies are in place on an international level and recommendations for an Orphan Drug Policy (OPD) in Canada.

Discussion

Using Critical Social Theory (CST) as a framework allowed me to illuminate the struggles of the parents and authenticate their reality by exposing their experiences of marginalization, not just by focusing on individual parent coping skills but analyzing the broader system issues (social and political factors) such as health policies and how that

impacted on their experiences. This study led to uncovering the entire experience of these parents through four stages (starting with searching for a diagnosis and ending with waiting for ERT funding) and the exploration and description of what power differentials they were up against. Examples of the power differentials included: parents searching for a diagnosis and persisting by seeking second opinions when doctors said there was nothing wrong with their children. Once Health Canada approved the ERT drugs and based on the decision by the CDR, provinces decided not to pay for ERT. Parents fought back by accentuating their children's plight through letter writing campaigns and making their fight public through the media. This public action not only illuminates the clinical aspect of their fight but also highlights the much broader level issues that include provincial and federal health policies. Utilizing CST allowed me to uncover this dilemma. By having the parents recount their stories, it legitimized knowledge generation through human subjectivity (perceptions and experiences), thereby reinforcing CST epistemology, which states that "knowledge as truth is socially constructed" (Duchscher, 2000, p. 454) and the facts put forth by these parents are relevant because it is a lived experience (Duchscher). Looking at this phenomenon through the CST lens has helped me to analyze parents' experiences and to be aware of provincial and federal health policies, orphan drug policies, and how our drug policies differ compared to other developed countries. As a nurse it has lead me to explore and understand what influences health policy (i.e., advocacy groups and health policy economics) and the impact of these policies on the everyday lives of the children and their families.

I started with the research question "What is the lived experience of parents with children diagnosed with MPS waiting for federal drug approval and provincial/territorial drug funding for ERT." I thought parents would concentrate only on the aspect of waiting for

federal drug approval and provincial/territorial drug funding. However, as the parents' stories unfolded a picture of *waiting* emerged that covered a much greater length of time. What I found was the waiting started much earlier, as it started with waiting for a diagnosis for their child and then progressed to waiting for a treatment for their child through accessing a clinical drug trial. Even after the child received treatment, the children and parents continued to experience more waiting because in order to guarantee ongoing treatment with ERT, they all had to wait for their respective provincial/territorial government to approve funding for ERT or provide the ERT on a compassionate basis. The time for *waiting* for the provincial/territorial governments to approve ERT funding and/or provide it on a compassionate basis for my parents ranged from 1 year to 2 years. The parents in Stubblefield and Murray's (2002) study had a much shorter waiting period as their *waiting* started when their children were placed on the transplant list and had a range of 2 to 9 months.

As described in the findings, some parents in this study stated that they believed funding of ERT was part of the government's "fiduciary responsibility." The majority of parents experienced feelings of betrayal when talking about the provincial/territorial government's decision not to fund ERT. These parents were not only caring for their children but in addition they also had to fight for their child to remain on ERT. Similarly, Ray (2005) found that parents spent an inordinate amount of time on what she referred to as "navigating bureaucracy". While Ray focused on parents with chronically ill children trying to secure community services for their children at the local level, the parents in this study had to focus on making their plight for ERT funding known to the federal/provincial/territorial government levels. Examples of advocacy on the part of the parents included letter writing to

their federal Members of Parliament or their provincial/territorial counterparts, assembling at health ministers' summits and describing their child's plight to the media. Most parents felt that they could not sit back and wait until their provincial government decided on whether to fund ERT or not. They felt that they needed to be their child's advocate and that it was important for their respective health minister to meet and interact with their child in order for the health minister to realize that there was a real person they were saying "no" to when deciding on the "bottom line". Parents in both Ray's study and my study had to *navigate bureaucracy* but, at different levels. In my study they were trying to influence policy at a provincial and federal level, however the parents in Ray's (2005) study were trying to influence case managers at the local level to provide more services. Another difference is that in my study, influencing policy is a life and death situation, as the children need ERT to prolong their lives and improve their quality of life.

Another factor that sets this population apart from the ones described in Ray's study is the parents' experience of persistence and belittlement in trying to find a diagnosis for their children. The majority of the parents started their stories by revealing how long they had to wait for a diagnosis. Parents spoke at length of their journey in trying to secure a diagnosis and the toll it exerted on their lives. They described a period of time that in some cases covered three to four years in seeking opinions from different physicians. The parents also described feeling belittled when they persisted in their quest for a diagnosis as they were told over and over again that nothing was wrong with their child. Ray (2005) found that once a diagnosis was made both parents and health professionals gained a sense of legitimacy, confidence and direction. Parents in my study felt a sense of vindication in their persistence

in trying to secure a diagnosis for their child, therefore the significance of finding a diagnosis appears to be common to both populations.

Parents in my study also experienced the phenomenon of "putting [their] lives on hold." The families had to travel to and from the paediatric regional centres where the clinical trials were being held and/or move closer to the paediatric regional centres. This period was one of upheaval and stress for the family as decisions had to be made about whether to take part in the clinical trial and move closer to the trial site or travel on a weekly basis to the paediatric centre. Parents needed to understand that they had two choices: the first choice being either to travel weekly or to move closer to the paediatric centres and relinquish their homes, employment and social supports; and secondly if they didn't agree to the clinical trial they would be passing up the opportunity of a treatment for their child that could possibly never be offered again. Although the majority of the MPS parents traveled back and forth on a weekly basis, there were two families that, in fact, relocated to where ERT was being offered. One parent discussed the upheaval the family felt when she and the child moved closer to the regional centre for treatment and left her spouse and the younger sibling behind. Another parent described how the decision was made to move the family permanently from their hometown to the city where the regional treatment centre was located. This parent described the loss of financial stability and employment and searching for new employment. Stubblefield and Murray's (2002) study on parents waiting for their child's lung transplant described a similar phenomenon as families in their study relocated to be closer to the paediatric treatment centres as well. These authors discussed that participants experienced a sense of anguish in leaving family members behind and the need for additional sources of support which in these cases, included other parents also waiting for their

children's lung transplant (Stubblefield and Murray). However, Stubblefield and Murray also reported that this support network developed by parents had its down side because as children underwent lung transplants and the families moved back home the families that had been part of the support network at the paediatric treatment centres and who were then left behind still awaiting transplant, experienced a sense of abandonment and thus adding another issue that they had to live through.

The phenomena of having to move closer to seek treatment for regionalized services may be more prevalent and taxing in paediatric care overall because of the stage the families are in (children in school, economic responsibilities such as house payments and the upheaval to other siblings). For example if seniors had to move or re-locate for treatment, their responsibilities may not be as diverse (i.e. work commitments, child care, school) as a young family's. Therefore, health care providers must consider not only the enduring stress of treatment but also the displacement of the affected child from their home, parent and siblings and subsequently, their increased vulnerability given the developmental stage the family is at.

Parents in my study also described "a lack of holism" on the part of the physicians. The parents were attempting to secure a diagnosis for their children and the lack of information that doctors relayed to parents because of the limited amount of knowledge doctors had regarding MPS. Parents described a feeling of frustration in that the doctors could only treat the children's symptoms instead of the disease. This lack of information limited the amount of decision making the parents had regarding their children's plan of care. The lack of decision making on the part of the parents fed into their feelings of helplessness and hopelessness. Parents spoke of "watching their child die" in front of their eyes.

Hummelinck and Pollock (2006) examined the information needs of parents of chronically ill children and found that parents presented with a great variety of information needs. They also reported that parents couldn't pinpoint their information needs, or what information they might need in order to better cope with treatment and the possibility of further deterioration of their children. The authors described that some parents actively sought out information while others resisted or avoided information as a type of positive coping strategy, in that they resisted information for fear of negative news. The authors explained this positive coping strategy as a means for the parents to selectively decide if they wanted to hear the bad news or not. Parents in this study expressed frustration, and in some cases anger, in that their concerns were not taken seriously when trying to secure a diagnosis for their child. Therefore having accurate and adequate information was important to both populations represented in these two studies.

Hope was another essence that emerged from my study. This essence was most prominent in the theme *A New Optimism,* but it could be found to a lesser degree within the other three themes. Parents began to express hope when they were informed their children were eligible to participate in ERT clinical trials. It didn't matter to them that this was an unproven treatment, this was an opportunity for them to "do something" for their children. Parents next expressed hope when they started to witness an improvement in their children's physical condition such as an improvement in oxygen saturation, improvement in the range of motion, a decrease in liver and spleen size and in the case of the younger children an increase in their height. Parents began to speak again of having dreams for their children. Even though the parents saw the improvements in their children's quality of life most of the parents realized that ERT was not a cure.

In a study by Barrera et al. (2005), that involved parents of children with cancer, the theme of *hope* emerged as a significant finding. This study included parents of children who had been enrolled in Phase 1 oncology clinical trials. The theme that emerged in Barrera et al.'s study in regard to hope was "hope for a cure or prolongation of the child's life." The parents included in Barrera et al.'s study were parents whose children were in the terminal stages of cancer whereas the children in my study were in stable condition and could eventually die from their disease without ERT. The parents in Barrera et al.'s study were in a different situation as they were grasping at any opportunity for a cure or prolongation of life because their child's death was much more imminent. Hope in relation to treatment outcomes for the parents in my study was closely linked to improving their child's quality of life rather than being viewed as their child's last chance for survival. This could explain why Barrera et al.'s study found parental hope to be a much more significant theme than in my study.

Policy Context: Implications of Findings

Using the Critical Social Theory as a lens in which to illuminate the struggles of parents with children diagnosed with MPS led to the substantiation of their experiences by exposing occurrences of marginalization, not just by focusing on individual family challenges but analyzing the broader system issues (social and political factors) such as health policies and how these policies impacted on their experiences. The phenomenon of waiting for ERT drug funding is an issue health care professionals ought to be aware of when caring for MPS affected children because parents in Canada struggle spiritually, emotionally, and financially not only because their children have a chronic debilitating disease, but also because of current government policies they are living out. The parents in this study have no choice but to be advocates for their children in order for their children to receive the only

treatment available: enzyme replacement therapy (ERT). This phenomenological descriptive study describes the experiences of these parents and illuminates the human factor (real life) in the complex social and political environment of health policy decision-making.

International Orphan Drug Initiatives

Canada is the only developed country without an Orphan Drug Policy (ODP), which translates into limited support for research and development into treatments for orphan diseases (CORD, n.d. [a]). With limited research support, Canadian patients with orphan diseases often do not have the same access to life-saving therapies (such as ERT) as patients in other countries (CORD, n.d. [a]). During the last twenty-five years the United States and Australia have instituted orphan drug programs, which provide incentives to biopharmaceutical companies that undertake the development and marketing of orphan products for orphan diseases (BIOTECanada, 2004). International orphan drug initiatives are discussed in this section (specifically the United States (USA) and Australia) and what these initiatives have garnered for these two countries economically.

In 1983, the US was the first country to implement and pass into law the *Orphan Drug Act* (ODA). The Act not only covers pharmacological and biological products but also medical devices and dietary or diet products (OrphaNet, n.d. [a]). The Office of Orphan Products Development (OOPD) was created and is housed within the Food and Drug Administration (FDA). The OOPD's mission is to promote the availability of safe and efficacious products for the treatment of orphan diseases (OrphaNet, [a]). The ODA provides incentives to pharmaceutical companies for orphan disease drug development such as, 1) market exclusivity for 7 years after marketing approval of drugs designated as *orphan drug* by the FDA; 2) a 50% research tax credit for clinical studies of orphan diseases undertaken in

the US; and 3) simplification of administrative procedures such as a decrease in the waiting time for approval and a reduction in registration fees (OrphaNet, [a]). With the advent of the *Orphan Drug Act* in the US, as of 2003, there have been more than 900 orphan drugs developed with more than 200 receiving market approval by the FDA (Iribarne, 2003).

Australia's *Orphan Drugs Policy* was implemented in 1997 and ensures the availability of a greater range of treatments for orphan diseases (OrphaNet, n.d. [b]). The main characteristics of the policy are as follows, 1) The Australian Therapeutic Goods Administration (TGA) uses information from the US FDA as part of the Australian evaluation process; 2) a waiver of application, evaluation and annual registration fees thereby, removing a major obstacle in making orphan drugs available to individuals. The TGA covers all costs of the orphan drug designation process; and 3) five year market exclusivity of drug (OrphaNet, [b]). The major difference in Australia is that research and development is not supported by grants or tax credits (OrphaNet, [b]). Australia designates an orphan disease as having prevalence of 2,000 affected individuals or less in the Australian population of 18 million (OrphaNet, [b]).

Australia's ODP is grounded in the American ODA where the TGA uses information from the OOPD (within the FDA) in making its decision of approving orphan drugs. I recommend that Canada, like Australia, base its ODP on the American Orphan Drug Act because presently, Health Canada bases its decision for approval of ERT drugs on clinical drug trials that originate in the USA. As discussed in the literature review the main problem of approving ERT funding for the provinces/territories is the recommendation that comes from the Common Drug Review (CDR). Critics of the CDR lament that orphan drugs should not be assessed by the CDR but by "a distinct and appropriate process" where drugs for

orphan diseases can be assessed without inequities (CORD, 2007). Using the information that the OOPD publishes, orphan drugs would then have the opportunity of being properly assessed and funded by the provincial/territorial drug plans.

The literature review identified two ideas: the first idea being an ODP advocated by the MPS Society of Canada and CORD; and the second idea being a *Catastrophic Drug Plan* identified in both Romanow (2002) and Kirby (2002). My recommendation would be to continue to lobby for an ODP and follow Australia's lead by basing a Canadian ODP on the American Orphan Drug Act. The ODP would be a more comprehensive and specific policy than the Catastrophic Drug Coverage Plan in that an ODP would specifically dedicate monies to research and development in the orphan diseases sector. Patients and their families would have more security in knowing that there would be funding for their children to receive ongoing ERT treatments apart from having to rely on the pharmaceutical companies' drug trials. Another advantage of an ODP would be that Canada would be able to establish the pricing of these drugs because these orphan drugs would be developed in Canada and not imported.

The Canadian Organization of Rare Diseases (CORD) has developed a position platform entitled "A Chance for Life Fund" (see Appendix E) reinforcing their stand on the development of an ODP for Canada. On February 29, 2008, CORD hosted the 1st Annual International Rare Disease Day in Ottawa, Ontario, Canada. Patients, family members, and friends of people with orphan disorders met with more than 60 Members of Parliament and the Canadian Senate to raise awareness of the inequities in healthcare available to Canadians who suffer from orphan diseases (CORD, 2008). The MPS Society of Canada (which

supported this study and assisted with recruitment) had representation at this event. In addition, one of the speakers at this event was a child of one of the participants of this study.

Gaining an understanding of the lived experience of parents caring for children diagnosed with MPS and waiting for federal drug approval and provincial/territorial drug funding is beneficial because it gives an insight into the social inequalities that restricts a person from reaching their full potential (McEwan & Wills, 2002). This lived experience will bring awareness of constraints and power imbalances in society that affect areas such as access to care, care of the chronically ill, and access to orphan drugs.

Implications for Nursing Practice

When caring for MPS affected children, nurses informed of the constant worrying of waiting for ERT funding by the family will enhance the nurses' understanding of the stress experienced by the family and aid the nurses in their interaction with the family by being sensitive to the family's needs. Nurses have a responsibility to take this factor into consideration and attempt to support and understand a parent's worry and anguish in watching their child deteriorate physically and mentally, leading to a diminished quality of life. For example, nurses may not be able to fully understand the degree of frustration exhibited by parents when they are at a hospital visit and are told they will need to *wait* for their child's treatment to commence because the pharmacy department happens to be running late that particular day. Being aware of the continual *waiting* parents have dealt with up to that point will assist the nurse in supporting the parent and attempting to make their treatment visit as efficient as possible such as making sure that nursing procedures are completed on a timely basis such as insertion of peripheral intravenous lines or accessing port lines.

As MPS is a chronic illness, the relationship between the parent and the nurse is an important one. The establishment of trust between the nurse and child/parent/family is paramount for a caring relationship; therefore, communication is vital. Practicing family-centred care is essential, making sure to inform, discuss and plan the child's plan of care with the parents and if possible with the child. The nurse needs to remember that the parents and the child are the *experts* when it comes to MPS and having the family and the child express what are their priorities and necessary in the plan of care is of utmost importance. This type of communication intensifies the feeling of trust between the family and the nurse in that the family feels that the nurse values their input and that they are equal partners in the care of their child. The child and the family will be making the weekly trek to the hospital over many years, either until the child is transferred to an adult facility or to a clinic closer to their home. The nurse needs to be aware of the time, energy and turmoil the family experiences on a weekly basis of traveling to and from the hospital for the ERT and acknowledging to the parent that it is difficult to *put life on hold* in order to care for their child. The nurse can assure that the child's and parent's favourite spot in the clinic is available when the child comes in for the infusion to provide a sense of comfort and familiarity for the family. Other comfort measures may include having available the child's favourite movie, the child's favourite meal at lunch and the child's favourite game or arts and crafts activity. If the child needs to visit with other health care professionals every attempt should be made to schedule the appointments together as to not make the infusion day too disruptive or extra long.

It is essential for nurses to assess the needs of individual families and assist parents with acquiring resources and support. Because of the complexity of care that MPS affected children require, the nurse should seek collaboration from all other team members, such as

social work, physiotherapy and the child life therapist. Social work would be able to assist in acquiring lodging if they have relocated to be closer to a regional paediatric centre, and assist the family in exploring and applying for social services such as home care, drug plans and assistive devices. In collaboration with the social worker, the occupational therapist would assess the child for needs in the home such as wheelchairs, bathing and other mobility aids. The physiotherapist can educate parents about range of motion exercises for their child to assist with mobility. The child life therapist would be a welcome addition to the health care team in that they would engage the child in play therapy and alleviate stress for both the child and the parent during their visits to the hospital for ERT.

Nurses can link parents to organizations and resources in the community such as The MPS Society of Canada and CORD. These organizations can link MPS families together and can provide a sense of community for parents and siblings. Nurses could also seek resources from these organizations to learn more about the disease and the complexity of care that is required at home involving the family. Being aware of these advocacy groups and their campaigns in the community may inform the nurse about health policy and may lead the nurse to take a more active role in advocacy for patient care.

Implications for Further Research

At the present time the body of knowledge on MPS is focused on the medical aspect of the disease. Clinical trials with ERT have been ongoing for numerous years and there has been demonstrable improvement in children who have received the treatment; however, there have been no studies focusing on the holistic view of the child and their family. This exploration could lead to further research including investigation into the child's perspective of having this chronic debilitating illness, how they perceive themselves (self-image), and

what they feel about ERT. Further studies could also include research into parents' beliefs and values regarding genetic screening and counselling, and prenatal screening and diagnosis. The findings in these future studies on MPS would be beneficial not just to gain a greater understanding of the lived experience of patients and families living with MPS specifically but, they could explore fundamental beliefs, attitudes and perspectives of individuals living with a maladaptive genetic condition. These findings could inform a further understanding of the lived experience of other patients and families living with similar circumstances. The benefit to society would be a more nuanced understanding of the needs of such individuals and, so, a more informed and strategic approach to developing appropriate health and public policy could be developed.

Parents with children diagnosed with MPS residing in Canada live with the constant fear of having ERT funding terminated because the F/P/T governments are still negotiating how ERT will be funded and by whom. Although there are studies focusing on parental experiences with other chronic conditions, this study gave parents with children affected with MPS an opportunity to tell their stories and provided a detailed picture of what families must contend with while living with the Canadian government policies on orphan drugs. This experience is unique to parents in Canada as it describes their experiences of having to live out the policies set by the federal and provincial/territorial governments.

Limitations

The findings of this study reflect the lived experiences of interviewed parents of children with MPS receiving ERT at this particular time, for this particular group, in Canada, and therefore are not transferable to other populations. These findings are not transferable to parents of children with MPS waiting to begin ERT infusions because the children that would

commence ERT infusions presently would eventually be granted the funding on a compassionate basis and would be guaranteed the drug indefinitely. The findings in this study were also the view of one parent in the parental dyad (there was only one parent dyad that participated and they were interviewed separately). Interviewing the other parent may have provided another perspective within the family unit and proved an opportunity to compare mothers and fathers experiences.

Conclusion

This study explored the lived experience of parents with children diagnosed with MPS waiting for federal drug approval and provincial/territorial drug funding of ERT. Descriptive phenomenology was the method that guided this study and Critical Social Theory was the lens I used to illuminate this group's marginalization within the social and political environment. This allowed me to provide a detailed description of what it was like for parents whose children are diagnosed with an orphan disease that must wait many months and sometimes years before their children receive the appropriate drug treatment. Four themes emerged from this descriptive phenomenological study that describes the parent's experiences of waiting and include: *A New Journey; A New Reality; A New Optimism* and *A Road to Nowhere.* Also, given the frequency with which these families must interface with the health care system in all stages from pre-diagnosis, diagnosis and to treatment, this study illuminates the need for an Orphan Drug Policy in Canada and the important role nurses can play to support children and families.

APPENDIX A

RYERSON UNIVERSITY

The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis (MPS) Waiting for Federal Drug Approval and Provincial Drug Funding for Enzyme Replacement Therapy (ERT)

Dear Parent,

My name is Maria Maione and I am a Registered Nurse studying for a Master of Nursing degree at Ryerson University in Toronto. I have chosen to study the experience of parents/legal guardians with children diagnosed with MPS waiting for federal drug approval and provincial/territorial drug funding for Enzyme Replacement Therapy (ERT).

I am a pediatric nurse and became interested in this topic after caring for children undergoing ERT. I have cared for these children for the past 5 years and I have worked with families who have gone through the ups and downs of access to treatment for their child.

The purpose of my research study is to explore and describe a parent's every day experience of caring for a child who has Mucopolysaccharidosis (MPS) and the parent's experience of waiting for federal drug approval and provincial drug funding for Enzyme Replacement Therapy (ERT).

Presently, knowledge of MPS is focused on the medical aspect of the disease. Drug trials with ERT have taken place over many years and there has been obvious improvement in children who have received the treatment; however, there have been no studies looking at the MPS affected child and their family as a whole. This study, *The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis (MPS) Waiting for Federal Drug Approval and Provincial Drug Funding for Enzyme Replacement Therapy (ERT)* will give parents a chance to voice their hopes, fears, and frustrations. Stress on the family unit have a disturbing toll on the health of everyone, which in the long run can lead to further need on the health care system and familial problems such as marriage difficulties, sibling rivalry and financial hardships.

The Research Ethics Board at Ryerson University and the Board of Directors at The Canadian MPS Society have both approved this study. This letter has been sent to you on my behalf by The Canadian MPS Society to inform you about participation in this study.

Your story will be unique and your voice and your story is what are valued and most important to me and that is why I am approaching you.

I would like to interview 5 to 10 parents/legal guardians for this study. If you decide to participate, you would be interviewed at a prearranged time and place of your convenience. Interviews will be tape-recorded and will last approximately 30 to 60 minutes. Please be assured that your interviews would be kept strictly confidential. There would be no

identifiers that link you to any specific responses. The information that you provide would be maintained in a locked cabinet, and accessible only to the researcher. Participation is voluntary and you would be able to withdraw at any time. Withdrawal from the study will not jeopardize your present or future relationship with either The MPS Society of Canada or Ryerson University.

If you are interested in participating please contact me at 416-631-8323 or mmaione@ryerson.ca and I can provide more details about the study and answer any questions you may have before you decide to participate or not. Also, if you know of another parent/legal guardian who may be interested in participating please have him or her contact me.

Thank you for considering my request.

Sincerely,

Maria Maione, RN, BScN, (Master of Nursing student)
Ryerson University

RYERSON UNIVERSITY

Consent Agreement

Study Title: The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis (MPS) Waiting for Federal Drug Approval and Provincial Drug Funding for Enzyme Replacement Therapy (ERT).

You are being asked to participate in a research study. Before you give your consent to be a volunteer, it is important that you read the following information and ask as many questions as necessary to be sure you understand what you will be asked to do.

Investigators:

Maria Maione, RN BScN (Master of Nursing student)
Ryerson University
416-631-8323
mmaione@ryerson.ca

Dr. Karen Spalding, RN PhD
Thesis Supervisor
Ryerson University
416-979-5000 Ext. 6307
kspaldin@ryerson.ca

Thesis Committee Members:

Dr. Sherry Espin, RN PhD
Ryerson University
416-979-5000 Ext. 7993
sespin@ryerson.ca

Dr. Nancy Walton, RN PhD
Ryerson University
416-979-5000 Ext. 6300
nwalton@ryerson.ca

Purpose of the Study:

The purpose of this study is to explore and describe your every day experience of being a parent of a child who has Mucopolysaccharidosis (MPS) and your experience of waiting for federal drug approval and provincial drug funding for Enzyme Replacement Therapy (ERT).

Presently, knowledge on MPS is focused on the physiological aspect of the disease. Drug trials with ERT have taken place over many years and there has been obvious improvement in children who have received the treatment; however, there have been no studies looking at the MPS affected child and their family as a whole. This study, *The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis (MPS) Waiting for Federal Drug Approval and Provincial Drug Funding for Enzyme Replacement Therapy (ERT)* will give parents a chance to voice their hopes, fears, and frustrations. Stress on the family unit may have a disturbing toll on the health of everyone, which in the long run may lead to further need on the health care system.

I will be seeking to interview 5 to 10 parents who are willing to share their story with me.

The findings from this study will be used for my Master's thesis. Upon completion, a major research paper will be produced. Findings may be used for publication or presentation purposes.

Description of the Study:

Once ethics approval was gained from the Ryerson University Research Ethics Board (REB), The MPS Society of Canada allowed me to access parents such as you to participate in the study. At most 10 participants will be interviewed at a prearranged time and place, preferably at the parent's home or via telephone. Interviews will be tape-recorded and will last approximately 30 to 60 minutes. Your informed consent will be obtained at the start of the interview.

The following are examples of some of the questions I will be asking you:

- What has been your experience of waiting for drug approval and drug funding for Enzyme Replacement Therapy.
- What has been your experience in dealing with the provincial government with regards to funding Enzyme Replacement Therapy?
- What were your feelings when you learned your child was selected to take part in a drug trial?
- What impact has Enzyme Replacement Therapy had on your child's life and on your own life?

What is Experimental in this Study?

There are no experimental procedures used in this study. I will be gathering your stories for the purpose of examining them in detail in order to understand your experiences better and/or to discover more about your experiences and to note if there are any similar themes present amongst the stories.

Risks or Discomforts:

Parents volunteering to participate in the study will be asked to tell their stories about their every day experience of being a parent of a child who has Mucopolysaccharidosis (MPS) and your experience of waiting for federal drug approval and provincial drug approval for Enzyme Replacement Therapy (ERT).

Prospective informants who have extremely busy schedules and/or are trying to balance multiple personal and work life demands may feel stress at the thought of taking time to participate in interviews. To minimize stress, no coercion will be involved in the recruitment or participation of parents. Your involvement in all phases of the research project is optional. Your choice of whether or not to participate will not have an effect upon your current or future relations with Ryerson University and/or The Canadian MPS Society.

Because of the personal nature of the questions being asked, you may find that you are reflecting upon unpleasant memories while responding to questions during the interview. At any time, you may refuse to answer a particular question or stop participation altogether. You may also request a break at any time during the interview.

Benefits of the Study:

I cannot guarantee that participating in this study will result in any direct benefit to you. However you may find satisfaction in being given a chance to voice your concerns and tell your stories.

Additionally, there may be broader benefits to having these stories told through a study, publication and presentations, such as increased awareness of the challenges of parenting a child with MPS and the challenges of waiting for federal drug approval and provincial drug funding for ERT.

Confidentiality:

The consent will be e-mailed or mailed to you. When you have read the consent I will answer any questions you may have. At the interview session we will review the consent form together and I will ask you to sign the consent form. If the interview is held over the telephone, I will ask you to return the consent via mail prior to the interview in a self-addressed stamped envelope. A copy of the consent will be given to you to keep for your records.

Methods to ensure confidentiality will be as follows:

1. As a participant, you will be given a code. This code will be the only thing that links your signed consent form to your transcribed interview.
2. All audiotape equipment, audio recordings and transcribed interviews will be kept locked in a file cabinet or desk in the researcher's home office. Consent forms with your signature will be kept separately and in a secure place.

3. Data that is stored on the researcher's computer will be password protected.
4. Names of parents and children will be not be used in any publications or reports.
5. The researcher and the researcher's thesis committee members will be the only people that will have access to the data. Thesis committee members will not have access to the audiotapes and will only be viewing the coded transcripts.
6. The audiotape recordings will only be used to record the interview session and then to write out the stories onto paper. Audiotapes will be stored until completion of the final research paper and will then be destroyed. Transcriptions of interviews will be kept for a period of two years after completion of the graduate degree. The expected date of thesis completion is November 2007.
7. You will be given the opportunity, if you wish, to review, edit and provide feedback on your transcribed story prior to any publication. You will also have an opportunity to request a copy of the final research paper, if you wish.

Incentives to Participate:

All participants will be paid to participate in this study.

Costs and/or Compensation for Participation:

A monetary award of $50 will be given to you at the end of the interview session to cover for babysitting costs, travel and any other incidental costs you may encounter including your time.

Voluntary Nature of Participation:

Participation in this study is voluntary. Your choice of whether or not to participate will not have an effect upon your current or future relations with Ryerson University and/or The Canadian MPS Society. If you decide to participate, you are free to withdraw your consent and to stop your participation at any time without negative consequences. At any particular point in the study, you may refuse to answer any particular question or stop participation altogether. Again choosing to do so will have no negative consequences for you.

Questions about the Study: If you have any questions about the research now, please ask. If you have questions later about the research, you may contact:

Maria Maione, RN, BScN (Masters of Nursing student)
416-631-8323
mmaione@ryerson.ca

Or

Karen Spalding, RN PhD
Thesis Supervisor
416-9795-5000 Ext. 6307
kspaldin@ryerson.ca

If you have questions regarding your rights as a human subject and participant in this study, you may contact the Ryerson University Research Ethics Board for information.

Research Ethics Board
c/o Office of Research Services
Ryerson University
350 Victoria Street
Toronto, ON M5B 2K3
416-979-5042

Agreement:

Your signature below indicates that you have read the information in this agreement and have had a chance to ask any questions you have about the study. Your signature also indicates that you agree to be in the study and have been told that you can change your mind and withdraw your consent to participate at any time. You have been given a copy of this agreement.

You have been told that by signing this consent agreement you are not giving up any of your legal rights.

_____
Name of Participant (please print)

_____ _____
Signature of Participant Date

_____ _____
Signature of Investigator Date

Audiotape Agreement:

By signing below, you also indicate that you agree to be audio taped. You understand that your interview will be transcribed verbatim and audiotapes will be kept until thesis completion:

_____

Name of Participant (please print)

_____ _____

Signature of Participant Date

_____ _____

Signature of Investigator Date

I wish to be contacted at a later date in order to have an opportunity to revise, edit or provide feedback on my transcribed story.

I wish to be contacted at a later date in order to receive a copy of the final research paper, upon completion.

Contact information:

_____

_____

_____

_____

APPENDIX C

THE LIVED EXPERIENCE OF PARENTS WITH CHILDREN DIAGNOSED WITH MUCOPOLYSACCHARIDOSIS WAITING FOR FEDERAL DRUG APPROVAL AND PROVINCIAL DRUG FUNDING FOR ENZYME REPLACEMENT THERAPY

INTERVIEW GUIDE – DECEMBER 2006

Objective: To explore how parents/legal guardians of children diagnosed with MPS have dealt with the waiting for federal drug approval and provincial drug funding for ERT.

Questions and prompts:

1. What has been your experience of waiting for drug approval and drug funding for ERT.

2. What has been your experience in dealing with the provincial government with regards to funding ERT?
    a. Do you think the province is acting in good faith in trying to approve funding for ERT?
    b. Have you had the opportunity to meet with and make your opinions known to the provincial health minister?
3. How would you describe your experience as a parent of a MPS affected child?
    a. What kind of support do you have?
    b. Do you seek support from advocacy groups?
4. How did you feel when you were told your child had MPS?
5. How did you feel when your child was considered to take part in a clinical trial?
    a. Did you feel like your child was given a lifeline?
    b. What type of questions/concerns did you have in regards to the clinical trial?
    c. Did you have to travel to take part in this trial?
6. What impact has ERT had on your child's life and on your own life?

a. Has your child shown improvement with ERT?

b. How long has your child received ERT?

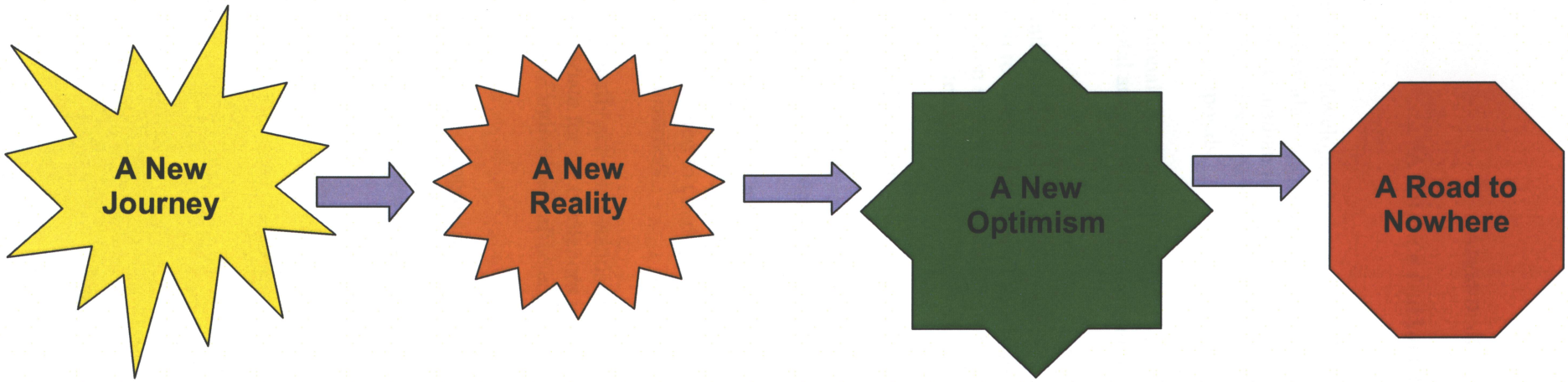
7. Imagine the Minister of Health is sitting across from you. What would you say to him/her?

Appendix D

The Lived Experience of Parents with Children Diagnosed with Mucopolysaccharidosis waiting for Enzyme Replacement Therapy

A Conceptual Framework

Themes and Essences



Essences

- Persistence
- Parents' feelings of belittlement
- A lack of Holism

Essences

- Double-edged Sword
- Guilt
- Protection of Child

Essences

- Hope
- Persistence
- Fear of the Unknown
- Putting lives on hold
- Regret

Essences

- Betrayal, frustration & fear
- "Government's fiduciary responsibility"

Waiting

APPENDIX E

"A CHANCE FOR LIFE FUND"

Canada is one of the only developed countries in the world that does not have an orphan drug policy. Not only does Canada not support research and development into treatments for rare and neglected disorders, Canadian patients with rare disorders often do not have the same access to life-saving therapies as patients in other countries. CORD requests immediate action on the following proposals.

- Establish a national (federal/provincial/territorial) "Chance for Life Fund" equivalent to 2% of the total annual public drug expenditure to be designated for therapies for rare disorders.

- Establish a multi-stakeholder Advisory Body, including treaters (medical caregivers) and patients, to recommend treatment access for life-threatening or serious rare disorders based on scientific standards and social values (humanitarian, ethical and compassionate criteria).

- Establish Centers of Reference for specific rare disorders, comprised of national and international experts, who will develop criteria for treating patients based on scientific evidence and patient impact and provide on-going surveillance into the real-world safety and effectiveness of these treatments on individual and group basis.

- Provide incentives through Orphan Drug Regulation and policy equivalent to those in the United States and European Union to assure Canadian organizations and researchers are motivated to conduct research and development into treatments for rare and neglected disorders.

- Ensure internationally accepted standards for conduct of clinical trials in rare disorders appropriate for the challenges inherent to very small patient populations (i.e. low number of individuals affected, limited long term data, lack of validated measures, etc.).

- Ensure Health Canada's progressive licensing framework provides appropriate support to the design of clinical trials for very small patient populations and appropriate review of evidence submitted from these trials.

http://www.raredisorders.ca/index.php

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