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The Parent Experience of Caring for a Child with Mitochondrial Disease

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Mitochondrial disease is a spectrum of progressive genetic disorders resulting from dysfunctions of cellular metabolism in the mitochondria that greatly compromise the lives of affected individuals, who are often children.
Purpose: This study described the parent experiences unique to caring for a child with mitochondrial disease.
Methods: Internet surveys were made available to parents of children with a known mitochondrial disease. Surveys included demographic items and two questionnaires: Parent Experience of Child Illness (PECI) and Pediatric Inventory for Parents (PIP). Descriptive data were collected and correlations calculated to determine relationships between the parent experience and stress.
Results: The majority of participants (n = 231) were mothers (95%) of children with mitochondrial disease around the age of 10 years (M = 9.85). Elevated scores were found in parent adjustment illness-related concerns regarding Guilt and Worry (M = 2.30, SD = .650), Sorrow and Anger (M = 2.09, SD = .730), Long-term Uncertainty (M = 2.56, SD = .690), and Emotional Resources (M = 2.36, SD = .615). Scores indicated elevated feelings of stress in terms of both difficulty and frequency. Significant correlations ($p < 0.01$) were found between parent illness-related concerns and parenting stress.
Conclusions: The results of this study suggest that parents of a child with mitochondrial disease feel a burden of responsibility that exceeds the typical caregiver role, see their child as fragile, and have concerns about their child's future. Identification of these concerns can assist nurses to better meet the needs of these parents and families.

MITOCHONDRIAL DISEASE IS not a single disease, but rather a spectrum of genetic disorders that result from problems with cellular metabolism in the mitochondria (Haas et al., 2007). Mitochondria are the "powerhouses" of the cell, responsible for creating more than 90% of energy needed to sustain life (United Mitochondrial Disease Foundation, 2011). Mitochondrial disease is caused by mutations in genes that encode proteins critical to mitochondrial function. Failure of the mitochondria to perform normally results in decreased energy production, cell injury or cell death, and eventual organ dysfunction or failure. Receiving the news of a mitochondrial diagnosis for a child can cause feelings of uncertainty and worry in parents as they navigate a complex health care system seeking answers to an equally complex condition. Little is known about the impact of parenting a child with a mitochondrial disease. The purpose of this study was to describe the lived experience of parents caring for a child with mitochondrial disease.

Understanding Mitochondrial Disease

Mitochondrial disease is considered to be a rare disease (Office of Rare Disease Research, 2007), with a reported prevalence of 1 in 3,000 to 5,000 children before the age of ten years (Haas et al., 2007; Kisler, Whittaker, & McFarland, 2010). However, it is estimated that 1 in 300 people may

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have a genetic mutation that could affect mitochondrial function (Codier & Codier, 2014). Mitochondrial diseases vary considerably in clinical manifestation, effects on organs, age of onset, and rate of progression (Noorda et al., 2007). Symptoms may appear at any age. These diseases are often progressive and debilitating, most often harming organs with high energy demands such as the central nervous system, skeletal muscle, heart and endocrine system (Dassler & Allen, 2014). Establishing a diagnosis of mitochondrial disease is a complex process that may take years. The life of the individual with mitochondrial disease can be greatly compromised, according to the degree and nature of mitochondrial malfunction.

Mitochondrial function is determined by hundreds of proteins; each protein is encoded by a particular gene. A mutation in any of those genes may cause its encoded protein to be defective, impairing mitochondrial function according to the protein's role in energy production. A variety of disease manifestations can result. Children with mitochondrial disease may display muscle weakness and/or muscle pain, exercise intolerance, heart failure or rhythm disturbances, movement disorders, dementia, stroke-like events, seizures, gastric reflux, severe vomiting, failure to thrive, constipation, diarrhea, blindness, deafness, droopy eyelids, diabetes, swallowing difficulties, heat and cold intolerance, poor growth, developmental delays, susceptibility to infection, lactic acidosis, and problems with the immune system, heart and kidneys (United Mitochondrial Disease Foundation, 2011).

The serious nature and erratic, complex clinical presentation of mitochondrial disease make it difficult to establish a diagnosis and contribute to increased stress and worry among parents and caregivers of these children. In general, parents of children with chronic illness report increased frustration and uncertainty about the origins of their child's suffering during the pre-diagnosis phase of an illness (Finnvold, 2010). The benefit of receiving a diagnosis extends beyond receiving specialized treatment, also affording parents the ability to label the condition and better cope with the situation (Finnvold, 2010).

Parents need support from health care professionals, including nurses, to manage the many demands and stressors of this set of rare diseases. Little is known, however, about the lived experience of caring for a child with a mitochondrial disease. By understanding the parental experience, nurses can become sensitive to the issues these families face and provide a therapeutic presence with personalized and holistic care.

Mitochondrial Genetics

Mitochondria are tiny cellular organelles found in the cytoplasm of the cell. Consistent with their role in energy production, they are more plentiful in cells with high energy demands. Mitochondria are distinctly different from other organelles because they have their own DNA (Kisler et al., 2010). Mitochondrial DNA (mtDNA) contains genes that encode a number of proteins critical to mitochondrial function; a mutation in one of these genes can cause mitochondrial disease. The mitochondria in a single cell may not be genetically identical, a phenomenon known as heteroplasmy. Therefore, it is possible for a cell to contain both healthy and genetically defective mitochondria. When such a cell divides, the mitochondria segregate randomly into the daughter cells, so that the ratio of healthy to damaged mitochondria is not fixed. Heteroplasmy contributes to wide variability and limited predictability of signs and symptoms in mitochondrial disease (Saneto & Sedensky, 2013).

Mutations in mtDNA are not the only cause of mitochondrial disease, however; DNA in the cell nucleus (nuclear or nDNA) also contains genes necessary for mitochondrial function. A cell's ability to generate enough energy to support normal cellular activities therefore depends on functional genes in both types of DNA, and mutations in any of these genes can cause mitochondrial disease (Dassler & Allen, 2014). Disease-causing mutations can occur spontaneously or be inherited from one or both parents. The inheritance pattern depends on location of the mutation.

Because a child inherits mitochondrial DNA only from the mother, mtDNA mutations follow a maternal inheritance pattern (Codier & Codier, 2014). A mother with a mtDNA mutation will pass that mutated gene to all of her children. Not all of her children, however, will necessarily become symptomatic, and each affected child may display different disease manifestations (Boles & Mason, n.d.).

When mitochondrial disease is caused by an inherited mutation in nDNA, it is most commonly transmitted in an autosomal recessive pattern (Boles & Mason, n.d). In this case, each parent has a mutated copy of the same gene but is not affected because they also have a normal copy of the gene. In these families, each child has a 25% chance of inheriting the defective gene from both parents and manifesting the disease (Boles & Mason, n.d). Unless families have had genetic testing, they may not know if their child's disease is due to a new or inherited mutation, or if it was inherited from one or both parents.

Clinical Presentation

Children with mitochondrial disease have a complicated clinical diagnosis. Children usually appear normal at birth or may demonstrate mild symptoms that are not always associated with a disease process (Read & Calnan, 2000). The variability in clinical manifestations, multiple organ involvement with rapid deterioration, and lack of a reliable biological marker make diagnosis difficult and frustrating to clinicians (Haas et al., 2007). An example of a disputed diagnosis concerning mitochondrial disease recently occurred at Boston Children's Hospital, when 16-year-old Justina Pelletier was placed in state child protective custody after being referred for a gastrointestinal problem related to her mitochondrial disease. Her "perplexing set of symptoms" divided specialists and raised suspicion of medical child abuse (Swidey & Wen, 2014). Justina remained in state