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Original Article

Stress and coping of parents caring for a child with mitochondrial disease

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ABSTRACT

Mitochondrial disease comprises a group of rare, genetic, life-limiting, neurodegenerative disorders known to affect children. Little is known about disease-related challenges, parental stress, and coping when caring for a child with a mitochondrial disease. Purpose: This study explored disease-related characteristics and parental stressors and coping behaviors related to caring for a child with mitochondrial disease. Methods: Internet surveys were posted on known mitochondrial disease websites for parent completion. Surveys included demographic items and two questionnaires: Coping Inventory for Parents (CHIP) and Pediatric Inventory for Parents (PIP). Descriptive data were collected and correlations used to determine relationships between parenting stress, coping, and demographic variables. Results: The majority of participants ($n = 231$) were mothers (95%) of children with mitochondrial disease around the age of 10 years ($M = 9.85$). On average, children had 6 organs involved ($M = 6.02$) and saw 7 different specialists ($M = 7.49$); 61% were hospitalized in the past year. Significant correlations ($p < 0.05$) were found between parenting stress and parent age, parent income, parent education, child age, child age at diagnosis, presence of developmental delays, number of hospitalizations, number of medical visits, number of organs involved, and number of specialists seen. Significant correlations were also found between parenting stress and coping behaviors such as family integration, social support and understanding health care. Conclusions: The ability to identify disease-related challenges, stressors, and coping strategies in parents of children with mitochondrial disease is novel and can assist nurses to provide disease-sensitive, family-focused care and improve child health outcomes.

Mitochondrial diseases comprise a group of rare, genetic, incurable, neurodegenerative disorders that cause metabolic energy disturbances resulting in progressive cell deterioration and organ failure (Haas et al., 2007). Mitochondrial diseases are genetic, i.e., they occur when mutations in nuclear or mitochondrial DNA disrupt the production of proteins necessary for energy production by the mitochondria. Symptoms can present in utero, at birth, in infancy or childhood, or later in adulthood (Codier & Codier, 2014). The parent experience of caring for a child with mitochondrial disease is unique. The serious and unpredictable nature of this illness likely presents an overwhelming challenge for families, especially parents who are the primary caregivers for their child. Parents need support to manage the many physical, cognitive and psychosocial demands and stressors of this rare, erratic, and debilitating disease. Very little is known about the stressors and coping behaviors of parents caring for a child with mitochondrial disease (Noorda et al., 2007). The study described in this paper explored the experiences of those parents by examining illness-related parenting stressors and coping behaviors unique to this population. Knowledge gained from this study can assist health care professionals to provide disease-sensitive, family-focused care and lead to better health outcomes for their children.

Although classified as a rare disease by the National Institutes of Health (Office of Rare Disease Research, 2007), high mortality rates and underreporting make the exact prevalence of mitochondrial disease elusive (Haas et al., 2007; Kisler, Whittaker, & McFarland, 2010). Evidence suggests a risk of developing mitochondrial disease anywhere from 1:3000 to 1:5000 before the age of ten years (Haas et al., 2007; Kisler et al., 2010). However, medical experts believe mitochondrial disease is more prevalent than once thought, with estimates that one in 300 people worldwide may have a genetic mutation that could potentially affect mitochondrial function (Codier & Codier, 2014).

0.1. Mitochondrial genetics

Mitochondria are cellular organelles responsible for energy production and are the only cellular organelles with their own DNA (mtDNA), distinct from the nuclear DNA (nDNA) that is present in every cell (Naviaux, 1997). Both mtDNA and nDNA contain genes that encode proteins required by mitochondria for energy production, and mutations in any of those genes can impair mitochondrial function (Kisler et al., 2010; Naviaux, 1997). Mutations in nDNA are more prevalent,

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accounting for 70% to 75% of mitochondrial disease in children, and are associated with earlier onset of symptoms, more severe symptoms, and earlier mortality than disease caused by mtDNA mutations (Balcells & Turco, 2008; Boles & Mason, (n.d.); Kisler et al., 2010). Inheritance of mitochondrial disease varies according to the type of mutation. Because paternal mtDNA is lost at fertilization, all mtDNA is passed from mother to child. Disease associated with mutation in mtDNA therefore follows a maternal inheritance pattern, while disease caused by mutation in the nDNA of either parent is transmitted in an autosomal dominant or recessive pattern. Occasionally, mitochondrial disease is not inherited at all but results from a new mutation in the child's DNA. Families who have not had genetic testing to identify the disease-causing mutation may not know the transmission pattern.

0.2. Diagnosis

Mitochondrial diseases are difficult to diagnose, lack biological markers, and present with nonspecific signs and symptoms (Haas et al., 2007). Children with mitochondrial disease typically appear normal at birth and develop mild developmental delays or subtle neurological symptoms that worsen over time (Read & Calnan, 2000). Mitochondrial diseases are usually progressive and typically involve organs with high energy demand, such as brain, central nervous system, muscles, heart, kidney, eyes, ears, endocrine, liver and gastrointestinal tract; however, any organ or tissue can be affected (Haas et al., 2007; Kisler et al., 2010). Generally, the involvement of three or more organs without a unifying diagnosis should raise suspicion for presence of mitochondrial disease, as should symptoms that worsen with an acute illness or present in atypical fashion (Haas et al., 2007, p. 1327; Read & Calnan, 2000). Mitochondrial diagnostic criteria comprise a combination of clinical symptoms, metabolic and imaging studies, skeletal muscle morphology and biochemical investigations of skeletal muscles (Wolf & Smeitink, 2002).

0.3. Treatment

No definitive treatment has been identified for mitochondrial disease. Strategies for disease management depend on the type and severity of mitochondrial involvement. Goals for managing mitochondrial disease range from preventing physiologic stress to acute symptom management (Parikh et al., 2009). Scientific support is growing for the use of vitamin supplementation as a means to promote mitochondrial health in the absence of a cure or a definitive way to halt disease progression, despite a lack of evidence that vitamin treatments are effective.

A multidisciplinary approach by health professionals is needed to address the complexity of care required to manage mitochondrial illness. Communication between parents and multiple health care providers is essential (Kisler et al., 2010). Families of children with mitochondrial disease not only travel long distances to obtain metabolic services, but they often see multiple specialists, requiring frequent trips between home, school, clinics and hospitals. Parents carry the burden of having a child with a chronic and potentially life-limiting condition, and many also suffer financial strain. At the same time they may be grieving the loss of the healthy child they once knew. The child's level of functioning may fluctuate from day to day, with continual care plan adjustments requiring close interactions between parents and health care professionals (Kisler et al., 2010).

1. Review of literature

The stress associated with mitochondrial disease is particularly challenging and requires successful coping behaviors. Previous research suggests that caring for a child with a chronic illness is predominately stressful for the primary caregiver, who in most cases is the mother (Ratliffe, Harrigan, Haley, Tse, & Olson, 2002). Therefore, it is useful to compare and contrast the experience of parents of a child with mitochondrial disease with that of parents who have children with other chronic diseases. When compared to mothers of children with PKU, another inherited metabolic disorder, mothers of children with mitochondrial disease suffered greater strain and worry, their children required a significantly greater number of health care services, such as the involvement of more specialists and more hospitalizations, and overall these families experienced increased financial burden (Read, 2003). Additionally, mothers of children with mitochondrial disease, when compared to parents of children with intractable epilepsy, had significantly higher caregiver burden and lower quality of life, with greater levels of depression and anxiety (Kim et al., 2010). Mothers of children with a mtDNA disorder also reported guilt and anxiety associated with possible maternal inheritance, influencing their perception of caregiver burden and ability to handle the stress of caring for a child with mitochondrial disease (Kim et al., 2010). These results strongly suggest a link between psychological distress and burden of care, although not causality; the authors recommend finding strategies to assess emotional elements of caregiving to ease anxiety, guilt and caregiver burden in mothers of children with mitochondrial disease (Kim et al., 2010).

Compelling evidence indicates that helping parents manage stress leads to better health outcomes for children with chronic disease. There is a strong relationship between parental stress and child health outcomes, with parent and child psychological reactions being significantly correlated; the higher the parental stress, the greater the level of psychological distress for the child (Grootenhuis & Bronner, 2009; Moola, 2012). A distressed parent is less emotionally available to his or her child (Santacroce, 2002). Family functioning and support, along with disease pathophysiology, prognosis, and severity of illness, influence the child's developmental outcomes (Allen, Vessey, & Schapiro, 2010). Parents spend much energy balancing medical care concerns, activities of daily living, financial obligations, and travel for medical appointments, all which have been associated with increased stress and decreased quality of life for parents (Grootenhuis & Bronner, 2009). Understanding the experiences of parents can help provide better health outcomes to children with chronic conditions and improve their quality of life (Moola, 2012).

Few studies have explored the needs and problems of children and families living with mitochondrial disease, particularly compared to the number of clinical and biochemical studies of mitochondrial disease (Noorda et al., 2007; Read, 2003). What has been demonstrated is that information about disease progression, parental use of coping behaviors, and the presence of support influence the family's ability to process information (Noorda et al., 2007). Therefore, health care professionals are charged with providing current, credible health information to parents of children with mitochondrial disease and assisting with planning effective care to improve quality of life for parents and children (Noorda et al., 2007). Understanding factors that contribute to or alleviate stress in parents of children with mitochondrial disease could provide health care professionals with the tools they need to develop interventions that support the needs of the mitochondrial disease family.

2. Methods

The purposes of this descriptive, exploratory and cross-sectional study were to 1) identify family demographics, disease-related characteristics, pediatric illness-related parenting stress and coping strategies in parents caring for children with mitochondrial disease, and 2) explore the relationships between family demographics, disease-related characteristics, pediatric illness-related parenting stress, and coping strategies in parents caring for children with mitochondrial disease.

This study was granted exempt status by the Institutional Review Board of Washington State University. Informed consent was obtained prior to beginning data collection. Permission to use webpages and social media for survey collection was granted by webpage administrators.