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Parent Recommendations for Family Functioning With Prader–Willi Syndrome: A Rare Genetic Cause of Childhood Obesity

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Purpose: Prader–Willi syndrome (PWS) is the most common genetic cause of childhood obesity. Neonates have hypotonia and may fail to growth and develop. Within a few years, behavioral problems occur along with insatiable hunger (hyperphagia) and the potential for excessive weight gain. The purpose of this study was to identify how families function when they have a child with PWS.
Design and Methods: This qualitative descriptive study was based on 20 face-to-face, audio-taped interviews with parents. They were asked to identify family responses to PWS and offer recommendations. Data were transcribed, coded and analyzed for commonalities and themes.
Results: There were twelve identified themes with two overarching themes of 1) taking action and 2) caring for oneself and family. Taking action was focused on achieving what was best for the child with PWS. Caring for oneself and family attempted to assure that all in the family were healthy, content, and living a fulfilling life under their circumstances.
Conclusions: This study revealed parental insight as to how they learned to creatively cope with a stressful family life. There was a recognition of inevitable sacrifice and the need for changes in expectations so as to help the child with PWS flourish while also focusing on the needs of all the members of the family.
Practice Implications: Nursing and health care providers should be aware of these issues in the provision of anticipatory guidance to families contending with this genetic disorder.

APPROXIMATELY 17% OF children and adolescents in the United States (U.S.) are overweight, representing over 12.5 million children (cdc.gov, 2015). Causes include poor nutritional habits, sedentary lifestyles, or underlying medical conditions, such as hypothyroidism. Prader–Willi syndrome (PWS) is a rare genetic disorder that often leads to excessive weight gain. It is the most common of genetic causes of severe childhood obesity. An individual with PWS goes through progressive stages of the disorder. As a newborn the child is typically hypotonic and fails to gain weight and grow. For unclear reasons, in a few years the child develops an insatiable hunger with weight control issues. Other problems include dysmorphic features, delayed growth and development, intellectual disability, and a range of behavioral problems. Prevalence is estimated at 1:15,000 (PWSUSA, 2015). This study explored how parents function with a child with PWS.

Review of Literature

Rare Diseases

Nearly 7,000 identified rare diseases affect over 6 million people in the U.S. A disease is termed "rare" if it affects < 200,000 people (European classification is 1 in 2000). The worldwide incidence of most rare diseases remains unclear. Classification,

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tracking, and records of disease progression and treatment are not commonly maintained. Afflicted families may be isolated from others with similar diseases (National Institute of Health & Office of Rare Diseases, 2015 (rarediseaseinfo.nih.gov).

Genetic Testing and Incidence

PWS was first described in 1956 by endocrinologists, Prader, Labhart, and Willi. It was recognized in 1981 as the first microdeletion syndrome. More recently it has been referred to as a genomic rather than a genetic condition because it involves genomic imprinting. There are 23 chromosomes that are normally inherited from each parent. The etiology of acquiring PWS has been identified as with chromosome 15. Children normally inherit one copy of chromosome 15 from each parent. PWS is attributed to the loss of function or deletion of specific genes in a particular region of chromosome 15. Parent specific gene activation or inactivation is caused by the phenomenon of genomic imprinting. There are four genetic forms of PWS that arise from either maternal or paternal causes. Approximately 75% of PWS cases result from an absence or failure of paternally inherited genes on the proximal long am of chromosome 15. An estimated 20% of PWS cases have two maternal copies of chromosome 15 (no paternal copies). The remaining 5% may occur as an abnormal translocation of chromosome 15. Re-occurrence within the same family accounts for only 1% of PWS cases. There are no gender or racial differences that have been identified. Incidence is 1:8,000–1:29,000 of live births. Estimates of population prevalence are 1:52,000–1:76,000. The incidence may be underestimated due to limited genetic testing (Chen, Visootsak, Dills, & Graham, 2007; McAllister, Whittington, & Holland, 2011; Pogson, 2012).

> Genomic Imprinting is the differential expression of a gene or genes as a function of whether they were inherited from the male or the female parent (e.g., a deletion on chromosome 15 that causes Prader-Willi syndrome if inherited from the father and causes instead Angelman's syndrome if inherited from the mother. Mosby's Medical Dictionary, 8th edition. © 2009, Elsevier. Genetics Home Reference: http://ghr.nlm.nih.gov/condition/prader-willi-syndrome.

Three Phases of PWS Development

Hypotonia characterizes the first phase. Neonates have poor sucking ability and as a consequence poor growth. They often can present as a failure-to-thrive. Dysmorphic features include a triangular mouth (thin upper lip/downturned mouth corners), almond shaped eyes, small hand/feet, hypogonadism and respiratory issues.

The second phase occurs 1–6 years later. The child is noted to have delayed speech and motor development. An insatiable appetite (hyperphagia) occurs with excessive weight gain if diet is not highly controlled. Other developments may include hypopigmentation, short stature, small hands and feet, early dental caries, strabismus, developmental delays, and maladaptive behaviors (Boyall, 2013; Chen et al., 2007).

The third phase continues in the older child with symptoms lasting throughout life. Hyperphagia remains present with ongoing developmental delays. Mild behavioral problems may range from obsessive compulsions to more troubling psychiatric manifestations. Mood stabilizing medications may be needed (Yearwood, McCulloch, Tucker, & Riley, 2011). Additional traits may include articulation difficulties, scoliosis, osteoporosis, delayed sexual development, infertility, and/or mild intellectual deficits. Diabetes and hypertension can result from uncontrolled weight gain (Chen et al., 2007. Strengths may be found within reading, vocabulary, or long-term memory and skills such as visual–spatial aptitude. Some children display an interest and proficiency in jigsaw puzzles or word search games. Ongoing insatiable hunger combined with an unusually low metabolic rate and decreased caloric requirement is common. An inability to vomit has been described in some children which can be dangerous if excessive intake has occurred (Chen et al., 2007; Boyall, 2013). Hypotonia contributes to reductions in physical activity. Weight control is a primary concern among associated co-morbidities such as diabetes and psychiatric disorders (Yearwood et al., 2011). Medications such as growth hormone are being used to help control obesity and improve motor development (Aycan & Baş, 2014).

Family Functioning

Family functioning is defined for the purpose of this paper as how family members provide essential aspects of family well-being such as support, love, caring, security, a sense of belonging, open communication, value, respect, and self-esteem in meeting the individual needs of all family members. This definition was drawn from normal family functioning by the American Academy of Pediatrics (2015). A review of literature on this topic does not reveal any directly related studies. Scales have been identified that measure aspects of functioning such as quality-of-life in other pediatric conditions. The goal of this study is not to measure but to identify how parents best function from their unique perspective.

Need for Research

Professional recommendations for managing behavior and food-related issues are available for parents. However, much less information is accessible concerning how parents and families actually cope with the challenges of PWS and the range of strategies they employ. This study has attempted to describe family functioning from a parental perspective and identify how parents have risen to their challenges.

Method

Purpose/Design

The purpose of this study was to describe how families function while living with a child diagnosed with PWS.