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In the Dark: Challenges of Caring for Sons with Klinefelter Syndrome

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The purpose of this mixed method study was to describe family management challenges for parents who have sons with Klinefelter Syndrome (KS). Standardized survey results showed that stress, quality of life and family management struggles varied by parent age. When interviewed, parents described feeling uninformed and without support to make decisions about managing their sons' KS. Parents reported that a lack of guidance and case coordination created barriers in caring for their sons throughout childhood. Given the prevalence of KS, health care providers need to be prepared to provide comprehensive evaluation and anticipatory guidance for KS boys and families.

KLINEFELTER SYNDROME (KS) is a common genetic condition that affects only males and is caused by the presence of an extra X chromosome. The prevalence of KS is approximately 1 in 450–600 male births (Bojesen, Juul, & Gravholt, 2003; Herlihy, Halliday, Cock, & McLachlan, 2011) and may be rising (Morris, Alberman, Scott, & Jacobs, 2008).

Physical characteristics and symptoms of KS are highly variable and include tall stature, wide arm span, small testes, androgen deficiency, breast development, and azoospermia (Simpson et al., 2003; Zeger et al., 2008). Most affected children appear normal at birth with phenotypic features becoming increasingly apparent during pubertal development when testicular failure typically begins (Wikstrom & Dunkel, 2008). KS is associated with language-based learning disabilities, (Graham, Bashir, Stark, Silbert, & Walzer, 1988; Rovet, Netley, Keenan, Bailey, & Stewart, 1996) as well as behavioral, (Ross et al., 2012) psychiatric (Geschwind, Boone, Miller, & Swerdloff, 2000) and psychosocial problems (Boks et al., 2007; van Rijn, Swaab, Aleman, & Kahn, 2008; van Rijn et al., 2013). A number of health risks are associated with KS, including diabetes, cardiovascular disease and osteoporosis (Bojesen, Juul, Birkebaek, & Gravholt, 2006). Reports also show that boys and men with KS suffer low self-esteem, poor quality of life and increased risk for depression compared to the general population. (Turriff, Levy, & Biesecker, 2011). KS can affect different aspects of health throughout the life span including both physical and psychosocial parameters. Little is known about how families of affected males manage the diagnosis. Parents of boys with KS are understandably concerned and confused about what to expect regarding the health of their affected sons. Since few studies of those with KS involve children, there is little evidence to guide clinicians or parents on how to care for affected boys or how to disclose and explain this condition to them during childhood and adolescence (Close, Smaldone, Fennoy, Reame, & Grey, 2013).

How parents think about a genetically-based chronic condition like KS may shape the way the family manages the needs of the affected child (Gallo, Hadley, Angst, Knafl, & Smith, 2008; Knafl, Knafl, Gallo, & Angst, 2007). The needs of a boy with KS often include a complex interplay among medical, pharmaceutical, psychosocial and educational

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management issues. For parents, management of their son's health is further complicated by the paucity of evidence-based guidelines regarding treatment and family support for KS. The purpose of this study was to describe family management challenges as parents try to meet the needs of their sons with KS.

Theoretical Framework

This study was guided by the Family Management Style Framework (FMSF) shown in Fig. 1 (Knafl, Deatrick, & Havill, 2012). The FMSF provides a structure for understanding how family members manage having a child with a chronic condition including how family members define, manage and perceive the consequences of their child's chronic health condition. The first aim of this study was to explore stress, family quality of life and family management style in parents of sons with KS. A second aim was derived from the "major component" sections of the framework. We described how parents define the situation of having a son with KS, their management of health, education and psychosocial issues and how they perceive the consequences of this condition on the family including stressors, worries and unmet needs. A summary of study variables with constructs found in the FMSF are shown in Table 1.

Design

We conducted a concurrent triangulated mixed method study to explore and describe the experiences of parents who have a son with KS. The qualitative approach was Interpretive Description (Thorne, Kirkham, & MacDonald-Emes, 1997). Interpretive Description incorporates the use of theoretical frameworks such as the FMSF, methods of sample selection and data analysis to conduct investigations into human health and illness experiences (Thorne, 2008; Thorne et al., 1997).

Methods

This study was approved by the Yale University Human Research Protection Program prior to data collection. All participants provided written informed consent. In depth semi-structured interviews and online questionnaires were conducted between December, 2012 and March, 2013.

Participants

A purposive sample of 40 parents participated in the study. Parents were recruited from a KS national advocacy association known as the Association of X and Y Chromosome Variations ©. Participants were eligible if they were English-speaking, had a KS-affected son between the ages of birth and 26 years and had access to a telephone and/or a computer. Participants were chosen using maximum variation to reflect the spectrum of parents who had sons of varying ages, differing levels of symptom severity, length of time since diagnosis, timing of diagnosis and parents whose sons were informed or not informed about their diagnosis. Sample size was determined based upon purposive sampling used in mixed methods research (Teddlie & Yu, 2007). The sample allowed for the minimum sample size required to detect differences in the survey data and to accommodate the sampling strategy for collection of interview data. Using perceived stress scores as a primary outcome measure with a two-tailed t test, 80% power with alpha at .05 would require a sample size of 33 participants assuming a large effect size.

Semi-Structured Interview Guide

Using the FMSF and a review of the literature on families who have children with genetic conditions, we developed a semi-structured interview guide that focused on the parent experience of having a son with KS. The guide consisted of items developed from the "Components" section of the FMSF concerning parental experience of raising a son with KS, information management, management of health and social issues and the perceived consequences of having a son with KS. Sample interview questions are shown in Table 2.

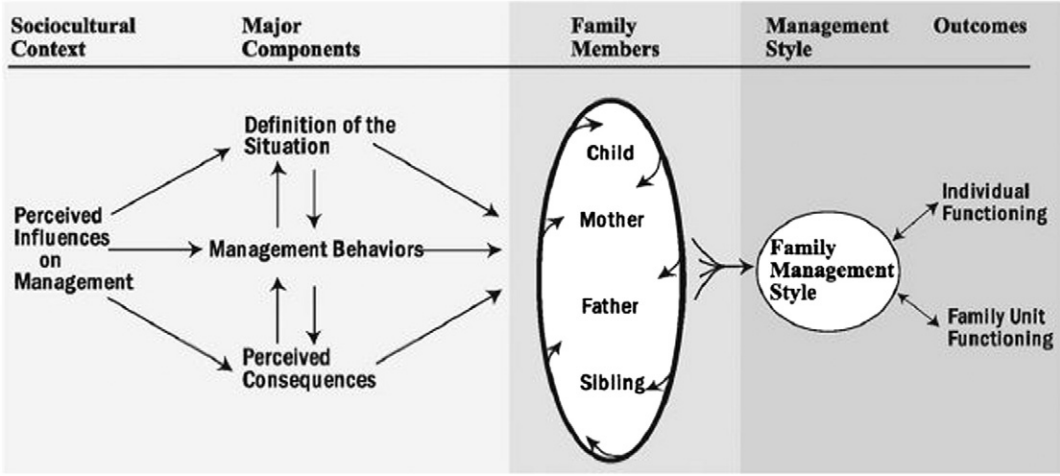

Figure 1 The Family Management Style Framework reprinted with permission from Knafl, K, Deatrick, J. and Havill, 2012 in the Journal of Family Nursing 18 (1), 11–34.