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"It Was an Overwhelming Thing": Parents' Needs After Infant Diagnosis With Congenital Adrenal Hyperplasia[1]

Kyla L. Boyse MS, RN, CPNP[a,*], Melissa Gardner MA[b], Donna J. Marvicsin PhD, PNPBC, CDE[a], David E. Sandberg PhD[b,c]

[a]*School of Nursing, University of Michigan, Ann Arbor, MI*
[b]*Child Health Evaluation and Research (CHEAR) Unit, University of Michigan, Ann Arbor, MI*
[c]*Division of Child Behavioral Health, Department of Pediatrics and Communicable Diseases, University of Michigan, Ann Arbor, MI*

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This study characterizes the experiences and expressed needs of parents following diagnosis of their newborn with congenital adrenal hyperplasia (CAH). Six parents (four mothers and two fathers) were interviewed about how they learned about CAH and its management, followed by qualitative data analysis. Coding of transcripts revealed several themes, including health communication problems, a lack of medical home and decision support, and a desire for parent-to-parent social support. Findings have implications for how family-centered health care is delivered following an unexpected newborn diagnosis.

> "Most families enter the world of illness and disability without a psychosocial map to navigate the normative (i.e., common and expectable) strains" (Rolland & Walsh, 2006, p. 529)

Background

Parenting a newborn with a complex health condition presents special challenges. The process of establishing the parent–child relationship in infancy can be complicated by a neonatal intensive care unit admission and uncertainties about future growth and development. How parents adapt ultimately affects the child's and family's well-being (Fisher, 2001; Rolland & Walsh, 2006). Contextual factors, including the family's interactions with the child's health care providers and their personal social network, influence how parents accomplish adaptive tasks (Knafl, Deatrick, & Havill, 2012).

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* Corresponding author: Kyla L. Boyse, MS, RN, CPNP.
E-mail address: kylaboys@umich.edu.

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Newborn screening (NBS) represents a major accomplishment in health care implementation. Current recommendations for all U.S. states and territories are to screen for a uniform panel of 31 conditions (Watson, Mann, Lloyd-Puryear, Rinaldo, & Howell, 2006; U. S. Department of Health and Human Services, Health Resources and Services Administration, Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, 2013). With the benefits that accompany early detection of serious medical conditions also come the challenges to parents of having their infant unexpectedly diagnosed, shortly after birth, with a life-threatening or life-limiting condition. Ideally, the medical home would coordinate the care of infants identified through NBS and work with specialists to assist the family in understanding and managing their child's condition (American Academy of Pediatrics, Newborn Screening Authoring Committee, 2008). However, in reality, primary care physicians do not always feel prepared to manage follow-up after a positive NBS (Kemper, Uren, Moseley, & Clark, 2006). As a result, positive adaptation for these infants and families may be compromised (McAllister, Sherrieb, & Cooley, 2009).

One of the conditions in the uniform NBS panel is congenital adrenal hyperplasia (CAH), which is typically identified through NBS and confirmatory diagnostic testing or, in the case of affected girls, due to masculinization of the genitalia. "Classic" CAH, inherited as an autosomal recessive genetic disorder, affects males and females in equal numbers and is potentially life-threatening. Mutations of genes for enzymes that mediate the biochemical steps of adrenal gland steroidogenesis result in insufficient cortisol and aldosterone production, and excessive androgen production (Speiser et al., 2010). The resulting hormonal imbalances, if untreated, can lead to electrolyte imbalances, dehydration, vomiting, cardiac arrest and shock. Incidence of the classic form of CAH ranges from 1:10,000 to 1:20,000 births (Speiser et al., 2010). Medical treatment of CAH involves hormone replacement of glucocorticoids and mineralocorticoids. Clinical management has been described as a "difficult balance between hyperandrogenism and hypercortisolism" (Speiser et al., 2010, p.4140). Yet, when a newborn is diagnosed with CAH, parents are expected to quickly learn about the required routine and emergency medical management so they can care for their child at home. Parents of girls may also experience pressure to decide whether to opt for genital surgery.

Although much is known about parents caring for children with special health care needs in general, little is known about parents' experiences and needs in learning to care for their infant after his or her unexpected diagnosis with CAH. The aim of this qualitative study was to characterize early parent caregiver experiences and needs in CAH, with a focus on contextual factors that can be modulated through education and support delivered by health care providers or via the NBS system.

Method

Participants

Four mothers and two fathers from four separate families agreed to participate in, and completed, a larger study to pilot Web-based educational resources for parents of newborns and young children identified by NBS. Participants were recruited through one state's NBS program via an invitation letter sent to families by the medical director of the program. At the time of enrollment, two of the four families also had an older child with CAH. The first affected child (two girls, ages 5 and 6 and two boys, ages 8 and 11) was the focus of the current study in order to investigate the parents' experiences when CAH was completely unfamiliar to them.

Five parents identified as "White" and "non-Hispanic," and one as "Asian or Pacific Islander." One parent had a high school education, two had some college, two were college graduates, and one had a graduate degree. Three worked as paid employees, two were self-employed, and one was a full-time parent and caregiver in the home.

Design

The study's qualitative descriptive design (Sandelowski, 2000) was well suited to the aim of characterizing parents' experiences and needs. While not strictly adhering to the phenomenological methodology, the design had what Sandelowski described as "phenomenological overtones," in that the aim was to describe parents' common experiences in receiving their newborn's CAH diagnosis and learning to care for their baby. To that end, both what parents experienced and the context in which they experienced it were emphasized in data collection (Creswell, 2007). The design was retrospective, with parents reflecting on the time after the birth of their first affected child.

Procedure

In individual, semi-structured telephone interviews that lasted 45–90 minutes, parents were asked how they learned about CAH and its management, including contextual factors in the process. They were also asked to comment on how they feel their needs could have been better met during that time. Interviews were audio-recorded, transcribed and coded. Study procedures were approved by the institutional review board, and all participants provided signed informed consent.

Data Analysis

Conventional content analysis (Hsieh & Shannon, 2005) was used to produce a description of parents' experiences organized by theme. Data analysis consisted of identifying