ARTICLE

Parental Reflections on the Diagnostic Process for Duchenne Muscular Dystrophy: A Qualitative Study

Roxanna M. Bendixen, PhD, OTR/L, & Amy Houtrow, MD, PhD, MPH

ABSTRACT

Purpose: Duchenne muscular dystrophy (DMD) is a rare neuromuscular disease with no known cure. We sought to update over 30 years of research reporting on the diagnostic delays in DMD.

Methods: Through personal interviews, this study qualitatively explored parents' experiences regarding receipt of the DMD diagnosis and the guidance for care provided. Thematic analysis identified themes and provided answers to the research questions being addressed.

Results: Four themes emerged: (a) *Dismissive* illustrates little consideration of parent concern in the diagnostic process; (b) *Limited Knowledge* describes misunderstandings about clinical signs, recommended screenings, and testing to achieve a diagnosis of DMD; (c) *Careless Delivery* reports on the manner in which the diagnosis was given; and (d) *Lack of Guidance* describes the follow-up that occurred after the diagnosis.

Conclusion: Despite marked medical progress over the past several decades, substantial barriers to arriving at the diagnosis of DMD and the provision of care guidance remain. J Pediatr Health Care. (2016) ■, ■-■.

KEY WORDS

Clinical decision-making, delayed diagnosis, neuromuscular disorder, parent-report, rare disease

INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most common X–linked recessive fatal muscle disease in children that affects 1 in 3,500 to 6,000 live born males worldwide (Romitti et al., 2015; Theadom et al., 2014). DMD is suspected when young boys display atypical gait patterns and difficulty with physical activities, such as running and climbing stairs. Early clinical signs of DMD may include pseudohypertrophy of calf muscles and the presence of the Gower's sign—a maneuver whereby boys use their hands to walk up their legs to assist them in rising to stand from the floor (Bushby et al., 2009). The route to confirming a diagnosis of DMD is dependent on the availability of rapid and reliable testing, which must be interpreted alongside the clinical presentation of symptoms of DMD (Bushby et al., 2009). Since the early 1980s, creatine kinase (CK) levels have been used as an effective biochemical marker for early detection of DMD (Crisp, Ziter, & Bray, 1982). Because

Roxanna M. Bendixen, Assistant Professor, Department of Occupational Therapy, University of Pittsburgh, Pittsburgh, PA.

Amy Houtrow, Associate Professor and Vice Chair, Department of Physical Medicine and Rehabilitation, Children's Hospital of Pittsburgh, Pittsburgh, PA.

Conflicts of interest: None to report.

Correspondence: Roxanna M. Bendixen, PhD, OTR/L, Department of Occupational Therapy, University of Pittsburgh, 5020 Forbes Tower, Pittsburgh, PA 15260; e-mail: bendixen@pitt.edu.

0891-5245/$36.00

http://dx.doi.org/10.1016/j.pedhc.2016.09.002

boys with DMD typically express elevated CK levels ranging from 5,000 to 150,000 IU/L (Rodino-Klapac, Mendell, & Sahenk, 2013), a normal CK level rules out the diagnosis, whereas an elevated CK level warrants further workup and a prompt referral to a neurology specialist (Bushby et al., 2009; Verma, Anziska, & Cracco, 2010).

Despite the significant progress made in identifying DMD and the straightforward diagnostic pathway (Bushby et al., 2009; Ciafaloni et al., 2009; Verma et al., 2010), more than three decades of research continues to report on the protracted nature of reaching a definitive DMD diagnosis (Aartsma-Rus, Ginjaar, & Bushy, 2016; Ciafaloni et al., 2009; Crisp et al., 1982; Firth, 1983; Green & Murton, 1996; Holtzer et al., 2011; Marshall & Galasko, 1995; Parsons, Clarke, & Bradley, 2004; van Ruiten, Straub, Bushby, & Guglieri, 2014). Past research has shown that parents of boys with DMD often report concerns when their sons are between the ages of 6 months and 3 years because their child is not meeting or regressing in certain developmental milestones (Ciafaloni et al, 2009; Crisp et al., 1982; Firth, 1983; Green & Murton, 1996; Holtzer et al., 2011; Marshall & Galasko, 1995; Parsons et al., 2004). The American Academy of Pediatrics (AAP) describes the importance of eliciting and attending to parental concerns about their child's development, especially when motor delays are pronounced and/or progressive (American Academy of Pediatrics, 2015; Noritz, Murphy, & Neuromotor Screening Expert Panel, 2013). This is a core concept of family-centered care—the belief that health care providers and the family are partners, working together in an effort to be responsive to the family's needs and choices (Committee on Hospital Care and Institute for Patient- and Family-Centered Care, 2012). Knowing the value of eliciting key clinical information about a child's motor development from the child and his/her parent(s), we sought to update previous research and to further explore and understand the diagnostic experiences of families with boys with DMD. We were specifically interested in qualitatively studying (a) the shared experiences of parents regarding receipt of a diagnosis of DMD from their health care providers and (b) after the diagnosis, the resources or guidance for care that were provided to parents by their health care provider(s). A greater understanding of parental experiences of diagnosis and care recommendations of DMD may lead to more appropriate surveillance and earlier referrals/interventions for children showing motor delays.

A greater understanding of parental experiences of diagnosis and care recommendations of DMD may lead to more appropriate surveillance and earlier referrals/interventions for children showing motor delays.

METHODS

A qualitative approach was used to explore parents' shared experiences in the context of receiving the diagnosis of DMD for their children and the guidance provided after the diagnosis (McCaslin & Scott, 2003). Parents of boys with DMD were recruited through two ongoing DMD natural history studies at large academic facilities in the United States, one in the Southeast (Florida) and one in the Northeast (Pennsylvania). If the child was currently participating in the DMD natural history study, clinicians associated with the original studies contacted parents and provided information and an institutional review board–approved flyer describing our study. Fifteen parents from various geographic areas in the United States responded to the flyer and contacted the principal investigator of this study. Written informed consent was obtained from all 15 parents. The first author had limited connection with parents during the two natural history studies but had worked with the clinicians associated with the studies; the second author had no connection or knowledge of the participants or the clinicians involved. All parents fully participated in semistructured telephone interviews from their homes (Table 1). The first author conducted all interviews. All boys with DMD (N = 15) were living at home, and diagnosis for boys occurred between 3 and 10 years ago.

The interview guide was initially developed by the first author, and it received review and discussion from qualitative experts before initiation into the study. Sample interview questions used to elicit in-depth information regarding parents' pre- and post-diagnosis experiences with health care providers and the DMD diagnosis for their children, along with additional examples of parents' responses, are provided (Table 2). Telephone interviews lasted approximately 1 hour, and were audio-recorded and transcribed standard verbatim, which omits filler words such as "um," "you know," and "like." After a thorough review of the written transcriptions for data quality assurance, audio recordings were deleted to protect participant confidentiality.

Thematic analysis was used to identify patterns of meaning across the data and provide answers to the research questions posed (Boeije, 2010; Braun & Clark, 2006; Patton, 1980). Initially, the interviews were methodically read multiple times to allow for increased familiarity with the datasets (Shenton, 2004). The datasets were then hand-coded line by line to identify important patterned responses relevant to