HHS Public Access

Author manuscript

J Pediatr Health Care. Author manuscript; available in PMC 2018 May 01.

Published in final edited form as:

J Pediatr Health Care. 2017 ; 31(3): 285–292. doi:10.1016/j.pedhc.2016.09.002.

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

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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 parent's 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: 1) *Dismissive* illustrates little consideration of parent concern in the diagnostic process; 2) *Limited Knowledge* describes misunderstandings about clinical signs, recommended screenings and testing to achieve a diagnosis of DMD; 3) C*areless Delivery* reports on the manner in which the diagnosis was given; 4) *Lack of Guidance* describes the follow-up that occurred following 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.

Keywords

Neuromuscular disorder; rare disease; delayed diagnosis; parent-report; clinical decision-making

Introduction

Duchenne muscular dystrophy (DMD) is the most common X-linked recessive fatal muscle disease in children that affects 1 in 3,500–6,000 live born males worldwide (Theadom et al, 2014; Romitti et al, 2015). 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, as well as the presence of the Gower's sign - a maneuver where boys use their hands to walk up their legs

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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 1980's, creatine kinease (CK) levels have been used as an effective biochemical marker for early detection of DMD (Crisp, Ziter, & Bray, 1982). Because boys with DMD typically express CK levels ranging from 5,000 to 150,000 IU/L (Rodino-Klapac, Mendell & Sahenk, 2013), a normal CK rules out the diagnosis while an elevated CK warrants further workup and a prompt referral to neurology (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; Verma et al, 2010; Ciafaloni et al, 2009), more than 3 decades of research continues to report on the protracted nature of reaching a definitive DMD diagnosis (Aartsma-Rus, Ginjaar & Bushby, 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 due to their child 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 describes the importance of eliciting and attending to parental concerns about their child's development, especially when motor delays are pronounced and/or progressive (Noritz & Murphy, 2013; American Academy of Pediatrics (AAP), 2015). This is a core concept of family-centered care - the belief that healthcare providers and the family are partners, working together in an effort to be responsive to 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 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: 1) the shared experiences of parents regarding receipt of a diagnosis of DMD from their healthcare provider, and 2) following the diagnosis, the resources or guidance for care that were provided to parents by their healthcare 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 demonstrating motor delays.

Methods

A qualitative approach was used to explore parent's shared experiences in the context of receiving the diagnosis of DMD for their child and the guidance provided following 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 U.S., one in the southeast (Florida) and one in the northeast (Pennsylvania). If their child was currently participating in the DMD natural history study, clinicians associated with the original studies contacted parents and provided information and an IRB approved flyer describing our study. Fifteen parents from various geographic areas in the US responded to the flyer and contacted

the PI 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 semi-structured telephone interviews from their homes. The first author conducted all interviews. All boys with DMD (N=15) were living at home and diagnosis for boys ranged between 3 and 10 years ago.

The interview guide was initially developed by the first author, and received review and discussion from qualitative experts prior to initiation into the study. Sample interview questions used to elicit in-depth information regarding parent's pre- and post-diagnosis experiences with healthcare providers and the DMD diagnosis for their child, along with additional examples of parents' responses are provided (Table 2). Telephone interviews lasted approximately one hour, and were audio-recorded and transcribed standard verbatim, which omits filler words such as 'um', 'you know' and 'like'. Following 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 poised (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 the research questions. Observations of patterns within the data were synthesized and reduced to themes (Boeije, 2010; Braun & Clarke, 2006). Data relevant to each theme was reviewed and discussed, then refined and rephrased to more clearly describe the narrative of our parents. Data analysis began following 10 parent interviews whereby themes developed. An additional five interviews were conducted to ensure that our analysis had reached the point of no new themes. Qualitative findings were then presented to experts in qualitative data analysis for review and discussion of the themes. Agreement across reviewers provided confidence that we had achieved data saturation, identified appropriate themes, and posed thoughtful interpretations (Guest, Bunce & Johnson, 2006; Patton, 1980).

Results

Parents of boys with DMD described their pre- and post-diagnostic experiences regarding how they presented their earliest concerns to their healthcare providers, the diagnosis, and the follow-up that occurred. Within our data an overarching theme of "*Communication Breakdown*" emerged. Our families report not feeling listened to about the symptoms they observed, feeling like the diagnostic information was carelessly delivered, and feeling that their provider didn't communicate guidance. Within *Communication Breakdown*, four themes are presented – *Dismissive, Limited Knowledge, Careless Delivery, and Lack of Guidance*.

Dismissive

The *Dismissive* theme illustrates that parent's felt their concerns about their child's motor skills were not sufficiently addressed and there was little consideration of parent report in the diagnostic process. One father stated, "*The pediatrician told us that we were just worrying and we just needed to calm down because we were overly worried parents and our son was fine. I even pointed his calves out, but we were just made to feel it was our imagination, and we shouldn't be silly*." Another parent discussed how their pediatrician "*… was just very laid back about everything, he said it is no big deal he is just delayed in his gross motor skills, he has low muscle tone. That is pretty much the diagnosis we got – low muscle tone."* Our interviewees provided reports of being told they were overreacting and their child was *"just slow"* in their motor development. One parent "*felt relieved because this was someone who was a medical professional telling us we don't have anything to worry about, so we went home and we were elated. But the symptoms didn't go away."* Interviews also provided accounts of parents taking their child to multiple providers who did not take their concerns seriously. A father stated, "*So he was about 3-1/2 or 4 years old and he was not keeping up with other kids at the playgrounds. We knew nothing except that his legs looked really strong…but it was odd because he was so weak in the same token. So we started doing computer research…and the computer kept indicating this deadly form of muscular dystrophy. The pediatrician told us we were just worrying."* It was 8 more months and 2 different neuromuscular clinics before his son was diagnosed. One mother reported on their two year process, "*We had the one pediatrician he started out with, then he ended up seeing a developmental specialist, then I took him to another pediatrician because we weren't getting any answers from the first pediatrician, and she recommended that he see a neurologist. So he had actually seen 2 neurologists before we finally saw the third one who made the diagnosis."*

Parent's quotes expressed their feelings of worry and anxiousness about their child's health and well-being, and that their concerns were being minimalized and dismissed by their healthcare providers.

Limited Knowledge

Limited Knowledge describes parents' report of their healthcare provider's misunderstandings and misperceptions about clinical signs of DMD and the recommended screening and testing to achieve a diagnosis. Parents discussed their personal efforts to educate themselves about their son's signs and symptoms and pass the information onto their child's pediatrician.

> The experience with the healthcare providers was just terrifying and horrible. We felt we knew more than the doctors. Either they had never heard about DMD or it just wasn't a main topic for them that they knew in and out. In fact, the physical therapist recommended that my son get a CPK blood test. So I made the appointment with the pediatrician, he had no idea what I was talking about.

Numerous parents discussed their frustration that their sons were put through expensive and unnecessary testing. This emerged as a sub-theme, "ineffective testing." One mother stated, *"When he was about 10 months old they recommended an MRI…but he failed the sedation*

so we couldn't have the MRI." This mother went on to report that a confirmative diagnosis had not been given, and a few years later she worked with another neurologist. "*And then the second time around, the neurologist that we saw who thought he had a mild form of cerebral palsy recommended the MRI. But it was just an unneeded test when they could have done a simple blood test to rule out…you know they should have noticed that he was performing the Gower's maneuver and having difficulty with stairs, can't run, can't jump."* Eight of our parents reported a wait of 2 or more years before reaching a final diagnosis of DMD.

Our parent interviews provided numerous accounts of limited knowledge and understanding of the current diagnostic strategies for this rare disease.

Careless Delivery

Careless Delivery reveals concerns regarding how the diagnosis was delivered to parents. Nine parents told of the abruptness in which they were notified, and the lack of empathy received following the diagnosis.

> So I was working with a client and the phone rang and it was a nurse from [the clinic] who told me that the test came back positive without even warning me. I would think that a doctor would actually take the time to call and maybe throw a little bit of psychology into it, and understand that this is a parent, and you are telling them that their son has a terminal illness, so I think that the telephone call was inappropriate.

Parents discussed the inability to talk further with their doctor following the diagnosis. As one mother stated, *"This was all told to us on the phone on a Friday afternoon when there was nothing we could do. We were lost like there was nobody to contact besides the pediatrician who called to check on us and said, well, call me on Monday and we can talk again. So that was distressful."* Parents reported that the diagnosis was delivered with limited preparation or sensitivity, *"And I remember thinking what do you mean muscular dystrophy? Because they never prepped me at all or prefaced it with anything. They didn't even say we think it is muscular dystrophy, they just said you have an appointment at the muscular dystrophy clinic, and you know that changed my world from that day on."* Another parent, who had been researching DMD and knew what the devastating diagnosis would mean for their family, reported being at the doctor's office when they received the news, but it didn't come from the doctor *"…and the person at the front counter told us it was DMD, but she didn't know what DMD stood for, and then we finally had to say the words."* A few parents acknowledged that providers have limited experience giving the diagnosis of DMD. As one mother said, *"…they give the diagnosis so little that they don't even know what to do."*

Most of our parents reported how the uncaring delivery of their child's diagnosis of DMD caused much distress. This recurring theme belies the importance of supporting families and ensuring that adequate information is provided and collaborative relationships established among all members of the health care team.

Lack of Guidance

Lack of Guidance sheds light on the process and follow-up that occurred after the diagnosis of DMD was revealed. Fourteen of our parents stated that they received little guidance following the diagnosis and limited resources to assist in managing the impact of the disease, indicating a clear pattern from the interviews. Parents report being told there is little they can do for their sons. "*The neuromuscular neurologist pretty much gave us the diagnosis and said how grim it was, and there was really nothing that we could do, and we should go home and love him because there is no cure."* Parents felt that their child's pediatricians were not up-to-date on current information, *"I don't even know the words to describe it, but kind of like a whirlwind of misinformation…nobody was on the ball with what new therapies were out there."* When resources were provided to parents, these resources appeared to be limited, *"We thought they kind of focused more on whether or not we could send him to camps and that sort of thing, and we were kind of in a daze at the beginning so we weren't really interested in camps. There was kind of a standard that they went by, like this is what we can do for everyone, and we just didn't feel that was progressive enough or forward thinking enough."* Importantly, parents also discussed the benefit of the resources they did receive, *"I was thinking in my head I wanted to have more kids and so she helped me work through that in referring us to the geneticist, but I mean still with resources, I would say she did tell us about MDA but that was it."*

Within our theme of *Lack of Guidance*, the sub-theme of "*Self-education and Self-advocacy*" emerged. Parents reported how they researched available resources and moved forward with obtaining needed healthcare for their child, *"We got physical therapy from the beginning; it wasn't due to the doctor telling us we needed it though. Not even from the pediatrician who I put on a pedestal cause she is awesome. That was more on my part from reading online and then being able to find the Early Intervention program."* Advocating for their child often meant changing providers in order to receive adequate care. *"We ended up switching neurologists…you know it was hard to switch from her because she had such compassion and love for the children."* One father discussed their decision to seek care through a specialty clinic alone; he acknowledged, "*We don't have a pediatrician…it is odd to have a son with a serious condition and not have a pediatrician."* Eight of our families were currently traveling out-of-state for healthcare for their son. Parents provided stories of connecting with experts in DMD and the empowerment they felt once their child was receiving appropriate care and attention, *"The physicians who are educated about DMD and are involved with my son are like a family, there is no wall or patient-doctor barrier. When you go for a visit it is a whole different feeling. These are the best friends that I never wanted to have."*

Parents reported little guidance in the provision of basic care standards or where to obtain optimal care for their child. Importantly, parents who discovered strong, collaborative health care teams felt empowered by more informative clinical decision-making and established plans of care.

Discussion

Through personal interviews with parents of children with DMD, our data revealed that many pediatric healthcare providers failed to incorporate parental concerns into the diagnostic process, had limited knowledge to guide the medical work up and, after delivering the diagnostic news, provided little guidance to assist families in dealing with the impact of this devastating disease. As reported by the families in this study, the lack of attention to parental concerns, along with limited experience and expertise, may hinder a provider's clinical judgment and likely lead to diagnostic delays. These concerns may also lead to a collapse of family-centered care and a loss of trust in the doctor-patient-family relationship (Committee on Hospital Care and Institute for Patient- and Family-Centered Care, 2012; Council on Children with Disabilities (CCP), 2006)

It is not surprising that pediatric healthcare providers may have limited clinical knowledge or expertise regarding the diagnosis of a rare disease, such as DMD. In fact, a few of our parents recognized the paucity of information on DMD. Despite the limited exposure to the disease itself, healthcare providers do have specific tools in their armamentarium to help guide the diagnostic process. The AAP recommends developmental surveillance and screening for motor delays in all children, and has provided algorithms that focus on early identification of children with developmental disorders and motor delays (AAP, 2015; CCD, 2006; Noritz & Murphy, 2013). The AAP recommends that parental concerns mandate serious attention and that developmental surveillance include "eliciting and attending to the parents' concerns about their child's development" [Noritz & Murphy, 2013, pg. e2019]. For a life-threatening and progressive disease like DMD, the use of developmental surveillance for early detection and diagnosis is crucial to the initiation of comprehensive care, which improves health outcomes and can help mitigate negative family impacts (Baiardini et al, 2011; Bushby et al, 2009; Bushby et al, 2010; Poysky & Kinnett, 2008; Rodino-Klapac, et al, 2013).

Our data also revealed limitations in the use of evidence-based resources for the management of DMD and deviations from family-centered care ideals (Committee on Hospital Care and Institute for Patient- and Family-Centered Care, 2012). The pediatric medical home has a responsibility to identify and address developmental concerns in children and initiate treatment planning and management (CCP, 2006). Yet numerous families did not receive resources or guidance for disease management following receipt of the DMD diagnosis, creating cynicism within the patient/provider relationship and the eventual abandonment of the primary care medical home for care only in subspecialty clinics. Although the lack of a medical home is a concern, once parents located specialty clinics they reported how *"comforting"* it was to have a knowledgeable and experienced team to work with to move in the direction of action for their child.

The results of this study are disconcerting given the literature over the past three decades. More than 30 years ago, Crisp et al (1982) reported on delays in the diagnosis of DMD, concluding these delays could be corrected by listening to parent's concerns, early motor screening, and CPK testing. Firth (1983) qualitatively reported that parents described early motor delays in their sons and sought advice from their pediatricians, but were often told

they were unnecessarily concerned and sent home with a reassurance that the symptoms were within the normal range of development. Ten years later, diagnosis for DMD remained at a 2-year delay, and parent's specific concerns were still being dismissed (Green & Murton, 1996; Marshall & Galasko, 1995). In fact, in more recent studies, parents continue to notice symptoms more than 2 years before a definitive diagnosis is provided (Holtzer et al, 2011; Parsons et al, 2004; van Ruiten et al, 2014). Similarly, our participant group reported a large range in time since their son had been diagnosed (3–10 years), yet the communication breakdown was prevalent and diagnostic delay changed little over this time period. In line with each of these past studies, parents report going to numerous healthcare providers for the diagnosis and care their child needs, and results reiterate that CPK levels are sensitive biochemical markers for early detection of DMD and can play a pivotal role in establishing a DMD diagnosis.

Importantly, the difficulties with a timely diagnosis appear to be systemic and not just related to the primary pediatrician. Our families discussed other specialists, such as developmental pediatricians and neurologists, who also failed to promptly make a diagnosis – a diagnosis that in retrospect seemed obvious to our families. Clearly, these families were confused and angered by the perceived failure of the diagnostic process. Yet the reality is that for every one boy with DMD, a pediatrician may have encountered hundreds of children with other developmental disabilities. Potentially, time involved in "delaying the diagnosis" may have been used by the pediatric healthcare provider to follow guidelines for gathering more developmental information, performing more screening evaluations, and referring to therapy services before obtaining blood work. Despite established DMD standards of care and clinical guidelines, there remains a continued need for education of health professionals.

Limitations

This study has a few limitations. First, although our study participants lived in eight different states, most of our information came from families living in the southeast and northeast. Second, our conclusions are based solely on parental interviews, hence, we are only presenting one view of the diagnostic experience – the view of a parent who is struggling with the reality that their child has a lethal disease. Parents may be negatively associating their child's pediatric healthcare provider with this traumatic event. Moreover, recall bias may have occurred, as it has been documented that parents do not necessarily absorb or recall information provided in clinical encounters accurately, especially around the time of diagnosis (Committee on Hospital Care and Institute for Patient- and Family-Centered Care, 2012). Nonetheless, under the tenets of family-centered care, special attention is needed to assure that complicated medical information is delivered in a timely, unbiased and accurate manner that is conducive to understanding and retaining.

Conclusion

Multiple prior studies on the diagnostic process of DMD have demonstrated that providers dismiss parental concerns, that the presence of developmental delay didn't appropriately arouse clinical suspicion, and that providers did not conduct the simple tests necessary to arrive at the diagnosis. Our study illustrates that 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. In line with family-centered care principles, our data demonstrate the essential nature of the families' voices in the identification and management of DMD. Future studies should explore pediatricians' experiences and knowledge about early identification and management of DMD and move forward to develop strategies to improve the diagnostic process and continuum of care of DMD and maximize quality of life of boys with DMD and their families.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

We would like to express our thanks to the parents for participating in this study and to Kelsey Iglesias, MOT, who assisted with data collection. The project described was supported by Award Number 5K01HD064778 from the Eunice Kennedy Shriver National Institute of Child Health & Human Development. The content is solely the responsibility of the authors and does not necessarily represent the official views of the Eunice Kennedy Shriver National Institute of Child Health & Human Development or the National Institutes of Health. The funder had no involvement in the study design, data collection, data analysis, manuscript preparation and/or publication decisions.

Abbreviations

DMD	Duchenne muscular dystrophy
CK	creatine phosphokinase
MDA	Muscular Dystrophy Association

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Table 1

Parent Demographics

Parent Demographics (N=15)	# (%)
Age, gender, and marital status	
≤ 30 years old	2 (13.3)
30 – 50 years old	12 (80.0)
≥ 51 years old	1 (6.7)
Females	12 (80.0)
Males	3 (20.0)
Married/long-term commitment	12 (80.0)
Divorced/separated	3 (20.0)
Race/Ethnicity	
Caucasian	12 (80.0)
African American	1 (6.7)
Hispanic/Latino	1 (6.7)
Other	1 (6.7)
Location and household income	
Urban	5 (33.3)
Suburban	8 (53.4)
Rural	2 (13.3)
≤ $50,000	3 (20.0)
$51,000 – $100,000	8 (53.4)
➢$100,000	4 (26.6)
➢	
Education	
High school or less	3 (20.0)
College or technical school	8 (53.4)
4-year college degree or beyond	4 (26.6)

Table 2

Semi-structured interviews: Sample interview questions to elicit in-depth information regarding parent's pre and post experiences with healthcare providers before and after receiving the DMD diagnosis for their child, along with additional examples of parents' responses.

Interview Guide	Additional Examples of Parental Responses to Interview Questions
1. I would like to talk about your experience of receiving a DMD diagnosis for your child, specifically your experience with your healthcare provider. Who first gave you the diagnosis of DMD and what was that experience like?	*"We came to realize that the clinic physician was not so much a specialist as someone who was a doctor just kind of assigned to that. Not a doctor who was very passionate about helping with this extremely serious disease."* *"You know I can't think of any way to candy coat it. It was just something we had to endure. But I think that where I was when that phone call came just made it really difficult. I had no time to swallow all that. My son was with me and I had to be happy and smile and play with [name]; there was nowhere to run."* *"It took to long, and it is really hard when you don't know what is happening with your son, you start thinking yes, he has Duchenne, but sometimes you can think no maybe it is not. But really you don't want to accept that your little boy has Duchenne muscular dystrophy."*
2. What were some of the strengths and weakness of your healthcare provider during that experience?	*"As far as the initial diagnosis, the neurologist that we initially went to, I really liked her, she showed a lot of compassion and was very knowledgeable about the diagnosis."* *"He pretty much told us, well he's two years old; we wouldn't do anything right now, go home and love him, and you know, let him be a boy. Well, that obviously is not really very acceptable."*
3. What if anything did you really cherish or appreciate about your healthcare provider and what if anything were you not pleased about?	*"We are now surrounded by what I would call a pit crew of doctors…it is the most comforting thing because he is followed so intensely by them, as well as by physicians here at the [clinic].* *"So the doctor had said it will be a couple of weeks before we get full confirmation that it is Duchenne. Well 2 weeks went by, then 3, 4, 5 and 6 weeks. I was getting really annoyed and angry. It was like put is out of our misery here, we need to know 100 percent. I finally get a call back from my pediatrician's office and find out that the results had been sitting on the doctor's desk for 4 weeks."*
4. How did this experience with the healthcare provider affect you and your family and your view of the diagnosis?	*"So we just felt hopeless as we went through the whole lengthy process."* *"So the neurologist where we were at said I'm not a specialist on Duchenne muscular dystrophy but I will help you find one of the best."*
5. What resources did your healthcare provider give you? What did you do after you received the information from your healthcare provider?	*"I think if I didn't search on my own, I would have been lost, so I feel like I can relate to parents saying they don't have enough information."* *So there is a lot of focus on research and there is a lot of focus on fundraising, which are both needed, but the day to day parenting stuff is really what is not really out there."* *""From that moment on it was pretty much rolling up our sleeves and trying to figure out where to go from there."*