

ARTICLE

A mixed methods study of age at diagnosis and diagnostic odyssey for Duchenne muscular dystrophy

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The delayed diagnosis of Duchenne muscular dystrophy (DMD) may be an ongoing problem internationally. We aimed to ascertain age at diagnosis and explore parents' experiences of the diagnosis of DMD in Australia. Using mixed methods, data were collected from laboratory and clinical record audits of testing for DMD in Victoria and Tasmania, interviews and a national survey of parents regarding their experiences from first noticing symptoms to receiving a diagnosis. The audits revealed that the median age at diagnosis for DMD was 5 years ($n=49$ during 2005–2010); this age had not changed substantially over this period. Fourteen parents interviewed reported age at diagnosis ranging from 2 to 8 years with a 6 month to 4 year delay between initial concerns about their child's development and receiving the DMD diagnosis. Sixty-two survey respondents reported the median age at diagnosis was 3 years and 9 months, while the median age when symptoms were noticed was 2 years and 9 months. Parents experienced many emotions in their search for a diagnosis and consulted with a wide range of health professionals. Half the survey respondents felt that their child could have been diagnosed earlier. Despite advances in testing technologies and increasing awareness of DMD, the age at diagnosis has remained constant in Australia. This mixed methods study shows that this diagnostic delay continues to have a negative impact on parents' experiences, places families at risk of having a second affected child and may have a deleterious effect on affected children's treatment.

European Journal of Human Genetics (2015) 23, 1294–1300; doi:10.1038/ejhg.2014.301; published online 28 January 2015

INTRODUCTION

Duchenne muscular dystrophy (DMD), the most common muscular dystrophy of childhood,¹ is degenerative, life-limiting² and incurable.³ Affected boys have increasing muscle weakness, cardiorespiratory and orthopedic complications,⁴ and are at a risk of cognitive, behavioral and language difficulties. DMD can initially present with global developmental delay.⁵ DMD is caused by dystrophin gene variants⁶ which are inherited from female carriers in approximately two-thirds of cases² or occur *de novo* in the remaining one-third,⁷ although a recent estimate suggests that the incidence of *de novo* variants may be as high as 50%.⁸

Historical studies suggest an average age of diagnosis of DMD of ~5 years in the United States and Europe, this figure remaining relatively stable over 26 years.^{9–14} Age of diagnosis in Australia has not been previously published. Ciafaloni *et al.*¹⁴ observed a 2.5 year delay between parents first noticing symptoms and ultimately receiving a DMD diagnosis for their child. An earlier diagnosis would mitigate the impact of the diagnostic odyssey on parents⁹ and facilitate access to a range of health interventions, including corticosteroid treatment and physiotherapy.⁴ As novel treatments for DMD become available, maximal benefit will be derived from their use early in the disease course.^{15–17} Early diagnosis also enables genetic counseling to assist parents in their reproductive planning.¹⁴ Despite previous studies reporting diagnostic delay in DMD,^{9,11,14} there are scarce data regarding this process and the associated emotional challenges. An in-depth understanding of parental experiences of the current

diagnostic process is essential in determining the role of population screening for DMD. Here we report findings of an Australian study exploring the age at DMD diagnosis through three different data collection methods, and investigating parents' experiences of the diagnosis of DMD in their child.

METHODS

A mixed methods triangulation approach was used in this study, in which interpretation of results are mixed and equal weight is given to both the qualitative and quantitative data.¹⁸ We used qualitative data from interviews to develop a quantitative survey, both of which examined parents' experiences of the process of diagnosis. We further triangulated these data with audit data from Victoria and Tasmania to determine age at DMD diagnosis. The Royal Children's Hospital (RCH), Melbourne, Human Research Ethics Committee granted ethics approval (HREC30136B).

Audit of DMD testing

Diagnostic testing for DMD in Victoria and Tasmania is performed by statewide laboratories: the Victorian Clinical Genetics Services (VCGS) provides genetic testing and the State Neuropathology Service (SNPS) analyses muscle biopsies. The RCH Neuromuscular Clinic provides multidisciplinary management for Victorian and Tasmanian boys with DMD, and clinicians in the VCGS provide genetic counseling. Information on all DNA tests performed for DMD in these two states between 2005 and 2010 was obtained from VCGS. 2005 was chosen as the audit starting time point because the more sensitive multiplex-ligation probe amplification (MLPA) technique was implemented for DMD in that year (pers. comm., Dr Desiree du Sart, VCGS). Exclusions included tests

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Received 26 September 2014; revised 5 December 2014; accepted 16 December 2014; published online 28 January 2015

ordered for: (1) female patients with neuromuscular symptoms (rare for females to have DMD and tests likely requested to exclude DMD); (2) male patients over 10 years old (DMD symptoms usually obvious before 10 years and tests likely ordered to diagnose Becker muscular dystrophy); (3) children previously diagnosed with DMD on muscle biopsies (MLPA likely requested to confirm the diagnosis); and (4) determination of female carrier status for DMD. Inclusion criteria were therefore MLPA tests ordered between 2005 and 2010 for male patients aged ≤ 10 years showing clinical symptoms of DMD. These MLPA results were then cross-referenced with various databases and patient files: (1) RCH Neuromuscular Clinic patient database and files; (2) VCGS patient files; and (3) SNS database of results of muscle biopsy samples (immunohistochemistry). The following data were collected: (1) total number of DMD diagnoses; (2) MLPA test results (positive/negative); (3) type of testing to obtain DMD diagnosis; (4) age at DMD diagnosis; and (5) medical specialty of ordering physician.

Exploration of parents' experiences: interviews and survey

Parents to be interviewed were recruited between 2010 and 2011 via an invitation letter through the RCH Neuromuscular Clinic. Inclusion criteria were: parents ≥ 18 years old, able to speak and read English, with a DMD diagnosis in their child ≥ 1 year previously. Semi-structured interviews (face-to-face or by telephone), informed by a literature search, enabled questions to be guided by participants' responses.¹⁹ When couples participated, each parent was interviewed separately to explore their individual experience without the partner's influence, which allowed each parent to freely share views that may have differed from their partner's. Interviews were digitally recorded and transcribed verbatim. Transcripts were de-identified, participants assigned pseudonyms and transcripts were imported into NVivo 9 software (QSR International Pty Ltd, Melbourne, VIC, Australia) to facilitate data management. Interviews were conducted until no new themes emerged, that is, the data had reached saturation.²⁰

From the qualitative findings, an item bank of questions was generated to inform a survey for distribution to families of boys with DMD throughout Australia. The draft survey was piloted with the original interview participants, who provided feedback regarding: ease with which they could complete the survey; whether the survey items allowed them to provide accurate information to describe their experiences; length, layout and clarity of wording. The final survey included questions on: demographics; number of children in the family with and without DMD and relationship to the responder; initial symptoms of DMD noticed by parents or health professionals; the journey undertaken to find a diagnosis and contacts made with various health professionals; and views on potential changes to the way DMD may be diagnosed in the future. The data presented in this paper relate to the first four categories of questions; views on potential changes in the way DMD may be identified and diagnosed in the future will be presented elsewhere.

In 2012/2013, muscular dystrophy support associations throughout Australia emailed a link for an online version of the survey to the members who were parents of boys with DMD, and mailed out hard-copies when required. Descriptive statistics were used to analyze the data in Microsoft Excel. The age when symptoms were first noticed was determined by ordering the values from youngest to oldest, and taking the middle value as the median.

RESULTS

Description of participants

The audit revealed that 77 boys from Victoria and Tasmania underwent testing. Forty-nine were diagnosed with DMD between 2005 and 2010: 42 were diagnosed based on positive MLPA test results; 5 by muscle biopsy; and 2 because of an affected older brother, clinical findings of DMD and a persistently raised serum creatine kinase. The biopsy results of the five boys were reviewed by our colleagues working at the SNS, and the medical records of the two boys were reviewed by AK to confirm the DMD diagnoses. Interviews were conducted with 14 parents from Victoria (10 mothers, 4 fathers; there were 3 couples); 11 face-to-face and 3 by telephone. Data from the interviews therefore represented the experiences of 14 parents from 11 Victorian

families, with two boys in the same family having DMD. Sixty-two anonymous surveys were received from parents nationally. Respondent characteristics are described in Table 1. Not all questions were completed by all respondents.

Age of initial symptoms and at time of diagnosis

The median age at diagnosis for 49 boys as determined by the audit was 5 years (Figure 1a) and was constant over the study period. Approximately one-third ($n = 16$) were diagnosed between two and

Table 1 Characteristics of survey respondents ($N = 62$)

Characteristic	n (%)
<i>Relationship to the child with DMD</i>	
Mother	50 (80.6)
Father	12 (19.4)
<i>Relationship status</i>	
Married	45 (72.6)
Divorced	3 (4.8)
Separated but not divorced	2 (3.2)
<i>De facto</i> (living with partner)	8 (12.9)
Single	3 (4.9)
Partners, not living together	1 (1.6)
<i>Age range (years) at the time of survey</i>	
18–30	2 (3.2)
31–40	20 (32.3)
41–50	25 (40.3)
51–70	15 (24.2)
<i>Highest level of education attained</i>	
Year 11 or below ^a	11 (17.7)
Year 12 or equivalent ^a	12 (19.4)
Tertiary certificate/diploma/trade/apprenticeship	23 (37.1)
University qualification	15 (24.2)
Other	1 (1.6)
<i>Home state of residence</i>	
New South Wales	9 (14.5)
Queensland	9 (14.5)
South Australia	8 (12.9)
Victoria	33 (53.2)
Western Australia	3 (4.9)
<i>Number of children</i>	
1	5 (8.1)
2	22 (35.4)
3	23 (37.1)
> 3	12 (19.4)
<i>Reported age of child at diagnosis</i>	
0–1 year	2 (3.2)
1–2 years	8 (12.9)
2–3 years	10 (16.1)
3–4 years	13 (21.0)
4–5 years	13 (21.0)
5–6 years	6 (9.6)
6–7 years	5 (8.1)
> 7 years	5 (8.1)

^aYear 12 is the highest level of high school (matriculation) in Australia although students may leave earlier, most notably in year 11.

three years of age; notably, 10 of this group of children ($n=16$) were diagnosed by a clinician (general pediatrician or pediatric neurologist) in a tertiary hospital (Figure 1b). The median age at diagnosis was 4.5 years when tests were ordered by tertiary pediatricians compared with the median age of 5 years when the tests were ordered by community pediatricians. We conducted statistical analysis (Mann–Whitney) and did not find any significance in this difference, however the numbers are too small to have sufficient power with only 11 tests ordered by community pediatricians.

In the interviews, parents ($n=14$) were asked to recall the ages at diagnosis for their sons. These ages were verified with the audit data, showing a high level of congruence between parent reports and the audit data (Table 2). In most cases there was a delay between the time symptoms were first noticed (ranging from approximately 6 months to 4 years) and the diagnosis of DMD (ranging from 1

year 11 months to 7 years and 11 months, with the median age 3 years and 2 months based on their specific audit data— $n=11$). In the survey, parents reported that the age at which symptoms were first noticed was a median of 2 years and 8.5 months ($n=62$), with the median age at diagnosis being 3 years and 8 months ($n=62$).

Parents' experiences noticing symptoms and reaching a 'tipping point' to seek a diagnosis

Parents who were interviewed described taking a variety of pathways to obtain the DMD diagnosis, with three main emergent themes shown in Table 2. The transition from noticing symptoms to seeking a diagnosis was described as a period of watching and waiting, in which families described 'see-sawing' between reassurance and concern until they ultimately reached a 'tipping point'. Factors influencing parents' lack of self-confidence regarding their concerns included being a first-time parent, false reassurance from others, and recognizing the broad range of normal development. On the other hand, those who reached the tipping point sooner were not first-time parents, compared their child with their own other children or those of friends and relatives, and when others noticed symptoms in their child. Symptoms prompting parents to seek medical assessment were examined in the survey and those noted by $>20\%$ of parents are shown in Table 3; the most common initial signs were difficulties with running and climbing stairs. Fine motor skills, speech and language, cognitive skills and social function were less often a concern. Many parents discussed this as a time fraught with confusion and emotions:

'there was a fair amount of stress involved as well, I was feeling low': Pauline, mother of Caleb

'you feel so guilty...that you brought these children in the world and...they've got this disease to live with': Hilda, mother of Mark

Many of the feelings reported by the respondents changed during transitions between first noticing symptoms, seeking a diagnosis and after confirmed diagnosis (see Table 3).

Consulting with health professionals regarding concerns about development

In the interviews, parents described in detail their experiences leading up to their child's diagnosis. For 4 of the 11 families interviewed, the pathway was straightforward; the rest experienced more protracted journeys involving consultations with multiple allied health and/or medical professionals. Fifty-two parents (88%) surveyed reported consulting with a health professional about their concerns when they first noticed symptoms, including: primary care (76%: general practitioners and maternal child health nurses); secondary and tertiary care pediatricians (53%: community pediatricians and hospital-based general pediatricians and pediatric neurologists – NB in Australia a referral is required for this category); allied health professionals (42%: physiotherapists, speech therapists and occupational therapists); complementary health professionals (22%: chiropractors, naturopaths, dieticians and osteopaths). The frequency of consultations with health professionals was also recorded using the categories of 'once off', 'a few times (2–4 times)' and 'more often'. Thirty-one percent visited these health professionals 'more often' about symptoms in their child.

One aspect that was particularly frustrating for parents who were interviewed was the focus on referrals for early intervention for management of symptoms rather than obtaining a diagnosis, which often came at a later time:

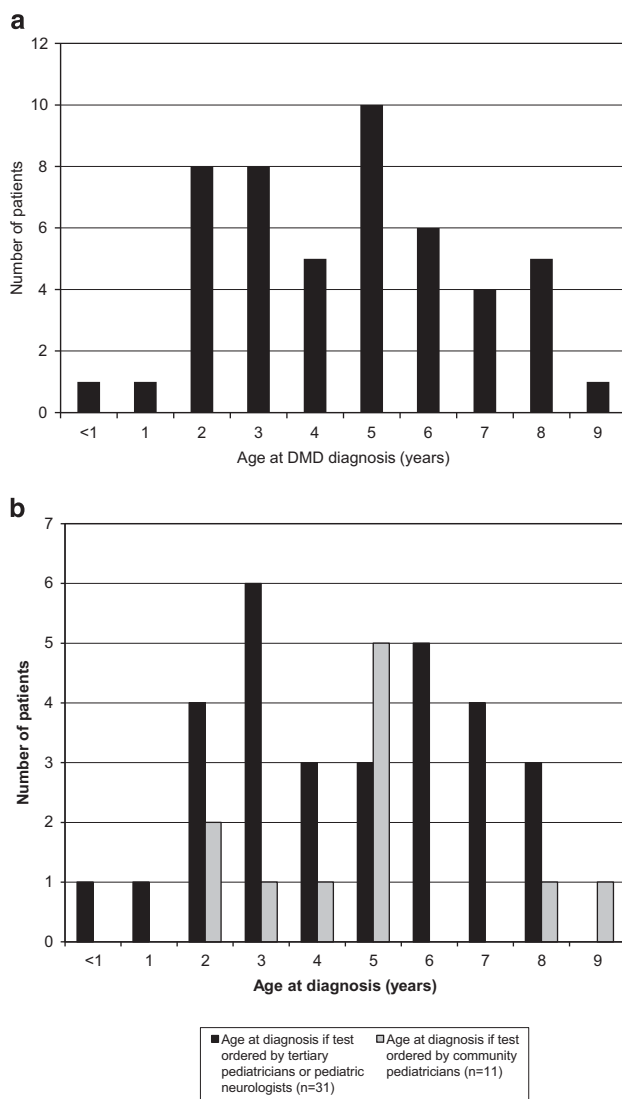


Figure 1 (a) Audit of clinical and laboratory records: age at DMD diagnosis for all tests ordered 2005–2010 in Victoria and Tasmania ($N=49$). Median=5 years. (b) Audit of clinical and laboratory records: age at DMD diagnosis for tests ordered by pediatricians and pediatric neurologists ($N=42$). Median age at diagnosis for tests ordered by paediatricians in tertiary hospitals or paediatric neurologists=4.5 years. Median age at diagnosis for tests ordered by community paediatricians=5 years.

Table 2 Age at diagnosis and parents' experiences of the journey to diagnosis

Participant pseudonym	Pseudonym of son(s) with DMD	Parent-reported age at diagnosis	Audit age at diagnosis	Age at which parents first noticed symptoms	Time between noticing symptoms and obtaining diagnosis	Themes about parents' experiences of the journey to diagnosis with illustrative quotes from participants
Hilda	Mark	'seven and a half'	Unavailable ^a	'about four'	3 years 6 months	Noticing symptoms and reaching the 'tipping point' "I remember just being frustrated ...I just thought ...'is it me being silly or am I not handling this child properly?' ...I just thought... wait and see what happens ...until I just thought one day, 'enough's enough'."
Janice	Robert	'a month before his eighth birthday'	7 years 11 months	'say three and four'	3 years 11 months to 4 years 11 months	Lucy
Laura	Christopher	'just before he turned three'	3 years 2 months	'eight months'	2 years 6 months	
Lucy	Victor	'I'm pretty sure he was only two'	2 years 6 months	'about six months'	2 years	
Mabel	Gabriel	'three, three and three quarters'	3 years 7 months	'about a year old'	2 years 7 months	
Mary	David and Simon	'five and a half and three and a bit'	David—4 years 5 months Simon—2 years 5 months (diagnosed on David's biopsy)	Did not notice symptoms	Not applicable	Actively searching for a diagnosis: experiencing frustration "..."[we went] to the local GP, and ... she put us on to go... see the first [specialist]... about his feet... 'cause [he] just wasn't walking and [then it] was his ears because of his balance, and then [he] got his eyes tested. So there was quite a few [specialists] over a span of about a year..."
Samantha	Jeff	'a week before his second birthday'	1 year 11 months	'at about ten months'	1 year 1 month	Ben
Ted	James	'could have been three, three and a half'	3 years 8 months	'six months' before diagnosis (2 years 6 months)	6 months	
Ben	Stanley	10/11/2005 (3 years 3 months) ^b	3 years 3 months	No age given	Unable to be determined	
Melissa		10/11/2005 (3 years 3 months) ^b	3 years 3 months	'13 months'	2 years 2 months	Receiving false reassurance from health professionals "..."the paediatrician did not even... look at Duchenne's. He just kept saying there's nothing wrong with the fact that my son walked on his toes and [was] falling a lot. 'Nup! There's nothing wrong with him. [so] we took him to a new paediatrician who diagnosed him (snaps fingers) like that."
Jake	Caleb	'just a fraction over two'	2 years 2 months	'one or so'	1 year 2 months	Janice
Pauline		'two and two months or something'	2 years 2 months	'about nine or ten months'	1 year 4 months to 1 year 5 months	
Keith	Roger	'when he was two and a half'	2 years 8 months	No age given	Unable to be determined	
Melanie		'nearly two and a half'	2 years 8 months	No age given	Unable to be determined	

^aMark's name was not found on the Victorian Clinical Genetics Services database of tests ordered for Duchenne muscular dystrophy. Mark resided in Perth at the time of his diagnosis; consequently his diagnostic tests may have been performed by a laboratory in Western Australia.^bBen and Melissa quoted the actual date at Stanley's diagnosis and this date was corroborated by data from the audit.

Table 3 Parents' report of symptoms first noticed and their feelings at that time and while seeking and following the diagnosis

Description of symptoms reported by more than 20% of parents	Data collected in the survey N = 62 (%)	Words and phrases describing how	When first	While seeking	After
		parents felt at various stages, Data collected in survey N = 62	noticing symp- toms n = 53 (%)	diagnosis n = 55 (%)	confirmed diagnosis n = 59 (%)
Difficulties with standing	16 (26)	Stressed	18 (34)	33 (60)	45 (76)
		Frustrated	16 (30)	28 (51)	31 (53)
Difficulty with getting up from a seated position	23 (37)	Anxious	24 (45)	39 (71)	39 (66)
		Confused	15 (28)	25 (45)	27 (46)
Poor co-ordination	14 (23)	Guilty	5 (9)	12 (22)	41 (69)
		Sad	10 (19)	23 (42)	56 (95)
Poor balance	23 (37)	In denial	8 (15)	5 (9)	11 (19)
Large calves	26 (42)	Unsure	21 (40)	25 (45)	30 (51)
Complaining of tired legs	16 (26)	Angry with the medical system	10 (19)	13 (24)	19 (32)
		Pressured by health professionals	4 (8)	5 (9)	10 (17)
Difficulties with walking	22 (35)	Pressured by family or friends	7 (13)	7 (13)	16 (27)
		Optimistic about the future	16 (30)	11 (20)	27 (46)
Falling down often	27 (44)	Pessimistic about the future	4 (8)	14 (25)	37 (63)
Clumsy	13 (21)	Worried about my child's development	30 (57)	24 (44)	37 (63)
Tiring easily	23 (37)	Questioning my parenting abilities	14 (26)	11 (20)	19 (32)
Difficulties running	40 (65)	Questioning my own concerns	8 (15)	11 (20)	21 (36)
Difficulties hopping	34 (55)	Concerns dismissed by others	15 (28)	16 (29)	15 (25)
Difficulties climbing stairs	38 (61)	Had a gut instinct something was wrong	37 (70)	26 (47)	10 (17)

'...the speech therapist wasn't doing anything, 'cause he wasn't getting any better so (I went) back to my doctor, and she referred me to a pediatric speech person, and he said, 'he seems to understand everything but it's just taking time', and then I went back to my doctor and said that wasn't good enough, and then she referred me to (early intervention centre)...that was horrible...you just keep getting referred from one place to the next person...it was frustrating.'; Melissa, mother of Stanley

Parents described a tenacious approach to seeking a diagnosis in the face of false reassurances. Of the 52 parents who consulted with a health professional, 83% reported at least one reason for consulting a health professional about their concerns: 46% 'compared their child with other children who were healthy'; 37% had 'someone else noticing symptoms'; 29% 'compared their child with other siblings' while 25% noticed 'more or new symptoms'.

Perceptions about the timing of the diagnosis

For all participants, perceptions about the timing of diagnosis were complex. From the survey, views on an earlier diagnosis were mixed: of 57 respondents, 51% indicated that they felt their child could have been diagnosed earlier while 49% did not have this view. A χ^2 -test was performed to examine whether this response was associated with such variables as parental gender, age, relationship status or education but there were no statistical differences observed, likely because of the small numbers of respondents in each group. Parents in the interviews raised both pros and cons about the possibility of earlier diagnosis.

'There's no easy answer because nobody wants to be told at any stage their child has a terminal illness', Janice, mother of Robert.

Benefits included validation of a reason for the symptoms, ability to access specific treatments sooner, preparing for financial and practical issues in the future, and informing reproductive planning. Parents felt that receiving a diagnosis before symptoms are obvious, however,

could be confronting or negatively impact on bonding, and that having to go through a process of noticing symptoms and seeking a diagnosis allows time to gradually accept the diagnosis.

'...for a year of so [Caleb] was a...normal baby and [we had] normal expectations and, we eventually found out [the diagnosis... I don't know if I...would have wanted to know...straightaway [at birth].'; Jake, father of Caleb

'...one of the things about learning at two, with Jeff, was that [we had] a two year old who still wanted to play with his dad...and... those years...will allow a parent to develop acceptance [of the diagnosis].'; Samantha, mother of Jeff

Nevertheless, parents felt it was important that the diagnosis was not too late and that perhaps a time later in infancy or early childhood could be preferable.

DISCUSSION

This study investigates both the age of diagnosis of DMD and parents' experiences of the process in Australia, and gives a rich perspective of these issues. There continues to be a delay in diagnosis of DMD through standard clinical practice; audit data showed a median age at the diagnosis of 5 years in Victoria and Tasmania, Australia, which has not substantially changed in the last 25 years. International centers report similar findings.⁹⁻¹⁴ Parent-reported age at diagnosis was remarkably similar to the actual age at diagnosis, for boys ($n=11$) of parents who were interviewed, suggesting that parents' recollections were accurate. However, it should be noted that overall the median age of diagnosis self-reported by the parents in the interview and survey is lower than the age identified by the audit. Given that the self-reported ages from the two groups of parents are more similar, one possibility is that parents who were interviewed or completed the survey represent a biased group as one would expect that the audit data more accurately reflect the time of diagnosis. These parents may also have been more proactive in seeking a diagnosis.

Parents reported consulting a range of different types of health professionals, including complementary health practitioners, when they first started noticing symptoms. The audit data, however, indicated that if boys were referred to pediatricians or pediatric neurologists in tertiary hospitals, testing tended to occur at an earlier age.

Delays in the diagnosis of DMD place families at risk of having a second affected child.^{11,14} These delays also affect access to steroid therapy, the only treatment shown to alter the course of DMD, which is usually recommended from the time that motor skills plateau in DMD, typically occurring between the ages of 4 and 6 years. There is increasing evidence that early treatment affects the outcome in DMD,^{4,21,22} and early access to therapies will become increasingly important as novel, more effective treatments become available.^{15,17} Other studies have also shown that delays in DMD diagnoses are frustrating and stressful for parents particularly when they are given false reassurance and/or alternate/incorrect diagnoses.^{9,23,24} Indeed, these experiences have been mirrored by parents of children with a range of other genetic conditions including fragile X syndrome (FXS),²⁵ Klinefelter syndrome²⁶ and childhood spinal muscular atrophy (SMA).²⁷

Diagnostic delay in DMD, and other genetic conditions, may occur for several reasons. First, health professionals may not recognize developmental delay given the wide normal range in early childhood.²⁸ The heterogeneity of presentations may slow the recognition of symptoms related to these conditions.²⁹ Diagnostic delays may occur due to a lack of awareness and knowledge amongst health professionals, particularly those in the community. Also, children who are initially managed by allied health professionals may not be promptly diagnosed with DMD as allied health professionals (eg, physical, occupational or speech therapists) cannot order screening tests for DMD,¹⁴ or because models of care may focus on managing symptoms rather than ascertaining a diagnosis.

Parental factors may also have a role in the timeframe of diagnosis of DMD. During interviews, parents described struggling between feeling that 'something was wrong' and feeling that the symptoms were 'not a big deal' until they reached a 'tipping point' of finally searching for a diagnosis; parents who reach this 'tipping point' earlier may receive earlier diagnoses. It is also possible that parents who were assertive and persistent might be more likely to persevere in searching for a cause for their child's symptoms.

Half the parents in the survey felt that their child could have been diagnosed earlier and, although there was frustration about the delay between noticing signs and ultimately receiving a diagnosis of DMD, parents reported appreciating having that period with their child who appeared normal and healthy before signs of DMD were evident. Strategies to earlier diagnosis could include: raising greater awareness of DMD through continuing professional development and education of health professionals with guidelines that are widely disseminated; screening of children at various ages, such as in infancy (1–2 years of age), and which might be targeted to children showing very early signs of developmental delay; newborn screening, which is currently offered as a clinical service in Antwerp, Belgium and, until late 2011, was available in Wales, United Kingdom.³⁰ Newborn screening for conditions in which there are limited treatment options is controversial due to concerns about the impact a positive screening result may have on the parent–child relationship.³¹ Welsh data suggest that a DMD diagnosis at birth had not negatively impacted bonding; however, those parents felt that their sons were 'more precious' and more likely to describe their sons as 'cuddly' compared with parents from the general population.³² Further research is needed to explore

options for screening that allow the benefits of earlier identification of DMD and other conditions presenting in early childhood, while minimizing the potential for harms. For some conditions, such as FXS and SMA, carrier screening is also an option, but would be less useful in DMD because of the high rate of sporadic variants.

A number of study limitations must be acknowledged. Assumptions were made to identify patients tested for the purpose of diagnosis of DMD in the audit (ie, excluding records from children > 10 years). This may have underestimated the average age at DMD diagnosis. Sample bias may have resulted from only proactive parents and those members of muscular dystrophy support associations participating in the study. Non-members may have differing experiences and opinions. A response rate for this study could not be determined because invitations to join the study were sent by the support associations and the RCH Neuromuscular Clinic as per the requirements of the ethics committee and these groups were unable to provide the number of invited eligible parents.

CONCLUSION

The age of DMD diagnosis has not significantly decreased internationally nor in Australia. Parents' experiences and the key factors contributing to the diagnostic delay for families have been described. Clearly strategies need to be put in place to assist in minimizing the diagnostic delay. A variety of approaches, including population-based screening, may be required and evaluated that also consider parental psychosocial impacts.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

ACKNOWLEDGEMENTS

We are grateful to participating families, to Daniella Villano and Anita Mach of the RCH Neuromuscular Clinic for distributing invitation letters and for helping with the clinical audit. We thank the following for distributing the survey to their members: Muscular Dystrophy Australia; Muscular Dystrophy New South Wales; Muscular Dystrophy Tasmania; Muscular Dystrophy Western Australia; Duchenne Foundation and Muscular Dystrophy South Australia. We thank Boris Struk for his encouragement and support of the study and to the Victorian Government's Infrastructure Support Program.

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