Peer reviewed article
Do males diagnosed with Duchenne muscular dystrophy (DMD) unnecessarily suffer bowel and bladder problems?

Abstract
Forty-eight males aged 4-28 years diagnosed with Duchenne muscular dystrophy (DMD) participated in an exploratory study to investigate the type and extent of reported bowel and bladder problems. Using parental interviews, this study aimed to identify the types and incidence of bowel and bladder voiding problems and explore associations with mobility, eating and drinking patterns. One third of the study group reported urinary and bowel urgency and constipation, and did not urinate during a school or workday. Urinary frequency and hesitancy, bowel pain and discomfort were reported in a quarter of the group. The high levels of reported urinary problems, especially in the transition group, as well as the lack of association between bowel problems and mobility, were of interest.

Introduction
Duchenne muscular dystrophy (DMD) is the most common of the muscular dystrophies. It is an inherited x-linked recessive condition affecting approximately 1:3300 male births. It causes progressive weakness of limb and trunk musculatures, some cognitive effects and cardiomyopathy. Scoliosis is common to almost all people diagnosed with DMD after losing independent ambulation. Sphincter muscles are largely spared and there is no recognised link between bladder dysfunction and DMD.

Therapists working with people with DMD throughout Queensland, Australia noted that a considerable number of them had voiding problems; this led to the current study. While there is a wide variety of literature on the development, aetiology, diagnosis and progression of DMD, a systematic review of the literature found there was a dearth of literature relating to practical aspects of humans diagnosed with DMD, their lifestyle and bowel and bladder voiding. Since there was limited published research reported in the literature, an investigation was undertaken to identify the nature and extent of bladder and bowel voiding problems among boys and young men with DMD. Parker and colleagues investigated an adult DMD population (n=25) over 7 years and reported a need for improved services. Therefore the current study was undertaken with a view to developing better resources and care protocols for clients and their families.

This paper reports on urinary and bowel continence problems reported among boys and young men diagnosed with DMD and any relationships these may have with mobility and some aspects of diet.

The function of the digestive and urinary tracts in humans with DMD is not generally thought to be affected until the very late stage of DMD or as a result of surgery or scoliosis. Until recently, there was mainly circumstantial evidence from the occasional occurrence of bladder paralysis, paralytic ileus and gastric dilation. Caress and colleagues investigated seven boys aged 7-14, diagnosed with DMD, who experienced urinary incontinence. Most had undergone spinal fusion and were found to have problems with bladder wall sensitivity and sphincter control. The authors concluded that the urinary incontinence was most likely due to severe scoliosis or complications from spinal surgery. The problems in these cases were treatable.

Urinary and bowel continence issues
A 2003 study identified 46 from a group of 74 males aged 3-31 years diagnosed with DMD as having urinary problems. Both ambulant and non-ambulant males reported urinary problems, including day and night-time incontinence, nocturia, urinary frequency, urge urinary incontinence and stress incontinence. Urinary hesitancy affected older participants, with a mean age of 16.7 years.

Bowel evacuation dysfunction is known to be a problem for people with neuromuscular diseases, especially in the non-ambulant phase. Both constipation and diarrhoea have been reported in DMD and it has been suggested that both increased colonic transit time and pseudo-obstruction are implicated. These may be caused by atrophy and fibrosis of the intestinal smooth muscle. A single case study in 1996 reported on acute gastric dilation in a young man with DMD.
Mobility issues

DMD causes progressive muscular weakness which is first evident in the larger muscles of the body. Early milestones are normally reached, though motor and speech milestones may be delayed. Gait abnormalities, difficulties running and climbing, toe-walking and frequent falls may lead to diagnosis of the condition, often in the toddler or pre-school years. As the condition progresses, the boys enter a transition period when walking is still possible, but slow, with frequent falls and increasing fatigue, and rising becomes difficult. Until recently, walking ceased on average at age 10 years. However, the use of prescribed corticosteroids, in recent times, has substantially lengthened the period of transition. Decreasing independence mobility leads to increased dependence for daily functions like toileting.

Dietary issues

Malnutrition and obesity are reported to be equally common in young adults with DMD, each occurring in about 44% of individuals, with younger boys tending to experience obesity and older ones malnutrition. The author’s observations in the field indicate a high incidence of fussy eating, with limited variety of foods consumed. Dehydration has been linked to several health issues, including constipation and UTI. To prevent dehydration, the Australian Government Department of Health and Aging recommends consumption of 1.5 litres of fluid daily.

Methodology

The Mater Human Research Ethical Committee approved this Montrose Access study. A non-experimental study design was employed. It involved surveying parents through the use of a telephone or face-to-face interview. In total, 48 interviews were conducted. Selection criteria included that the participants (i.e. children of parents interviewed) were current clients, or past-clients of Montrose Access, aged between 4-28, and had been diagnosed with DMD. Montrose Access is the main service provider for this population in Queensland. The parents were contacted, informed of the purpose and design of the project, and invited to participate. Those who agreed to participate signed and returned a consent form. The boys diagnosed with DMD were invited to sign an assent form.

A semi-structured interview method was used. The young men with DMD answered all the questions they were able to and a parent answered the remainder, which included questions relating to early development and school results. In these cases, the parent was also asked to verify some of the answers. The purpose of the questionnaire was to identify toileting problems of people diagnosed with DMD, including identification of bladder and bowel voiding problems in relation to mobility and eating habits.

The selection of urinary voiding variables was based on common abnormalities of the urinary system, including frequency, enuresis, hesitancy, urgency, incontinence and stress incontinence. Young men diagnosed with DMD may reduce fluid intake to avoid the need to toilet during their work or school day and therefore this was included as a variable. Other symptoms reported in the pilot study included bowel urgency and faecal incontinence. These were reported to be separate from diarrhoea. In order to determine chronicity, participants were asked to report the frequency of these problems over the last 2 years.

A pilot survey format was trialled with 10 clients diagnosed with DMD. Feedback was sought on content, layout and wording. The final questionnaire consisted of a combination of open-ended questions, forced choice (yes/no) and 5-point scales (from never to always).

The statistical data, including the bowel and bladder voiding variables and the three factors, were analysed using Statistical Package for the Social Sciences (SPSS version 13) software. Normality of variables was assessed and, as a result, all bowel and bladder variables, as well as eating factors, were collapsed from five to three categories (frequently, occasionally and never) to achieve normality and usable cell sizes. Bowel and bladder problems were also combined into two composite bowel and bladder groups. Data were analysed with 3-by-3 analysis of variance (ANOVA) and Tukey’s post hoc tests to understand the relationship between the groups.

Results

The 48 participants surveyed were aged between 4-28 years, with a mean age of 14.7 years (SD=5.4). Participants lived at home with their families. One participant was below school age. Thirty-four percent of participants attended a special school or spent a significant part of the week in a special education unit. Twenty-four participants were from Brisbane or Ipswich, 16 from rural Queensland, six from the Gold Coast and two from the Sunshine Coast. Twenty eight percent of participants used prescription corticosteroid treatment to delay the progressions of muscle weakness. Nineteen participants (40%) had spinal fusion surgery.

Prevalence of voiding problems

Over one third of the participants were reported to have problems ‘over half the time’ with daytime urinary urgency, constipation, voiding postponement for over 6 hours or longer and daytime bowel urgency. Between a quarter and a third of the participants were reported to have problems ‘over half the time’ with daytime urinary hesitancy, urinary frequency and bowel pain. Nocturnal bowel and bladder problems and bowel or urinary incontinence were reported in less than 20% of the cohort. Bowel problems other than diarrhoea were not reported in children younger than 9 years. Urinary hesitancy was reported in participants 12 years and over (Table 1).
Prevalence of factors
Of the 48 participants, 36 were non-ambulant, used an electric wheelchair and were unable to weight-bear. Nine participants were in the transition stage, being ambulant, but intermittently used a mobility aid such as an electric scooter or manual wheelchair for longer distances. No participant used callipers or walking devices. Three participants were competent walkers.

Forty-four percent of the participants drank less than a litre of fluid daily. Fifty-six percent drank in excess of 1 litre of fluid daily. Sixty percent of participants had average food consumption and 15% above average. Twenty-five percent of the participants ate less than average and some were reported to eat only one meal a day. Although exact content of diets was not assessed for this survey, 29% of participants were reported to be fussy eaters who consumed a limited variety of foods.

Relationships between bladder problems and mobility
When urinary problems affecting more than 20% of participants were combined into a composite score, a significant variance was found between the means of the composite urinary scores and the mobility scores (F(2,45)=3.543, p=0.037). As mobility decreased, overall urinary problems increased (Figure 1).

When analysis of variance was performed on separate urinary problems, significant differences were found between the mean of mobility and the means of urinary hesitancy (F (2,45)=5.536, p=0.007) and voiding postponement (F(2,45)=3.462, p=0.040). Variance between urinary frequency and mobility was also significant (F(2,45)=3.847, p=0.029). The transition group experienced the main problem with frequency, whilst urinary hesitancy was experienced by the non-ambulant group. As expected, the non-ambulant group employed the strategy of voiding postponement to avoid toileting during their day at school or work (Figure 2). Voiding postponement was significantly related to reduced drinking (F(3,44)=5.075, p=0.010).

Relationships between bowel problems and diet
Constipation was highly correlated with bowel pain (p=0.001) and bowel urgency (p=0.011) and these three variables were therefore collapsed to form a composite bowel problem variable. A significant association was found between the means of the bowel composite scores and the fussy eater group (F(4,43)=3.936, p=0.008). Post hoc analysis showed that participants who were fussy eaters experienced greater problems with bowels (Figure 3). Specifically, post-hoc analysis showed fussy eaters experienced more frequent bowel pain (F (4,43)=3.433, p=0.041) and bowel urgency (F(4,43)=4.320, p=0.005).

Qualitative results
Twelve of the 17 participants reporting constipation had sought medical advice for this problem. Of this group, five participants used stool softeners or laxatives regularly, and three had received whole bowel irrigation to resolve faecal impaction. Four used enemas or suppositories regularly. Two participants reported that they used to have constipation, but that this was no longer a problem after 18 years of age. One of these attributed this to improved drinking habits. The other had no explanation. Antibiotics were thought to cause constipation in the short-term for two participants, and bowel urgency in another.

Two participants reported regular UTIs as a result of voiding postponement and two participants required exceptionally frequent toileting after school. Only three participants had sought medical advice for urinary problems: one was successfully treated for bed wetting with medication and two had been assessed and treated for UTI. None of the participants had urodynamic assessments performed.

The importance of good positioning and appropriate equipment was mentioned by seven of the parents. For example, one mother said, “He can’t start the flow if he is sitting wrongly”. Urinary hesitancy was overcome in several cases by reclining the chair, thus widening the hip angle.

Of 19 participants with spinal fusion, a surgical procedure to manage deformity, only two participants reported bowel or bladder problems following surgery. One participant used enemas for some months following surgery. Another reported that, since surgery he had, “...no sensation, no desire to go; no indication of the needs to go until after it happens”. One participant reported that he had difficulty releasing urine since the development of his scoliosis, and had found that this could be relieved by leaning back.

Anxiety or mental discomfort played a part in hindering effective voiding in four participants. Parent comments included: “He wouldn’t use public toilets and had to work to overcome this”, and “Listening to music helps him to relax and concentrate with urinary hesitancy problems”. Thirteen parents reported that effective bowel and bladder evacuation was hindered by the fact that their sons needed assistance and equipment with toileting. Some parents reported that school staff would call them from home or work to assist with their sons toileting, particularly with bowel movements.

Discussion
The three most surprising findings of this study were the high levels of urinary problems reported, the lack of association between bowel problems and non-ambulance, and the high levels of bowel and bladder problems experienced by the transition group.

The overall frequency of reported bowel and bladder voiding problems in this study’s cohort of DMD boys in Australia was found to be significant; the frequency of urinary problems
in particular supports MacLeod’s findings \(^2\) of high incidence among people diagnosed with DMD, regardless of their age. Urinary urgency, described as a strong and immediate desire to urinate, was the most frequently reported problem, experienced by 40% of the cohort over the last 2 years. This problem was experienced in the daytime rather than the night.

There were no significant associations between urgency and other mobility or dietary factors reported from the youngest participant (4 years) through to those aged 25 years. However, as part of the urinary problems composite score, it contributed to an association with mobility scores and with an increase in urinary problems associated with a decrease in mobility. In practice, management of toileting issues once boys are non-ambulant becomes more complex \(^3\), \(^11\), and urinary urgency further exacerbates this situation. Medically, urinary urgency may be associated with bladder conditions as identified by MacLeod \(^1\), and requires referral to a general practitioner initially. In this study, no participants had sought medical advice for urinary problems, except when they were acute and associated with UTI.

One third of the cohort reported urinary voiding postponement. While voiding postponement is not intrinsically a problem, it can contribute to significant problems. Reducing fluid intake can impact on bladder and bowel health as well as pulmonary health \(^2\). People diagnosed with DMD are advised to drink sufficient fluids to prevent thickening of the pulmonary mucus associated with the risk of pneumonia \(^13\), \(^24\). Parental comments indicated problems resulting from this practice included UTIs and urinary frequency after school. Robson & Leung \(^21\) comment that incontinence, frequency, and urgency are common symptoms of urge syndrome and that voiding postponement may play a causative role.

Urinary hesitancy was experienced by almost a third of the cohort. As in MacLeod’s study, younger (under 12 years old) participants did not experience hesitancy \(^2\). Partial urethral obstruction or altered bladder tone could be associated with this complaint \(^19\) and medical referral is indicated. In the current study, no participants sought medical investigations for urinary problems apart from bed-wetting or UTIs.

Urinary hesitancy could also be related to psychological factors if the presence of an assistant during toileting procedures prevents sufficient relaxation \(^11\). Physical factors such as positioning or lack of abdominal muscle strength may also contribute to this problem \(^6\).

Urinary frequency, reported by a quarter of the cohort, was significantly associated with mobility; however, it was
Table 1. Prevalence and t-test of urinary and bowel voiding problems experienced half the time or more in the last 2 years and mean age of subjects (n=48).

<table>
<thead>
<tr>
<th>Bowel and bladder dysfunction</th>
<th>n</th>
<th>%</th>
<th>One-sample test</th>
<th>Age range of incidence</th>
<th>Mean age of incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Daytime urinary urgency</td>
<td>19</td>
<td>40</td>
<td>t=2.98 *</td>
<td>4-25</td>
<td>14.85</td>
</tr>
<tr>
<td>Constipation</td>
<td>17</td>
<td>35</td>
<td>t=6.61 *</td>
<td>9-25</td>
<td>15</td>
</tr>
<tr>
<td>Voiding postponement</td>
<td>16</td>
<td>33</td>
<td>t=6.09 *</td>
<td>9-25</td>
<td>16.44</td>
</tr>
<tr>
<td>Daytime bowel urgency</td>
<td>15</td>
<td>31</td>
<td>t=5.19 *</td>
<td>4-25</td>
<td>14.07</td>
</tr>
<tr>
<td>Urinary hesitancy</td>
<td>13</td>
<td>27</td>
<td>t=6.50 *</td>
<td>12-25</td>
<td>17.85</td>
</tr>
<tr>
<td>Daytime urinary frequency</td>
<td>12</td>
<td>25</td>
<td>t=7.03 *</td>
<td>8-25</td>
<td>14.17</td>
</tr>
<tr>
<td>Abdominal pain or discomfort</td>
<td>12</td>
<td>25</td>
<td>ns</td>
<td>9-25</td>
<td>14.58</td>
</tr>
<tr>
<td>Faecal incontinence</td>
<td>9</td>
<td>19</td>
<td>t=9.18 *</td>
<td>10-25</td>
<td>14.89</td>
</tr>
<tr>
<td>Nocturnal urinary frequency</td>
<td>7</td>
<td>15</td>
<td>ns</td>
<td>8-25</td>
<td>17.57</td>
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<tr>
<td>Diarrhoea loose bowels</td>
<td>5</td>
<td>10</td>
<td>ns</td>
<td>6-18</td>
<td>11.6</td>
</tr>
<tr>
<td>Nocturnal urinary incontinence</td>
<td>3</td>
<td>6</td>
<td>ns</td>
<td>14-19</td>
<td>16.37</td>
</tr>
</tbody>
</table>

*Sig. (2-tailed)<0.005, df=47
the transition group that experienced the greatest problem with frequency rather than the non-ambulant group. This may indicate a social-emotional basis as this period involved increasing awareness of physical decline. The transition group also experienced more urgency and hesitancy than the walkers, but less than the non-ambulant group. Robson & Leung suggest that children with urge syndrome normally squat to cope with the urge to urinate, whilst children with DMD are limited in their ability to squat due to muscle weakness, causing the need to attend to toileting more frequently. Once the boys are in wheelchairs, there is the added embarrassment of needing to ask for assistance to toilet and requiring more time, suitable equipment and space. This may mean they postpone their voiding and consequently experience urgency and hesitancy.

Constipation was reported by 35% of participants, considerably higher than the 15% reported in the general population. Constipation is often associated with lifestyle issues of activity levels and diet, but may also be caused by colon disorders including irritable bowel syndrome or drug induced including from calcium supplements or laxative abuse. Symptoms may include abdominal pain and this variable as well as bowel urgency was highly correlated with constipation. Based on the literature, constipation is expected to increase with reduced mobility; however, in this study, there was no significant association found between bowel problems and mobility. However, there was a significant association between bowel problems and diet, specifically fussy eating and reduced fluid intake.

Fussy eaters consumed a limited variety of foods and tended to experience bowel pain and bowel urgency. Participants with reduced fluid intake experienced bowel urgency. Bowel urgency is often associated with diarrhoea, however, in this study diarrhoea was reported by only 10% of cases, whilst urgency was reported by 31%. Other possible causes of increased reports of bowel problems in this population may include higher levels of anxiety brought on by dependency in toileting procedures and inability to wipe small leakages of faecal matter independently during urination.

A limitation of this study was its reliance on parent reporting and therefore a dependence on parent perspectives and memories. The questions about diet were general and did not elicit information on dietary fibre content specifically. Medical verification and follow-up were not within the scope of this study. A similar study, broadened to incorporate more specific dietary, medical and psychosocial factors, specifically anxiety and mental discomfort, would be most valuable.

**Conclusion**

The survey found that up to 40% of males diagnosed with DMD experienced bowel and bladder problems and only the latter were statistically associated with non-ambulance. No participants had sought medical advice or assessment for urinary problems of hesitancy, frequency and urgency. A high percentage of non-ambulant participants practised voiding postponement during the day, with significant association between this and reduced
drinking. Bowel problems were not statistically associated with non-ambulance but there was a significant association with dietary factors.

It is recommended that young people diagnosed with DMD who experience voiding problems be offered holistic care that includes referral for medical examination and follow-up, dietary advice, support at home and school with toileting routines, and provision of appropriate equipment. To assist young men diagnosed with DMD, and parents and teachers to learn to manage faecal and urinary continence issues associated with DMD, a booklet with the title Some guidelines for management of toileting issues in boys with Duchenne muscular dystrophy has been prepared as an outcome of this study.

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References
22. SPSS Version 13. Chicago, USA.

Nominations open for the Australian and New Zealand Continence Journal Editorial Committee

The past two years have been an exciting time for the development of the Journal. The introduction of peer review articles has resulted in increased international inquiry and a greater presence in abstracting databases. These changes have been led by a dynamic Editorial Committee.

The current committee will spill as of the Continence Foundation of Australia Annual Meeting in November. The committee currently comprises eight members and nominations are sought from clinicians from the disciplines of medicine, nursing and physiotherapy residing in Australia and New Zealand.

The Editorial Committee – led by Journal Editor, A/Prof Peter Dietz and supported by Production Editor Jacinta Miller – oversees the organisation, compilation, production and quality of the Journal. The Editorial Committee term is for 2 years. The committee meets via teleconference four times a year.

A copy of the Editorial Committee Terms of Reference is available from the Production Editor. Nominations forms are also available from the Production Editor and are to be returned via fax (03) 5493 7755.

Nominations close Friday 29 September 2006.

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