SHORT REPORT

Bladder dysfunction in Duchenne muscular dystrophy

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Although bladder function is thought to be unaffected in Duchenne muscular dystrophy, 46/88 boys interviewed had urinary problems. Nine underwent video urodynamics, showing in eight a small capacity, hyperreflexic bladder, and in the ninth (post spinal surgery) hyperreflexia and detrusor sphincter dyssynergia. Urinary dysfunction is a treatable feature of DMD.

The main clinical phenotype in Duchenne muscular dystrophy (DMD) is of progressive weakness of limb and trunk musculature, a degree of cognitive impairment, and later, cardiomyopathy. There is no recognised association between bladder dysfunction and DMD. As a number of patients admitted to urinary problems when asked, we decided to investigate in detail urinary symptoms in DMD.

METHODS

Eighty eight males with DMD (aged 3–31 years), attending the neuromuscular clinic, were included in the study. A questionnaire was sent to them or their carers and followed up by telephone.

Those who reported urinary symptoms were interviewed and examined to exclude other causes of urinary dysfunction. Renal and bladder ultrasound pre and post micturition, and video urodynamics were then offered to define the pattern of lower urinary tract dysfunction.

RESULTS

Eighty four per cent (74/88) replied, of whom 46/74 (62%) were concerned about urinary problems; these are summarised in table 1. All 22 boys with daytime urinary incontinence had been continent at some stage and the problem had become apparent later. Twelve with nocturnal incontinence had been previously dry at night. Twenty used continence aids but only eight had been referred to a continence advisor. Of the 50 who were no longer able to walk independently, only 38 had some adaptation to their bathroom. Although 38 boys had learning difficulties, in only four were they severe enough to affect acquisition of continence.

Urinary problems occurred in young, ambulant boys as well as wheelchair dependent patients, with a mean age of 10.8 years (range 3–25years). In all but one patient, who suffered hesitancy post spinal surgery, the problem had not been brought to medical attention previously. Day and night time incontinence, nocturia, urinary frequency, urgency and stress incontinence were found at a mean age of 10.3 years (range 3–25 years). Urinary hesitancy affected an older group with a mean age of 16.7 years (range 11–22 years).

Table 1 Urinary symptoms experienced by 46/74 males with DMD (aged 3–25 years)

Urinary symptoms	Number of boys affected	Ambulant	KAFOs*	Wheelchair dependent	Spinal jacket	Spinal surgery
Daytime incontinence	22	11	3	8	1	0
Urinary frequency	14	5	2	7	4	1
Urinary urgency	18	7	4	7	4	0
Nocturnal enuresis	21	12	4	5	3	0
Nocturia	22	6	5	11	6	0
Stress incontinence	5	2	0	3	0	0
Urinary hesitancy	5	0	0	5	3	2

*Ischial weight bearing knee-ankle-foot orthoses.

Table 2 Symptoms of boys having urodynamic investigations

Boys	Age (y)	Daytime incontinence	Urinary frequency	Urinary urgency	Enuresis	Stress incontinence	Nocturia
1	7	✓	✓	✓	✓		✓
2	8	✓	✓	✓	✓	✓	
3	9	✓	✓	✓			
4	9	✓		✓	✓		
5	10	✓		✓	✓		✓
6	10	✓	✓	✓	✓		
7	10	✓	✓	✓	✓		
8	11	✓		✓	✓		
9	11	✓		✓	✓		
10*	14		Reduced with hesitancy				

*Symptoms developed post spinal fusion surgery.

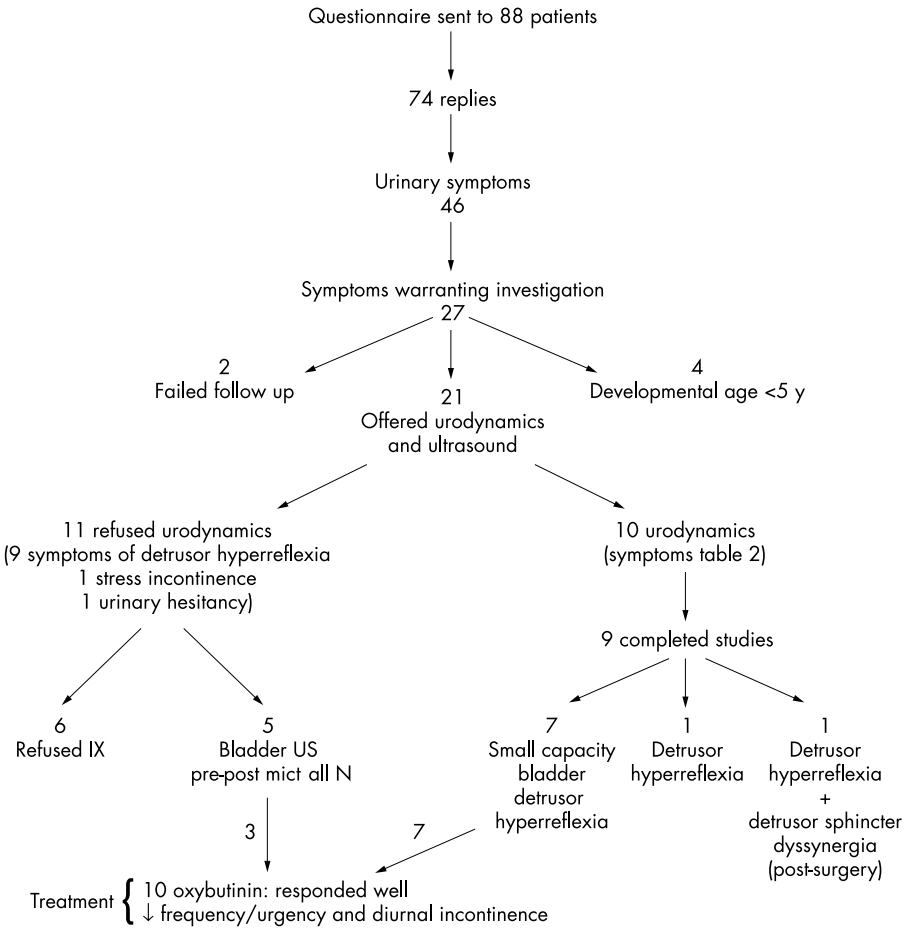

Figure 1 Results of investigations.

Symptoms were severe enough to warrant further investigation in 27 patients (fig 1). Of these, two failed follow up, and four had a developmental age of less than 5 years and were observed only. Pre and post micturition bladder and renal ultrasound scan and videourodynamics, were offered to 21. Ten had both investigations, five ultrasound only, and six refused investigation. All ultrasound scans were normal, with complete bladder emptying. Table 2 shows symptoms in the 10 who underwent urodynamics. One study was incomplete. The remaining nine were abnormal, with hyperreflexic detrusor contractions in eight, of whom seven also had a small capacity bladder. One patient (number 10 in table 2) had detrusor hyperreflexia and detrusor sphincter dyssynergia, having developed urinary symptoms following spinal surgery.

Of the 11 patients who refused urodynamics, nine had symptoms consistent with detrusor hyperreflexia, one had stress incontinence, and one had urinary hesitancy.

Ten patients were treated with oxybutinin (seven post urodynamics) and all responded well to treatment, reporting a reduction in frequency, urgency, and diurnal incontinence, and improvement in quality of life. Those with nocturnal incontinence became dry.

DISCUSSION

We were surprised by the frequency of urinary symptoms in DMD, particularly in the younger, ambulant boys, who appear to have an acquired problem with reduced bladder capacity and detrusor hyperreflexia. Previous studies have concentrated on older boys, when problems with immobility, constipation, scoliosis, spinal surgery, and depression may be implicated. For example, Caress and colleagues[1] carried out videourodynamic investigations in seven boys age 14–17 years, most of whom had undergone spinal fusion. They reported uninhibited, high pressure detrusor contractions in four, associated with detrusor sphincter dyssynergia in two, which was treated with anticholinergic medication. Electromyography studies of the pelvic floor were normal. Boland and colleagues[2] carried out a retrospective cohort study of 33 boys with DMD during their second decade, and found only two boys with urinary symptoms both of whom had urinary retention.

The neurological basis for this dysfunction is difficult to explain, except where symptoms clearly follow spinal surgery. Myopathic involvement of the detrusor would be expected to reduce the strength of contractions and cause a large capacity, flaccid bladder. Weakness of striated muscle of the pelvic floor, with impaired contraction, would account only for stress incontinence, as reported in five of our boys. Disturbance of neural control, affecting somatic or autonomic pathways, is therefore most likely. Early autonomic dysfunction has been reported in DMD in relation to the heart, with increased sympathetic activity associated with a decrease in vagal tone[3]; however, this would be expected to cause a hypotonic bladder with difficulty emptying, the opposite of our findings. Tests of autonomic function in skin have not shown any abnormality in DMD.[4] Dystrophin isoforms are expressed in the brain,[5] so that their absence in DMD could be implicated in impaired bladder control as well as cognitive difficulties. Mild learning difficulties were apparent in three of those who underwent urodynamics, although it is recognised that at least selective learning difficulties affect most DMD boys.[6]

Identifying and treating urinary dysfunction in DMD is important; symptoms are often assumed by parents to be part of the condition. We would recommend a history and examination to exclude other causes of urinary dysfunction. If symptoms are suggestive of detrusor hyperreflexia, a pre and post micturating bladder ultrasound should be carried out to ensure complete bladder emptying, and a trial of oxybutynin considered.

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Sadly, Dr Borzyskowski died on 17 October 2002

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IMAGES IN PAEDIATRICS

An unusual life threatening cause of torticollis in a child

An 8 year old girl was admitted to hospital because of fever, neck stiffness, and enlarged cervical lymph nodes. A diagnosis of lymphadenitis was made and she was treated in hospital two days with intravenous cefuroxime followed by oral cephalexin. Five days after discharge, the girl was readmitted to hospital for severe torticollis. She was not able to move her head but had no other abnormal neurological signs. The white blood cell count was 14.2 × 10^9/l and serum C reactive protein 23 mg/l, increasing within two days to 97 mg/l.

Magnetic resonance imaging (MRI; see fig) revealed potentially life threatening atlantoaxial subluxation (1) with atlas rotation of 40°, inflammatory changes in the atlantoaxial joint (2), and enlarged cervical lymph nodes (3)—Grisel's syndrome, an infectious process of the atlantoaxial joint.[1] She was treated with intravenous cefuroxime and clindamycin, analgesics, muscle relaxants, and neck support for nine days. Cefuroxime axetil, clindamycin, and neck support were continued for five weeks at home. MRI obtained three months later showed a normal atlantoaxial joint and the girl was asymptomatic.

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