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The prevalence of faecal incontinence in myotonic dystrophy type 1

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Abstract

Faecal incontinence is recognised as a feature of myotonic dystrophy along with other symptoms of bowel dysfunction, but its prevalence is poorly defined. We have surveyed 152 unselected myotonic dystrophy patients. We identified issues with bowel control in 104 (68% of the study population). Forty-eight (32%) reported faecal incontinence in the 4 weeks prior to completion of the questionnaire. Fifty-six patients (37%) reported having to change their lifestyle because of incontinence issues at some point in the prior 4 weeks. This study shows a high frequency of life-changing symptoms in a large unselected, cohort of patients with myotonic dystrophy type 1, and highlights lower gastrointestinal symptoms as an important issue for further research.

Keywords: Myotonic dystrophy type 1; Faecal incontinence; Bowel dysfunction.

1. Introduction

Myotonic dystrophy type 1 (DM1) [1] is an autosomal dominant multi-system disorder. The range of clinical features is well described [2–4]. The clinical picture is dominated by the consequences of muscle involvement, with associated risks of respiratory failure and cardiac arrhythmias. More recently the major impact of brain involvement on individuals and families has been recognised [5,6]. However, in addition, a major feature causing distress for patients and families relate to symptoms of bowel dysfunction. This has long been acknowledged but our understanding of its overall prevalence, severity and optimal management remain poorly understood [7,8].

The entire GI tract may be affected, with dysphagia and the risks of aspiration a major cause of morbidity. Disorders of gut motility are also well described and have been reviewed [7,9]. Anal incontinence has been reported in up to 30% of patients [8]. However the overall prevalence of lower bowel dysfunction and faecal incontinence has not been estimated in a large unselected population. This feature has a severe impact on patients and families' quality of life [10] and it is likely the overall prevalence of such debilitating symptoms may have been underestimated because of embarrassment.

2. Methods

This study ran during the period April 2016 until August 2017. Patients attending the myotonic dystrophy management clinics based in Edinburgh and Glasgow were approached. These clinics offer appointments to all known affected individuals in their own geographical area. 152 consecutive responses were obtained during this period of time. The questionnaire was handed to the patient prior to the clinic

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The Vaizey Incontinence Score

In the Prior 4 weeks

	Never	Rarely	Sometimes	Weekly	Daily
1. Incontinent for solid stool	0	1	2	3	4
2. Incontinent for liquid stool	0	1	2	3	4
3. Incontinent for gas	0	1	2	3	4
4. Alteration in lifestyle	0	1	2	3	4

5. Need to wear a pad or plug	No scores 0, Yes scores 2
6. Taking constipating medicines	No scores 0, Yes scores 2
7. Lack of ability to defer defecation for 15 minutes	No scores 0, Yes scores 4

Definitions - **Never**, no episodes in the past four weeks; **rarely**, 1 episode in the past four weeks; **sometimes**, >1 episode in the past four weeks but <1 a week; **weekly**, 1 or more episodes a week but <1 a day; **daily**, 1 or more episodes a day.

Total maximum score possible 24.

Fig. 1. The Vaizey incontinence score.

appointment and completed independently without advice or collaboration. There was no selection on the basis of severity of disease nor place of residence. The study was approved by the South East Scotland Regional Ethics Service.

We applied a previously published questionnaire, the Vaizey (or St Mark's modified) incontinence score [11] (Fig. 1) to assess the prevalence and severity of faecal and flatus incontinence, as well as the impact on day to day activities. This scale was chosen as it contains enquiries we felt most relevant to the reported symptomatology of DM1. Many of the other pre-existing scales are gastrointestinal disease focussed on or were designed for post traumatic or post surgical problems and so were considered less appropriate for this population and the questions we wished to answer [12].

Demographic details, including muscle impairment rating scale score (MIRS) [13] were recorded by the myotonic dystrophy specialist clinician conducting the clinic appointment. Additional clinical management was recorded from that collected on the basis of the Scottish Myotonic Dystrophy Group clinical standards [14]. Statistical analysis was undertaken using the Statistical Package for the Social Sciences (SPSS, Version 24.0; IBM 2015). Non-parametric tests were used, as most data were not normally distributed. Mann-Whitney U tests were used for comparisons between groups. Correlations were presented using the Spearman's rho correlation coefficient. Values were considered significant at $p < 0.05$. Analysis of covariance (ANCOVA) was used where group comparison required adjustment for additional covariates.

3. Results

3.1. Study cohort

Demographic details of the study cohort are summarised in Table 1. A total of 152 responses were obtained, 70 from female subjects and 82 from males. Age was not significantly different between male and female groups ($p = 0.393$). Among female respondents, 31 (44.3%) were nulliparous. The cohort was heterogeneous with regard to muscle weakness with a

Table 1
Demographics of study cohort.

n.	152
Age in years: mean (range)	45.1 (17 to 75)
Female: number (% total sample)	70 (46.1%)
Females who were nulliparous: number (% total female sample)	31 (44.3%)
MIRS* score: ratio 1:2:3:4:5	6: 16: 52: 67: 11

*MIRS, Muscle Impairment Rating Scale [13]. 1 = no signs, 2 = minimal signs, 3 = distal weakn*ess, 4 = mild to moderate proximal weakness, 5 = severe proximal weakness.

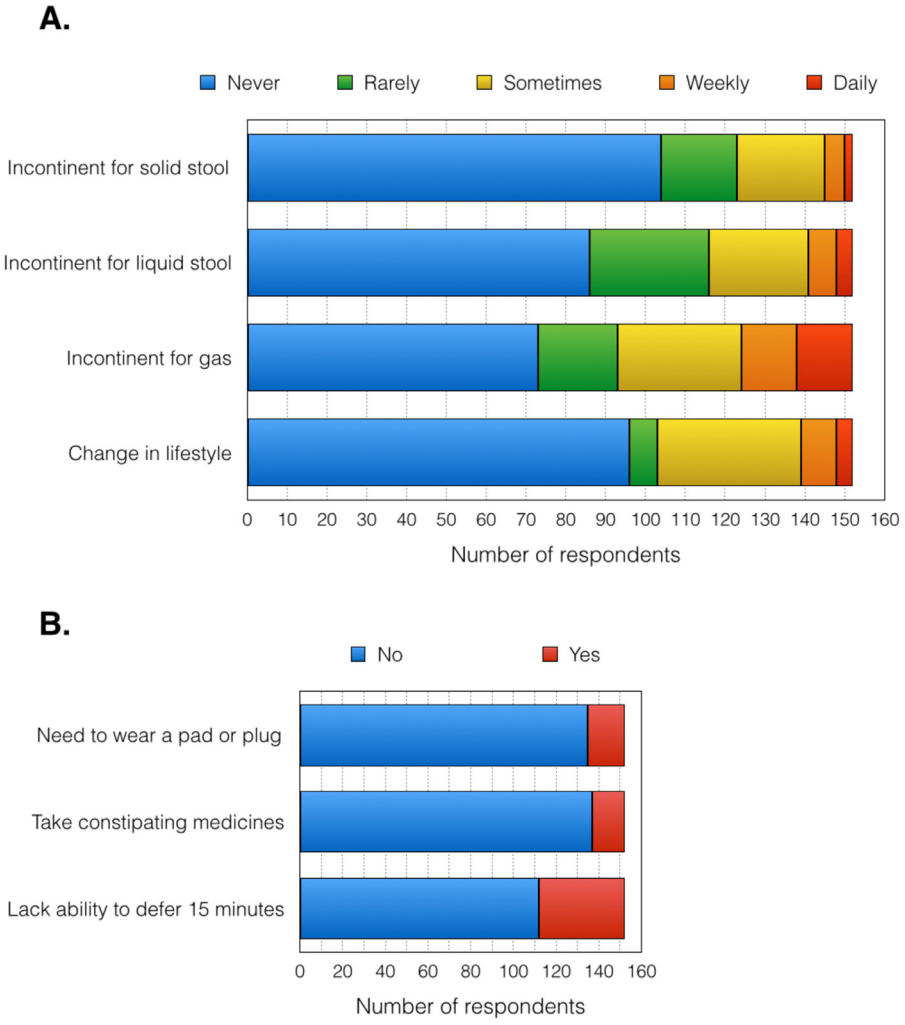

Fig. 2. Summary of responses to items 1 to 4 (A) and 5 to 7 (B) of the Vaizey incontinence score.

modal MIRS score of 4, indicating the majority of participants had established muscle symptoms of DM1.

3.2. Prevalence and characteristics of incontinence symptoms

The mean total incontinence score for the cohort as a whole was 4.8 (range 0–20), from a maximum possible score of 24. Forty-eight individuals had a total score of 0, implying that the remaining 104 respondents (68%) had experienced symptoms in the preceding four weeks.

Responses to the individual items within the incontinence questionnaire are summarised in Fig. 2. In the four weeks leading up to assessment, 48 (31.6%) of respondents had at least one episode of incontinence for solid stool, 66 (43.3%) for liquid stool, 79 (52.0%) incontinence for gas and 56 (36.8%) had changed their lifestyle as a result of bowel symptoms.

Furthermore, 17 (11.2%) of respondents had to use a pad or plug in the preceding 4 weeks, 15 (9.9%) had used constipating medicines and 40 (26.3%) had experienced difficulty in deferring defecation for 15 min.

3.3. Clinical correlates of total incontinence score

Total Vaizey score did not correlate with age ($p=0.768$; $r_s=-0.024$), and showed no significant sex effect (mean 4.93 in males versus 4.64 in females; $p=0.854$). A significant positive correlation was observed, however, with MIRS score ($p=0.010$; $r_s=0.208$). The range of scores for each MIRS category was widely variable (Fig. 3), suggesting peripheral muscle strength alone is not strongly predictive of continence problems.

The influence of previous pregnancy on bowel symptoms was also explored (Table 2). Parous female respondents were, on average, older than nulliparous females ($p=0.001$), but the groups were not significantly different with respect to MIRS score. There was no significant difference in mean total Vaizey incontinence score between the two groups ($p=0.947$). The difference remained non-significant after

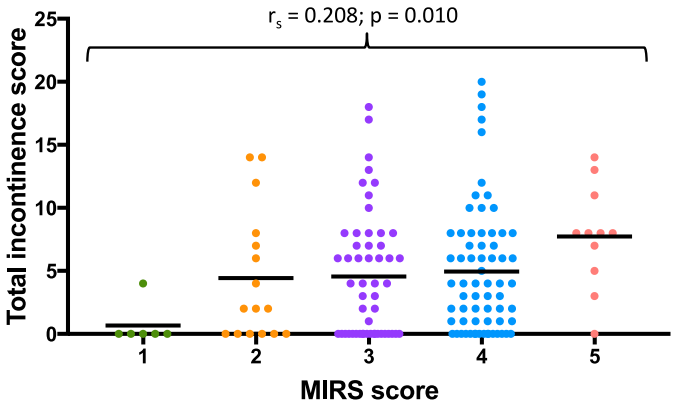

Fig. 3. Total Vaizey incontinence Score categorised by muscle impairment rating scale (MIRS). Horizonal bars indicate the mean score for each group.

Table 2
Group comparison of DM1-affected women with a history of pregnancy and those without.

	Nulliparous female (n.31)	Parous female (n.39)	P
Age in years: Median (range)	36 (21–74)	49 (25–74)	0.001
MIRS score: Median (range)	3 (1–5)	4 (1–5)	0.647
Vaizey incontinence score: Median (range)	4 (0–20)	3 (0–14)	0.947

adjustment for differences in age by one-way ANCOVA ($p = 0.427$).

4. Discussion

We report the findings of the first large survey of faecal incontinence in 152 unselected patients with myotonic dystrophy type 1. The results support the view often expressed by patients, families and support groups that faecal incontinence is a common and life-changing symptom in DM1. Overall 68% reported problems with bowel control. Fifty-six patients (37%) reported having to change their lifestyle at some point in the 4 week period prior to the study. Forty-eight (32%) reported faecal incontinence in the 4 weeks leading to completion of the questionnaire. These figures are similar to the 30% prevalence reported by Ronnblom [8] in a Scandinavian questionnaire -based study involving 40 patients. It was commented by these authors that 25% of their patients thought they faecal incontinence was the worst of the symptoms they recognised. Bellini and co-workers reported that 66% had ‘occasional’ incontinence and 10% were incontinent once or more per week [7].

The impact of faecal incontinence for the day - to - day life and work of any individual is catastrophic with effects on domestic, social and work life [10]. Bowel dysfunction is not specifically addressed in MuSQoL, or the SF-36. It is a component of the MDHI [15] but there remains no Myotonic Dystrophy specific assessment tool for faecal incontinence. The tool used in this work was purely for the purpose of assessing prevalence of symptoms and measuring the impact will require the development of DM-1 specific tools. It is possible the impact of faecal incontinence has long been under recognised by those managing the disorder and may require the development of a specific measurement tool.

There are a number of caveats to this work, Neither the scale we elected to use nor other questionnaires have been widely used nor validated in DM-1 to date. This study did not include a control population. There is evidence to suggest that faecal incontinence may be more prevalent in the general population that many would anticipate with rates (albeit in a gastroenterology clinic setting) of up to 4.5% of patients experiencing incontinence on a weekly basis [16]. We elected to allow completion of the survey in private given the embarrassment associated with faecal incontinence. However, the lack of discussion or explanation of the questions may have distorted data. A number of patients commented on the question regarding “Lack of ability to defer defecation for 15 min” as being unclear whether a yes or no response was appropriate when they were unable to control defecation. We are also unclear as to how honest patients were when completing the survey. It remains possible that embarrassment has still led to an underestimate of the prevalence of this symptom; indeed in previous work on non-DM1 patients it was estimated that only 5 - 27% of patients with faecal incontinence disclosed the problem to others [17]. The patients who attended the clinic may however be a more motivated cohort. Although the questionnaire was offered to all patients attending the clinic, not all patients known to have a mutation in our geographical area attend the clinic. It is therefore possible we may have under-representation from the very mildly affected as well as severely affected. We continue to work toward seeing all patients, but this study does not reflect the problems of the entire population.

We identified a significant trend towards increasing severity of bowel symptoms with increasing skeletal muscle weakness. This is intuitive, since more severe skeletal muscle disease would imply a greater likelihood of smooth muscle involvement also, with potential impact on gut motility and sphincter tone. The contribution of mobility, affecting patients’ ability to respond promptly to the call to stool, was not specifically assessed. Despite this trend, Vaizey scores varied widely within MIRS groups 2 to 5, which is in keeping with previous authors’ observations that primary muscle weakness alone is not strongly predictive of the severity of gastrointestinal symptoms in any individual with DM1 [7,8].

Considering additional risk factors, although all patients may be at risk of pelvic floor weakness secondary to muscle involvement in their DM1 women who had a previous pregnancy were no more severely affected by bowel symptoms than their nulliparous counterparts, suggesting this to have only a minor role in causation. We were unable to explore disease duration, as our clinical population did not

have age of onset recorded. However, our opinion has been, and indeed remains, that such figures are extremely difficult to properly estimate and subject to observer bias and leading questions.

This work was based on our management protocol and so did not record such issues as associated medical conditions or prior trauma, or as stated above past or present drug use. We did not explore additional possible associations with other features of myotonic dystrophy.

Prior studies on the aetiology of large bowel dysfunction and faecal incontinence suggest multi-factorial mechanisms with evidence for abnormalities of pelvic floor changes as well as changes in anal tone [18,19], small and large bowel dysmotility [20,21]. There was evidence of bacterial overgrowth in 65% of 20 patients studied by Tarnoplosky [22] with a 'good response to the use of Ciprofloxacin in 70%. Finally Ronnblom [23] identified evidence of bowel acid malabsorption in 12 or 20 patients studied. In addition there may in some patients be contributions from anxiety and depression as well as the effects of central nervous system changes related to DM1 including frontal disconnection. The better management of an individual patients symptoms will require an awareness of all these potential mechanisms.

In conclusion, taking into consideration the caveats mentioned above, this survey confirms that faecal incontinence is very common in myotonic dystrophy patients. This highlights a need for future studies to explore the pathogenesis of this important symptom, and to develop standardised and validated means of measuring the impact of incontinence issues in individuals with myotonic dystrophy.

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Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.nmd.2019.05.009.

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