Falls and stumbles in myotonic dystrophy

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J. Neurol. Neurosurg. Psychiatry 2006;77;393-396; originally published online 30 Sep 2005;
doi:10.1136/jnnp.2005.066258

SHORT REPORT

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Objective: To investigate falls and risk factors in patients with myotonic dystrophy type 1 (DM1) compared with healthy volunteers.

Methods: 13 sequential patients with DM1 from different kindreds were compared with 12 healthy volunteers. All subjects were evaluated using the Rivermead Mobility Index, Performance Oriented Mobility Assessment, and modified Activities Specific Balance Confidence scale. Measures of lower limb muscle strength, gait speed, and 7-day ambulatory activity monitoring were recorded. Subjects returned a weekly card detailing stumbles and falls.

Results: 11 of 13 patients (mean age 46.5 years, seven female) had 127 stumbles and 34 falls over the 13 weeks, compared with 10 of 12 healthy subjects (34.4 years, seven female) who had 26 stumbles and three falls. Patients were less active than healthy subjects but had more falls and stumbles per 5000 right steps taken (mean (SD) events, 0.21 (0.29) *v* 0.02 (0.02), $p = 0.007$). Patients who fell ($n = 6$) had on average a lower Rivermead Mobility score, slower self selected gait speed, and higher depression scores than those who did not.

Conclusions: DM1 patients stumble or fall about 10 times more often than healthy volunteers. Routine inquiry about falls and stumbles is justified. A study of multidisciplinary intervention to reduce the risk of falls seems warranted.

A fall may be defined as a loss of balance resulting in coming to rest on the floor or ground or on another object below knee level[1]; a stumble can pragmatically be defined as losing balance but regaining it before a fall occurs. Falls potentially cause loss of confidence, restriction of activity, and injury[2] but, in the elderly, multidisciplinary evaluation and measures may reduce falls.[3 4] A third of subjects of 65 years or over have ⩾1 fall per year,[5] but there is little information on patients with muscle disease. Patients with myotonic dystrophy type 1 (DM1) may have reasons to be at higher risk of falls, including critical fixed weakness in specific muscle groups (for example, knee extensors), combinations of muscle weakness (knee extensors and ankle/toe dorsiflexors), myotonia, visual impairment, behavioural or cognitive factors, hearing, balance and postural stability, head/neck posture, and cardiac dysfunction. We sought to compare DM1 patients and healthy volunteers prospectively with respect to their frequency of falls and stumbles, confidence, and overall ambulatory activity, and to investigate the association with relevant clinical features.

METHODS

Subjects

Thirteen successive patients with DM1 from different families were recruited from our muscle clinic. Inclusion criteria were: patients aged ⩾18 years; genetically confirmed diagnosis of DM1; from separate households in or near Cardiff; and sufficient cognitive and behavioural function to provide informed consent and participate. Twelve healthy volunteers were recruited from the local community. Exclusion criteria were a known history of any (other) major locomotor or neuromuscular disorder recognised to cause falls.

The study received local research ethics committee and Hospital Trust R&D approval. All subjects gave written informed consent.

Data collection

The muscle clinic record was used to extract retrospective information about previous illnesses, cardiac disorders, blood pressure, dizziness, blackouts, epilepsy, ocular problems, other problems recognised to contribute to falls or loss of consciousness, and other factors relating to mobility—for example, alcohol intake and drug treatment.

A brief questionnaire relating to past mobility and falls was administered. The Hospital Depression and Anxiety Scale (HADS)[6 7] and the Rivermead Mobility Index (RMI)[8] were scored. The modified activities-specific balance confidence scale (ABC-UK)[9] was used to measure falls/confidence related quality of life,[10] and the Performance Oriented Mobility Assessment (POMA) to measure balance and mobility skills.[11 12] Maximum voluntary isometric muscle strength (mean of three right and left repetitions) was measured using a wall mounted dynamometer; muscle groups tested were hip extensors/abductors, knee flexors/extensors, and ankle dorsiflexors/plantarflexors. The intratest class correlation coefficient[13] of the method was 0.9 (Busse ME, unpublished data).

Self selected gait speed and stride length were measured over the middle two metres (excluding acceleration/deceleration phases) of a 10 metre walkway, using digital video recording.[14] Right step count was recorded using the Step Watch™ step activity monitor (SAM) (Cymatech, Seattle, Washington, USA) attached to the right ankle[15 16] and programmed with sensitivity settings appropriate to the individual's height, cadence, and gait speed verified by visual inspection. All subjects wore the device over seven consecutive 24 hour periods, removing it only for bathing. Activity indices extracted were total steps/day, mean daily 24 hour step count (averaged over seven days), highest step counts sustained over any continuous 60 minute period (sustained activity), mean of highest of any (non-continuous) 30×1 minute periods (peak activity), and the proportion (%) of time spent inactive.

An intensive monitoring technique was used to count and classify falls.[17–19] Stamped addressed postcards asking all subjects to report events were provided for return weekly for 13 weeks with a final postcard at six months. Non-return of a

Abbreviations: ABC-UK, modified activities-specific balance confidence scale; DM1, myotonic dystrophy type 1; HADS, Hospital Depression and Anxiety Scale; POMA, Performance Oriented Mobility Assessment; RMI, Rivermead Mobility Index

Table 1 Demographics, gait, mobility, and lower limb strength in the study populations

Variable	DM1 (n = 13*; 7f) Mean	SD	Volunteers (n = 12†; 7f) Mean	SD	p Value
Age and size					
Age (years)	46.5	1.68	34.4	1.73	0.002
Height (m)	1.68	0.08	1.73	0.08	0.1
Weight (kg)	75.6	16.2	77.4	15.8	0.8
Body mass index (kg/m^2)	27.1	6.7	25.7	5.2	0.6
Gait indices					
Speed (m/s)	0.83	0.36	1.47	0.16	<0.001
Stride (m)	0.98	0.32	1.53	0.15	<0.001
Cadence (steps/min)	97.3	19.7	114.6	8.08	0.01
Activity indices					
Mean daily step count‡	3445	1967	6324	1222	<0.001
Peak 30 min (steps/min)	32.7	11.2	52.5	4.7	<0.001
Sustained 60 min (steps/min)	13.0	8.5	24.8	8.0	0.002
Time inactive (%)	80.5	9.2	72.1	4.9	0.01
Strength (N)					
Ankle dorsiflexion	54.3	28.5	180.6	67.6	<0.001
Ankle plantarflexion	109.4	65.8	313.1	126.3	<0.001
Knee flexion	95.6	50.7	143.7	63.6	0.06
Knee extension	169.5	72.9	274.4	137.0	0.03
Hip extension	141.0	57.8	262.0	145	0.02
Hip abduction	96.5	53.8	194.8	91.6	0.006
Mobility scales					
RMI (0–15)	11.3	3.9	15.0	0	0.002
POMA (0–28)	20.6	8.4	28	0	0.008
ABC-UK (%)	60.6	32.6	98.8	1.4	<0.001

*n = 11 for strength and mobility scales data.
†n = 12 for strength and mobility scales data.
‡Daily right step count averaged over seven days.
ABC-UK, modified activities-specific balance confidence scale; DM1, myotonic dystrophy type 1; f, female; N, newtons; POMA, Performance Oriented Mobility Assessment; RMI, Rivermead Mobility Index.

card or a reported fall led to the subject being contacted by telephone or provided with a postal debriefing questionnaire to collect further information. Subsequently two researchers agreed the fall category as extrinsic, intrinsic, non-bipedal, or non-classifiable.[1]

RESULTS

Patients and volunteers were similarly matched for sex, height, weight, and body mass index, but the volunteers were younger (table 1). Patients had significantly more comorbidities for cardiac disorder, eye problems and cataract, and complaints of impaired balance, and they had higher scores on the depression subscale of HADS. Eight patients and four volunteers were taking therapeutic drugs (four patients and one volunteer, all on sedative drugs). One patient died in the later stages of the study.

Patients had a significantly slower self selected walking speed, shorter stride length, and reduced cadence compared with healthy volunteers (table 1). Patients were more inactive, had lower mean daily step counts (average 54% that of volunteers), and lower peak and sustained walking activity. Muscle strength was reduced in all groups, most prominently in ankle dorsiflexion and plantarflexion.

Patients had significantly impaired mobility on the RMI and the POMA, and reduced confidence in relation to mobility issues and falling on the ABC-UK. From the retrospective questionnaire, patients more often used assistance than volunteers when walking outdoors (6 of 13 *v* 0 of 13, respectively; p<005), tripped more than once/week (5 of 13 *v* 0 of 12; p = 0.04), or fell and injured themselves more than once per six months (6 of 13 *v* 0 of 12; p<0.001). One patient regularly wore ankle-foot orthoses.

During weekly data collection the number of patients and volunteers who reported stumbles was similar (table 2) but more patients had a fall (NS). Eleven patients had 161 events (127 stumbles and 34 falls), while 10 volunteers had 29 events (26 stumbles and three falls). When falls and stumbles were expressed in relation to activity, patients had 0.206 (0.288) events per 5000 steps taken (mean (SD)), compared with 0.021 (0.021) in healthy volunteers (p = 0.007).

In volunteers falls were usually from extrinsic causes (for example, a slippery surface) in an unfamiliar outdoor environment, while in patients intrinsic causes were more likely (legs ''giving out'', ''unsteadiness'', misperception of the environment) (table 2). Stumbles followed a similar pattern although many could not be classified in patients. Two patients attended hospital following falls; neither was admitted or sustained a fracture. In weeks 14–26, following which most subjects (10 of 12 patients and 10 of 12 healthy volunteers) returned a single report card, two patients reported further falls, with one fracturing a finger; four patients and four volunteers reported stumbles.

Associations tested across all subjects indicated that, although self confidence about falling (ABC) was correlated with predicted liability to fall (POMA), there was no correlation between these scores and the actual number of stumbles or falls. Ankle dorsiflexion and plantarflexion, knee extension, and hip abduction strength were inversely correlated overall with the rate of falls and stumbles per 5000 steps (Spearman *r*, p<0.01).

Within the patient group alone ankle plantarflexion and dorsiflexion strength had the highest correlations with gait speed (Spearman *r* = 0.92, p<0.001, n = 10, and *r* = 0.75,

Table 2 Stumbles and falls in the study populations

Events 13 weeks	DM1 (n = 13; 7f) Event	DM1 (n = 13; 7f) No event	Volunteers (n = 12; 7f) Event	Volunteers (n = 12; 7f) No event	p Value (Fisher's exact test)
N who stumbled	10	3	9	3	1.00
No of stumbles	127	–	26	–	
No who fell	6	7	2	10	0.202
No of falls	34	–	3	–	
No who stumbled or fell	11	2	10	2	1.00
	Mean	**SD**	**Mean**	**SD**	**p Value (Mann-Whitney test)**
Falls/person	2.62	4.56	0.25	0.62	0.17
Falls/5000 steps	0.076	0.167	0.002	0.006	0.15
Stumbles/person	9.62	19.25	2.17	2.37	0.19
Stumbles/5000 steps	0.130	0.219	0.019	0.021	0.04
Falls+stumbles	12.23	22.82	2.42	2.31	0.06
Fall+stumbles/5000 steps	0.206	0.288	0.021	0.021	0.007
Classification of cause	**No of subjects**		**No of subjects**		**p Value (χ^2)***
Extrinsic	1		6		0.004
Intrinsic	7		1		
Non-bipedal	2		0		
Combination	0		3		

*Excluding unclassified falls.
DM1, myoclonic dystrophy type 1; f, female.

p<0.008, n = 11, respectively) and mean daily step count ($r = 0.56$, p = 0.09, n = 10, and $r = 0.59$, p = 0.06, n = 11). Compared with seven non-falling DM1 patients, six who did fall had lower mean (SD) RMI scores (8.3 (3.4) *v* 13.9 (2.2); p = 0.005), were more likely to use aids or a person to assist mobility indoors (3/6 *v* 0/7) or outdoors (5/6 *v* 1/7) (Fishers exact test, p = 0.07; p = 0.08), had a slower self selected gait speed (0.61 (0.38) *v* 1.02 (0.24) $m.s^{-1}$; p = 0.05), and had higher depression scores (8.7 (3.3) *v* 3.7 (2.4); p = 0.01). A backward regression analysis suggested that self selected gait speed accounted for 47% of the variance of falls and stumbles per 5000 right steps (p<0.001).

DISCUSSION

Although younger, volunteers had considerable numbers of falls and stumbles, emphasising the importance of studying a control population. However, DM1 patients were far more likely (approximately 10-fold) to fall and stumble, even though they were active for only about 20% of the 24 hour day compared with 28% in volunteers, walked more slowly, and had substantially lower intensities of activity (table 1). Some patients maintained high activity levels despite a propensity to stumble or fall.

Volunteers stumbled or fell for obvious extrinsic causes. Why patients fell or stumbled was often unclear: questioning yielded no evidence for an association with preceding loss of consciousness, and confidence did not appear relevant. As expected, visual acuity was reduced and depression more common in the patient group; both factors[20] have been associated with falling in the elderly. Patients who had falls were less mobile by RMI, had slower self selected gait speeds and higher depression scores, but did not have notably different vision compared with those who did not. Medication was not an obvious factor.

Distal weakness combined with knee and hip weakness may particularly predispose to loss of pillar support following minor sudden perturbations in stance. While adaptive strategies may mitigate this risk these might fail if there is mood or cognitive impairment: frontal lobe function and visuospatial domains would perhaps be worthy of further study. Only one patient was wearing ankle-foot orthoses in this patient sample and this requires further systematic investigation.

This study provides insufficient data to measure the absolute likelihood of serious falls or resulting injury in DM1 patients, but there seems no reason to believe that either are less likely than in older people; comparison with other neuromuscular disorders would be of interest. The impact of osteoporosis is unclear in patients with muscle disease, although the benefits of muscle activity on bone mineral density are recognised.[21] However, DM1 patients are at relatively high risk of falls and stumbles and therefore of injury. These events should form part of routine follow up inquiry, and patients may benefit from a multifactorial evaluation and multidisciplinary interventions similar to elderly people.[22]

ACKNOWLEDGEMENTS

We are grateful to the patients and healthy volunteers who participated in the study. We thank Khaled Fallatah, Dr Iris Musa (School of Healthcare Studies), and Ms Sarah Williams for assistance with data collection. CMS is supported by a grant from the Myotonic Dystrophy Support Group. MEB is supported by a grant from the Wales Office for Research & Development. JF-M is supported by a grant from the Muscular Dystrophy Campaign.

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Competing interests: none declared

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Received 23 February 2005
In revised form 21 July 2005
Accepted 22 July 2005
Published Online First 30 September 2005

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