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Bladder Function in Patients With Myotonic Dystrophy

Inge Thomsen Bernstein, Birgitte Bo Andersen, Jens Thorup Andersen, and Peter Arlien-Søborg

Departments of Urology (I.T.B., J.T.A.) and Neurology (B.B.A., P.A.-S.), Hvidovre Hospital, University of Copenhagen, Denmark

Ten patients with a clinically and neurophysiologically established diagnosis of myotonic dystrophy underwent urodynamic evaluation of the lower urinary tract. Eighty percent of the patients had urinary complaints by history, but we were not able to identify a homogeneous bladder dysfunction pattern by voiding and incontinence chart, flowmetry, cystometry, or sphincter electromyography. © 1992 Wiley-Liss, Inc.

Key words: neurogenic bladder dysfunction, lower urinary tract, voiding

INTRODUCTION

Myotonic dystrophy (MD) is an autosomally dominant hereditary multi-organ disease, characterized by myotonia and distal muscular atrophy, and in later stages by cataract, endocrine disturbances, testicular atrophy and infertility, mental retardation or dementia, progressive frontal alopecia, and disturbances in cardiac conduction. MD was first described as a unique variant of myotonia by Steinert [1909] and later reviewed by Thomasen [1948].

The prevalence is stated to be 2.5–5.5 per 100,000 [Olsen, 1989]. The disease may be visible at birth or may manifest later up to the age of 60–70 years, the average age of onset being 20 years. The earlier onset of the disease, the more servere is the course. The survival is reduced on an average by 25 years [Brumback, 1987].

The penetration of the MD gene is practicaly 100% by the age of 14 years. The MD gene has during the last years been localized to the region between the centromere and band 13,2 on the long arm of chromosome 19 [Eiberg et al., 1983; Shaw et al., 1985]. In the literature, disturbances have been described not only in the skeletal muscles but also in the cardiac and smooth muscles. Respiratory dysfunction, dysphagia, megacolon, and gallbladder dysfunction have been described, secondary to disturbances in the smooth musles [Harper, 1979].

According to our knowledge, bladder dysfunction in patients with MD has never been described. Many of our patients with MD have complained of abnormal

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Address reprint requests to Inge Thomsen Bernstein, Department of Urology, Hvidovre Hospital, University of Copenhagen, Mellemvangen 17, DK-2700, Copenhagen, Denmark.

micturition, and we therefore found it relevant to investigate the detrusor and sphincter function in a group of patients with MD.

MATERIALS AND METHODS[1]

Ten patients (5 females) ranging in age from 22 to 55 years (median 44 years) with a well-established diagnosis of MD, based on clinical findings, family history of MD, electromyography, and split lamp examination were included in the material. The patients had been suffering from MD for 2 to 33 years (median 16.5 years). All patients with MD attending the department of neurology at our hospital from 1982 to 1987 were included. None of the patients had had urogenital surgery except for sterilisation in 5 patients (3 females). After informed consent all patients had a complete neurological work-up assessing myotonia, muscle strength, and atrophy, grading the serverity from 1 to 4. Eye examination including split lamp and electromyography (EMG) from the peripheral musculature were performed in order to verify the typical myotonia pattern and assess the degree of myopathy. To exclude other diseases the patients were examined with blood chemistry (B-Haemoglobin, B-leucocytes, differential count, S-potassium, S-sodium, S-calcium, S-albumin, S-alkaline phosphatase, S-creatinine, fasting B-glucose, S-triglyceride, S-cholesterol, T3-T4-TSH, and S-creatinine kinase), electrocardiography (ECG), chest X-ray, and computer tomography of the brain.

After the clinical and neurological examined a urodynamic evaluation was performed. The patients were assessed subjectively and objectively using history, 3-day voiding and incontinence chart, urinary flowmetry, post voiding residual urine, and supine transurethral medium fill water cystometry (DISA cystometer, type 21C15, 14F transurethral catheter and 37°C isotonic saline) with simultaneous integrated sphincter electromyography (DISA EMG-amplifier, type 21C01, with surface electrocardiographic electrodes placed on the perineum in 6 patients and ring electrode mounted on the catheter in 3 patients).

RESULTS

Clinical and Neurological Examination (Table I)

All the patients had a positive family history, except one who was adopted without contact with the biological parents. Distal paresis, atrophy, and myotonia were present in all the patients, and in 4 with the most severe condition proximal affection was present too. Eight patients had cataract. Dementia and pathological EMG were present in 9 patients and 1 refused the examination. Blood chemistry, ECG, chest X-ray and cranial computer tomography did not reveal any other diseases explanatory for the pathological findings.

Symptoms

Four of 10 patients had by history "infrequent voiding syndrome" with more than 5 hours between urination [Lapides et al., 1968]. One suffered from urge and

[1]Methods, definitions, and units conform to the standards recommended by the International Continence Society, except where specifically noted.

TABLE I. Neurological Evaluation*

Patient No.	Sex	Age (years)	Duration (years)	DMR	PMR	Dem.	Fac.M	Cat.	EMG
1	f	22	2	+	n	+	+	n	y
2	f	28	8	+	n	+	−[a]	n	y
3	m	27	12	+ +	n	+	+ +	y	n
4	m	45	12	+ + +	n	+ +	+ + +	y	y
5	m	38	15	+ + + +	y	+ + +	+ + +	y	y
6	f	47	18	+ + +	y	+ +	+ + +	y	y
7	m	45	20	+ + +	y	+ +	+ +	y	y
8	f	48	28	+ + +	n	+	+ +	y	y
9	f	43	31	+ +	n	+	+	y	y
10	m	55	33	+ + +	y	+ +	+ +	y	y

*Neurological evaluation in 10 patients with dystrophia myotonica. f = female; m = male; DMR = distal muscle reduction; PMR = proximal muscle reduction; Dem. = dementia; Fac.M. = facies myotonica; Cat. = cataract; EMG = electromyography; y = yes; n = no. DMR, Dem., and Fac.M are graded in severity from + to + + + + .

[a]Patient 2 does not have dystrophia myotonica.

stress incontinence, 1 had slight urgency without incontinence, 1 had moderate urgency with episodic incontinence, 1 had obstructive symptoms in the morning, and 2 were indicated to have a normal micturition pattern.

Voiding Diary

Number of micturitions per day and voided volumes were registered correctly on the 3-day voiding and incontinence chart by 8 of the patients. Two patients had an abnormal voiding pattern; 1 had frequent micturitions with 30–40 micturitions/24 h (the patient with urge and stress incontinence), and 1 had few micturitions 3–4/24 h with large volumes exceeding 600 ml (1 of the patients who indicated to suffer from infrequent voiding syndrome).

Urodynamic Findings

Uroflowmetry was performed in all 10 patients (Table II). Three patients had intermittent flow pattern and 1 of them had a pathologic voided volume (1,200 ml). Maximum flow rate was reduced in 5 patients (one male).

Cystometry was performed in 9 patients (Table III, one did not want to participate in this part of the evaluation). Residual urine was measured with the catheter used for cystometry, and residual urine was found in 2 patients. First sensation (FS) and maximal cystometric bladder capacity (MCBC) was pathologic in 1 patient (FS: 650 ml, MCBC: >1,000 ml), One patient had detrusor hyperreflexia and this patient had FS immediately after onset of the bladder filling. All the patients had normal resting intravesical pressure, and 7 of the 9 patients were on request able to elicit a voluntary detrusor contraction. Sphincter electromyography was normal in all 9 patients.

DISCUSSION

All the patients fulfilled the diagnostic criteria for DM with the typical clinical neurological findings and a pathological EMG. We found, in accordance with the

TABLE II. Uroflowmetry*

Patient No.	Flow curve pattern	Maximum flow rate, ml/s	Voided volume, ml
1	Continuous	12.2	240
2	Continuous	18.5	190
3	Intermittent	11	210
4	Continuous	16.7	306
5	Continuous	33.5	348
6	Continuous	20	560
7	Intermittent	41	1,200
8	Intermittent	16.2	277
9	Continuous	28	257
10	Continuous	34	250

*Results of uroflowmetry in 10 patients with dystrophia myotonica.

TABLE III. Cystometry*

Patient No.	FS, ml	MCBC, ml	Detrusor contractions	Resting intravesical pressure, cm H_2O	Residual urine, ml
1	200	320	Voluntarily	8–10	10
2	125	200	Voluntarily	6	0
3	Missing				
4	250	300	Voluntarily	15	100
5	75	250	Voluntarily	10–15	0
6	250	375	None	4	0
7	650	>1,000	None	4	0
8	150	200	Voluntarily	18–20	0
9	75	250	Voluntarily	4–6	4
10	0	300	Uninhibited	14	250

*Results of water cystometry in 10 patients with dystrophia myotonica.

literature, variability of the clinical expression with a tendency of a more severe course the longer the disturbances had been present [Adams and Victor, 1985].

Urinary complaints are dependent on the tolerance of the individual patient, and comparison is difficult. Eight of the 10 patients (80%) indicated to have an abnormal micturition pattern. The voiding pattern was not uniform in our patients, but there seemed to be a preponderance of "infrequent voiding syndrome" in the history (4 of 10 patients). We were not able to assess this objectively except in 1 patient, who had a high compliance bladder on cystometry (MCBC > 1,000 ml) and a pathologic functional bladder capacity (1,200 ml) on the flowmetry. The other 3 patients, who claimed to have "infrequent voiding syndrome," had a normal voiding chart. One patient with urgency and episodic incontinence had inhibited detrusor contractions during the whole cystometry, which could be the cause of the incontinence. All other patients had normal cystometry. Six patients had a pathological urinary flow curve. Two had both an intermittent flow pattern and a reduced maximum flow rate, but only one patient had obstructive symptoms. Unfortunately his intravesical pressure is unknown as the patient would not participate in cystometry.

In conclusion, patients with DM have different urinary complaints with a preponderance of infrequent voiding syndrome, but we were not able to identify a homogeneous bladder dysfunction pattern in the 10 patients with DM using history, 3-day voiding and incontinence chart, flowmetry, and cystometry with simultaneously integrated sphincter electromyography.

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