Faecal incontinence due to atrophy of the anal sphincter in myotonic dystrophy : a case report

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Abstract

Myotonic dystrophy or Steinert disease is an autosomal dominant multisystemic disorder with variable penetrance. The genetic defect is an amplified trinucleotide repeat in the 3-prime untranslated region of a proteinkinase gene on chromosome 19. Severity of symptoms increases with the number of repeats. Patients with myotonic dystrophy often present with gastrointestinal motility problems, such as intermittent diarrhoea, constipation, and also faecal incontinence. The underlying physiopathological mechanism of faecal incontinence differs from classic soiling due to faecal retention. We present a girl with congenital myotonic dystrophy and faecal incontinence due to anal sphincter atrophy ; and give an overview of present knowledge on the pathophysiology of gastrointestinal problems associated with myotonic dystrophy. (Acta gastroenterol. belg., **2011**, 74, **88-90**).

Key words : myotonic dystrophy type 1, faecal incontinence.

Abbreviations

DM1 : Myotonic Dystrophy type 1
DMPK : Dystrophia Myotonica Protein Kinase

Introduction

Steinert disease or myotonic dystrophy-1 (DM1, OMIM #160900) is an autosomal dominant disorder caused by a mutantly amplified CTG repeat in the dystrophia myotonica protein kinase gene (DMPK) on chromosome 19q13.3, which encodes for myotonin.

The length of the repeated CTG sequence (5 to 30 repeats in normal individuals) correlates with the severity of the disease : mildly affected persons having 50 to 80 repeats, and severely affected individuals having 2000 or more copies [www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=160900, 1, 2]. The penetrance is incomplete and clinical expression varies enormously among patients with the same number of repeats. Clinical symptoms are more severe in early onset disease. The main clinical features of the adult form are myotonia (delayed muscle relaxation after voluntary contraction or electrical stimulation), muscular dystrophy with progressive muscle weakness, cataract, hypogonadism, frontal balding, and ECG changes, whereas congenital DM1 is characterized by severe hypotonia, facial diplegia, poor feeding, arthrogryposis and severe respiratory failure. Myotonia is never present in the first year of life, during which motor function gradually improves. If they survive, patients are confronted with serious cardiorespiratory sequelae and mental retardation. As they become older, the classic symptoms of the adult onset DM1 appear (3). DM1 with onset before the age of 12 years, the juvenile form, is marked by the involvement of organ systems other than skeletal muscle including the gastrointestinal system, with mental retardation also being a dominant feature.

Gastrointestinal motility problems are highly prevalent and may affect any level of the gastrointestinal tract from the pharynx to the anal sphincter. When the upper digestive tract is involved, symptoms vary from dysphagia, chest pain, regurgitation, heartburn, epigastric pain, dyspeptic symptoms, nausea and vomiting. Abdominal pain, bloating, diarrhoea, constipation and dyschezia are common signs of impairment of the lower gastrointestinal tract. Recurrent intestinal pseudo-obstruction has been described (4). Dysfunction of striated and smooth muscles, caused by the disease, is considered to be the underlying pathophysiological mechanism of these motility problems. Recently however, some studies have also suggested that neurological and neuroendocrine changes may play a role (5). The gastrointestinal problems, including anal incontinence, are often considered to be the most disabling consequences of DM1 and bowel problems may predate the classic neuromuscular symptoms, i.e. even before the diagnosis (6,7).

Case report

Our patient is born as the second daughter of a non-consanguineous marriage. At birth, she is referred to the neonatology unit because of marked hypotonia (floppy infant). The diagnosis of Steinert disease is genetically confirmed. She is heterozygous for 1000 CTG-repeats in the DMPK-gene on chromosome 19. Family screening confirms the diagnosis in her older sister (1000 repeats, but no congenital hypotonia), her mother (150-200 repeats) and maternal grand-mother (79 repeats). During the first months of life, hypotonia subsides and initial feeding difficulties disappear. Psychomotor development is markedly delayed. Neither stool problems nor

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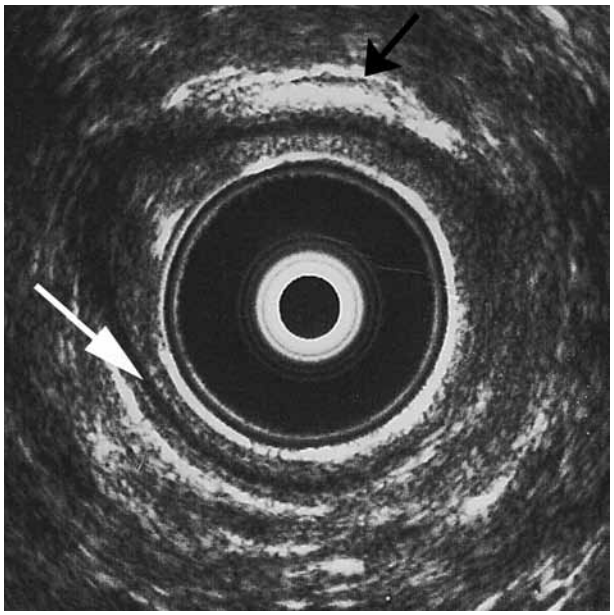

Fig. 1. — Intrarectal sonography showing only a very thin internal sphincter (incomplete black circle indicated by white arrow) and a small area of external sphincter (black arrow).

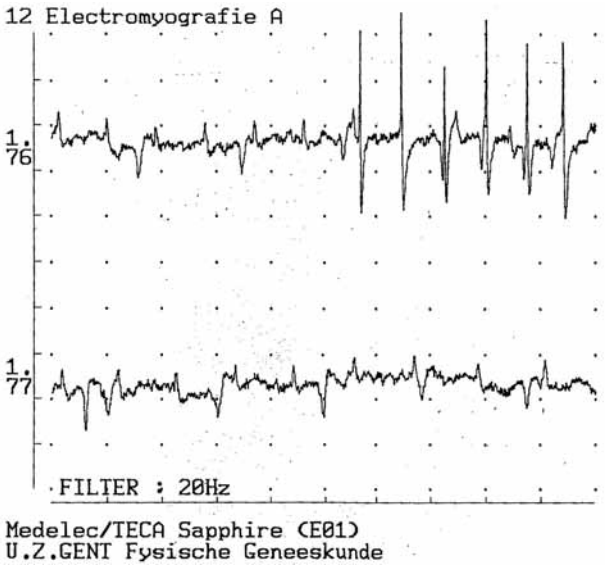

Fig. 2. — Electromyography recording by means of disposable concentric needle electrodes of 25 mm/30 G shows Complex Repetitive Discharges (sudden onset and stop of discharges) in the external anal sphincter.

constipation are noted in the first years of life. She has one to two bowel movements daily. At the age of 6 years, she presents at the paediatric gastroenterology department because of problems with toilet training. She has continuous leakage of stool and still needs diapers. She has mild mental retardation (developmental age 4 years). Unsuccessful attempts at toilet training have frustrated her and she is very anxious during the clinical examination which shows faecal impaction with a full rectum, apparently normal sensibility of the peri-anal region and a patent anus. Following an enema, she is started on osmotic laxatives (poly-ethylene glycol) and a strict toilet scheme with a stool diary. Anal manometry is not possible due to lack of cooperation and fear. Psychological support is proposed. She is lost to follow-up till the age of 8 years, when she returns with the same complaints. Faecal incontinence is becoming a real social handicap. She continued laxative use, and there are no signs of faecal impaction. Anal inspection shows an open anus, at rectal digital examination no sphincter force is felt, and the rectal ampulla is empty. No anal reflex can be elicited, but peri-anal sensibility is conserved. Further investigations are performed under general gas anaesthesia (no opiates, no muscle relaxants). An intrarectal sonography is performed (Fig. 1) which demonstrates a very thin internal sphincter (1 mm) but almost no external sphincter. Electromyography of the anus shows no basal activity, and complex repetitive discharges (formerly called pseudomyotonic discharges) indicative of a chronic severe mixed myopathic-neuropathic disorder (Fig. 2). Anal manometry fails to detect a distinctive sphincter pressure zone. Given these results, faecal continence can not be acquired. She is successfully started on a bowel cleansing program with regular retrograde enemas using luke-warm tap water to gain pseudo-continence.

Interestingly, her sister, who has the same number of CTG-repeats, did not present neonatal symptoms, is also mildly mentally retarded, but presents no continence problems and was toilet trained at the age of 4 years. Clinical examination of her anus is normal. As she presents no stool problems, no invasive investigations were done.

Discussion

This report describes a patient with DM1 with faecal incontinence due to both internal and external anal sphincter impairment. Although DM1 (as other neuromuscular disorders) is a rare cause of faecal incontinence, the multisystemic character of the disease and the need for appropriate gastroenterologic follow up should always be borne in mind.

In DM1, with onset at birth or during childhood, gastrointestinal disturbance is highly prevalent, with a prevalence of faecal incontinence of over 15% at the age of 5 years (bowel control is achieved in 97% of normal children by the age of 3 years) (7). This problem, being reported as the most burdensome and disabling by DM1 patients, increases over the years with up to 66% of adult DM1 patients suffering from occasional faecal incontinence and more than 10% reporting faecal incontinence one or more times a week (4).

Faecal continence is preserved by striated (external anal sphincter and puborectalis) and smooth muscle

(internal anal sphincter). It is well recognised that both types of muscle are affected in patients with DM1. In adults with the disease, it is known that both anal sphincters are involved pathophysiologically (8). The diminished strength is best explained by myopathic changes, whereas alterations in the anorectal inhibitory reflex are more likely an expression of an additional neurogenic defect (8,9). These data are consistent with our EMG-findings which showed alterations consistent with chronic myogenic and neurogenic injury. To date these anal electromyographic findings were never described in children with DM1.

A particular finding in this case concerns the near absence of anal muscle, as demonstrated by intrarectal sonography. Apart from the myotonia, DM1 is characterised by muscle weakness and wasting. This muscular atrophy usually affects facial and distal limb musculature (9). Given the fact that as an infant our patient had repeatedly a closed anus, and normal stooling pattern (no continuous soiling), but at the age of 8 years had a patent anus, with hardly an external sphincter demonstrable at ultrasonography and only a very slender internal sphincter of 1 mm, muscle degeneration probably occurred in the anal sphincter. This clinical feature has been described, and carries the risk of wrongfully being interpreted as a sign of sexual abuse (10).

Unstable CTG segregation analysis and genotype/phenotype studies have demonstrated that triplet number correlates with the severity of the symptoms and inversely with the age of onset (11,12). Although gastrointestinal dysfunction in general is not correlated with repeat size, anal abnormalities typically affect subjects with the congenital form, i.e. those patients with a larger CTG expansion than the adult variant (12). The CTG sequence in our patient consisted of 1000 repeats. As anal laxity is regularly reported, it seems that faecal incontinence might correlate with CTG-repeat length. Interestingly, illustrating the phenotypical heterogeneity of the disease, her sister, showing the same number of CTG-repeats, has neither faecal incontinence nor stool problems. It would have been interesting to perform an intrarectal sonography and an anal EMG in her sister. But as she is also mentally retarded, these invasive examinations can only be performed under general anaesthesia. As the results are without therapeutic consequences for her, she was not investigated because of obvious ethical reasons.

In conclusion, this case report describes anal sphincter atrophy as a cause of faecal incontinence in a patient with DM1. Faecal incontinence is one of the gastrointestinal motility disorders frequently encountered in DM1 patients, underlining the necessity of gastroenterologists being familiar with the disease.

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