Endometriosis as Initial Manifestation of Myotonic Dystrophy Type-2

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Though gynecological involvement (ovarian cysts, endometriosis, hypogonadism) has been occasionally reported in myotonic dystrophy type-2 (DM2),^{1,2} endometriosis as initial manifestation is unknown.

The patient is a 67-year-old female referred for nonsystemic vertigo and recurrent collapses for the last 10 months together with stocking-type sensory disturbances for the last 2 years and tingling of the toes bilaterally for the last 1 week. Her history was noteworthy for: endometriosis with severe abdominal pain 1 week prior to and 1 week postmenstruation since age 18 years requiring left ovariectomy at age 24 years; endometriosis of the colon with stenosis of the sigma requiring resection of the sigma at age 30 years leading to resolution of perimenstrual abdominal pain; volvulus at age 30 years requiring surgery; recurrent surgery for hernia cicatrix at ages 31, 32, and 34 years; steatosis hepatis upon liver puncture at age 43 years; hysterectomy, right ovariectomy, and urinary bladder lifting at age 50 years; diabetes since age 53 years; right cataract-surgery at age 54 years; laser therapy at age 55 years; hyperlipidemia since age 55 years; myotonic and pseudo-myotonic discharges at most investigated sites on needle electromyography from the anterior tibial muscle; inclusion body myopathy or DM2 (hypertrophic fibers, some atrophic fibers, rimmed and autophagic vacuoles, internalized nuclei) upon muscle biopsy because of hyper-CKemia and muscle weakness triggered by statins at age 56 years; cholecystectomy at age 57 years; hypoacusis since age 61 years; left cataract-surgery at age 64 years; chronic constipation since age 65 years; arterial hypertension since age 65 years; and recurrent collapses during exercise since age 67 years.

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Clinical neurologic exam at age 67 years revealed short stature, hypoacusis bilaterally (hearing devices), unrounded pupils, massively sore neck muscles, diffuse muscle weakness of upper limbs (MRC4-), diffuse wasting, positive pyramidal signs on the right side, weakness for right hip flexion (MRC5-), weakness for foot extension bilaterally (MRC5-), diffuse wasting, a subclonic left patella tendon reflex, reduced Achilles tendon reflexes, and hypoesthesia of all toes. There was a pes equino-excavatus. The Gower sign was positive. Creatine-kinase was normal. Cerebral magnetic resonance imaging revealed contact between the cerebelli anterior inferior artery and the vestibulo-cochlear nerves and nonspecific gliotic spots bilaterally. Cerebral computed tomography showed hyperostosis frontalis. Echocardiography revealed concentric thickening of the left-ventricular myocardium. Electrocardiogram was normal. On the Mini-Mental State Exam, she scored 27/30 points. Nerve conduction studies revealed axonal neuropathy of lower limbs: Ultrasonography revealed carpal tunnel syndrome bilaterally. Genetic work-up revealed a CCTG-repeat expansion of >300bp (>75 repeats) in *ZNF9*.

The presented patient is interesting for DM2 and endometriosis as initial manifestation of DM2. The diagnosis DM2 relied on the clinical presentation, electromyography, and genetics. The diagnosis was challenged by previously undescribed phenotypic features, such as occasionally mildly elevated serum lactate, short stature, and hyperostosis frontalis (►Table 1). Arguments for a causal relation between DM2 and endometriosis are that gynecological involvement has been previously reported in DM2 (►Table 1),³ that endocrine involvement is frequent in

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Table 1 Clinical manifestations of DM1 and DM2 in the presented case and the literature

Manifestation	Index case	DM1	DM2
Muscle			
Myopathic face	No	Yes	Yes
Limb muscle weakness	Yes	Yes	Yes
Clinical myotonia	No	Yes	Yes
Myalgias	No	No	Yes
Muscle wasting	Yes	Yes	Yes
Calf hypertrophy	No	No	Yes
Hyper-CKemia	Yes	Yes	Yes
Peripheral nerves			
Polyneuropathy	Yes	Yes	Yes
Autonomic dysfunction	No	Yes	No
Carpal tunnel syndrome	No	Yes	No
Brain			
Cognitive impairment	Yes	Yes	Yes
Behavioral abnormalities	No	Yes	No
Mental retardation	No	Yes	No
Daytime sleepiness	No	Yes	Yes
White matter lesions	Yes	Yes	Yes
Eyes			
Cataract	Yes	Yes	Yes
Low intraocular pressure	No	Yes	No
Pigmentary retinopathy	No	Yes	Yes
Epiretinal membranes	No	Yes	Yes
Endocrine			
Diabetes	Yes	Yes	Yes
Hyperthyroidism	No	No	Yes
Hypothyroidism	No	Yes	
Hyperhidrosis	No	No	Yes
Hypogonadism	No	Yes	Yes
Hyperparathyroidism	No	Yes	No
Abortus, stillbirth	No	Yes	No
Endometriosis	Yes	Yes	Yes
Osteoporosis	No	Yes	No
Short stature	Yes	No	No
Cardiac			
Conduction defects	Yes	Yes	Yes
Arterial hypertension	Yes	Yes	Yes
Myocardial thickening	Yes	Yes	Yes
Dilative cardiomyopathy	No	Yes	Yes
Noncompaction	No	Yes	Yes

(Continued)

DM2, that endometriosis has been reported in DM1/DM2 (► **Table 1**),^{4,5} and that no other first-degree relatives had endometriosis.

Overall, the phenotypic spectrum of DM2 is broader than anticipated and resembles DM1 in many aspects.

Table 1 (Continued)

Manifestation	Index case	DM1	DM2
Gastrointestinal			
Dysphagia	No	Yes	Yes
Dysmotility	Yes	Yes	Yes
Steatosis hepatis	Yes	Yes	Yes
Liver cirrhosis	No	No	Yes
Elevated GGT	Yes	Yes	Yes
Cholecystolithiasis	Yes	Yes	Yes
Nonalcoholic fatty liver disease	Yes	Yes	Yes
Bones			
Hyperostosis frontalis	Yes	Yes	No
Foot deformities	Yes	Yes	Yes
Small sella	No	Yes	No
Large air sinuses	No	Yes	No
Skin			
Frontal balding	No	Yes	Yes
Pilomatricoma	No	Yes	No
Others			
Hydroureter	No	No	Yes
Hyperlipidemia	Yes	Yes	Yes
Hyperuricemia	Yes	No	Yes
Thrombocytosis	No	No	Yes
Renal cysts	No	No	Yes
Low IgG, IgM	No	Yes	Yes
Lactic acidosis	Yes	No	No

Abbreviations: DM1, myotonic dystrophy type-1; DM2, myotonic dystrophy type-2; GGT, gamma-glutamyl transferase; IgG, immunoglobulin G; IgM, immunoglobulin M.

Ethical Approval

Informed consent was obtained from the reported patient.

Authors' Contributions

Both authors contributed equally (J. F.: clinical investigations, design, literature search, discussion, first draft; C. S.: clinical investigations, literature search, discussion, critical comments).

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Conflict of Interest

None declared.

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