# 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 cicatrica 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 pseudomyotonic 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.

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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