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DYSAUTONOMIC SYMPTOMS DUE TO PERIPHERAL NERVE INVOLVEMENT IN MYOTONIC DYSTROPHY TYPE 2 (DM2)

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Introduction

A peripheral neuropathy has been only seldom described in DM2 patients. Here we report a father and his son affected by DM2 manifesting peripheral neuropathy with dysautonomic symptoms among their cardinal disease features.

Patient 1

50 year-old male

•**Symptoms**: from age 36 erectile dysfunction. At age 37 episodes of cold sweating, palpitations and syncopes, but refused prophylactic ICD for severe ventricular tachyarrhythmias. Recently muscle stiffness and pain.

•**Neurological examination**: frontal balding, no muscle weakness or atrophy, reduced lower limbs deep tendon reflexes.

•**Blood tests**: CPK 719 UI/l (n.v. 20-200), hypogammaglobulinemia, normal glycemia, thyroid and sexual hormones.

•**EMG**: rare pseudomyotonic discharges.

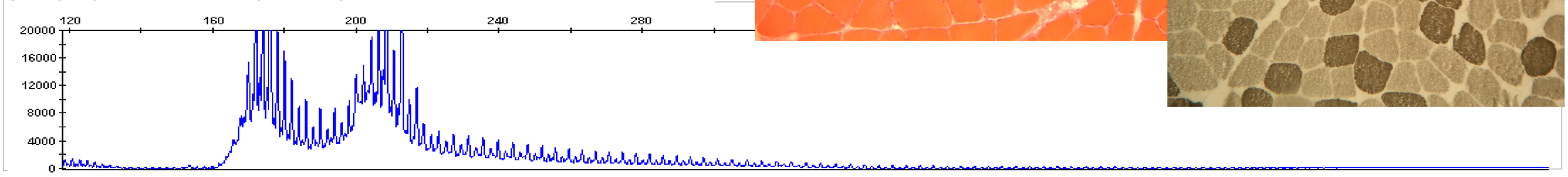
•**ENG**: sensory-motor prevalent axonal polyneuropathy.

•**Muscle MRI, brain MRI and neuropsychological tests:** normal.

•**Muscle biopsy**: normal.

•**Nerve biopsy**: chronic aspecific axonal neuropathy. Red congo stain negative.

•**24-hour Holter monitoring ECG**: severe ventricular tachyarrhythmias. He again refused prophylactic ICD pacing.



Patient 2

27 year-old son of patient 1.

•**Symptoms:** erectile dysfunction, recurrent episodes of cold sweating, palpitations and syncopes, muscle stiffness and pain.

•**Neurological examination**: mild frontal balding, handgrip myotonia, diffuse hyporeflexia.

•**Blood tests:** CK 490 UI/l (n.v. 30-170), normal glycemia, thyroid and sexual hormones.

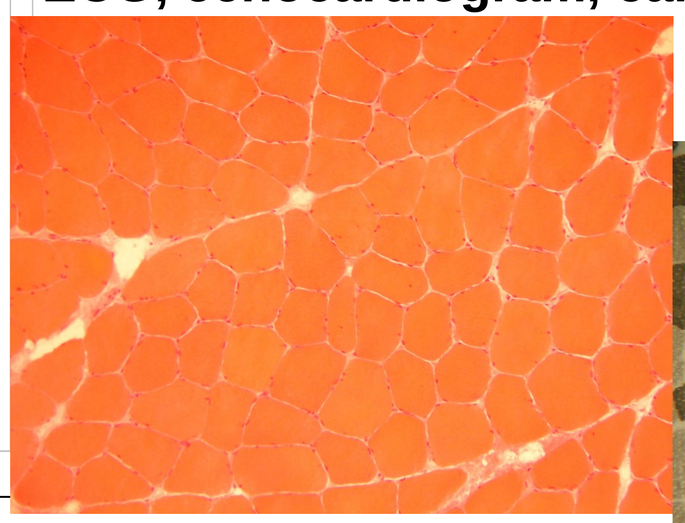
•**EMG**: pseudomyotonic discharges.

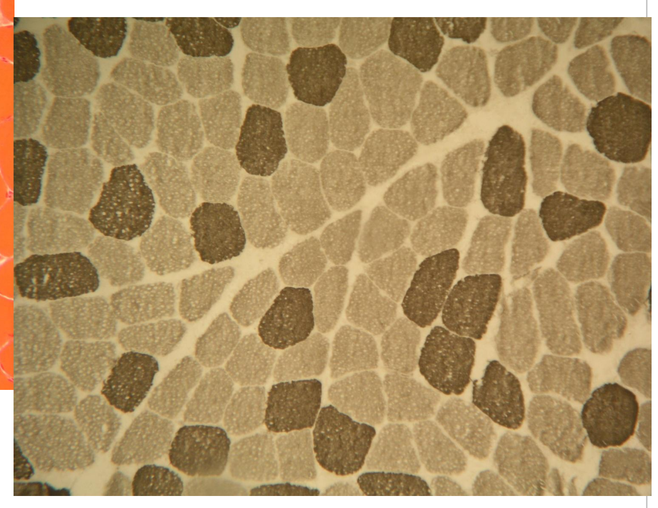
•**ENG**: sensory-motor prevalent axonal polyneuropathy.

•**Muscle MRI, brain MRI and neuropsychological tests**: normal.

•**Muscle biopsy (*figures*)**: normal.

•**ECG, 24-hour Holter monitoring ECG, echocardiogram, cardiac MRI**: normal.





In both cases, molecular testing for **FAP**, **CMT1B** and **CMTX1** resulted negative, whereas **DM2** testing was positive.

Conclusions

This report emphasizes that a sensory-motor mixed polyneuropathy with dysautonomic features may be part of the clinical manifestations of multisystem involvement in DM2.

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