

## **Proximal myotonic myopathy (PROMM): case presentation**

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Myotonia is a rare neurologic phenomenon, defined as persisting contraction despite attempts at relaxation. Myotonia does not involve spontaneous and involuntary muscle contractions; rather, forceful muscle contraction is followed by delayed relaxation (action myotonia) caused by prolonged excitation of the muscle membrane. Myotonia can also be elicited by direct percussion of the muscle (percussion myotonia). Prolonged waxing and waning electrical discharges with gradually declining amplitude and the characteristic 'dive bomber's sound' are induced by the insertion and subsequent manipulation of an electromyography needle (electrical myotonia) (1).

Myotonia is a sign of myotonic dystrophies (myotonic dystrophy type1- DM1; myotonic dystrophy type2- DM2, and proximal myotonic myopathy- PROMM), where it is combined with signs of myopathy and muscle dystrophy. Isolated myotonia is due to chloride, sodium or calcium channel disorders of the muscle membrane, or it is drug induced or associated with malignancy. It may be combined with malformations in the Schwartz-Jampel syndrome (1, 2).

Myotonia should be differentiated from other involuntary muscle contractions (Table 1).

### **Case presentation**

An eleven years old girl was admitted to neuropediatric ward of the Plovdiv Medical University Hospital because of stiffness of the thighs after sitting for a long time, feelings of narrow bands in the anterior part of the thighs, impossibility to move the legs during these episodes, and frequent falls after trying to walk. These complaints started about 6 mo ago with a frequency of about one daily, mainly before noon. She also complained of frequent stiffness of the legs when climbing stairs, rare falls after kneeling, stiffness of the hands (impossible opening of the fist after shaking hands), rare pains in the thighs. These symptoms increased during psychological stress. The girl was induced to stop her sportive activities after the onset of these complains. The premorbid and familial history were uneventful.

The clinical presentation revealed well delineated muscle mass of the legs, possibly hypertrophy and mild postural and intentional tremor. No other abnormalities in the general and neurological examinations were found.

The results of the myotonic tests were:

- Percussion myotonia- positive
- Slow flexion and extension of the fingers at onset; positive warming-up phenomenon.
- Eyelid lag when looking down (rarely)
- Slowness at the beginning of repetitive kneeling or climbing stairs; no muscle weakness.
- No stiffness after rapid getting up from bed
- Slowness of flexion and extension of fingers in the first 5-10 seconds after being immersed in cold water (15°C for 15 min)

The laboratory investigation revealed mildly elevated creatin kinase – 184 UI/L. The ECG, cardiac ultrasonography, serum electrolytes and calcium and eye examination were all normal.

EMG of m. vastus lateralis and m. deltoideus of the patient performed by two independent investigators showed myotonic discharges, duplets, triplets and fibrillations during insertion of the needle. "Dive bomber's" sound was present. There also myopathic signs – MAP with small amplitude (0.70-0.38mcV) and short duration (10-11ms). The EMG of the same muscles of the

father revealed myotonic discharges, duplets, triplets and fibrillations during insertion of the needle, but no typical “dive bomber's” sound. The EMG of the mother found no definite abnormalities. Molecular genetics gave negative results for DM2. The child was dismissed without specific medical treatment. Caution during surgery and no excessive physical exercise were advised.

### **Discussion**

The complains of the patient suggested myotonia as their explanation. The presence of percussion myotonia and the abnormal fingers flexion-extension test and cold water test ascertained this suggestion. Myotonia was proved by EMG.

The next step in diagnosis was to point the specific diagnosis. The presence of signs of muscle dystrophy like elevated CK and myopathic findings on EMG lead to diagnosis of myotonic dystrophy. This conclusion was supported by the subclinical myotonia of the father which supposes an autosomal dominant disorder. All myotonic dystrophies (DM1, DM2 and PROMM) display an AD mode of inheritance, while channelopathies are autosomal recessive.

The presentation with thigh weakness, muscle pain and muscle hypertrophy is typical for DM2 and PROM. As the genetic tests were negative for DM2 we assumed diagnosis is PROMM (3). PROMM is a rare disease with prevalence much less than 1:8000 with unknown genetic defect. Probably it is not a genetically homogeneous disorder unlike DM1 and DM2.

### **Conclusions**

A rare sign in childhood is reported – myotonia. The clinical presentation is important for delineating the disease pattern and EMG is crucial for proving myotonia. Molecular genetics is necessary for defining the disease entity.

### **References:**

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Table 1.

**Differential diagnosis of involuntary muscle contractions (1, 2)**

<b>Clinical Findings, Observations</b>	<b>Cramp</b>	<b>Contracture</b>	<b>Myotonia</b>	<b>Dystonia</b>
EMG	Normal action potentials	Silent	Myotonic discharges	Normal action potentials
Duration	Minutes	Seconds to minutes	Seconds	500 ms, sustained
Physical activity at onset of symptoms	Rest (exacerbated by exercise)	Forceful exercise	Forceful exercise, direct muscle percussion	Often action task specific at onset; later occurs at rest
Pain	+	+/- <sup>a</sup>	-	+/-
Warm-up phenomenon	-	-	+	-
Second-wind phenomenon	-	+/- <sup>a</sup>	-	-
Alleviation by muscle massage	+	-	-	-
Effect of focal curare injection	+	-	-	+
Pathophysiology	Neurogenic diseases (unstable membrane depolarization of motor axons)	Muscle metabolic abnormality (no energy to relax)	Muscle membrane ion conductance abnormality	Basal ganglia or brainstem dysfunction

<sup>a</sup>Present in glycogen metabolism defects.

+, present; -, absent; +/-, variable.