Myotonic dystrophy type 2 – a case report

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The myotonic dystrophies (DM) represent a group of autosomal, dominantly inherited disorders, caused by mutations on chromosome 19 (DM type 1 – Steinert's disease) or on chromosome 3 (DM type 2 - proximal myotonic myopathy, a milder phenotype). Clinical DM is usually defined by myopathy, amyotrophy, myotonia, in combination with ocular, cardiac and endocrine abnormalities. We describe the case of a 50 year-old female, early menopaused at 40, with a history of depression, dyslipidemia, occasional hypertension, whose clinical symptoms started ca. 9 years before, with muscle cramps in the lower limbs, myotonia, progressive gait problems with frequent falls and head tremor. She has a sister with similar gait problems and muscle pain and her father and daughter have head and upper limbs tremor. Clinical examination revealed limited walk perimeter, with difficulty initiating steps, dystonic head tremor (aggravated when right-turning her head), muscle weakness (predominantly neck flexors, upper and lower limb extensors), contraction and percussion myotonia in both hands, no amyotrophy and hypertrophy. Biological tests revealed a mildly elevated CK level after exercise. ECG, 24-hour Holter monitoring and cardiac ultrasound showed no abnormalities. Cerebral MRI examination was normal; lumbar MRI examination revealed L1-L5 herniated discs with mild lumbar stenosis. The electromyography showed myotonic discharges in both proximal and distal muscles and reduced MUP durations and amplitudes. We performed a left deltoid muscle biopsy, which revealed a myopathic pattern, including moderate variation in fiber size, angulated fibers, predominant atrophy of the type II fibers, increased number of fibers with central nuclei. The electromyography and biopsy findings helped establish the diagnosis of myotonic dystrophy type 2. Particular aspects of this case would be the presence of the dystonic head tremor (very rarely described feature of DM2), little to no cold aggravation, no "warm-up" effect and no amyotrophy/hypertrophy.