Title: mtDNA depletion syndrome type 11 - a case report.

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INTRODUCTION: mitochondrial DNA depletion syndrome type 11 (MTDPS11) is an autosomal recessive disease caused by pathogenic variants in *MGME1* gene, which encodes an exonuclease crucial for replication and maintenance of mtDNA. **CASE REPORT:** Female, 40 years-old, with right eyelid ptosis at 11 years of age with slow progression to bilateral, learning difficulties and frequent falls. Family history with no cases with similar phenotype. Investigation showed slightly increased CK, glucose intolerance and normal lactate. Electrocardiogram showed second-degree atrioventricular block. Spirometry revealed mild mixed-type ventilation disorder. Retinography evidenced atrophy and granular appearance of the retinal pigment epithelium. Magnetic resonance of the brain showed cerebellar atrophy. Muscle biopsy depicted red ragged fibers and COX-negative fibers, compatible with mitochondrial myopathy, Genetic study revealed a homozygous pathogenic variant in the MGME1 gene, consistent with MTDPS11 (c.862C>T; p.Gln288*). **PROPEDEUTICS**: Physical examination showed mild scoliosis, hyperextensibility of elbows, flat feet, bilateral eyelid ptosis and restriction of eye movements in all directions, except for partial sparing of bilateral abduction and downgaze, associated with diffuse muscle hypotonia and grade 4 strength for arm abduction and neck flexion. **RESULTS:** Our patient has a nonsense variant never previously described. Although the phenotype was clearly compatible with MTDPS11, two clinical characteristics were peculiar in this case: presence of only mild respiratory disorder, and presence of cardiac rhythm conduction disorder. **CONCLUSION:** we present a MTDPS11 case with a milder phenotype, with cardiac arrhythmia, and we also describe specific alterations on ocular motricity, contributing for the characterization of the disease.

KEY WORDS: Mitochondrial diseases; Ophthalmoplegia chronic progressive external; mtDNA; MTDPS11;