neuromuscular Diseases

CLINICAL AND DIAGNOSTIC APPROACH TO CONGENITAL OCULAR MYOTONIA

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Introduction and objective:Pathologies associated with defects in the SCN4A protein, which is part of the sodium channels, are rare autosomal dominant inherited diseases. Mutations of this gene have been associated with myotonia fluctuans, myotonia permanens, acetazolamide-responsive myotonia, hypocalcemic periodic paralysis, paramyotonia congenita of Von Eulenburg, atypical myotonia congenita, and hyperkalemic periodic paralysis.

Material and methods: A 1-year-old girl is under the care of the Child Neurology Service given her daily episodes of sustained contraction of both orbicularis oculi after blinking. These are triggered by several stimuli, without compromising consciousness. The physical examination shows mild hypotonia, muscular hypertrophy, need to support for standing and episodes of ocular blepharospasm. Among other tests, an electromyogram with repetitive stimulation, brainstem auditory evoked potential and visual evoked potential were performed.

Results:Medical interconsultations and complementary tests include Ophthalmology:hyperopia in both eyes;Blood analytics:CPK increase; MRI:no alterations. EMG:there is no electrodiagnostic evidence of a neuropathic or a myopathic process.Repetitive nerve stimulation in the right facial nerve with recording in the orbicularis oculi muscle: drop in amplitude at the level of the fifth and tenth potential at 3 and 20Hz;BAEP:bilateral hearing loss; VEP:normal;Genetics:heterozygosis of the pathogenic missense variant c.T3479A:p.I1160N in the SCN4A gene.

Conclusion:The main purpose of this case study is to describe an atypical case of congenital ocular myotonia associated with a rare mutation in the SCN4A gene. The sustained contraction of both orbicularis oculi and pathological repetitive stimulation were the main trigger for further genetic study. She is currently under treatment with carbamazepine, with satisfactory evolution.