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Instrumental assessment of vestibular function, speech, balance and gait alterations in autosomal recessive spastic ataxia of Charlevoix-Saguenay: a case report

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Introduction: Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is an early-onset ataxia characterized by cerebellar dysfunction, early spasticity, and sensory-motor polyneuropathy due to mutations in the SACS gene (13q11). So far no studies have instrumentally assessed vestibular function, gait, balance, and speech alterations in these patients.

Case presentation: A 36 years-old woman with diagnosis of ARSACS came to our attention. Symptoms began at age two, with mild and progressive worsening over the years. She was found to harbor a homozygous mutation (c.12232 C>T, p.Arg4078Ter) in the SACS gene. Neurologic examination showed spastic-ataxic gait, dysarthric speech, four limbs ataxia, and spastic hypertonia with lower limbs' hyperreflexia.
Brain-MRI showed atrophy of the superior vermis and anterior lobes of the cerebellum. Electroneurography study showed a mixed sensory-motor polyneuropathy. Vestibular evaluation found gaze-evoked and rebound nystagmus on horizontal and vertical gaze, symmetrical bilateral vestibular impairment mostly involving the horizontal semicircular canals and slight impairment of VOR suppression. Perceptual speech assessment revealed mild ataxic dysarthria with scanning speech, pneumophonic incoordination, phonation on residual air, irregular articulatory breakdowns, consonant and vowel distortions, and a variable speech rate. Acoustic analysis showed slow speech, an alterated diadochokinetic (DDK) regularity and reduced rate, a significant reduction of the maximum phonation time and a higher standard deviation of the power spectral density. Balance, measured with eyes open/closed on a firm/soft surface, was impaired in all conditions. Sway amplitude and area increased with task difficulty until the inability to stand on the soft surface with closed eyes. The request for a cognitive task worsened balance, as well as the getting up, sitting down, and turning phases of the instrumented TUG test, but not walking.

Conclusion: We report a case of ARSACS with peculiar clinical-instrumental findings including vestibular disfunction, ataxic dysarthria, balance and gait specific alterations.