IKBKAP

To assess vestibular function in patients with familial dysautonomia (FD), a hereditary sensory and autonomic neuropathy - caused by a mutation in the IKBKAP gene (c.2204 + 6 T>C) - and characterized by marked gait ataxia.

METHODS: Cervical and vestibular evoked myogenic potentials (cVEMPs and oVEMPs) were recorded from the sternocleidomastoid (SCM) and extraocular muscles in 14 homozygous patients, 2 heterozygous patients, and 15 healthy controls during percussion of the forehead.

RESULTS: cVEMP and oVEMP amplitudes were significantly lower, and peak latencies significantly delayed, in the FD patients. There were no differences in overall EMG during attempted maximal voluntary contractions of the SCM muscle, suggesting intact efferent function. The two heterozygotes with a minor haplotype missense (R696P) mutation in exon 19 of the IKBKAP gene had cVEMP responses less affected than the homozygous.

CONCLUSIONS: The founder mutation in the IKBKAP gene affects the development of vestibular afferent pathways, leading to attenuated cVEMPs.

SIGNIFICANCE: Vestibular abnormalities may contribute to the gait ataxia in FD [1].

1)
Gutiérrez JV, Kaufmann H, Palma JA, Mendoza-Santiesteban C, Macefield VG, Norcliffe-Kaufmann L. Founder mutation in IKBKAP gene causes vestibular impairment in familial dysautonomia. Clin Neurophysiol. 2017 Nov 26;129(2):390-396. doi: 10.1016/j.clinph.2017.11.010. [Epub ahead of print] PubMed PMID: 29289840.