Title
Auditory neuropathy is asymptomatic in one of two forms of autosomal dominant axonal Charcot-Marie-Tooth disease linked to 8P21 chromosome

Permalink
https://escholarship.org/uc/item/66w1b893

Authors
Butinar, D
Starr, A
Zidar, J
et al.

Publication Date
2004-09-01

License
CC BY 4.0

Peer reviewed
SC318
Auditory neuropathy is asymptomatic in one of two forms of autosomal dominant axonal Charcot-Marie-Tooth disease linked to 8p21 chromosome
D. Butinar,1 A. Starr,2 J. Zidar,3 D.-M. Georgiou3 and J. Vatovec5
1Department of Neurology, Institute of Clinical Neurophysiology, University Medical Center, Ljubljana, Slovenia, 2Department of Neurology, University of California Irvine, USA, 3The Cyprus Institute of Neurology and Genetics, Nicosia, Cyprus, 4Department for Otorhinolaryngology and Head and Neck Surgery, University Medical Center, Ljubljana, Slovenia

Hearing loss and deafness is recognized as an uncommon phenotypic variant in CMT. It can be found in both hereditary and sporadic forms of peripheral neuropathies due to associated auditory neuropathy. It is characterised by hearing impairment, abnormal auditory brainstem responses (ABRs) in the presence of normal cochlear receptor functions. We have searched for AN in 17 members of two families with the axonal dominant Charcot-Marie-Tooth (CMT) disease that showed linkage to the same chromosomal region at chromosome 8p21 and have no hearing complaint. DNA sequencing revealed a novel missense mutation in the neurofilament-light (NF-L) gene in family 1, while no mutation in the coding region of this gene has been identified in the family 2. In addition to the typical clinical and electrophysiological signs of the axonal CMT disease, three members of the second family exhibited also clinical and electrophysiological signs of the pyramidal and lateral lemniscus tracts involvement. Auditory measurements included ABRs, distortion product otoacoustic emission (DPOAE), tonal audiogram and speech audiogram. In eight members of the family 2, ABR’s were not elicitable or abnormal, while DPOAEs and speech audiograms and gap detection were normal. The members of the family 1 had completely normal test results. We conclude that, in addition to the pyramidal tract involvement, a subclinical AN is an additional attribute that differentiates the two families. It can be speculated that the diseases in these two families, despite being linked to the same chromosomal locus, may be dissimilar, i.e. caused by mutations in different genes.