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Poster P17

Genetic distribution of inherited peripheral neuropathies in Türkiye

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Background: Inherited neuropathies encompass a spectrum of diseases, including subtypes of Charcot-Marie-Tooth disease (CMT), hereditary neuropathy with liability to pressure palsies (HNPP), hereditary sensory neuropathy (HSN), distal hereditary motor neuropathy (dHMN), and other rare and complex neuropathies. Expanding our understanding of the genetic landscape in these populations is essential for accurate diagnosis and improved patient care.

Methods: The distribution of genetically diagnosed patients with CMT, dHMN, and HSN, from 314 families (370 patients), is evaluated. Patients with HNPP or other complex neuropathies were excluded from the analysis.

Results: The most frequent subtype was CMT1 (135 families), followed by CMT4 (45 families), dHMN (33 families), CMT-I (31 families), CMT2 (27 families), AR-CMT2 (26 families), and HSN (11 families. Interestingly, biallelic variants were responsible for the disease in 110 families. Parental consanguinity was noted in 117 patients. Pathogenic variants were identified in 48 different genes. The most frequent variation was PMP22 duplication in all cohort, followed by the pathogenic variants in GJB1, MFN2, SH3TC2, GDAP1, HINT1, and SORD, respectively. Regarding subtypes, SORD-neuropathy was the leading cause in dHMN, whereas RETREG1 variants were the most frequent in HSN.

Conclusion: This study highlights the significant genetic heterogeneity of inherited neuropathies, emphasizing the predominance of biallelic variants in our cohort, which contrasts with findings from similar studies in European populations. These results underscore the importance of examining genetic variations across diverse populations to gain a deeper understanding of the underlying genetic architecture.