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Charcotâ€“Marieâ€“Tooth neuropathy: clinical phenotypes of four novel mutations in the MPZ and Cx 32 genes

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Abstract

Charcotâ€“Marieâ€“Tooth Hereditary Neuropathy is a heterogeneous syndrome associated with mutations in several different genes including peripheral myelin protein 22, myelin P0, connexin 32, and early growth response 2. There is considerable variability in the phenotypic expression of this syndrome and the relationship of this variability to mutation genotypes requires extensive analysis. Here we describe the phenotypes and genotypes of four new mutations underlying the Charcotâ€“Marieâ€“Tooth syndrome and document segregation with disease. Four families with Charcotâ€“Marieâ€“Tooth were ascertained, examined, and evaluated electrophysiologically. Each family had peripheral blood DNA screened for mutations in myelin protein 22, myelin P0, and connexin 32. Two families were found with new mutations in the myelin P0 gene: S140T in the extracellular domain and K236del in the cytoplasmic domain. All families showed

segregation of the mutations with the Charcotâ€“Marieâ€“Tooth phenotype as did a new family with the rare G163R mutation in the membrane domain. A 49-year-old man with the S140T mutation demonstrated conduction block on electrophysiological testing. A family with a novel S49P mutation in the connexin 32 gene had a neuropathy with very slow nerve conduction. These new mutations in the myelin P0 and connexin 32 genes help to clarify the pathophysiology of the clinical Charcotâ€“Marieâ€“Tooth syndrome. The S140T mutation in myelin P0 can be associated with conduction block and Charcotâ€“Marieâ€“Tooth should be part of the differential diagnosis of that phenomenon. Mutations in the cytoplasmic domain of myelin P0 can cause clinical neuropathy. The S49P mutation in the connexin 32 gene can produce aspects of a demyelinating type of X-linked hereditary neuropathy.

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Keywords

Neuropathy; Charcotâ€“Marieâ€“Tooth; Myelin Pzero; Comnexin 32

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