The MPZ gene provides instructions for making a protein called myelin protein zero. It is the most abundant protein in myelin, a protective substance that covers nerves and promotes the efficient transmission of nerve impulses.

Charcot-Marie-Tooth disease (CMT) is a hereditary monogenic peripheral nerve disease. Variants in the gene encoding myelin protein zero (MPZ) lead to CMT, and different variants have different clinicalphenotypes. A variant site, namely, c.389A > G (p.Lys130Arg), in the MPZ gene has been found in Chinese people. The pathogenicity of this variant has been clarified through pedigrees, and peripheral blood-related functional studies have been conducted.

Whole-exome sequencing and Sanger sequencing were used to detect the c.389A > G (p.Lys130Arg) variant in the MPZ gene in family members of the proband. Physical examination was performed in the case group to assess the clinical characteristics of MPZ site variants. The expression of MPZ and phosphorylated MPZ in the blood of 12 cases and 12 randomly selected controls was compared by RT-qPCR, Western blotting, and ELISA.

The proband and 12 of her family members presented the AG genotype with different clinical manifestations. The expression of MPZ mRNA in the case group was increased compared with that in the control group, and the levels of MPZ and phosphorylated MPZ in peripheral blood were higher than those in normal controls.

The heterozygous genotype of the c.389A > G (p.Lys130Arg) variant in the MPZ gene mediated the increase in MPZ and phosphorylated MPZ levels in peripheral blood and was found to be involved with CMT [1].

1)
Hao X, Li C, Lv Y, Zhou T, Tian H, Ma Y, Ding J, Li X, Wang Y, Wang L, Yang P. MPZ gene variant site in Chinese patients with Charcot-Marie-Tooth disease. Mol Genet Genomic Med. 2022 Feb 17:e1890. doi: 10.1002/mgg3.1890. Epub ahead of print. PMID: 35174662.