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CMT4G: Rare Form of Charcot-Marie-Tooth Disease in Slovak Roma Patient

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Abstract : The Roma (Gypsies) is a transnational minority with a high degree of consanguineous marriages. Similar to other genetically isolated founder populations, the Roma harbor a number of unique or rare genetic disorders. This paper discusses about a rare form of Charcot-Marie-Tooth disease – type 4G (CMT4G), also called Hereditary Motor and Sensory Neuropathy type Russe, an autosomal recessive disease caused by mutation private to Roma characterized by abnormally increased density of non-myelinated axons. CMT4G was originally found in Bulgarian Roma and in 2009 two putative causative mutations in the HK1 gene were identified. Since then, several cases were reported in Roma families mainly from Bulgaria and Spain. Here we present a Slovak Roma family in which CMT4G was diagnosed on the basis of clinical examination and genetic testing. This case is a further proof of the role of the HK1 gene in pathogenesis of the disease. It confirms that mutation in the HK1 gene is a common cause of autosomal recessive CMT disease in Roma and should be considered as a common part of a diagnostic procedure.

Keywords : gypsies, HK1, HSMN-Russe, rare disease
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