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Determination variations encountered on KATP protein encoding genes in Raynaud's phenomenon cases

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Abstract

Raynaud's phenomenon (RP) is a vascular disorder characterized by recurrent vasospastic response of the fingers and toes to cold or emotional stimuli. Classically ischemia, deoxygenation and hyperemia are the sequence of a typical attack. RP is a relatively common disorder in worldwide population with the prevalence of 3.3% to 22%. ATP-dependent potassium channels (KATP) containing Kir6.1 and SUR2A proteins (KCNJ8/ABCC9 genes), particularly in the regulation of vascular tone in the coronary arteries has a critical role and deficiency or defects in the function can cause vasospasm associated with Prinzmetal's angina. It would be important to determine whether variations of KATP genes related to Raynaud's phenomenon is thought to be associated with vasospasm. It is believed that the studies describing mechanisms involved in the pathogenesis of inherited vascular disorders offers the best opportunity for investigation of the early stages of pathogenicity and diagnosis of Raynaud's phenomenon and associated other diseases. The purpose of this study, KATP channel which is gene coding of run across mutations in vasospasm associated with Raynaud's phenomenon in patients to determine the characterization and investigation of mutation frequency. In our study; the cases with Raynaud's phenomenon, the relation between the variation in the KCNJ8/ABCC9 genes (S422L/V734I) was examined. 50 subjects who were diagnosed with Raynaud's phenomenon (patient group) and 50 healthy subjects (control group) were included in the study. Variations were determined using the Tetra-Primer ARMS PCR method. KATP channel protein variants analysed for possible correlations among Raynaud's phenomenon were not observed in patient and control groups.

Keywords: Raynaud's Phenomenon, KATP Channel Proteins, KCNJ8/ABCC9 Genes, S422L/V734I Variants.