GENETICS AND GENODERMATOSES

A NOVEL MUTATION IN THE SLCO2A1 GENE IN A CHINESE FAMILY WITH PACHYDERMOPERIOSTOSIS

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Background: Pachydermoperiostosis(PDP) is a rare autosomal recessive condition characterized by 3 major symptoms: pachydermia, periostosis, and digital clubbing. Two genes have been associated, HPGD and recently SLCO2A1. Until now, only few cases with mutations in these genes have been reported.

Objective: We aimed to identify the genetic cause of the Chinese family with PDP.

Methods: Standard clinical and laboratory evaluation was carried out. We screened for the gene mutation in the family members using whole Exome Sequencing and Sanger Sequencing. And we discuss the clinical features and known mutations of previously reported cases identified through a PubMed literature review.

Results: According to the classic clinical definition, the proband and his father in one Chinese family were diagnosed with the complete and incomplete form of the PDP respectively. And We identified a novel splice site mutations(c.861+1G>A) in the SLCO2A1 gene.

Conclusion: Our findings expand the spectrum of SLCO2A1 mutations and provide clues to the phenotype–genotype relations involved in PDP.

Keywords: Pachydermoperiostosis; SLCO2A1 Gene; Mutation; Phenotype