GENETICS AND GENODERMATOSES

A RARE CAUSE OF RECURRENT LEG ULCERS, IDENTIFIED BY A NOVEL MUTATION IN PEPD GENE: PROLIDASE DEFICIENCY

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Background: Prolidase deficiency (PD) is a rarely seen hereditary disorder of aminoacid metabolism, whose incidence is 1-2/1.000.000. It is inherited in an autosomal recessive manner first described in 1968.1 This disease results from many different types of mutations in the peptidase D (PEPD) gene encoding prolidase, an enzyme which breaks down iminodipeptides that have proline and hydroxyproline. 3
Herein, a 20 years-old male patient diagnosed with PD through PEPD gene analysis will be presented. The mutation detected in this case has never been reported in the literature yet.

Observation: A 20-years old male patient was admitted to our clinic with the complaint of recurrent wounds in both legs since his childhood. He had situs inversus totalis in his personal history of disease. Dermatological and physical examination revealed that there were ulcers on both legs and low anterior hairline, synophrys, high arched palate and poliosis . Laboratory and radiological findings were microcytic, hypochromic anaemia, thrombocytopenia, hypoalbuminemia, hypergammaglobulinemia and splenomegaly.
Recurrent and recalcitrant leg ulcers history since his childhood accompanied by anemia, thrombocytopenia, hypergammaglobeulinemia, splenomegaly and several dismorphic face findings prompted us to suspect prolidase deficiency. Thus, PEPD gene sequence analysis was performed and the results revealed a biallelic deletion in this gene (NM_000285.3 c.580delG (p.Val194Phefs*16) Homozygous). This deletion which was characterized by a single nucleotid deletion that led to frameshift mutation and the coding of a stop codon prematurely was evaluated as a cause of PD. With these clinical and laboratory findings, the patient was diagnosed with PD.

Key message: We report a patient diagnosed with PD, a very rare disorder, which is difficult to think of in a daily routine practice and a novel single nucleotid deletion of the PEPD gene.