

PROLIDASE DEFICIENCY: REPORT OF TWO BRAZILIAN SIBLINGS WITH ASSOCIATED RENAL DISORDER

CAMILA MATZENBACHER BITTAR; MERCEDES VILANUEVA; DIEGO MIGUEL; CAROLINA SOUZA; CRISTINA NETTO; FRANCISCO VERONESE; IDA SCHWARTZ

Objectives: This report describes two sibs (male and female) with prolidase deficiency (PD). We discuss some clinical aspects of PD among these two patients born from a consanguineous couple. Prolidase deficiency is a rare recessive disorder caused by mutations in the prolidase gene and characterized by severe skin lesions. Symptoms are usually multisystemic and its hallmarks are ulcerations of the skin, chronic dermatitis, recurrent infections, an unusual facial appearance and splenomegaly. The pathophysiology of PD is still poorly understood. Methods: We have been following these two patients, who were diagnosed as by a TLC and ion-exchange-chromatography of aminoacids compatible with PD, in Giannina Gaslini Institute, Genova, Italy. Results: Patient 1: male 27 yo, started with splenomegaly in the first month of life, and with a secondary hemolytic anemia. Splenectomized since 1989 (partial splenectomy). He developed recurrent infections, erythematous dermatitis and severe progressive ulcerations of lower extremities and the motor and mental development is normal. The diagnosis of PD was made at 9 yo of age. Laboratory examination revealed hypergammaglobulinaemia, specially Ig E. At the moment, he is on nephrological evaluation due to early onset of proteinuria since 2007. Subsequently it was made a renal biopsy, showed Focal Segmental Glomerulosclerosis (FSGS) at 26 yo. His psychomotor development was normal. Patient 2: female 24 yo, showed her first lesion at infancy as well, but appear to have a more localized disease, with the scalp and axillary more affected. She has smaller leg ulcers than her brother. A scalp biopsy was made recently and showed psoriatic lesions. Laboratory analysis also revealed hyperimmunoglobulin E. Renal evaluation evidenced mild proteinuria.