GENETICS AND GENODERMATOSES

PAPILLON-LEFÈVRE SYNDROME: A NEW OBSERVATION

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Background: Papillon-Lefèvre syndrome (PLS) is a genodermatosis of autosomal recessive inheritance due to mutations in the cathepsin C gene responsible for rare immune deficiency. PLS combines diffuse palmoplantar keratoderma (KPP) and periodontal disease with edentation leading to premature loss of temporary and permanent teeth.

Observation: We report the case of a 13-year-old girl from related parents. The girl went to a pediatric dermatology consultation for erythematous-like lesions of the elbows and knees associated with diffuse palmoplantar keratoderma, which has been evolving since the age of 5. She also reports a loss and abnormal mobilization of her teeth during infancy. She is currently complaining of poor oral health and repeated dental infections despite rigorous oral hygiene. Clinical examination revealed severe gingivitis, superior canine mobility, lower lateral incisor drop, diffuse orange-like palmoplantar keratoderma, and erythematosquamous elbow injury without a sweating disorder. She received a local treatment based on emollients, keratolytic (salicylic vaseline) and dermocorticoides that made her PPK very discreet. She is also treated by a stomatologist who put her on antibiotic related to dental care.

Key message: PLS is a rare immune deficiency often characterized by a diffuse PPK and severe gingivitis causing abnormal tooth loss and alveolysis. This syndrome is related to a loss of function of cathepsin C responsible for colonization of the gingival biofilm by anaerobic microorganisms. The treatment is based on oral retinoids in severe forms or local keratolytics in moderate forms associated with stomatological support.