Olmsted syndrome (OS) is a rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma (PPK) and periorificial keratotic plaques with severe pruritus of lesions. Diffuse alopecia, constriction of digits, and onychodystrophy also has been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes (1, 2). The following mutations are actually known: TRPV3 gene (G573S; 607066.0001), G573C (607066.0002), W692G (607066.0003).

Case Report

Infant, with good growth and development, starts dermatosis at 2 months of age, located in palms, soles, axillary folds, popliteal fossa, neck and periorificial sectors (perineal-gluteal and genital, perioral, perinasal and external auditory canal), characterized by erythemato-squamous plaques with intense red erythema and hyperkeratosis with thick, yellowish scale, without itching. No deafness was found.

We propose an Acrodermatitis enteropathica (deficiency of zinc absorption) because of the location of lesions and the value of zinc in the lower limit of normality. He was treated with zinc sulphate 1mg/kg/day, without improvement. Dr. R. Ballona (Peru) suggested the diagnosis of Olmsted syndrome and a biopsy was performed. Acitretin was prescribed with little improvement. Molecular diagnostic study is in process.

Histopathology: Psoriasiform hyperplasia, granulous layer with coarse granules, parakeratotic hyperkeratosis, high nucleus/cytoplasmic ratio, even in suprabasal layers.

Clinical features were characteristic of OS. Other differential diagnoses were ruled out because the patient had periorificial hyperkeratosis without deafness. Zinc treatment was not effective discarding acrodermatitis enteropathica and microorganisms were not found (chronic mycosis). Light and ultrastructural features are in agreement with augmented mitotic activity (3). Thick clumps of cytokeratin in basal and spinous layers were found. Suprabasal mitosis are in concordance with previously increased mitotic activity reported (3). We propose that a strong attachment between layers in mucous stratum, alterations, mainly in desmosomes and hemidesmosomes, may contribute to hyperkeratosis. The chronic (inflammatory) mechanism that leaves to digital constriction, which in turn may progress to autoamputation of fingers and toes is still unknown.

Discussion & Conclusion