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Patient With Malformations of the Face, Mouth, and Fingers: Type I Orofaciodigital or Papillon-Léage Psaume Syndrome

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Sr. Director:

Type I orofaciodigital syndrome (OFDS), or Papillon-Léage Psaume syndrome (Mendelian inheritance in man 605041), is characterized by the presence of orofacial and digital malformations.

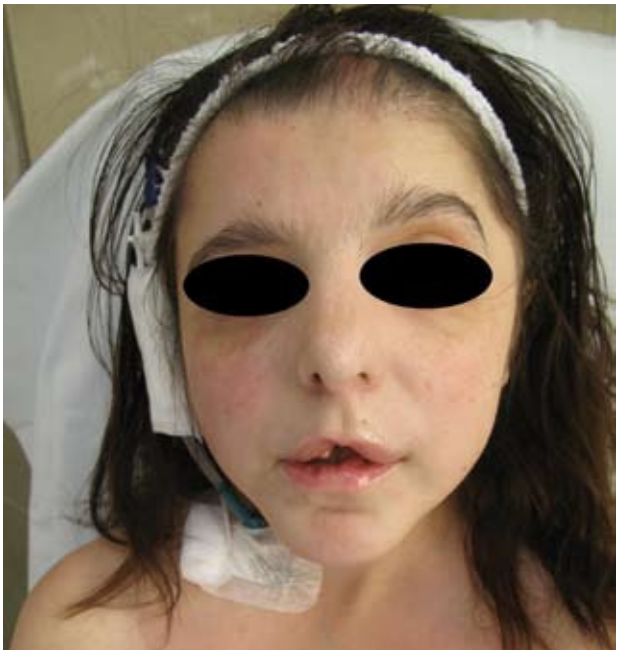

Figura 1. Abnormal facies with hypertelorism, a broad nasal root, a midline cleft of the upper lip, tooth loss, and diffuse alopecia.

The patients often develop dermatologic disorders during the early months of life. The dermatologist can play a key role in establishing an early diagnosis. The disease is sporadic in 75% of cases, while the remainder show an X-linked dominant inheritance. It is lethal in males, and is therefore only observed in females. Its incidence is of 1 in 50 000 live births.[1] The gene responsible for the disease is found on the short arm of the X chromosome (Xp22.2-22.3). The gene product of this fragment may play a fundamental role in organogenesis.[2]

We present the case of a 29-year-old woman, admitted to the nephrology department due to vascular rejection of a kidney transplant. She was seen for severe generalized pruritus and marked skin dryness during the hospital admission. The important findings on physical examination were an abnormal facies with hypertelorism, a broad nasal root, low-set ears, a cleft upper lip, and the absence of several teeth (Figure 1). She also presented brachydactyly of the fingers of both hands and of some of the toes (Figure 2). The hair was thin, dry, and fragile, with areas of lower hair density most evident at the line of implantation on the forehead.

The patient had been born by normal vaginal delivery after an uncomplicated pregnancy. There was no consanguinity between the parents and karyotyping was 46XX normal. She had multiple phenotypic

abnormalities present since birth, in particular a trilobulated tongue with an accessory frenulum, dental abnormalities, hypoplasia of the alar cartilages, brachydactyly, and areas of diffuse alopecia on the scalp. She had undergone multiple reparatory operations during childhood. The progressive loss of teeth made it difficult to perform basic activities such as mastication or speaking. At 17 years of age she developed renal failure due to the presence of multiple cysts, finally requiring kidney transplant. She also presented cutaneous xerosis and pruritus, exacerbated by the deterioration in renal function. Treatment with oral antihistamines and the use of emollients was recommended.

In 1954, Papillon-Léage and Psaume described a syndrome with facial, oral, and digital malformations.[3] Later, the heterogeneous group of syndromes that included malformations at these sites was given the name OFDS.[4] Thirteen variants have been described, and the identification of each one is difficult as they have common characteristics and traits. The syndrome described by Papillon-Léage and Psaume is known as type I OFDS, and is the most common variant. Only types I and II have skin alterations.[5] The abnormalities in the oral cavity are the most constant and characteristic. Patients can have a lobulated tongue; a short or accessory frenulum; abnormalities of the hard and soft palate, of the floor of the mouth, and of the gums; supernumerary teeth; and the absence of teeth. A midline cleft of the upper lip is characteristic. The facial abnormalities include a prominent forehead, hypertelorism, a broad nasal root, hypoplasia of the alar cartilages and of the jaw, micrognathia, and low-set ears.[6] The most common alterations of the digits are clinobrachysyndactyly and polydactyly. The radiologic image of the phalanges shows an irregular reticular radiolucency in the middle part of the bones, a finding considered pathognomic of type I OFDS.[7] Mental retardation or abnormalities of the central nervous system are present in 50% of patients. The prognosis is determined by the degree of organ involvement. Up to 15% present renal cysts that can lead to terminal renal failure.[8] Hepatic and pancreatic cysts can also develop.

The dermatologic alterations are particularly useful for establishing an early diagnosis. The appearance of multiple, milia-like cysts on the scalp, face, and dorsum of the hands is common in the first years of life. These resolve leaving pinpoint scars in the affected areas.[9] More than half of patients present diffuse alopecia with thin, dry, fragile hair. Broken hair at the hairline is a typical finding. The patients also present cutaneous xerosis and dermatoglyphic abnormalities of the digits.[10]

An early diagnosis is essential in a newborn infant with malformations. The finding of facial, oral, and digital abnormalities associated with skin alterations should lead to a suspicion of the diagnosis of type I OFDS. The milia-like cysts are only present in this variant. A multidisciplinary approach is necessary. Reparative surgery of the malformations should be performed in the early years of life to avoid sequelae in the patient's development and maturation. Periodic dental controls should be performed to prevent dental loss. Dermatologic care requires the frequent use of emollients and a balanced diet that prevents nutrient deficiencies. In addition, periodic controls of renal and liver function should be performed to detect polycystic disease early.

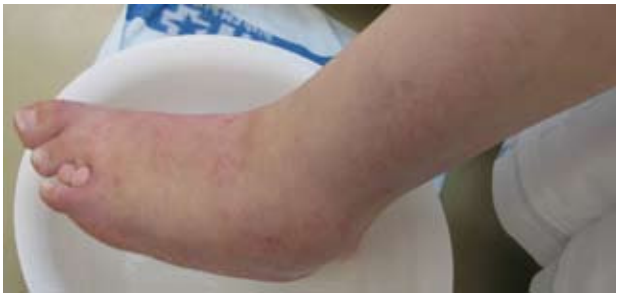

Figure 2. Brachydactyly of the third and fourth toes of the left foot.

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Conflicts of Interest
The authors declare no conflicts of interest.

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Refractory Subacute Cutaneous Lupus Erythematosus With a Response to Efalizumab

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To the Editor:

Subacute cutaneous lupus erythematosus is a specific form of lupus that occurs with annular or polycyclic erythematous plaques in areas exposed to sunlight.[1] Conventional treatments include the use of sunscreens, topical and oral corticosteroids, sulfones, antimalarial drugs, and immunosuppressive agents including cyclosporine, azathioprine, or methotrexate.[1]

We present the case of a 54-year-old woman—allergic to heparin, with a history of arterial hypertension, deep vein thrombosis in the lower right leg, and bilateral pulmonary thromboembolism resulting from an ankle fracture—who had been monitored for subacute cutaneous lupus erythematosus for 25 years. Since the initial diagnosis, the patient has presented continual outbreaks of cutaneous lesions consisting of annular erythematous plaques with a shiny surface and clear margin around the eyebrows, and on the cheeks and upper back (Figure 1). There were no associated systemic symptoms and tests for antinuclear antibodies (ANA) and anti-Ro antibodies were positive. The condition progressed with successive poorly controlled cutaneous outbreaks that were treated with medium to high potency topical corticosteroids, topical calcineurin inhibitors, oral prednisone (minimum of 15 mg on alternate days up to 60 mg daily), and systemic agents (hydroxychloroquine, azathioprine, methotrexate, and cyclosporine). All forms of treatment had been suspended due to ineffectiveness, poor tolerance, or adverse effects. As the daily dosage of 30 mg of prednisone could not be reduced, subcutaneous efalizumab 1 mg/kg/week was administered and the skin lesions resolved after 2 months (Figure 2). The patient was symptom free at the end of 4

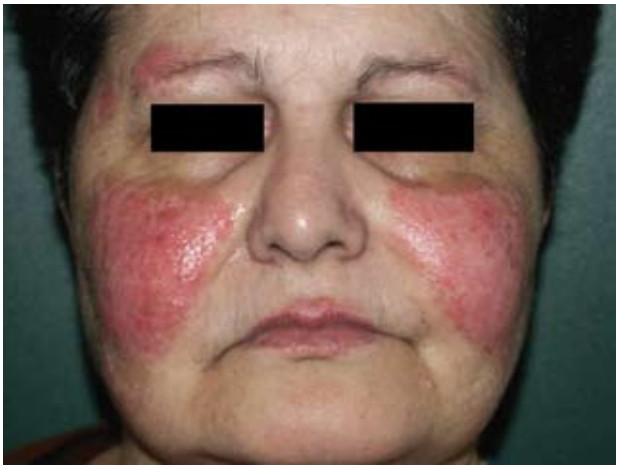

Figure 1. Clearly defined infiltrated erythematous plaques on the eyebrows, cheeks, and upper lip.

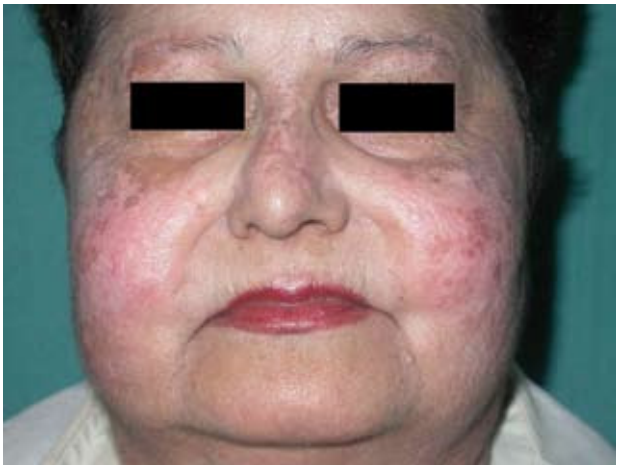

Figure 2. Resolution of the lesions after 2 months treatment with efalizumab.