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CASE REPORT

A four-month old male patient, born from non-consanguineous parents, coming from the countryside of São Paulo, was referred to our clinic due to chronic diarrhea, protein-energy malnutrition and skin lesions. At admission, he presented exulcerations covered with honey-colored hematic crusts on the anterior chest, upper limbs, dorsal area, face and especially on the scalp. The patient also had a cleft palate, deformity of the ears, decreased amount of eyelashes and eyebrows, besides erythematous papules on lower limbs, micropenis, dystrophy on all nails and personal history of surgery for ankyloblepharon (Figures 1-3). Skin biopsy result suggested epidermolysis bullosa.

Given the clinical manifestations we diagnosed Hay-Wells Syndrome. Foam dressings (Mepilex ®) on the scalp and topical antibiotics in areas with exulcerations and exudation were instituted.

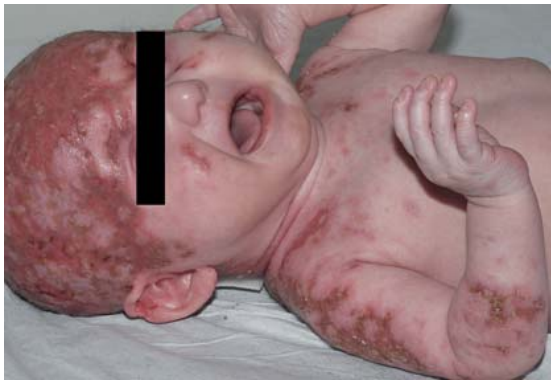

FIGURE 1: Exulcerations covered with honey-colored hematic crusts mainly on the scalp, chest, and upper limbs, besides right-ear deformity

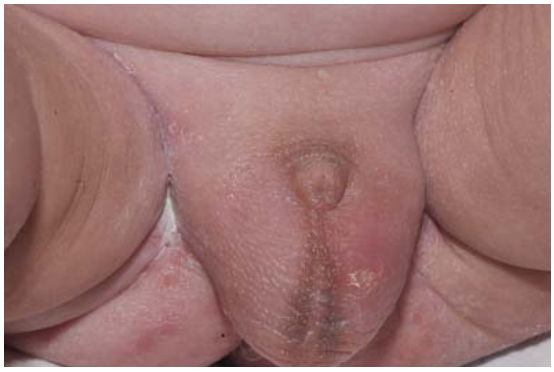

FIGURE 2: Detail of genital region showing micropenis

DISCUSSION

Hay-Wells syndrome is a rare syndrome caused by a specific subtype of mutations in p63 gene, which encodes a transcription factor involved in embryonic ectodermal development.[1] At least six syndromes caused by p63 mutations are recognized: Hay-Wells syndrome (AEC), ectrodactyly ectodermal dysplasia clefting syndrome (EEC), Rapp-Hodgkin syndrome (now considered synonymous with AEC syndrome), limb mammary syndrome (LMS), acro-dermato-ungual-lacrimal-tooth syndrome (ADULT) and ectrodactyly syndrome(Split-hand/Split-foot malformation).[2]

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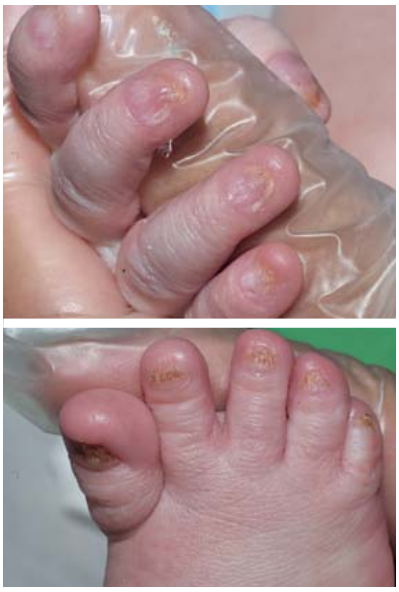

Figure 3: Ungual dystrophy in hands and feet

Changes are evident at birth and characterized by ectodermal abnormalities of the skin, teeth, hair and nails, associated with ankyloblepharon, and cleft palate and/or lip.[3] Over 90% of all affected infants will present erythrodermia and superficial erosions, involving the scalp. Abnormal granulation tissue is common, with colonization and recurrent bacterial infections, besides decrease in body hair, malformed external ears and recurrent otitis media. Nails may be normal or appear thick, hiperconvex, absent or dystrophic.

It is not possible to differentiate a sporadic mutation from germinal mosaicism in parents, thus there is a risk that future children will be affected.[4]

Clinical features are not specific and erythrodermia with desquamation and exulcerations in the postnatal period can lead to misdiagnosis of epidermolysis bullosa, as occurred in this case.[5]

Therapy should be multidisciplinary and focused on ankyloblepharon and oral cleft corrections, management of infections, in addition to local care with appropriate dressings and gentle removal of scales and crusts. Treatment of exulcerated lesions is key to improve the quality of life of patients, alleviating pain, accelerating healing and preventing recurrent episodes of bacterial colonization and infection. In this sense, we chose to use a semi-occlusive dressing of the foam class. These dressings can be cut out in the exact shape of the wound and act by a combination of exudate absorption and moisture loss through evaporation. On the other hand, they maintain thermal insulation and a moist environment. These dressings can be changed every four days. The prognosis is good, with improvement of skin lesions according to age, however it is necessary to provide genetic counseling for the parents.[5] ❑

Abstract: Hay-Wells syndrome or AEC (Ankyloblepharon, Ectodermal dysplasia and Cleft lip and palate syndrome) is a rare ectodermal disorder. The treatment is aimed to prevent clinical complications. We describe the case of a four-month old male patient with erosions on the scalp, trunk and arms, trachyonychia, deformity of the ears, micropenis, cleft palate, decreased eyebrow and eyelash hairs, in addition to antecedents of surgical correction of ankyloblepharon. The importance of the correct diagnosis is emphasized, besides the investigation of the associated diseases, treatment of complications and genetic counseling of the parents.
Keywords: Cleft palate; Ectodermal dysplasia; Skin diseases, genetic

REFERENCES

1. Hay RJ, Wells RS. The syndrome of ankyloblepharon, ectodermal defects and cleft lip and palate: an autosomal dominant condition. Br J Dermatol. 1976;94:277-89.
2. Sawardekar SS, Zaenglein AL. Ankyloblepharon-Ectodermal Dysplasia- Clefting Syndrome: A Novel p63 Mutation Associated with Generalized Neonatal Erosions. Pediatr Dermatol. 2011;28:313-7.
3. Clements SE, Techanukul T, Lai-Cheong JE, Mee JB, South AP, Pourreyron C, et al. Mutations in AEC syndrome skin reveal a role for p63 in basement membrane adhesion, skin barrier integrity and hair follicle biology. Br J Dermatol. 2012;167:134-44.
4. Barbaro V, Nardiello P, Castaldo G, Willoughby CE, Ferrari S, Ponzin D, et al. A novel de novo missense mutation in TP63 underlying germline mosaicism in AEC syndrome: Implications for recurrence risk and prenatal diagnosis. Am J Med Genet A. 2012;158A:1957-61
5. Rosa DJ, Machado RF, Martins Neto MP, Sá AA, Gamonal A. Hay-Wells syndrome: a case report. An Bras Dermatol. 2010;85:232-5.

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