



GENETICS AND GENODERMATOSES

BLOCH-SULZBERGER SYNDROME - AN X-LINKED GENODERMATOSIS

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Background: Bloch-Sulzberger syndrome is a rare, dominant, X-linked genodermatosis that has both cutaneous and extracutaneous manifestations. It affects ectodermal and mesodermal tissues such as skin, eyes, hair, teeth and the central nervous system. There are around 800 registered cases worldwide, and the estimated incidence is around one in 40,000 children. This condition mainly affects newborns of the female sex, being lethal, most of the times, when it affects the masculine sex, occurring spontaneous abortion in the majority of cases.

Observation: Patient, female, 1 month of age, presented with papules, vesicles and crusts in upper and lower limbs, with onset since birth. Healthy child, without other comorbidities, born at term, of normal delivery and without intercurrents. On physical examination, she had vesicobulous and crust lesions with a linear distribution along the Blaschko lines in upper and lower limbs and in lower intensity on the right lateral side of the trunk. Her mother said that at birth she had erythematous lesions similar to those of the patient, but that she had a spontaneous resolution of the condition, without investigation at the time. Clinical findings led to the diagnosis of Bloch-Sulzberger syndrome in the vesicle-muscular stage with transition to the verrucous stage. The patient was referred for multidisciplinary follow-up with ophthalmology and neuropediatrics. Boricated water was used to clean the lesions and emollients. The child, with 7 months of life, presented evolution of the vesicobulous and verrucose lesions for hyperchromic spots following Blaschko lines. Currently, the patient is under multidisciplinary outpatient follow-up and occupational therapy.

Key message: The first manifestations occur in the neonatal period and progress through several well-defined stages, which may occur concomitantly or sequentially. There may be extracutaneous manifestations and the involvement of a multidisciplinary team is of paramount importance in patient management.

