GENETICS AND GENODERMATOSES

IS YELLOWISH KERATODERMA A CLUE FOR THE DIAGNOSIS OF SJOGREN LARSSON DISEASE?

A Zaouak [1] - G Abdessalem [2] - S Abdelhak [2] - H Hammami [3] - S Fenniche [3]

Habib Thameur Hospital, Department Of Dermatology, Habib Thameur Hospital, Tunis, Tunisia [1] - Institut Pasteur De Tunis, Laboratory Of Biomedical Genomics And Oncogenetics, Tunis, Tunisia [2] - Habib Thameur Hospital, Dermatology Department, Tunis, Tunisia [3]

Background: Sjogren Larssen syndrome (SLS) is a rare autosomal recessive genodermatoses characterized by ichthyosis, spastic diplegia or tetraplegia, and mental retardation. It belongs to the spectrum of syndromic ichthyosis and is characterized by defective activity of fatty aldehyde deshydrogenase (FALDH), caused by mutations in the ALDH3A2 gene. More than 90 mutations of the ALDH3A2 gene have been reported for SLS. We report a case of SLS in a 20-month-old Tunisian boy with a common mutation of ALDH3A2 gene manifested in a new altered allele.

Observation: A 20-month-old boy, originating from Tataouine in the south of Tunisia and born of a consanguineous marriage, was referred to our department for a congenital ichthyosis. He was born as a collodion baby at 36 weeks of gestation. Dermatological examination revealed a generalized skin xerosis and an ichthyosiform hyperkeratosis predominant in his flexural areas and in his lower abdomen. The skin had a yellowish, dark brown appearance. He also had a yellowish palmoplantar keratoderma. He had an impaired psychomotor development. His neurological examination revealed an axial hypotony and spastic diplegia. DNA sequencing confirmed the diagnosis of SLS and revealed a new allelic form of mutation in ALDH3A2 gene. Treatment with emollients and keratolytics partially improved the patient's skin condition.

Conclusion: In summary, yellowish keratoderma associated to ichthyosis and neurological abnormalities could be a clue for the diagnosis of Sjögren Larsson disease. Our patient helps expanding the clinical and genetic spectrum of the SLS. This should be useful for carrier detection, genetic counseling and prenatal diagnosis.