GENETICS AND GENODERMATOSES

TYROSINEMIA TYPE II : THREE CASES REPORT

Samia Djoudi [1] - *Farid Ait Belkacem* [1] - *Dalila Bouharati* [1]

Hospital, Dermatology, Algiers, Algeria [1]

Background : Tyrosinemia Type II (Richner-Hanhart Syndrome, Oculocutaneous Tyrosinemia) is a rare autosomal recessive disorder caused by deficiency of tyrosine aminotransferase (TAT) and results in palmar and plantar hyperkeratosis, herpetiform corneal ulcers, and intellectual disability. We report 3 cases of this syndrome from the same family.

Observation : Femal patient 27 years old, from a consanguineous marriage of first degree, consults in march for a focal painful palmoplantar hyperkeratosis appearing since the age of 9 months. The painful and focal lesions character made us think tyrosinemia type II .The plasma tyrosine values is 211mg/l.
His brother 18years old and her sister 8 years old have similar lesions wich are less extensive and less painful, while the plasma tyrosine level are respectively 195 mg/l and 268mg/l.The urinary succinylacetone dosages are negative.
The ophthalmological examination does not detect any abnormality except hypermetropy in the younger sister.
All three patients report a summer improvement in lesions, something we have constated despite the fact that a restrictive diet has not been followed by the siblings.

Key message : Richner-Hanhart syndrome must be evoked before any painful palmoplantar hyperkeratosis. The diagnosis is easily confirmed by a plasma tyrosine . Our observation is original because of the notion of summer improvement of lesions, which has never been reported in the literature in our knowledge.