

Picture Stories

Richner-Hanhart Syndrome: A case report of an 11 month old female

Biplab Maji¹, Sandipan Dhar¹, Apurba Ghosh¹, Surupa Basu¹

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Case report

An 11-month-old girl had intermittent redness of eyes, excessive lacrimation and photophobia from 3 months of age and thickening of the soles for past 4 months. She was alert, active with a resting heart rate of 96/min and normal vital parameters. She had multiple tender hyperkeratotic plaques over the soles (Figure 1), but no lesions on the palms.



Figure 1: Hyperkeratotic plaques over soles

Eye examination showed central corneal clouding (Figure 2) with dendritic ulcer. She had no gum bleeding, oral ulcers or bony anomalies. She was developmentally delayed. Her sensory functions and other systems were normal.

¹*Institute of Child Health, 11 Bireswari Road, Kolkata 700017, India*

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Figure 2: Central corneal clouding



Figure 3: Resolved skin lesions over soles

Blood counts, liver and renal function tests, thyroid function tests and alpha-fetoprotein levels were within normal limits. Skiagram of chest and both knees and ultrasonogram of the abdomen were normal. Serum tyrosine level was 2657 μ mol/L (normal <550 μ mol/L), phenylalanine 2034.8 μ mol/L (normal <165 μ mol/L), with normal levels of other amino acids in serum. A diagnosis of Richner Hanhart syndrome was made and the child was started on a diet low in phenylalanine and tyrosine. On follow-up of the child after 2 months, the eye and skin lesions were found to be resolved (Figure 3) and the child was doing well.

Discussion

Richner Hanhart syndrome or tyrosinaemia type II (Oculo-cutaneous tyrosinaemia) is an autosomal recessive disorder resulting from the deficiency of the enzyme tyrosine aminotransferase (TAT)¹, which is located on chromosome 16q22.1-q22.3. Ocular lesions, painful palmo-plantar hyperkeratosis and occasionally mental retardation are the cardinal features of this disease. High blood levels of tyrosine result in deposition of tyrosine crystals in the cornea and give rise to the ocular changes². The eye symptoms develop by 2 weeks of age and include redness, lacrimation and photophobia. The eye signs are corneal clouding with central or paracentral corneal opacities, dendritic ulcers and corneal scarring. Skin manifestations usually begin after the first year of life, but may occur as early as the first month of life. The skin manifestations are well demarcated, gradually progressive, painful, non pruritic hyperkeratotic papules and plaques involving the soles and palms and associated with hyperhidrosis. The pain in the soles may be severe enough to prevent ambulation³. Mental retardation occurs in less than 50% of patients. Investigations reveal high urinary tyrosine levels with associated high plasma tyrosine levels estimated by tandem mass spectrometric assay. It is rarely necessary to perform a liver biopsy for TAT assay⁴. Dietary restriction of phenylalanine and tyrosine is the key step in the management of these cases⁵. In isolated skin lesions, retinoids can be prescribed either alone or in combination with dietary therapy⁶. In children with severe plantar hyperkeratosis where ambulation is difficult, autografts can be placed over these lesions as the disease spares autografts⁷. The threshold level of tyrosine for appearance of clinical manifestation is reported as 1000 μ mol/L. It is reasonable to keep the blood tyrosine level at 600 μ mol/L though there is no clear statement in this regard⁴. Richner Hanhart syndrome is a very labile disease, and with good dietary restriction of

phenylalanine and tyrosine, there is rapid & complete resolution of the eye and skin lesions⁴.

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