EPIDEMIOLOGY

A CASE REPORT OF EPIDERMOLYTIC HYPERKERATOSIS TYPE NPS-3 IN A MOTHER AND A CHILD

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Background: Epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosiform erythroderma is a rare autosomal dominant disorder characterized by an early onset, with erythroderma and bullous lesions, leading to severe generalized hyperkeratosis in adulthood. Mutations have been found in keratin 1 and keratin 10 genes. The clinical manifestations of EHK present striking heterogeneity and at least six clinical phenotypes have been identified.

Observation: We report on a case of EHK in a 14-year-old girl with erythroderma, erosions and blisters on the entire body surface at birth and generalized hyperkeratosis but without severe palm amd sole involvement in the later stage. On the basis of clinical and histopathologic findings, the diagnosis of EHK type NPS-3 was made.
The patient had been coming to the clinic for check ups but soon after we lost contact when she stopped attending. About three months ago, she came back to our clinic this time with a 6 month old baby. Her child also displayed the same symptoms and we were able to confirm the same diagnosis as the mother.

Key message: This case emphasizes the importance of genetic counselling and prenatal diagnosis to ensure that parents at risk are properly informed.