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Photo vignette

Infantile epidermolytic ichthyosis with prominent maternal palmoplantar keratoderma

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Abstract

Epidermolytic Ichthyosis (EI) is a rare autosomal dominant genodermatosis. Although an inherited disorder, 50% of cases represent novel mutations. This disorder presents as a bullous disease in newborns progressing to a lifelong ichthyotic skin disorder. Other manifestations include palmoplantar keratoderma (PPK). EI results from mutations in the keratin 1 and keratin 10 genes. Phenotypic variability is seen in affected individuals based on the genotypic mutation. We present a mother and her newborn son with EI and prominent PPK in the mother, which also developed in the child at a few months of age. Genotype analysis was performed on the newborn child who was found to harbor a mutation in the keratin 1 gene. This family demonstrates the phenotypic expression of PPK associated with keratin 1 gene mutations and illustrates the importance of genotype-phenotype correlation in this disorder.

Case synopsis

A 2-day-old baby boy was transferred from an outside hospital for evaluation of blisters. The patient had an overall unremarkable delivery. Approximately 24 hours after delivery, it was noted that the patient was developing flaccid vesicles and bullae accentuated in areas of pressure and trauma (Figure 1). The patient had been afebrile and feeding well without any signs of systemic toxicity. The patient was placed empirically on ampicillin and gentamicin as well as acyclovir for possible infectious etiologies. All cultures were negative. The patient's mother was known to have a keratoderma-like skin disorder of the palms and soles (Figure 2). The patient's mother also reported that she had similar blisters when she was born. Biopsies were obtained for further evaluation of the blistering eruption. The clinical differential diagnosis included other bullous disorders, in particular epidermolysis bullosa simplex.



Figure 1. Blistering and several areas of denuded skin in the infant patient

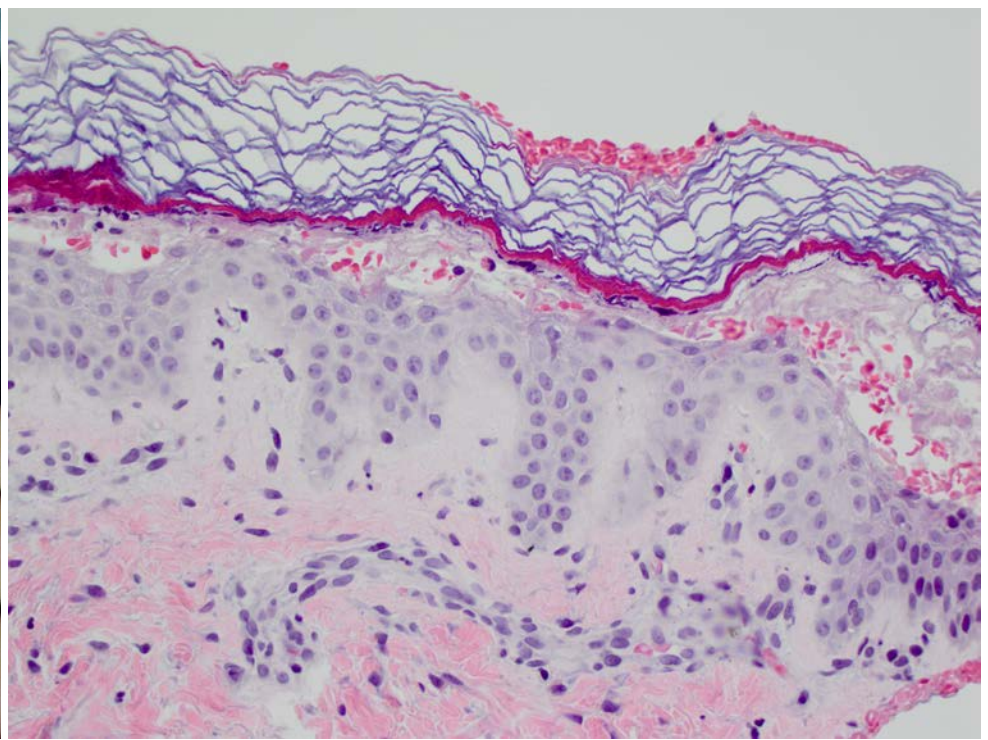


Figure 2. Diffuse thickening of the palmar aspect of the mother's hand. **Figure 3.** Subcorneal vesicular dermatitis, consistent with epidermolytic hyperkeratosis – 40X

Skin biopsy revealed a subcorneal vesicular dermatitis, consistent with EI (Figure 3). Given these findings, genetic tests were later performed and showed positivity for a mutation in the KRT 1 gene (I479T mutation) and negativity for mutation in the KRT 10 gene. This confirmed the diagnosis of EI with prominent PPK; the patient began to develop PPK at just a few months of age.

Diagnosis: Epidermolytic Ichthyosis with prominent palmoplantar keratoderma

Discussion

Epidermolytic Ichthyosis (EI) is a rare autosomal dominant genodermatosis. Rare cases of autosomal recessive inheritance have been reported. Approximately 1 in 200,000 to 1 in 300,000 individuals worldwide are recognized as affected [1]. Both genders are affected equally. EI is caused by heterozygous mutations in the genes encoding keratin 1 (KRT1) and keratin 10 (KRT10) [2]. KRT1 mutations are associated with severe palmoplantar keratoderma (PPK) but KRT10 mutations spare the palms and soles; this gene is not expressed in these locations [3]. EI presents at birth with erythroderma, erosions, peeling, with potential widespread areas of denuded skin. Over time the blistering and erythema decrease and severe hyperkeratosis become prominent, with characteristic furrowed hyperkeratosis accentuated in flexural areas. In the neonatal period sepsis and fluid/electrolyte imbalances can be life-threatening. It is a disfiguring disorder that has tremendous impact on patients' quality of life and social interactions.

Key histologic features include dense orthokeratotic hyperkeratosis, prominent acanthosis, hypergranulosis, and cytolysis of the suprabasal and granular layers leading to small intraepidermal blisters. Marked intracellular vacuolization of keratinocytes with dense clumps of keratin intermediate filament are seen [1].

Prenatal diagnosis can be performed when an underlying mutation has been identified in family members [4]. Work-up includes skin biopsy to differentiate between other vesiculo-bullous and erosive disorders in neonates, including various forms of epidermolysis bullosa and disorders such as staphylococcal scalded skin syndrome.

Treatment in the neonatal period includes management in an intensive care nursery to treat dehydration and electrolyte imbalance while providing protective isolation. In cases of sepsis, broad-spectrum antibiotics should be used. Protective padding and lubricants usually promote rapid healing of denuded skin. In children and adults, treatment focus is on reducing hyperkeratosis by using keratolytic creams, topical tretinoin, vitamin D preparations, and other emollients. Oral retinoids can drastically reduce hyperkeratosis as well as frequency of infections, but can increase epidermal fragility and blistering. Low initial doses with careful monitoring are advised [5].

Phenotypic variability is seen in affected individuals based on the genotypic mutation in EI. Previous cases and studies have described the phenotypic finding of PPK being associated with the genetic keratin 1 gene defect. Our case presents a mother and her newborn son with EI; prominent PPK was noted in the mother and was later evident in the infant at just a few months of age. The mother likely exhibited a de novo mutation. Genotype analysis of the neonate showed a mutation in the keratin 1 gene. The patient's mother was presumed to harbor the same mutation. This case presents an educational overview of a rare ichthyotic congenital disorder. This family demonstrates the phenotypic expression of PPK associated with keratin 1 gene mutations and illustrates the importance of genotype-phenotype correlation in this disorder. It also emphasizes the importance of taking a complete family history, which can assist in establishing the correct diagnosis.

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