Blistering Babies: A Case Report Of A Family With Epidermolysis Bullosa

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Topic: Clinical GeneticsFirst Author: Meenakshi Sigireddi, MD, MPH; Icahn School of Medicine at Mount Sinai, Department of Genetics & Genomic SciencesCo-Authors: B. D. Webb MD, Icahn School of Medicine at Mount Sinai, Department of Genetics & Genomic SciencesSession Type: PosterAbstract title: Blistering Babies: A case report of a family with Epidermolysis BullosaSkin erosions and blistering in the neonate are common and often multifactorial in etiology. Newborn skin is different in structure and function than adult skin, predisposing them to infectious, traumatic and autoimmune damage (Nischler et al.). Heritable disorders that present with increased skin fragility can also be seen in the neonatal period. We present a case of skin blistering in a male patient born at 33 weeks gestational age. The child was born to a couple of Brazilian descent, with no history of consanguinity. The couple has another child, a 10 year old who has suffered from skin blistering since birth, and was found to have epidermolysis bullosa. Dystrophic epidermolysis bullosa (DEB) is an inherited condition involving mechanical stress-induced blistering of the skin and mucous membranes. DEB is caused by biallelic or heterozygous pathogenic variants in the COL7A1 gene encoding type VII collagen, which is involved in dermal-epidermal adhesion (Shinkuma).On examination at day of life 5, our patient was found to have blistering on the dorsal surfaces of his bilateral upper and lower extremities in various stages of healing. The remainder of his physical exam was unremarkable. His prenatal history was notable for genetic testing via chorionic villi sampling, which was conducted in Brazil. The report suggested that he was a carrier of the c.5009G>A/p.Gly1670Asp (G1670D) variant (maternally inherited) in the COL7A1 gene. His brother's genetic testing revealed that he carried the aforementioned maternal variant, as well as an additional c.5132_5133ins5 variant from his father.Both parents have no history of skin blistering. As our patient was found to be a heterozygous carrier of the maternal variant on CVS testing, with a mother who was unaffected, we repeated genetic testing to confirm results, which revealed his prior testing was accurate, and that the newborn only carried the maternal variant. Our patient was discharged shortly thereafter with an uncomplicated NICU stay. On evaluation at 2 months of age, he was found to have no skin blistering or erosions, with no interim lesions since birth. We propose that rather than a mild presentation of autosomal dominant dystrophic epidermolysis bullosa, our patient may have suffered from a transient form of neonatal blistering (secondary to skin infection or neonatal pemphigus) as he has remained asymptomatic since initial evaluation.

Unaffected mother with variant 1 (chr3:48580624, NM_000094.3:c.5009G>A, p.Gly1670Asp (G1670D)), unaffected father with variant 2 (c.5132_5133ins5), affected older brother with both variants (compound heterozygous), and proband only has variant 1 and no variant 2 but with transient phenotype of DEB. Inheritance is not clear, could be AD or AR as described in OMIM (https://omim.org/entry/131705), or maybe parents were with similar transient phenotypes at neonatal stage but without detailed medical records.

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