Anales de Pediatría 99 (2023) 360–361

analesdepediatría

www.analesdepediatria.org

IMAGES IN PAEDIATRICS

Ichthyosis as a manifestation of fetal gaucher

Ictiosis como manifestación de enfermedad de gaucher fetal

Francisco A. Palazón-Rico[a,*], **Jose María Lloreda-García**[b]

[a] *Servicio de Pediatría, Hospital Universitario Santa Lucía, Cartagena (Murcia), Spain*
[b] *Unidad de Neonatología, Hospital Universitario Santa Lucía, Cartagena (Murcia), Spain*

Received 20 January 2023; accepted 22 March 2023
Available online 25 October 2023

We present the case of a male term neonate with no family history of interest, who had an uncomplicated birth and an Apgar score of 9–10. He was the second-born child of non-consanguineous parents. There was no previous history of miscarriage or abnormalities in prenatal care tests.

The salient findings of the physical examination were a dry, erythematous skin with desquamation and diffuse petechiae (Figs. 1 and 2) associated with hypotrophic appearance, jaundice and weak sucking.

The patient was admitted to the intensive care unit, with subsequent detection of coagulopathy, cholestasis and persistent thrombocytopenia despite multiple transfusions. The results of cultures and serological tests were negative. The peripheral blood smear examination revealed the presence of vacuolated lymphocytes. The postnatal radiograph and ultrasound examinations detected hepatosplenomegaly (Fig. 3).

Ichthyosis associated with a genetic disorder was suspected, and the differential diagnosis included foetal Gaucher disease, Chanarin-Dorfman syndrome and NISCH syndrome. The enzyme test for Gaucher disease ordered on day 8 post birth evinced the absence of glucocerebrosidase. The evaluation continued with an ichthyosis panel, adding molecular tests for Gaucher disease and NISCH syndrome.

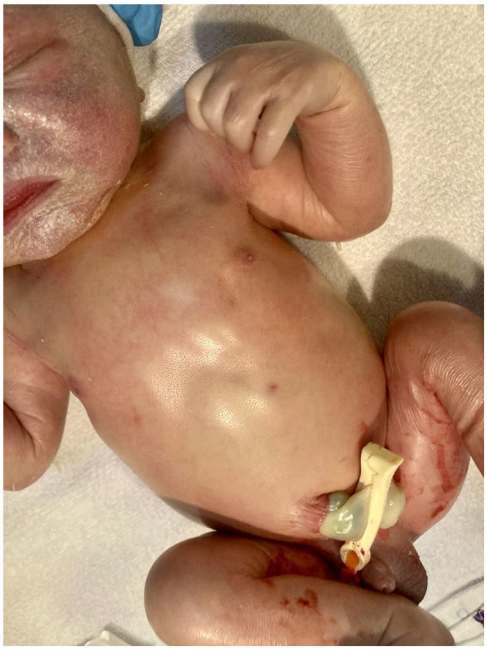

Figure 1 Appearance at birth.

This resulted in the detection of two heterozygous variants of the *GBA* gene (p.Pro430Leu and p.Leu483Pro, which were present in the parents). The patient died 2 months later during a respiratory infection.

DOI of original article: https://doi.org/10.1016/j.anpedi.2023.03.009
* Corresponding author.
E-mail address: palaxote@gmail.com (F.A. Palazón-Rico).

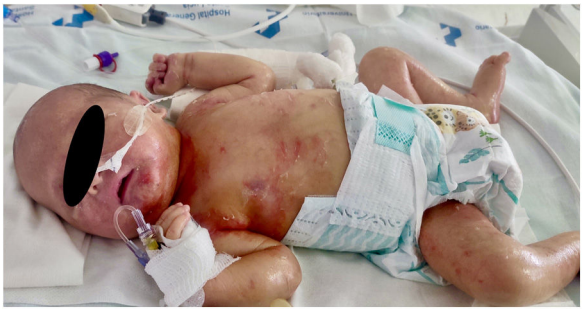

Figure 2 Day 6 post birth.

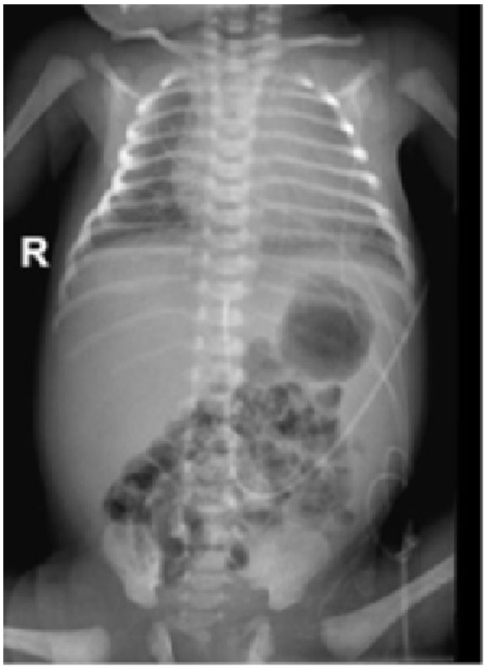

Figure 3 Hepatosplenomegaly.

Foetal Gaucher disease[1] has an incidence of less than 1 case per 10^6 live births and a prevalence of nearly zero on account of the lack of effective treatment, early mortality and forms of disease leading to miscarriage or hydrops fetalis. In the rest of cases, it is associated with hepatosplenomegaly and sustained cytopenia and may present as an ichthyosiform syndrome.[2]

It is caused by changes in the *GBA* gene (1q21), resulting in glucocerebrosidase deficiency. Forms associated with the p.Leu483Pro variant have a poorer prognosis.[3]

Awareness of the association between thrombocytopenia and ichthyosis is important in order to diagnose affected patients and provide genetic counselling to families.

References

1. Pastores GM, Hughes DA. Gaucher Disease. In: Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 2000. p. 1993–2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1269/
2. Schmuth M, Martinz V, Janecke AR, Fauth C, Schossig A, Zschocke J, et al. Inherited ichthyoses/generalized Mendelian disorders of cornification. Eur J Hum Genet. 2013;21:123–33, http://dx.doi.org/10.1038/ejhg.2012.121, published online 27 June 2012.
3. Stirnemann J, Belmatoug N, Camou F, Serratrice C, Froissart R, Caillaud C, et al. A review of gaucher disease pathophysiology, clinical presentation and treatments. Int J Mol Sci [Internet]. 2017;18(2):441, http://dx.doi.org/10.3390/ijms18020441. Available from:.