

Bilateral Ectropion and Eclabium: Clues for Diagnosis of Congenital Ichthyosis

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Full-term newborn, female, daughter of non-consanguineous parents, family history not significant, transferred to the neonatal intensive care unit on the first day of life due to diffuse desquamatory erythroderma, in flaps, accompanied by eversion of the eyelids (ectropion) and of the lips (eclabium) (Fig. 1). She was covered by a membrane of thick, rigid, smooth and bright skin (colloid type), with areas of fissuration and disperse erosions all over the tegument (Fig. 2). No other associated malformations were observed, nor alterations of the hair or nails. Presumptive diagnosis of congenital ichthyosis was made (from the Greek *ichthyo* – fish and *ōsis* – abnormal condition), collodion baby type presentation, and support measures were started (humid atmosphere with strict thermal control, hypercaloric nutrition, ocular oxybuprocaine, vaseline gauze as a dermal emollient and prophylaxis with fluconazole). At eight days old she began oral acitretin (retinoid) with a dose of 0.5 mg/kg/day, until she was two months old. The ulcerated lesions gradually improved, evolving into a typical pattern of lamellar ichthyosis. The Next Generation Sequencing (NGS) panel for associated genes revealed two mutations in heterozygosity, *c.1787_1794del* (*p.Pro596Hisfs*9*) and *c.406_408del* (*p.Glu136del*), in the gene *ALOX12B*. After two years of monitoring, she still applies emollients daily, with discreet xerosis and an excellent cosmetic result.

The paradigm of serious congenital ichthyoses changed in recent years with the early introduction of systemic retinoids, which increase survival rate and reduce the severity of the sequelae. However, simple measures like the use of soothing oils in the daily bath and the liberal use of emollient creams continue to be the basis of the treatment. The early recognition of this rare, but potentially fatal disease, allows the rapid introduction of support measures and the start of appropriate treatment, improving the prognosis of affected newborns.



Figure 1. Bilateral ectropion and eclabium at four days old.



Figure 2. Ulcerated / fissured lesions at two days old.

Keywords: Ectropion/congenital; Ichthyosis/congenital; Ichthyosis/therapy; Infant, Newborn; Lip/abnormalities

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WHAT THIS REPORT ADDS

- Eversion of the eyelids (ectropion) and of the lips (eclabium) present at birth is a very suggestive sign of congenital ichthyosis.
- The use of emollient oils in the daily bath and the liberal use of emollient creams continue to be the basis of the treatment of congenital ichthyoses.
- The early use of systemic retinoids, namely acitretin, improves the prognosis of the affected patients.

Conflicts of Interest

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Protection of human and animal subjects

The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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