Harlequin Ichthyosis: A case image from Syria

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Abstract

Harlequin ichthyosis is a rare autosomal recessive congenital ichthyosis with a distinct phenotypic appearance. We report a male baby with harlequin ichthyosis who was born to consanguineous parents that have a previous female baby who was diagnosed with harlequin ichthyosis.

CASE IMAGE

Harlequin Ichthyosis: A case image from Syria

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Abstract

Harlequin ichthyosis is a rare autosomal recessive congenital ichthyosis with a distinct phenotypic appearance. We report a male baby with harlequin ichthyosis who was born to consanguineous parents that have a previous female baby who was diagnosed with harlequin ichthyosis.

KEYWORDS

Harlequin ichthyosis, Harlequin fetus, autosomal dominant.

CONSENT FOR PUBLICATION

Written informed consent was obtained from the patient's parents to publish this report in accordance with the journal's patient consent policy.

1 | CASE PRESENTATION

A 2.5 kg male baby was born via cesarean section to a 25-year-old female, Gravida 5 Para 3, at 37 weeks of gestation.

The parents are maternal first cousins who have only one healthy female child, and a prior history of a female baby who was diagnosed with harlequin ichthyosis.

At the time of birth, the entire body of the baby was covered with armor-like scale plates with deep fissures that split the scales. The hands and feet were covered with thickened plate-like encasement. Other evident

features included severe bilateral ectropion, bilateral eclabium with a fixed-open mouth, flattening of the nose, rudimentary external ears, partial hair loss on the scalp and absence of eyelashes and eyebrows [Figure 1 & 2].

Adequate nutrition, broad-spectrum intravenous antibiotics, emollients, fluids and electrolyte were performed.

A dermatology consultation was done, with advice to add Acitretin to the treatment plan.

Anyway, the baby succumbed on the sixth-day of life without changing his treatment plan or receiving Acitretin.

To the best of our knowledge, this is the first case of harlequin ichthyosis to be reported in Syria.

[**FIGURE 1**]On day 1 of life, (A) the entire body is covered with armor-like scale plates with deep fissures, (B) bilateral ectropion, eclabium, flat nose.

[**FIGURE 2**]

On day 2 of life, (A) shedding of the thick plates and changing of their color to pale yellow, (B) partial hair loss on the scalp.

2 | DISCUSSION

Harlequin ichthyosis is a rare disorder which is inherited in an autosomal recessive manner. It is caused by mutations in the ABCA12 gene. The disease incidence is 1 in 300 000 births. The histological hallmarks include an extensive hyperkeratosis in the stratum corneum, and abnormal or absent lamellar bodies in the granular layer.[1]

Early administration of systemic retinoids, in particular acitretin, usually improves the clinical status. [2]

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CONFLICT OF INTEREST

The authors have no conflicts of interest to declare.

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ETHICS APPROVAL AND CONSENT

TO PARTICIPATE

Not applicable.

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