



Harlequin ichthyosis: The third babies with harlequin ichthyosis in a family

To the Editor,

The third baby with harlequin ichthyosis (HI) of a family who lost their two babies because of HI previously, but did not receive genetic counselling was presented to emphasize the importance of prenatal diagnosis and genetic counselling.

A female baby who was born by normal vaginal delivery with a birth weight of 2 600 g at the 34th gestational age as the third living baby from the fifth pregnancy of a 32-year old woman was admitted to the neonatal intensive care unit. No consanguineous marriage was reported in the familial history. It was learned that the patient had a healthy brother and a sister, but her two sisters were lost in the neonatal period because of HI. It was learned that these babies were born by normal vaginal delivery from the first and third pregnancies of the mother and they could live only for 18 and 3 days, respectively. The mother did not carry risk factors including hypertension and diabetes mellitus in this pregnancy and her previous pregnancies and did not have any pyretic disease. Although the family was recommended to receive genetic counselling, since they lost their two children because of HI previously, they did not receive genetic counselling and the mother did not attend follow-up visits during this last pregnancy. On physical examination of the patient, yellow-white thick plaques separated from each other with deep clefts which appeared like a shield wrapping the skin surface tightly were observed (Figure 1). Severe ectropion was observed in the eyes and eclabium was observed in the lips (Figure 2). The auricles were not developed, the external auditory canal could not be observed bilaterally, the ears and nose appeared hypoplastic, only the vibrissa could be observed. Fish mouth finding was present. The hands and feet were hypoplastic and necrotic towards the ends. There were contractures in the extremities (Figure 3). Complete blood count findings were as follows WBC: 17 500/mm³, hematocrit: 53% and platelets: 286 000/mm³. Biochemical tests were found to be normal. The patient whose respiration was normal was placed in a humidified incubator and intensive fluid treatment was started by venous catheter. The skin was humidified with frequent vaseline applications. The body temperature was monitored closely. Ampicillin and amikacin treatment was started after obtaining blood culture samples. Oral retinoid treatment was started at a dose of 1 mg/kg/day. The patient was fed orally by orogastric route from the first day. Despite intensive fluid treatment and enteral feeding in the follow-up, marked weight loss developed. The patient was lost on the 8th day because of sepsis despite appropriate antibiotic treatment and compliance with hygiene rules.

Harlequin ichthyosis is an autosomal recessive disease. Mutations in the ABCA12 gene have been found (1). It is a very rare but generally fatal keratinization disorder (2). The main problem has been reported to be abnormality in lamellar granules which have an important role in desquamation (3). Preterm delivery is generally present. Plaques which wrap the skin like a shield and deep clefts are observed in this disease. Ectropion is observed in both eyes (the eyelids are turned outwards). Eclabium (the lips are turned outwards), fish mouth appearance and flattened ears and nose are present. The ears and nose appear hypoplastic. The hair and nails are not developed. The hands and feet are hypoplastic and necrosis may develop in the ends of the extremities because of the shield-like structure of the skin. Range of motion is limited in the joints.

Patients with harlequin ichthyosis should be followed up in full-fledged intensive care units and with a multidisciplinary approach. The skin should be humidified very frequently to prevent fluid loss. Use of humidified incubators, compliance

Address for Correspondence: Çapan Konca, Department of Pediatrics, Adıyaman University Faculty of Medicine, Adıyaman, Turkey.
E-mail: dr.capan@hotmail.com

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Figure 1. General appearance of the patient



Figure 2. Marked eclabium and ectropion on the face of the patient

with hygiene rules, appropriate fluid treatment, providing heat balance and protection from infections are the main points in treatment. These patients are usually lost in the first days of life because of severe fluid loss, disruption of heat balance and sepsis (4). Vitamin A treatment gives good results in terms of skin findings, decreases hyperkeratosis and improves ectropion and eclabium (5). In survivors after the neonatal period, a very good neonatal intensive care and use of vitamin A have been reported to be efficient (6).

Currently, it has been reported that the mutations in the genes which cause to this disease can be found by molecular methods, families can be given genetic counselling about pregnancy planning or family planning and gene-based therapies will be possible in the near future (7).

Conclusively, prenatal diagnosis of HI syndrome which may mostly be fatal in the neonatal period is possible and genetic



Figure 3. Hypoplasia in hands and feet and necrotic appearance towards the ends

counselling should be absolutely given to families who have babies with HI.

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Mehmet Tekin, Çapan Konca, Zela Kahramaner, Aydın Erdemir
Department of Pediatrics, Adıyaman University Faculty of Medicine, Adıyaman, Turkey

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