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Case report **Open Access**

HARLEQUIN ICTHYOSIS: A RARE CASE REPORT

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ABSTRACT

Harlequin ichthyosis is the most severe form of congenital ichthyosis. It is a rare autosomal recessive disorder (1:300,000). The vast majority of affected individuals are due to mutation in the ABCA12 gene, which cause a deficiency of the epidermal lipid transporter, resulting in hyperkeratosis and abnormal barrier function of skin. Infants are very susceptible to metabolic abnormalities and infections. They usually do not survive for very long. We report here a case of a newborn with harlequin ichthyosis of consanguineous parentage who had a history of similar birth previously.

Key words: Harlequin ichthyosis, ABCA12 gene mutations, bad prognosis

INTRODUCTION

Ichthyosis is a heterogeneous family of skin disorders, harlequin ichthyosis being the most severe[1]. Due to excessive keratinization, at birth, the skin is very hard and thick, forming a dense “armor”-like scale all over the body. The skin forms large diamond shaped plates separated by deep fissures. The protective skin barrier is compromised and the infant is very susceptible to infections. These skin abnormalities also affect the shape of the eyelids, nose, mouth, and ears. A restriction of movement of the arms and legs is present. Breathing difficulties and respiratory failure may also occur [2,3]. The disorder has an ominous prognosis and affected babies usually die in first days to weeks of life [4].

CASE REPORT

After birth a female baby was admitted in our SNCU for severe asphyxia and skin deformity. The subject was born by vaginal delivery to a 26-year-old second gravid mother. There was history of consanguinity and their first baby was born with similar skin condition. The pregnancy was not monitored. The birth parameters were: weight 2570 g, length 45 cm, head circumference 34 cm, and chest circumference 31 cm. Physical examination revealed that the skin was thickened, hard, with yellowish and leathery white regions, split irregularly by deep erythematous fissures. The infant also had ectropion and chemosis, the nose and ears are flattened, lips are everted and gaping with fixed, open mouth and sparse hair. The limbs were in a semi-flexed position, with flexion contractures at elbows and knees and limited mobility. The dorsum of the hand and feet was oedematous. Finger and toes though formed there tip were gangrenous (Fig 1a & b). Baby died within 15 minutes due to asphyxia.

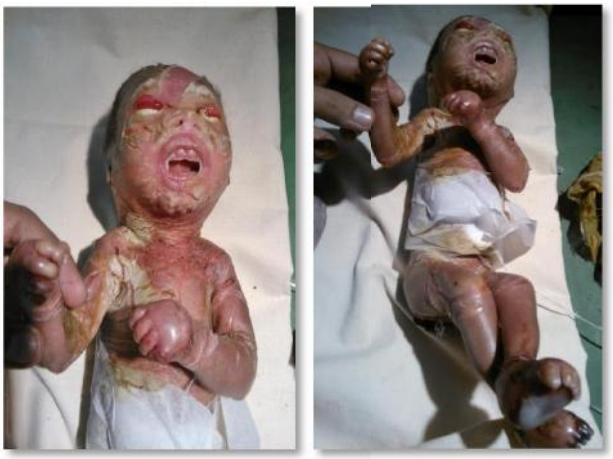

Fig 1a &b: showing characteristic skin thickening, scaling with erythematous fissure. There is also ectropion, eclabium, oedematous hands and feet, gangrene of fingers and toes tip.

DISCUSSION

Harlequin icthyosis is a rare hereditary disorder with an incidence of 1 in 300 000 births[4]. It is also called ‘ichthyosis congenita’ or ’keratosis diffusa foetalis’. The inheritance is autosomal recessive and there is no male to female preponderance [4,5]. Pathophysiology of this genetic defects lies on mutation of ABCA12 gene. These changes prevent the formation of lipid bilayers in the stratum corneum and result in hyperkeratosis and abnormal

barrier function which leads to characteristic clinical findings[6].

The condition can be diagnosed in-utero by fetal skin biopsy, anmiocentesis or chorionic villus biopsy (for early DNA-based diagnosis) and by 3 Dimensional Ultrasonography. Preimplantation genetic testing can also be done [7]. Postnatally, diagnosis is made by the pathognomonic appearance, and can be confirmed by skin biopsy from any cutaneous sites, which show abnormalities in the structure of lamellar granules and in the expression of epidermal keratin. Genetic testing for ABCA12 gene mutation is the most specific diagnostic test[2] .

Initial treatment of this condition includes high fluid intake to avoid dehydration from transepidermal water loss and use of a humidified heated incubator. Creams or ointments are used to keep the skin soft and hydrated. Keratolytic agents will promote peeling and thinning of the stratum corneum. Lubrication of the cornea in cases with ectropion prevents corneal drying. Nevertheless, infants given these therapies almost invariably succumb to their disease from sepsis, inability to feed, and inadequate ventilation[3]. Oral retinoids (1 mg/kg/day) therapy has changed the scenario. With its use quality of life being improved and survival well into childhood had been documented [8]. Infants receiving retinoids must be monitored for toxic effects. All survivors have had severe icthyosis as an outcome; some have intellectual impairement also. Genetic counseling for the families is mandatory [9]. Parents of affected children also need support.

CONCLUSION

In conclusion, this case has been reported for its rarity and to create awareness among pediatricians to identify and treat the condition.

Conflict of interest: Nil

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