“HARLEQUIN ICHTHYOSIS ” : A CASE REROR T

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Background

Harlequin ichthyosis (HI) is an extremely rare and most severe form of autosomal recessive congenital ichthyosis. Incidence of the disease is nearly 1 in 300,000 live births.

The disease might be lethal at birth and the affected babies are often premature. The condition is caused by mutation of the ABCA12 gene resulting in impaired lipid transport in the outermost layer of the skin, the epidermis.

Case Report

A 30-year-old primigravida admitted with preterm, premature rupture of membrane at her period of amenorrhoea of 29weeks.

The mother had received regular prenatal care. Polyhydramnios, absence of stomach bulb and soft tissue at temporal bone lateral angle of the eye was noted in the last ultrasound examination at 28 weeks of pregnancy.

Case Report

A female baby with Harlequin ichthyosis was born via normal spontaneous vaginal delivery at 29weeks and 4 days with the birth weight of 1.6kg. Apgar scores at the 1st and 5th minutes were measured as 6 and 8 respectively. Parents had a distant relation and had no family history of any inherited skin disorder.

Physical examination of baby revealed that the skin was thickened, hard, with yellowish and leathery white region, split irregularly by deep erythematous fissures.

The infant also had ectropion, rudimentary ears, nasal hypoplasia, eclabium and fixed, open mouth. Fingers and toes were hypoplastic. The limbs were in a semiflexed position, with flexion contractures at elbows and knees and limited mobility

Management

The baby was nursed in neonatal intensive care unit and placed in a humidified incubator with cardiorespiratory monitoring. Antibiotic therapy and conservative treatments were given. Baby died one day later.

Conclusion

There is no definitive treatment for this disease; supportive medical treatment may prolong the survival of these patients. Harlequin ichthyosis has been linked to mutation in the ABCA12 gene; therefore, geanetic counseling and mutation screening of this gene should be considered especially in families with a consanguinity marriage.

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