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Prenatal diagnosis and ultrasound features of Harlequin Ichthyosis

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Objective

A 30 year old lady in her first pregnancy was referred for flattened facial profile with suspected thickening of the fetal skin on prenatal ultrasound at 29 weeks of gestation. She previously enjoyed excellent health. Anomaly scan at 20 weeks was normal. Ultrasound at 30 weeks of gestation showed retracted fetal lips with exposure of gum and flattened nose. Harlequin Ichthyosis was suspected. The couple was counseled about the possible difficulty in setting intravenous line and skin condition. Follow up scan showed satisfactory growth, while flattened facial profile remained the same.

Methods

The patient underwent medical induction of labour for preterm rupture of membranes at 35 weeks of gestation. A baby boy weighing 2405 grams was delivered by Cesarean section for failed induction. The baby suffered from catheter related sepsis, eye infection and joint contractures in the neonatal period. Genetic work up confirmed the diagnosis of Harlequin Ichthyosis, with both parents found to be heterozygous carriers of ABCA12 gene mutation.

Results

Harlequin ichthyosis is one of the three major types of autosomal recessive congenital ichthyosis. The other two are lamellar ichthyosis and congenital ichthyosiform erythroderma. Preimplantation genetic diagnosis is possible following identification of ABCA12 gene. Features on prenatal ultrasound includes flat nose, short neck, thick lips, eclabium, hypertelorism, open eyes ectropion and fixed flexion of extremities. The foot length could be short because of restrictive dermopathy.

Conclusion

The use of systemic retinoids accelerates the shedding of hyperkeratoic plates and improves scaling. Together with enhanced moisturization, electrolyte monitoring in early neonatal period, sepsis prevention and physiotherapy, long term survival of 81% has been reported.