CASE REPORT

HARLEQUIN FOETUS - CONGENITAL ICHTHYOSIS - A CASE REPORT

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ABSTRACT: Congenital ichthyosis, also called harlequin foetus is a lethal autosomal recessive disorder resulting from a keratinizing disorder. Harlequin Ichthyosis is a very rare condition and the exact incidence is not known ⁽¹⁾. Here is a case report of Mrs. X G2P1D1 at 27 weeks of gestation whose previous baby had Harlequin Ichthyosis and died two months after birth, now the present pregnancy scan report showing features of Harlequin Ichthyosis. The disorder is fatal with neonates suffering from dehydration, infection (sepsis), restricted breathing due to plating. The disease has been known since 5th April 1750 and was first described in the diary of a cleric from Charleston, South Carolina.

KEYWORDS: Harlequin Ichthyosis, autosomal recessive, dysmorphic facies, skin biopsy.

INTRODUCTION: Ichthyosis is regarded as a disorder of keratinization or cornification and it is due to abnormal differentiation or metabolism. It is a very rare condition. There are many types of Ichthyosis and they are classified based on their appearance and their genetic cause ⁽²⁾. The term Harlequin Ichthyosis derives from newborns facial expression and the triangular and diamond shaped pattern of hyperkeratosis. The newborn mouth is pulled wide open, mimicking a clown's smile ⁽⁵⁾. The underlying genetic abnormality in Harlequin Ichthyosis has been identified as a mutation in the lipid-transporter gene ABCA12 on chromosome 2. The presence of homozygous mutation in affected individuals supports an autosomal recessive pattern of inheritance ⁽⁴⁾. Immuno histo cytochemical examination of the skin reveals characteristic abnormalities in the structure of lamellar granules and in the expression of epidermal keratin ⁽³⁾.

CASE REPORT: Mrs. X, 23 years old, G2P1D1 at 27 weeks with previous neonatal death at 2 months after birth now diagnosed to have Ichthyosis by ultrasound scan. Patient conceived after ovulation induction, took her folic acid tablets. Dating scan was done at 12 weeks; blood sugars evaluated and were within normal limits. She had a 2nd degree consanguineous marriage. Her 1st baby died 2 months after birth, Autopsy examination and skin biopsy showed Ichthyosis.

Examination: Her general condition was fair, she was afebrile, weight - 72 kgs, height - 156.5 cms, no pallor, no icterus, no pedal edema. Blood pressure – 100/70 mm of Hg. Urine albumin absent. Chest, cardiovascular system, CNS examination was within normal limits. On per abdomen examination she had a 28 week size pregnancy with transverse presentation, liquor was clinically adequate, foetal heart rate (FHR) was 146/min regular, uterus was relaxed.

Investigation: Routine biochemistry and thyroid function test were within normal limits. Dating scan – Single live intrauterine gestation corresponding to 12 weeks + 6 days, placenta anterior, normal liquor, NT - 2.3mm, Nasal Bone seen, Ductus venosus normal flow.

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Anomaly scan - Single live intrauterine gestation corresponding to 22 weeks, placenta anterior, liquor normal, estimated foetal weight – 540 gms, upper limb – complete opening and closing of hands not seen, lower limb – abnormal toes seen in feet, foot measured 3.7 cms (mean 3.9 cms).

Abnormal curvature of toes
Foot measured 3.7 cms(3.9 Cms)

Repeat scan @ 26 weeks - Abnormal facial profile - Dysmorphic facies, abnormal curvature of toes, complete opening of hands not seen, fetal mouth persistently open, features suggestive of Ichthyosis.

Abnormal curvature of foot @ 26
weeks

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Dysmorphic facies
foetal mouth persistantly open

DISCUSSION: Mrs. X showed anomaly scan with features of incomplete opening and closing of hands, abnormal toes seen in feet, abnormal curvature of toes and foot measuring 3.7 cms (mean 3.9 cms). In view of the previous history of Harlequin Ichthyosis and the present finding the possibility of evolving Ichthyosis was considered. Repeat scan was suggested after 4 weeks. In addition to the above features, the fetus showed dysmorphic facies and wide open mouth. Couple was informed about the fetal condition and the need for follow up monitoring. At 30 weeks of gestation, Mrs. X had preterm premature rupture of membrane and delivered a 1.3 kg boy baby. Baby cried immediately after birth, placenta weighed 390 gms. Physical assessment of the fetus at birth was done and revealed open mouth, dysmorphic facies, abnormal curvature of toes, incomplete opening and closing of hands, skin peeling.

Opened mouth
flattened nose

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Incomplete opening and closing of hands
Skin peeling noted

Abnormal curvature of toes

complete image of foetus with ichthyosis

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Baby died on day 1 of life due to dehydration and sepsis. Skin biopsy was taken to access the histologic characteristics of the cells. They revealed hyperkeratotic skin cells. Genetic testing was also done and revealed Harlequin Ichthyosis. Postnatal counselling was given to the couple. The chance of 25 – 50 % recurrence ⁽⁶⁾ of this autosomal recessive condition was also explained.

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