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Case Study

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RECURRENT FOETAL HARLEQUIN ICHTHYOSIS: A RARE CASE REPORT

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ABSTRACT

Harlequin ichthyosis is a rare and most severe form of congenital ichthyosis characterised by generalised thickening and splitting of the skin. It is a genetic disorder with autosomal recessive inheritance. Incidence of the disease is 1 in 3,00,000 live births and the foetuses presenting with this disorder have a very high mortality rate. It is most commonly seen in consanguineous marriages and is associated with ABCA 12 (adenosine triphosphate-binding cassette A12), gene mutation. Hence genetic screening, prenatal diagnostic testing and counselling of susceptible parents must be considered. I report a case of 35 year $G_2P_{1+0}(l_0)$ at 36 weeks of gestation with breech presentation and previous history of lower segment caesarean section (LSCS), who presented to the emergency in labor. It was an unbooked case. Emergency LSCS was done with extraction of a male child with birth weight 3kg with thick scales present all over the body with APGAR of 4/6 at 1 minute and 5 minutes respectively. Clinical diagnosis of harlequin ichthyosis was made and the baby was managed in neonatal ICU but died on 3rd day of life. This case report highlights the presentation of ichthyosis in a neonate, associated risks, complications and its management

KEYWORDS: *Harlequin ichthyosis, genetic disorder, skin thickening, consanguineous.*

INTRODUCTION

The word ichthyosis is derived from a greek word *ichthys*, meaning fish, referring to the cutaneous scaling that is characteristic of these disorder, which resembles the scales of fish. Ichthyosis represents a large group of cutaneous disorder characterised by abnormal cutaneous differentiation. Harlequin ichthyosis (HI) is one of the most severe form of congenital ichthyosiform dermatoses, and it primarily affects the skin.[1] In HI there is severe thickening and scaling of skin of entire body along with ectropion, hypoplastic nose and fingers, open mouth and joint contractures. Histologically, it is characterized by the existence of extracellular lipid material in the stratum corneal layer of the epidermis.[2] In these patents the barrier function of the skin is compromised leading to decreased ability to protect from bacterial, chemical and mechanical assault as well as to prevent from transepidermal water loss. The most common cause of death amongst the affected neonates is infections secondary to deep skin fissures and respiratory failure due to limited chest wall expansion because of contractures and thickened epidermis leading to impaired ventilation. HI is an autosomal recessive disorder triggered by mutation in ABCA 12 gene, which is responsible for skin development. Consanguinity (marriage amongst relatives) between parents has a role to play in its etiology.[3]

CASE REPORT

A 35 year $G_2P_{1+0}(L_0)$ at 36 weeks of gestation presented to the emergency of a tertiary care hospital in labor. She was married for the past 5 years with her husband being her maternal first cousin (consanguineous marriage). She had a history of emergency lower segment caesarean section (LSCS) done 2 years back for non progress of labor (NPOL) with extraction of a female child with birth weight 2.75 kg at a private hospital. The patient gave history that the child was born with some skin disease with abnormal skin texture and died one day after birth. She did not have any records of her previous pregnancy. She conceived spontaneously 1 year after her last child birth. It was an unbooked and unsupervised pregnancy and she reported to the hospital for the first time in labor. At admission her vitals were stable. On per abdominal examination height of uterus (HOU) was corresponding to the period of gestation (POG) with breech presentation and foetal heart sound was regular, 140/ minute. On per vaginal examination cervix was 4-5 cm dilated, 60% effaced with membranes intact and presenting part breech at 0 station. Decision for emergency LSCS was taken in view of previous LSCS with breech presentation. Intraoperatively there were no adhesions, lower uterine segment was well formed and bladder was not advanced. A live male child was extracted out with birth weight 3kg with APGAR 4/6 at 1 minute and 5 minutes respectively. The body of the neonate was covered with thick yellowish white scales with

erythematous fissures splitting the scales and extending deep into the dermis. The scales and fissures were more marked at the flexion points. Other features were ectropion, eclabium with wide open mouth and protruded tongue, nasal hypoplasia and rudimentary pinna. Eyebrows and eyelashes were absent but scalp hair were present and normal (figure1). All four limbs were rigid and semi flexed with underdeveloped toes and fingers (figure 2). The baby was evaluated by paediatrician and clinical diagnosis of harlequin ichthyosis was made. The baby was intubated at the time of birth and shifted to neonatal ICU. Thorough systemic examination was done which revealed no other abnormalities. There was no history of maternal drug exposure in antenatal period. The skin was cleaned with normal saline and topic emollients were applied. The baby was started prophylactically on intravenous antibiotics to prevent sepsis. Artificial tears were used to prevent drying of eyes. The baby was closely monitored, but despite all supportive management he died on 3rd day of life due to septicaemia and respiratory distress. The parents were counselled regarding the nature of inheritance of this disorder and were advised to undergo genetic screening and prenatal diagnostic testing prior to next conception.

Figure 1: Features of harlequin ichthyosis in a neonate.

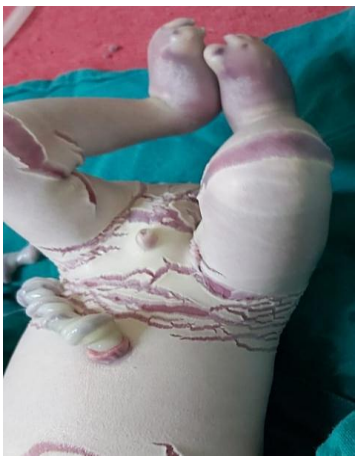

Figure 2: Rigid and semiflexed limbs with underdeveloped toes.

DISCUSSION

Harlequin ichthyosis is a rare form of genetic disorder due to mutation in ABCA 12 (adenosine triphosphate binding cassette transporter, subfamily A, member 12) gene on chromosome 2q33-q35 resulting in premature termination of protein translation. This gene is responsible for transporting lipids to cells that form the epidermis and the normal development of the skin.[4] At birth the neonate is covered with hyperkeratotic armour like yellowish white scales with deep red fissures extending to the dermis resembling joker like skin hence the name harlequin ichthyosis.[5] Infants with HI might have microcephaly, ectropion and eclabium. External auditory meatus and nostrils appears rudimentary and immature.[6] All these features were present in the present case report. Prenatal diagnosis is crucial for prevention and management of this disorder. Hence, family history, pedigree analysis, consanguinity between parents and history of similar disorder in family is important. In this case the parents had consanguineous marriage and had history of HI in previous child birth. Genetic screening to look for mutated gene in the parents with previous history of ichthyotic infant is beneficial and the parents may be counselled for assisted reproductive techniques and prenatal testing in future pregnancies for early detection of the disease. The same was advised to the parents in this case. Amniocentesis and an ultrasonography particularly of the foetal mouth at 17 weeks of gestation has shown to yield decisive results.[7] In the postnatal period the diagnosis can easily be made on clinical examination and gross appearance of the neonate though skin biopsy may reveal structural abnormalities of lamellar granules and epidermal keratin expression. Almost half of the infants born with this condition do not survive due to sepsis, respiratory failure, dehydration, water and electrolyte imbalance, hypothermia and renal failure.[8] Infants with compound heterozygous mutation survive more than those with the homozygous mutation. Moreover, advancement in the postnatal care and treatment has improved the prognosis of the disease.[9] Management of neonates with ichthyosis necessitates a multidisciplinary approach. Initial management involves humidified incubator to keep skin moist, use of petroleum based emollients, intravenous antibiotics to prevent sepsis and adequate hydration to maintain water and electrolyte imbalance. Oral retiniods has been proven to increase survival rate as it helps in shedding the hyperkeratotic encasement and causes a phenotypic switch. Acitretin, is the retinoid of choice because of its shorter half life and favourable side effect profile.[10] Until the deep fissures heal, sepsis is the leading cause of death in these patients. Supportive therapy is the mainstay of treatment, however, these neonates do not have a very good prognosis. Genetic counselling, genetic screening and prenatal diagnosis must be advised to susceptible parents which may help reduce the physical and mental impacts of the disorder on the parents.

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