



A case of harlequin fetus, ichthyosis congenita

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Objective

Harlequin type Congenital Ichthyosis or Harlequin Ichthyosis (HI) is a very rare congenital disorder with an autosomal recessive inheritance. This disease is the most severe representative of the genotype of abnormal keratinization due to the formation of beta keratin (a normal constituent of the skin of reptiles), instead of the normal alpha keratin. This is the most difficult, extreme and lethal form of ichthyosis. Most newborns die in utero and the surviving ones are usually delivered preterm with thickened, yellow - colored, armor - like skin, with fissures that divide the skin into polygonal sections. Malformations feature a thick hard outer armor up to 10mm in thickness, which breaks in utero, resulting in fissures to the chorion. The thick hard outer layer on the face prevents breathing and swallowing and the thick hard outer skin layer on the extremities leads to contractures and deformities of limbs, nose, ears and eversion of the lips. The incidence is estimated to be less than 1/1.000.000. Harlequin ichthyosis is an autosomal recessive condition. Heterozygotes (carriers) are asymptomatic. Death usually occurs due to dehydration, infection (sepsis) and restriction of breathing and the newborns rarely survive more than a few days. The condition of those who survive the first few days progresses to nonbullous ichthyosiform erythroderma. We report a new case of HI in a preterm male infant with normal prenatal screening results, born to non - related parents, who had two previous normal term - delivered children.

Methods

This is a case report.

Results

A 26 year old patient was referred due to preterm labor at 34 weeks of gestation, after preterm premature rupture of membranes. She had two previous normal pregnancies with normal vaginal births in term. This pregnancy was routinely controlled with normal prenatal screening results. Microbiological testing of vaginal samples was normal. A preterm male baby was born via normal spontaneous vaginal delivery. His birth weight was 2160 gr and the length was 42 cm. His Apgar score was 7/7. On examination the newborn showed typical appearance of congenital (Harlequin) Ichthyosis fetus. The skin was thickened and fissured, he had flattened rudimentary external ears bilaterally with obliterated ear canals, bilateral ectropion, bilateral eclabium and deformed hands appearing as claws. The newborn was transferred to the Intensive Care Unit. The patient's condition deteriorated, despite the supportive care, he developed severe cardio - respiratory failure and unfortunately died on the 4th day post delivery. An autopsy of the newborn was performed, which produced the following macroscopic findings, confirming the primary postnatal clinical diagnosis of Ichthyosis congenita (Harlequin type): "An autopsy of a 4th day old premature newborn male fetus, weighing 1600 grams and with a body length of 42 cm. On external inspection, presence of irregular diamond - shaped flakes of skin, each separated by fissures in different directions. Bilateral ear lobes are rudimentary, there is a complete obliteration of the left ear canal and an incomplete obliteration of the right, which is passable by 0.5 cm. Present bilateral ectropion and eclabium. Visible deformities on the hands that give the appearance of claws. The main findings confirm a rare congenital disease, whose main features are visible on external examination, with a high mortality rate due to limited respiratory movements with subsequent hypoventilation and respiratory failure as the immediate cause of death".

Conclusion

Harlequin - type Ichthyosis congenita is the most severe and lethal form of congenital ichthyosis. It is one of the three major types of congenital ichthyosis with autosomal recessive inheritance. The other two types are lamellar ichthyosis and non - bullous congenital ichthyosiform erythroderma. Detection of this disease is difficult with ultrasound, due to the rarity of this disease and its late gestational phenotypic expression. In patients, who have a family history of previously afflicted infants or in case of consanguinity, the diagnosis of HI should be based on amniocentesis, chorionic biopsy and fetal skin biopsies under fetoscopy. The most common prenatal ultrasound findings associated with HI are: a large open mouth (eclabium), absence of typical ear and nasal morphology, minimal fetal movements with an abnormal position of hands

and feet and ectropion. Other unspecific ultrasound signs include hyperechogenic amniotic fluid, intrauterine growth restriction, short extremities and polyhydramnios.

