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Unfollowed Pregnancy and Unexpected Result: A Harlequin Neonate

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Authors' contributions

This work was carried out in collaboration between all authors. Author IK was involved in the conception, design and data collection as well as preparation of the manuscript, revision of the article at various stages and preparation of the final draft. Author SO contributed in design and manuscript preparation. Author FFV contributed in approval of the final document. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Background: Prenatal consultation of a couple and medical follow-up of a pregnant woman is an essential part of health care while absence of these is associated with significant perinatal morbidity and mortality. Herein, a non-followed pregnancy resulted birth of a neonate with harlequin ichthyosis is presented. Harlequin fetus is rarely seen autosomal recessive disorder and is the most severe form of the congenital icthyosis, charachterized by thickened, hyperkeratotic skin with deep fissures over the entire of the body. However, diagnosis in first trimester is difficult, high risk patients for autosomal recessive disorders can be identified with prenatal consultation or diagnosis can be suspected with second or third trimester ultrasonography. Thus, an unfavourable outcome can be averted.

Keywords: Harlequin; ichthyosis; autosomal.

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1. INTRODUCTION

Harlequin icthyosis (HI) is a very rare, lethal disease with the estimated prevelance less than 1 in a 1.000.000 [1,2]. The mortality rate for HI is extremely high, however, survival for several months or years has been reported in very few cases [3]. The disease is characterized by thickened, hard and hyperkeratotic skin on the entire body. Gapping lips (fish mouth), ear and nose hypoplasia are specific appearances for HI. Inelasticity of the skin and hypoplastic digits cause flexion deformities of all joints [4]. In addition, pulmonary surfactant deficiency may result in breathing difficulties and respiratory failure [3].

Prenatal diagnosis is important in order to terminate pregnancy as early as possible. 2D or 3D ultrasound in first and second trimester can lead physician to suspect the diagnosis [5]. Prenatal diagnosis based on examination of fetal skin biopsy samples can be made in the second trimester [6].

Herein, a case of HI who was born to the unfollowed pregnant was presented.

2. CASE REPORT

A 19-year-old woman, gravida 1 was admitted at 34 weeks' gestation with frequent contractions. She and her 23-year-old husband had no significant medical or family history and there was no family relationship between them. She never had a visit to any hospital in her pregnancy. In our physical examination, breech presentation and 9 cm cervical dilatation and 80% effacement was detected. A female baby with HI, weighing 2130 gr with APGAR scores of 7, 8 at 1 and 5 minutes respectively was delivered by caesarean section. She had an extremely thick skin with deep fissures, hyperkeratinization of all body, ectropion, immature eyes and auricles, contractures of the fingers (Figs. 1 and 2). In the physical examination, no respiration problem was seen. The infant was followed up in neonatal intensive care unit. Tetracycline eye ointment was used. Intensive skin care by daily moisturising and greasing was undertaken. Despite intravenous fluid, partial parenteral nutrition and prophylactic antibiotics, respiratory insufficiency was begun on the 2nd day of follow-up and fetus died on the 3rd day.

An informed consent form for publication of the case was obtained from parents.

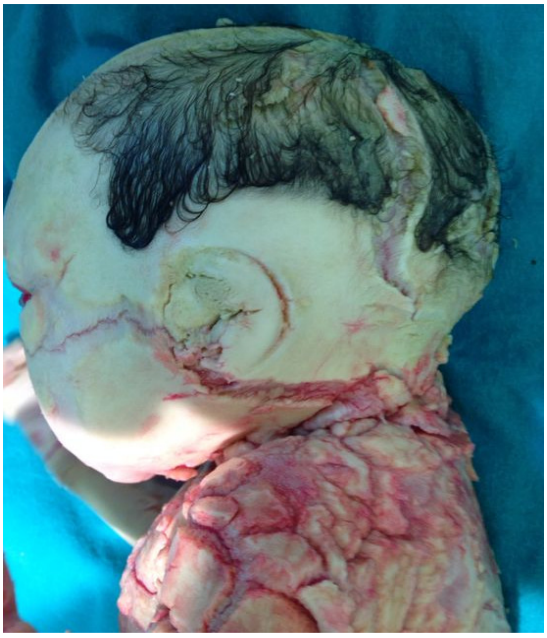

Fig. 1. Harlequin baby-immediately after birth

Fig. 2. Deep cracked skin, open wide mouth, abnormal eyes, and flatted nose and ear

3. DISCUSSION

HI is an extremely rare, inherited autosomal disorder [5]. Mutations in the ABCA12 gene have been reported for most of HI patients [7].

Because of high mortality, early detection of the disease is crucial. Early diagnosis enables patients to create a timely termination of pregnancy [8]. Therefore, prenatal diagnosis would be the first step. Detailed family history should be obtained. Consanguinity between the parents or the presence of other skin disorders would guide for early diagnosis. For these

couples, prenatal diagnosis is possible at the present time through direct sequence analysis and restriction enzyme digestion analysis using the fetal genomic DNA from amniotic fluid cells in second trimester [9]. In addition, two- or three-dimensional ultrasonography may show polyhidramnios, flexed extremities, short digits, open, immobile mouth and these characteristic findings should raise suspicion [10]. Sonographers should be aware of the signs observed at routine two-dimensional ultrasound examination in order to ensure appropriate referral for diagnosis.However, ultrasonography might not be applicable because of delayed phenotypic expression and the rarity of the disease [11]. Families with a history of HI should be tested for sequence analysis of ABCA12 or should undergo skin biopsy at 24 weeks of pregnancy [12,13].

In our case, although the family history was negative for the occurrence of any congenital skin disease in both the paternal and maternal line, the lack of prenatal consultations precluded to the possibility of discovering, through methodical ultrasound investigation, potential abnormalities indicative of congenital malformations. In this sense, the miss opportunity represented by the lack of systematic prenatal visits, has been instrumental in determining the unfortunate pregnancy outcome, with grave consequences for the baby and his family.

ETHICAL APPROVAL

Not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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