

CASE REPORT

Prenatal Diagnosis of Congenital Harlequin Ichthyosis with Two- and Three-Dimensional Ultrasound in the Third Trimester



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Harlequin ichthyosis (HI) is a rare congenital fetal skin keratinization disorder with an autosomal recessive inheritance. HI has specific sonographic features in the antenatal period. Several reports of prenatal sonographic diagnosis of HI have demonstrated its characteristic facial features that include a persistently open mouth and ectropion, with echogenic amniotic fluid and restriction of limb movements. Here, we report the case of a fetus in the 3rd trimester with these syndromic features identified by two- and three-dimensional sonography. Our ultrasonographic observation of membranes arising from the skin surface is particularly noteworthy. No prior case in which HI has been characterized by three-dimensional ultrasonography has been reported.

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Introduction

Harlequin ichthyosis (HI) is a rare and lethal fetal anomaly. At birth, a newborn infant with HI has thickened, yellow-colored, armor-like skin with fissures that divide the skin

into polygonal sections. Facial anomalies include ectropion (eversion of the eyelids), eclabium (eversion of the lips), and a large, round, and wide open mouth. The nose and ears are hypoplastic. The limbs are usually short and held in a fixed, flexed position [1].

The first case of prenatal diagnosis of HI was reported in 1983 [2]. As the patient had a family history of two previously affected infants, the diagnosis of HI was based on fetal skin biopsies under fetoscopy. However, the use of this traumatic method for the prenatal diagnosis of HI is limited to pregnant women with a previously affected

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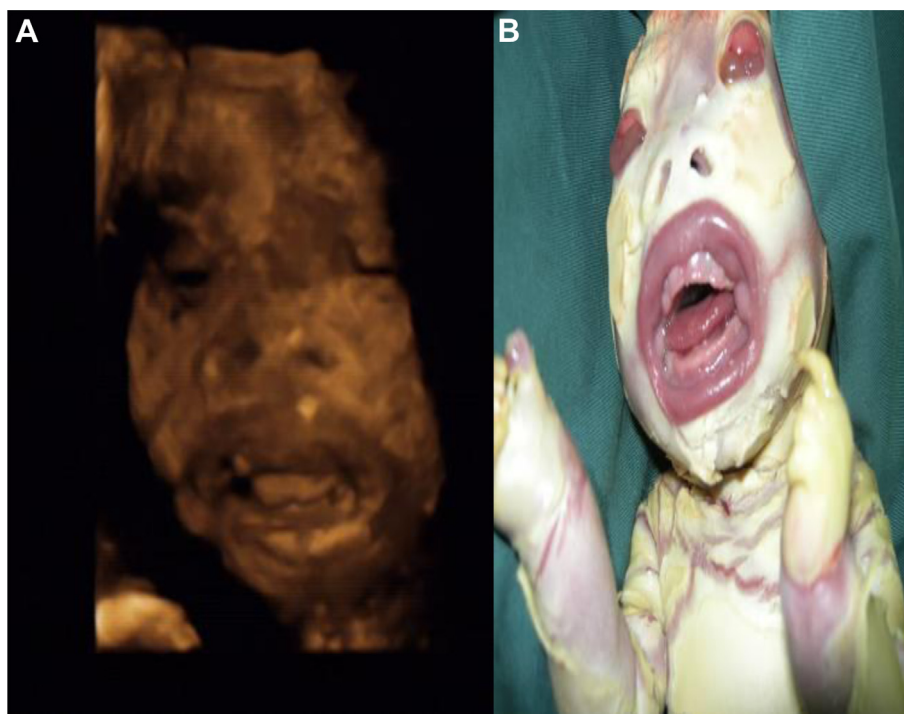


Fig. 1 Characteristic facial features of HI. (A) Three-dimensional image of the fetus with an open mouth, a protruding tongue, and open eyes and (B) photograph of the neonate upon delivery. HI = harlequin ichthyosis.

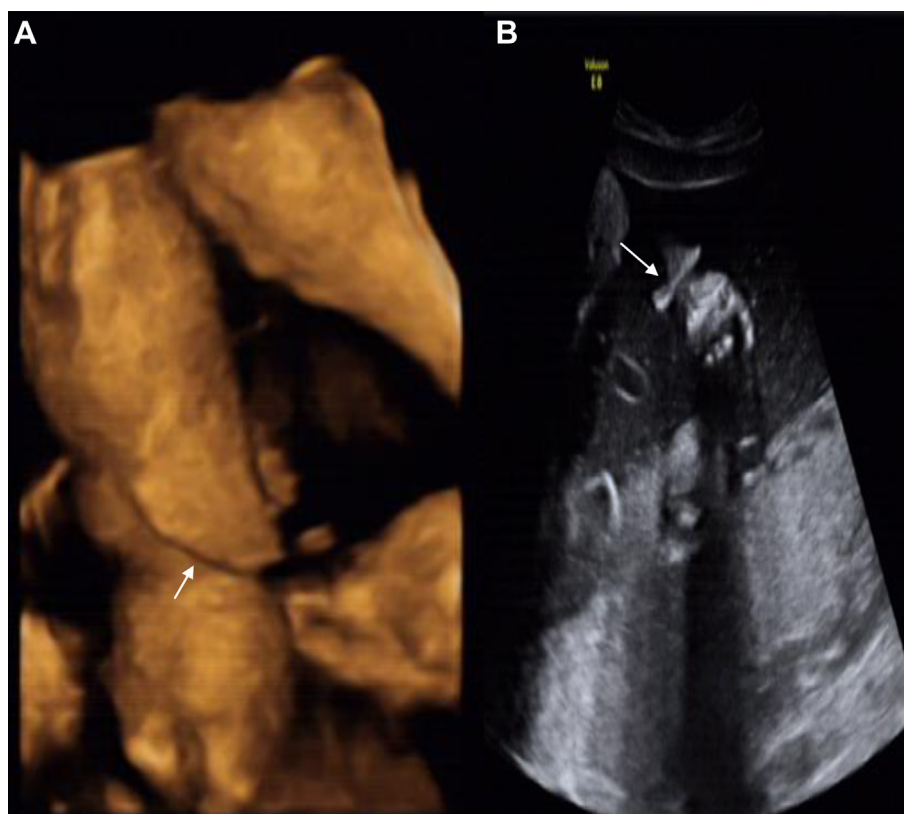


Fig. 2 Membrane arising from the skin surface of the fetus: (A) three- and (B) two-dimensional views. Membrane location is indicated by arrows.

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