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PICTURE OF THE MONTH

Prenatal Diagnosis of Low-set Ears with Asymmetrical Microtia in the First Trimester

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ABSTRACT

Initially, the external ears are in the lower neck region, but with the development of the mandible, they ascend to the side of the head at the level of the eyes. Low-set ear is one of the features often associated with genetic disorders, and external ear defects are significant because they are often associated with other malformations. The problem may occur as a symmetrical condition but an asymmetrical condition is not rare, in which one side of the face is maldeveloped. This defect varies in severity; however, it always includes maldevelopment of the ear and the mandible. Recent advanced 3D HDlive ultrasound enables us to demonstrate fetal external ear position and development even as early as in the first trimester. The picture of the month clearly demonstrated low-set ears with asymmetrical development of the ear and face at 12 weeks of gestation. Early detection of ear abnormality leads to further genetic and morphologic investigation as well as to proper management and counseling.

Keywords: Asymmetrical, Ear, Fetus, First trimester, Low-set, 3D ultrasound.

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LOW-SET EARS WITH ASYMMETRICAL MICROTIA AT 12 WEEKS

Low-set ear is one of the features often associated with genetic disorders such as Down, Patau, Edwards, Turner, Noonan, Cri du chat, DiGeorge, and other syndromes. External ear defects, or hypoplasia, is significant from the standpoint of the psychological and emotional trauma it may cause and for the fact that it is often associated with other malformations. All of the frequently occurring chromosomal syndromes and most of the less-common syndromes have ear anomalies as one of their characteristics.[1]

Ear development is based on pharyngeal arch development. Pharyngeal arches are paired structures associated with the pharynx that contribute to the formation of the face, jaw, ear, and neck. The 1st pharyngeal arch appears at about the beginning of the 4th week and others are added more caudally later. The external auditory meatus develops from the dorsal portion of the 1st pharyngeal cleft. The auricle develops at the dorsal ends of the 1st and 2nd pharyngeal arches, surrounding the first pharyngeal cleft. These swellings (auricular hillocks) form the definitive auricle. Initially, the external ears are in the lower neck region, but with development of the mandible, they ascend to the side of the head at the level of the eyes.[1]

As fusion of the auricular hillocks is complicated, developmental abnormalities of the auricle are common. Maldevelopment of the pharyngeal arch may occur on both sides as a symmetrical condition. However, an asymmetrical condition is not rare, in which one side of the face is maldeveloped. It is referred to as hemifacial microsomia, the 1st and 2nd pharyngeal arch syndrome oral–mandibular–auricular syndrome, lateral facial dysplasia, or otomandibular dysostosis. This defect varies in severity; however, it always includes maldevelopment of the ear and the mandible. This is the second most common congenital facial defect after cleft lip/palate.

Owing to recent advances in 3D ultrasound, early human development has been demonstrated accurately,[2-5] and 3D/4D sonography moved the prenatal diagnosis of fetal anomalies from the 2nd to the 1st trimester of pregnancy.[6]

It has been quite difficult to demonstrate the external ear position by a 2D ultrasound. Even by 3D ultrasound it is hard to depict the external ear of small fetuses in the first trimester. Prenatal 3D HDlive[7,8] with shadowing small structure, however, enables us to demonstrate the ear position and development even as early as in the first trimester. The external ear can be demonstrated from early

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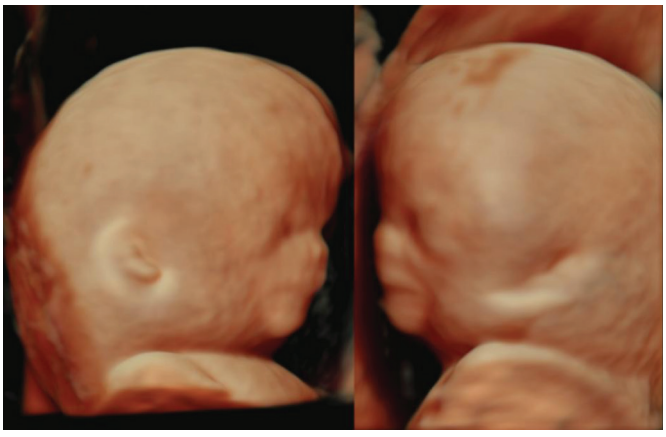

Fig. 1: Asymmetrical external ear development at 12 weeks of gestation. Both figures are from the same fetus of trisomy 21 at 12 weeks of gestation. Left figure shows the right low-set external hypoplastic ear, but in the opposite side of the profile (right figure), extremely hypoplastic external left ear is demonstrated. Facial structure appears to be asymmetrical, which is called hemifacial microsomia

gestation. The external ears rise from the embryonal neck level at 9 weeks of gestation to the level of the eye slant at 12 weeks according to fetal development. Figure 1 shows both right and left profiles of a fetus with trisomy 21 at 12 weeks of gestation. Both external ears are low set, but the development of the ear is quite different. Furthermore, facial appearance is also different and the left side of the pharyngeal arch appears to have been more affected than the right side. Thus, early prenatal ultrasound can depict facial development in detail. As mentioned above, ear abnormalities are often associated with other malformations; therefore, early detection of ear abnormality leads to further genetic and morphologic investigations as well as to proper management and counseling.

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