



CONGENITAL ABNORMALITIES BY ULTRASONOGRAPHY

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ABSTRACT

Aim: To evaluate congenital abnormalities (ear) with the help of ultrasonography.

Methods: Present study was performed at department of radiology, Gujarat Adani institute of medical science, Bhuj, Kutch, Gujarat. Study was performed Between July 2014 and August 2015 during which pregnant women with single fetus during 16–40 weeks of pregnancy were enrolled in the study. Fetal auricles and external auditory canal in the second trimester of pregnancy were evaluated by routine color Doppler ultrasound screening and systematic screening. Ultrasound images of fetal external ears were obtained on transverse-incline view at cervical vertebra level and mandible level and on parasagittal view and coronal view at external ear level.

Results: Five fetuses had anomalous ears including bilateral malformed auricles with malformed external auditory canal, unilateral deformed external ear, and unilateral microtia. The detection rate of both auricles was negatively correlated with gestational age. Of the 584 fetuses undergoing a routine ultrasound screening, 579 had bilateral auricles. Of the 495 fetuses following systematic screening, all fetuses had bilateral auricles. The best time for fetal auricles observation with ultrasonography is 20–24 weeks of pregnancy.

Conclusion: Detection of external ear abnormalities may assist in the diagnosis of chromosomal abnormalities.

KEYWORDS

Auricle, Ear abnormalities, Fetuses, Ultrasonography

Introduction

Studies have shown that an incidence of 1 : 6000 until 1 : 6830 newborns has been reported for external ear malformations^{1–3}. Ear abnormalities are important in the diagnosis of a variety of congenital malformations or syndromes in newborns^{4–6}. For example, reduced ear length is the most consistent phenotypic characteristic of neonates for diagnosis of trisomy 21⁷. Therefore, dedicated examinations on fetal external ear are imperative. Ultrasound is generally considered to be a reliable noninvasive method for monitoring and assessing fetal growth and well-being as well as for the early diagnosis of specific disorders associated with the pregnancy⁸. Ultrasonography is, moreover, the method of choice for the diagnosis of congenital abnormalities of the fetus⁹. Its diagnostic performance of routine ultrasound examinations performed in the 1980s and 1990s has been described in several publications. Because of improved ultrasound technology offering better resolution and improvement in knowledge and experience of ultrasound examiners, detection rates of fetal malformations may have increased since the 1990s¹⁰. External ear has a relatively complex structure and shape that is species specific and is remarkably constant in its basic normal shape¹¹. Numerous studies have reported the utility of the US in the evaluation of fetal anatomy and abnormalities. In particular, this modality has been helpful in evaluation of facial abnormalities, hand abnormalities, club feet, skeletal dysplasia, and spinal malformations^{12–15}. To the best of our knowledge, there are only a few reports on the diagnosis of external ear

abnormalities by ultrasonography which mainly focus on auricular deformities indicative of chromosomal abnormalities^{16–18}. To evaluate congenital abnormalities (ear) with the help of ultrasonography.

Materials and Methods

Present study was performed at department of radiology, Gujarat Adani institute of medical science, Bhuj, Kutch, Gujarat. Ethical clearance was taken from the institutional ethics board and informed consent was obtained from all the participants. Study was performed Between July 2014 and August 2015. The median maternal age was 31.3 years. Gestational age, calculated from the last menstrual period and confirmed by measuring the fetal crown-rump length, was 16–40 (mean 24.5±1.3) weeks. Inclusion criteria were as follows: (1) women who were scheduled for routine fetal ultrasound examination between weeks 16 and 24 of pregnancy; (2) women referred for determination of gestational age or growth discrepancy reassurance because of a preceding miscarriage, lack of fetal movement, inability to detect a fetal heartbeat, or other miscellaneous reasons. Scans were performed by sonographers and interpreted by fetal-imaging specialists with comparable experience. The content of routine color Doppler ultrasonography mainly included the measurement of fetal biological indicators in the second and third trimester and basic morphological examination with a focus on the morphostructure of vital organs and screening for lethal or severe abnormalities. In addition, data on any obvious fetal abnormalities were recorded and demographic details and the findings of the

ultrasound examinations were entered into a computer database at the time of scan. Systematic screening was carried out aiming at the high risk pregnancy and doubtful fetal abnormalities in the second trimester observed in the routine ultrasound examination. Based on the results of routine ultrasound screening and systematic screening for fetus, detailed examination of fetal auricular anatomy was carried out at 16–40 weeks of gestation. In order to observe the bilateral auricles and external auditory canal of fetuses, the scan images of fetal external ear in different sections included transverse-incline view at cervical vertebra level (section A) and mandible level (section B) and parasagittal view(sectionC) and coronal view (section D) at external ear level 19. In all cases the ear length measurement was performed and the measurement of ear length was obtained between two points, from the apical part of the helix to the caudal part of the earlobe.

Statistical analysis

The data was coded and entered into Microsoft Excel spreadsheet. Analysis was done using SPSS version 15 (SPSS Inc. Chicago, IL, USA) Windows software program. Descriptive statistics included computation of percentages. For all tests, confidence level and level of significance were set at 95% and 5% respectively. The sensitivity, specificity, positive predictive value, and negative predictive value were calculated with corresponding exact 95% binomial CIs. In addition, the prevalence of congenital malformations (the proportion of infants affected) was calculated 20.

Results

Ultrasound scanning of normal fetal auricles between 16 and 40 weeks' gestational age displayed clear and bright field C- or S-shaped images with hyperecho. Scanning images of parasagittal view and coronal view at external ear level (section C and D) had a higher detection rate which can be used as standard observation sections for fetal external ear. The auricular positions between two auricles can be judged by the scanning images of sections A and B, but the imaging results were more affected by changes of fetal position, placenta, and amniotic fluid. Therefore, these two sections can be used as supplementary for section C and section D. Total 584 underwent a routine ultrasound examination and 495 fetuses underwent systematic. Of the 584 fetuses undergoing a routine ultrasound screening from 16 weeks' gestational age to the birth, 579 between 16 and 40 weeks' gestational age had bilateral auricles. Of the 495 fetuses following systematic screening in the second trimester of pregnancy, all fetuses had bilateral auricles. The detection rate of bilateral ears was negatively correlated with gestational age ($r = -0.911, p < 0.01$) and was highest between 20 to 24 weeks of gestational age. Table 1 shows the distribution of the duration of pregnancy at which the ultrasound examinations were performed.

Five cases of anomalous ears were screened out in the examination and none of them were associated with deformities of other organs. Of these one had a small bilateral microtia with malformed external auditory canal and one had unilateral auricle with abnormal morphology. The remaining three cases had unilateral microtia.

Table 1: Link of ear abnormality-related syndrome with chromosomal abnormalities

Syndromes	Chromosomal abnormality
Down syndrome	Chromosome 21
Patau syndrome	Chromosome 13
Trisomy 9	Chromosome 9
Jacobsen syndrome	Chromosome 11

Discussion

The development of the fetal ear is complicated; developmental disorders of the ears are not uncommon 21. In the development course of fetal ear, changes of ear size, shape, and position may result in deformity 22, 23. The results of our study show that size and shape of auricles and external auditory canal of aberrant fetuses can be visualized on ultrasound images including transverse-incline view at cervical vertebra level and mandible level, on parasagittal view and coronal view at external ear level, which was consistent with the findings of the literatures above. However, we did not find the difference between ear positions.

It is well known that the second trimester is the best period of prenatal ultrasonic screening for fetal malformation 23–25.The correlation analysis of auricular detection rate and gestational week in our study shows that the detection rate of two ears was negatively correlated with gestational age and was highest in 20–24 weeks which can be considered as the best period to detect external ear abnormalities. All the cases of external ear abnormalities screened out in our study were achieved in this period. In this study, five fetuses had anomalous ears; of these one had bilateral short auricles with unclearly external auditory canal and one had unilateral external with abnormal shape. The remaining three cases had unilateral microtia.

In the systematic examination of fetal auricles by color Doppler ultrasonography, although detection rate of ears is affected by placental amniotic fluid, the most important factor influencing the detection rate is fetal position. In the second trimester, changes in fetal movements are relatively large and fetal position is unfixed and thus bilateral external ears cannot be detected at a time. In this case, aerobic exercise during pregnancy may help the fetal movements and multiple ultrasound examinations will help locate and obtain images of two external ears.

One of the aims of the present study was to evaluate the efficacy of prenatal ultrasound screening for congenital abnormality. In this report, the detection rate of bilateral ears in the second trimester is highest in routine obstetric ultrasonography which can be used as one of the common examination items in the prenatal ultrasonography.

Although fetal external ear abnormalities belong to minor physical anomalies, developmental anomalies of the external ear are still be found in some genetic diseases such as trisomy 21 and trisomy 18 26–29. Among the 5 cases of ear abnormalities, 3 cases were confirmed as trisomy 21 (Down syndrome) through amniocentesis. Another case had similar ear abnormality with his grandfather. We have conducted postnatal follow up studies to these participants. Those fetuses that had showed normal ears in the prenatal ultrasound screen did have normal ears after birth, with only one exception of absence of auditory canal. In those cases of fetuses where auricle of pinna were not shown in the prenatal ultrasound screen, there was one case of unilateral absence of auricle of pinna after birth, the gestational age of which was 36 + 1 w.

Therefore, ultrasound screening of external ear can be used as one of the indicators of prenatal diagnosis of fetal chromosomal abnormalities which might help in decreasing the birth defects, and ultrasound screening of the external ear should ideally bemade in the period between 20 and 24weeks of gestation.

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