

Application of Ultrasound in Prenatal Diagnosis of Birth Defects

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Abstract: At present, prenatal diagnosis is simple, effective, uninvasive and repeatable, so that it has been recognized as the preferred method for prenatal screening of fetal malformations. Antenatal ultrasound has been widely used in clinic, and has made a great contribution to the screening of fetal malformations. In recent years, due to the standardization of the ultrasound and the increasing of intrauterine diagnosis technology by ultrasound-doctors, the birth of fetuses with abnormalities of central nervous system is decreasing year by year, and also the detection rate of fetuses with cardiovascular defects become higher and higher. As the ultrasound is widely used in prenatal intrauterine diagnosis, the safety of ultrasonography is also widely concerned. In China, the standard operation of the safety threshold of ultrasonic inspection in prenatal screening and diagnostic ultrasonography has been started.

Keywords: Prenatal Disgnosis, Supersonic Inspection, Fetal Malformations

Birth defects are abnormalities existed in the recognizable form or function of human embryos or fetuses before birth. Severe birth defects can cause the death of fetuses, infants and children, and also can lead to illness and long-term disability in children. The natality of Chinese defective children ranges from 2% to 16%, and there are from 800,000 to 1 million defective children in every year. That bring huge mental pain and heavy financial burden to the families and society. Ultrasonography as the preferred method for prenatal screening of the malformations of fetus, to reduce perinatal mortality and morbidity rate of fetus, the rate of prenatal diagnosis of abnormal fetal must be improved.

1. Overview of ultrasonic development

In 1970s, scientists developed type B ultrasound which could distinguish the white bone and the black liquid,

and should also accurately measure various biological lines of fetus to assess the growth and development^[1]. In early 1990s, color doppler provides the gray-scale imaging of blood flow direction and speed of overlay, and also could assess placenta, umbilical blood flow and the heart of fetus. That should more accurately diagnose the fetal malformations^[2]. The three-dimensional ultrasound and four-dimensional ultrasound can clearly show the clearly morphological images of organs, such as the facial image of cleft lip and palatoschisis^[3]. At present, Obstetric ultrasonography has developed from the original two-dimensional black-and-white ultrasound, spectrum doppler ultrasound, color doppler ultrasound to the three-dimensional, four-dimensional ultrasound.

2. The standard ultrasonic examination method

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Published at: <http://www.ijsciences.com/pub/issue/2017-11/>
DOI: 10.18483/ijSci.1478; Online ISSN: 2305-3925; Print ISSN: 2410-4477



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The best time for first ultrasonic examination is mixed. It is widely believed that 20-24 weeks are most appropriate time, because in this period the quantity of amniotic fluid and size of fetus is moderate. The influence affected by the attenuation of bone-echo is small, and the image is clear. In this period most of the fetal abnormalities could be detected. If the time for ultrasonic examination is too early and the fetus is too small, even if the abnormalities has appeared, because the lesion is small, it is difficult to be recognized by the ultrasound. If the examination time is too late, the fetus is too large, and the amniotic fluid is relatively reduced, some malformation is difficult for detected because of the position and activity of fetuses and fetal bone echo attenuation. Strictly complying with the relevant requirements, we examine the pregnant women by ultrasonography. The subjects were either supine or lateral position. At least nine standard aspects were checked in the middle pregnancy-period including the transverse section of cerebellum, the coronal section of lip-surface, the longitudinal section of the spine, four cavity section of heart, the long-shaft section of outflow tract of left and right ventricular, the cross section of two kidneys, the initial segment colour flow of lateral umbilical artery, the section of lower limbs and upper limbs. The Order of inspection is from head, lip face, spine, thoracic cavity, belly, the beginning of the navel-artery, upper limbs to the lower limbs.

3. The analysis of primary malformations and relevance ratio in prenatal diagnosis

The top 5 birth defects include cardiovascular deformity, central nervous system deformity, lip/palate cleft, lip and palate cleft, urinary system deformity, digestive system deformity in China^[4-5]. In recent years, the birth defects of some systems of fetuses have been declined year by year due to the standardization of ultrasonography and the improvement of the diagnostic techniques of the intrauterine. The diagnosis rate of cardiovascular defects, especially complicated congenital heart defects or anomalies, such as tetralogy of fallot, cardiac dysplasia, and large vascular

transposition, has been significantly improved^[6].

At present, domestic and foreign literatures have reported the detection rate of fetal malformations by antenatal ultrasound. The detection rate of pre-prenatal ultrasound in anencephalus was above 87%. The detection rate of pre-prenatal ultrasound in fetuses with Severe encephalocele was 77%. The detection rate in fetuses with open spina bifida by pre-prenatal ultrasound was 61% ~ 95%. The detection rate in fetuses with severe thoracic abdominal wall defect and viscera bareness by pre-prenatal ultrasound was 60% ~ 86%. The detection rate in fetuses with cleft lip and palate by pre-prenatal ultrasound was 26.6% ~ 92.5%. The detection rate in fetuses with Single cleft palate by pre-prenatal ultrasound was 0 ~ 1.4%. The detection rate in fetuses with diaphragmatic hernia by pre-prenatal ultrasound was 60.0%. The detection rate in fetuses with atrial septal defect by prenatal ultrasound was 0 ~ 5.0%. The detection rate in fetuses with ventricular septal defect by pre-prenatal ultrasound was 0 ~ 66.0%. The detection rate in fetuses with hypoplastic left heart syndrome by prenatal ultrasound was 28.0% ~ 95.0%. The detection rate in fetuses with tetralogy of fallot by prenatal ultrasound was 14.0% ~ 65%. The detection rate in fetuses with double outlet of right ventricle by pre-prenatal ultrasound was 70.0%. The detection rate in fetuses with monoarterial trunk by prenatal ultrasound was 67.0%. The detection rate in fetuses with gastrointestinal tract anomalies by prenatal ultrasound was 9.2% ~ 57.1%.

But antenatal ultrasound isn't all-powerful. Ultrasonic diagnosis belongs to morphological category, therefore the fetus must first have obvious morphological structure that can be distinguished by ultrasonic image, so that possible to be diagnosed. If the fetuses don't have obvious morphological changes or have only functional abnormality, that is difficult to diagnose by ultrasonic examination. As an imaging instrument, ultrasound has the limitation and dependence of the instruments. Ultrasound physicians' personal

experience and expertise, as well as many factors be likely to affect the prenatal detection rate of malformation fetuses, such as the personal experience and expertise of ultrasound physician, the fetuses themselves and the mothers.

In 1993, American association of obstetricians and gynecologists emphasized that no matter which ultrasound device was used, not matter in which stage of pregnancy, even if the most experienced ultrasound specialists make the thorough examination of the fetus, it is unrealistic and unreasonable to expect all fetal malformations to be detected. The diagnosis of severe fetal malformation is not easy to misdiagnose, however the diagnosis of the smaller tissue deformities which do not have obvious anatomical changes is easy missed due the age of the fetus, the position of the fetus and the posture of the fetus.

The accurate intrauterine diagnosis of fetal defects of dactylion, polydactylism, esophageal atresia, anal atresia, external ear abnormalities, pure cleft palate, ventricular septal defect and so on, is difficult by ultrasound examination. The rate of missed diagnosis in these defects is high. The misdiagnosis reasons are as follows:

(1) dactylion, polydactylism: In this group of cases, the rate of missed diagnosis is highest. The main reason is the low display rate of fingers and toes in prenatal diagnosis by ultrasound. The routine examine of hands and feet do not include the counting and observation of quantity. In addition to patients who have family history or pregnant women's special requirements for targeted screening. The image of hands and feet of fetuses are under the influence of various factors, such as position of the fetuses, gestational weeks, amniotic fluid, the hand with fistling state. Therefore, the hands and feet of fetuses become the blind area of ultrasonic examination, that bring about the misdiagnosis of the deformity of fingers or toes.

(2) Esophageal atresia and anal atresia: the ultrasound does not show the closed esophagus and anus directly,

therefore the prenatal diagnosis of these gastrointestinal anomalies is inferential. We only can judge by indirect signs of gastrointestinal tract obstruction, such as the decrease and disappear of gastric vesicles, colectasia and so on. Therefore, the diagnosis is short of specificity.

(3) External ear abnormalities and pure cleft palate: These are the main misdiagnosed facial deformity. The reason of misdiagnosis due to low display rate by ultrasound. Due to the influence of the posterior acoustic shadow of the bony alveolar process, the soft palate is difficult to display on conventional cut surfaces. Specific fetal position and examination methods are required for display. Therefore, it is difficult to diagnose cleft palate of fetuses by prenatal ultrasound, especially the soft cleft palate. (4) Ventricular septal defect: with the improvement of prenatal diagnosis techniques, the combined application of the four cavity section of heart, the long-shaft section of outflow tract of left and right ventricular and three vessels section, enhance the detection rate in fetal heart malformations by prenatal ultrasound. But it is affected by fetal movement, fetal position and other factors, causing the heart image to exchange the up and down position or exchange the left and right positions, right and left positions, that increased the difficulty in identifying fetal heart structures which leads to missed diagnosis^[7].

Beyond that, if the inspection is not careful or incomplete, that will result in missed diagnosis. If the inspection time is not chosen properly, that should also lead to missed diagnosis. In prenatal examination of fetal abnormalities, It is extremely important to select an appropriate time for ultrasonic examination. Normally, partial deformities can be detected before 12 weeks of gestation. The development of the middle and late period fetuses tends to improve, and the morphological features are remarkable. If the amniotic fluid is sufficient, we can find deformities more easily. In ultrasound screening of foetal birth defects, early detection and early

diagnosis of fetuses with birth defects, and early termination of pregnancy, that It is great significance to reduce the death rate of perinatal infants and to aristogenesis.

4.The reminder value of ultrasound for chromosomal diseases

Some common chromosomal diseases are often accompany with severe fetal morphological abnormalities. According to report, the fetus of 18 trisomic syndrome at least involves two or more abnormalities. Other chromosomal diseases, such as 13 trisomic syndrome and 5 trisomic syndrome,manifest multiple organ malformations and functional disorder.The application of ultrasound in prenatal detection of fetal congenital defects is of great importance in indicating fetal chromosomal diseases, reventing and reducing the birth of severe congenital defect^[8].

5.The safety analysis of fetal defect by ultrasound

In 1923 , for the first time radioactive rays was applied radiography for fetal imaging of pregnant women^[9], later,we found that radiation exposure can causesevere deformity,such asfetal microcephaly, fetal growth retardation, leucocythemia and malignant tumor so on^[10-11].International Society forUltrasound in Obstetrics and Gynecology (ISUOG) declared again^[12]:In fetal ultrasonography, it should be kept in mind that the intensity of ultrasonic should not be too strong, and the examination time should not be too long. And it should be limited to be used in pregnant women with medical indication, but should not be used for recreational purposes.In 1970 s ~19 80 s ,ScienceNews reported study of ultrasonography in the safety of prenatal fetal^[13].The damage of ultrasound to the embryo is shown that cell apoptosis can be caused when the output of the instrument is too loud and the inspection time is too long (more than 60 rain).

In China, the standard operation in safety threshold by ultrasonic check have been initiated, and the following principles are required: 1)The early, middle and late

inspections are advisable.2)In early pregnancy,we test embryo location, survive or not. Ultrasonic strength for normal pregnancy is within 10 mW/cm². Probe movement scan method must be no morethan 5 min. The fixed method should not exceed 2 min. We should use color Doppler Spectrum, doppler and other large ultrasonic diagnostic apparatus prudently.3)In mid-pregnancy and late-pregnancy: we should mainly observe fetal development,fetal position and fetal malformation.The inspection is required no more than 3 minutes per organ.

6.Outlook

Although antenatal ultrasound has been widely used in clinical practice, as a method of imaging examination, ultrasound is still dependent and limited.At present, Ultrasonic examinationcouldcan guide clinical treatment and termination of pregnancy in time, and the development direction should be focused on the unified examination plan and screening guideinthe future.With the developement of technology and the studying of more advanced hardware facilities, ultrasonography in the early-to-mid of pregnancy should reduce birth defectsfurther and improve the quality of newborn population.

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