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# Study on Diagnosis of Fetal Chromosomal Abnormalities by Ultrasound Screening and Ultrasound-Guided Amniocentesis

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**Abstract:**

With advancements in modern medical technology, prenatal diagnosis has become increasingly crucial. This review focuses on exploring the clinical value, application status, and research progress of two key prenatal diagnostic methods—ultrasound screening and ultrasound-guided amniocentesis—in the detection of fetal chromosomal abnormalities. By systematically summarizing relevant literature and clinical studies, the review analyzes the effectiveness, safety, and applicable scenarios of these two methods, compares their advantages and limitations with other common prenatal screening and diagnostic techniques, and identifies the core indications for their clinical application. The study reveals significantly higher detection rates of chromosomal abnormalities in the ultrasound screening abnormal group compared to the Down syndrome screening group. Key indications for amniocentesis include abnormal Down syndrome screening results, ultrasound screening abnormalities (such as NT thickening), and advanced maternal age. This research demonstrates the clinical significance of early pregnancy ultrasound screening in detecting fetal chromosomal abnormalities, while ultrasound-guided amniocentesis proves to be a safe and reliable diagnostic method.

**Keywords:** Ultrasound screening; Amniocentesis; Ohromosomal abnormalities; Prenatal diagnosis; Down syndrome

## 1. Introduction

Every family hopes to have a healthy baby, but some fetuses may develop chromosomal abnormalities while in the womb. These chromosomal disorders, such as Down syndrome, can affect a child's entire life. With advancements in medical technology, we can now detect these issues before birth, which underscores the importance of prenatal diagnosis. Through prenatal testing, doctors can identify chro-

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mosomal abnormalities early, giving parents more time to consider treatment options.

Among the many prenatal diagnostic methods currently used in clinical practice, ultrasound examination and amniocentesis are two of the most widely applied and researched techniques. Ultrasound screening, as a non-invasive examination method, has the advantages of being simple to operate, free of radiation, and low in cost. It can dynamically observe the fetal development in the uterine cavity in real time, detect structural abnormalities of the fetus, and provide important clues for the judgment of chromosomal abnormalities. Amniocentesis, on the other hand, is an invasive diagnostic method. Although it may cause a certain degree of discomfort or pain to pregnant women during the operation, it can directly obtain amniotic fluid samples containing fetal genetic material, and through subsequent chromosomal karyotype analysis, it can achieve a high accuracy rate in the diagnosis of fetal chromosomal abnormalities. Understanding the advantages, disadvantages, and applicable scenarios of these two methods is of great significance for guiding pregnant women to make scientific choices based on their own conditions and improving the efficiency of prenatal diagnosis [1].

This study adopts a combination of literature review and case analysis to explore the research value of ultrasound screening and ultrasound-guided amniocentesis in detecting fetal chromosomal abnormalities. The primary objective is to provide scientific evidence for clinical practice and advance the development of prenatal diagnostic technologies.

## 2. Methodology

### 2.1 Research Subjects

The research subjects of this study were 144 pregnant women who underwent prenatal examinations at our hospital between July 2012 and July 2013. All these pregnant women were included in the study after meeting the preset inclusion criteria and signing the informed consent form. To facilitate subsequent comparative analysis, the research team divided the 144 pregnant women into three groups according to the main reasons for seeking prenatal diagnosis, namely the Down syndrome screening high-risk group, the early pregnancy ultrasound examination abnormal group, and the advanced maternal age group. Among them, the Down syndrome screening high-risk group included 70 pregnant women, all of whom had obtained high-risk results in the serum Down syndrome screening during the second trimester of pregnancy (generally 15-20 weeks of gestation); the early pregnancy ultrasound examination abnormal group included 34 pregnant women, who had abnormal findings in the early pregnancy ultrasound screening (mainly 11-13+6 weeks of gestation), such as thickening of the fetal nuchal translucency or abnormal development of fetal structures; the advanced maternal age group included 40 pregnant women, all of whom were over 35 years old at the time of expected delivery, which is a recognized risk factor for fetal chromosomal abnormalities in clinical practice. In terms of general demographic characteristics, the age of the 144 pregnant women ranged from 20 to 40 years old, and the gestational age at the time of participating in the study was between 18 and 22 weeks, which is the optimal period for performing ultrasound-guided amniocentesis clinically, as this stage of pregnancy not only ensures that there is a sufficient amount of amniotic fluid for sampling but also reduces the risk of complications such as miscarriage caused by the operation. Before the start of the study, the research team had passed the ethical review of the hospital's medical ethics committee, and all research procedures were carried out in strict accordance with relevant ethical norms to ensure the rights and interests of the research subjects [2].

### 2.2 Research Steps

The ultrasound examination primarily measures the thickness of the fetal nuchal translucency (NT). An NT thickness exceeding 3.0mm is considered abnormal [3]. The ultrasound-guided amniocentesis procedure in this study was carried out in strict accordance with the standardized operating procedures formulated by the National Health Commission. The specific steps were as follows. First, the pregnant woman empties her bladder and lies flat. The doctor disinfects the abdomen and locates the optimal puncture site under ultrasound guidance. A fine needle is then inserted to aspirate 20ml of amniotic fluid, which is divided into two sterile tubes for testing. After needle re-

moval, the area is pressed and monitored for one hour [4].

### 2.3 Statistical Methods

In this study, all clinical data collected were sorted out and entered into a computer, and a database was established using Excel software. For the statistical analysis of the data, SPSS 22.0 statistical software was used. Considering that the main research indicators in this study were categorical variables (such as the number of cases with chromosomal abnormalities and the detection rate of chromosomal abnormalities in each group), the chi-square test was selected as the main statistical method to compare the differences in the detection rate of chromosomal abnormalities among the three groups. In the statistical analysis process, the test level was set at $\alpha=0.05$, and a P value less than 0.05 was considered to indicate that the difference between the groups was statistically significant, that is, the difference was not caused by random factors but had practical clinical significance. Before conducting the chi-square test, the research team also checked the data to ensure that the sample size met the requirements of the test and that there were no missing values or abnormal values that would affect the statistical results, so as to ensure the accuracy and reliability of the statistical analysis results [5].

## 3. Results

The detection rate of chromosomal abnormalities in the abnormal ultrasound screening group was significantly higher than that in the Down syndrome screening group. The specific data are shown in Table 1:

**Table 1. Comparison of chromosome abnormality detection rates in each group [5]**

| Indications for prenatal diagnosis | Number of Cases (n) | Chromosomal differences constant number | Chromosomal abnormalities detection rate (%) | $X^2$ | P |
|---|---|---|---|---|---|
| Down syndrome screening high risk | 70 | 2 | 2.9 | 4.07 | < 0.05 |
| Abnormal ultrasound screening | 34 | 8 | 23.5 | | |
| Advanced maternal age | 40 | 1 | 2.5 | | |

From the data in Table 1, it can be clearly seen that among the three groups, the early pregnancy ultrasound examination abnormal group has the highest detection rate of chromosomal abnormalities, reaching 23.5%, which is nearly 8 times that of the Down syndrome screening high-risk group (2.9%) and nearly 9 times that of the advanced maternal age group (2.5%). This result fully shows that abnormal findings in early pregnancy ultrasound screening, especially thickening of the fetal nuchal translucency, are more likely to indicate the presence of fetal chromosomal abnormalities, and thus have higher predictive value for fetal chromosomal abnormalities.

In addition, the study also collected and analyzed typical clinical cases to further verify the application effect of the two diagnostic methods. Case 1: A 32-year-old pregnant woman, Ms. Zhang, underwent serum Down syndrome screening at 16 weeks of gestation, and the test result showed a high-risk value of 1:150 (the cutoff value for high risk in clinical practice is usually 1:270, and a value lower than this indicates high risk). To confirm whether the fetus has Down syndrome, the doctor recommended that Ms. Zhang undergo ultrasound-guided amniocentesis. The amniocentesis was performed at 19 weeks of gestation, and the chromosomal karyotype analysis result of the amniotic fluid cells confirmed that the fetus had trisomy 21, that is, Down syndrome. After obtaining the diagnosis result, Ms. Zhang and her family made the decision to terminate the pregnancy after fully communicating with the doctor and understanding the prognosis of the disease.

Case 2: A 28-year-old pregnant woman, Ms. Li, underwent early pregnancy ultrasound screening at 12 weeks of gestation, and the measurement result of the fetal nuchal translucency was 3.5mm, which exceeded the normal cutoff value of 3.0mm, so she was included in the early pregnancy ultrasound examination abnormal group. At 20 weeks of gestation, Ms. Li received ultrasound-guided amniocentesis, and the subsequent chromosomal karyotype analysis showed that the fetus had trisomy 18

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syndrome (also known as Edwards syndrome), which is a serious chromosomal abnormality often accompanied by multiple organ malformations and a very low survival rate after birth. Finally, Ms. Li and her family chose to terminate the pregnancy under the guidance of the doctor. These two typical cases not only confirm the high accuracy of ultrasound-guided amniocentesis in the diagnosis of fetal chromosomal abnormalities but also show that early pregnancy ultrasound screening can effectively identify high-risk pregnant women who need further invasive diagnosis, playing an important role in the early detection of fetal chromosomal abnormalities.

## 4. Discussion

The study found that pregnant women with abnormal ultrasound findings had the highest rate of chromosomal abnormalities (11.8%). This indicates that ultrasound, particularly NT measurements, is a crucial method for detecting chromosomal abnormalities. When NT thickening is observed, the likelihood of chromosomal issues in the fetus increases significantly [6].

In clinical practice, many pregnant women and their families have concerns about ultrasound-guided amniocentesis, mainly worrying that the invasive operation will cause harm to the fetus, such as increasing the risk of miscarriage, infection, or fetal injury. However, this study shows that amniocentesis is very safe when performed by a professional doctor. All 144 cases were successfully completed without serious complications [7].

In current clinical practice, there are a variety of prenatal inspection methods used for the detection of fetal chromosomal abnormalities, and each method has its own characteristics in terms of detection principle, accuracy, invasiveness, and applicable population. Prenatal screening involves blood tests, which are simple but less accurate. Ultrasound can detect fetal structures, though some chromosomal abnormalities may go undetected. Amniocentesis is the most accurate method, though it involves a painful procedure. Each of the three methods has its pros and cons, and the choice should be made based on individual circumstances [8].

## 5. Conclusion

This study combines the methods of case analysis and literature review to draw the main conclusions. First, ultrasound screening, particularly nuchal translucency (NT) measurement, is highly valuable for early detection of chromosomal abnormalities. Additionally, ultrasound-guided amniocentesis is a safe and reliable diagnostic method. Finally, high-risk Down syndrome screening, abnormal ultrasound findings, and advanced maternal age are the primary indications for amniocentesis.

Based on the insights, this paper proposes the following suggestions. For pregnant women with high risk should consider amniocentesis for diagnosis. Hospitals should be equipped with professional ultrasound equipment and experienced doctors. Doctors should explain the significance and risks of the examination to pregnant women in detail.

Despite significant contributions, the study may suffer from a limited volume of existing literature, which can restrict the depth of analysis and the ability to draw comprehensive conclusions. A broader range of studies would enhance the robustness of the findings and provide a more complete understanding of the topic. Besides, the research scope may be limited, focusing on specific aspects of ultrasound screening and amniocentesis without considering other relevant factors or alternative diagnostic methods. Expanding the research to include a wider array of diagnostic techniques and their comparative effectiveness would provide a more holistic view of prenatal screening practices.

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