

Impact of Active Implementation of Prenatal Screening on Preventing Congenital Disabilities: A Study of 10,695 Pregnant Women

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Abstract: *Objective:* To explore the impact of active implementation of prenatal screening on preventing congenital disabilities. *Methods:* A total of 10,695 pregnant women at Chengyang District Maternal and Child Health and Family Planning Service Center underwent prenatal screening between January 2021 to December 2023. These women were divided into two groups: 18 to 30 years old and 31 to 45 years old. High-risk rates of Down syndrome (DS), Edwards syndrome (ES), and neural tube defects (NTD) were compared between the two groups of pregnant women. *Results:* Among the 10,695 pregnant women who received prenatal screening, 1,588 were identified as high-risk. Pregnant women aged 31 to 45 years old showed higher high-risk rates of DS, ES, and NTD compared to those aged 18 to 30 years old ($P < 0.05$). The high-risk group demonstrated elevated rates of DS, ES, and NTD diagnoses ($P < 0.05$). Additionally, the high-risk group's evaluation of spontaneous abortion rates exhibited higher levels, although not significantly different from the low-risk group ($P > 0.05$). Moreover, the high-risk group's evaluation of congenital disabilities in pregnant women showed a significantly higher rate ($P < 0.05$). *Conclusion:* Prenatal screening enables the early detection of potential fetal problems, facilitating the implementation of corresponding measures to reduce the incidence of congenital disabilities.

Keywords: Prenatal screening; Prevention of congenital disabilities; Clinical effect

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1. Introduction

Preventing congenital disabilities is crucial for ensuring the health and survival of babies while reducing the burden on families and society. Prenatal screening encompasses various aspects during pregnancy check-ups, requiring pregnant women to undergo numerous screening procedures during routine prenatal visits^[1].

Firstly, nuchal translucency (NT) thickness must be measured using color ultrasound technology between 11 and 14 weeks of pregnancy. An NT thickness value exceeding 2.5 mm warrants heightened vigilance for Down syndrome (DS) and the risk of congenital heart disease. Screening for Down syndrome typically occurs between 15 and 21 weeks of pregnancy. This involves assessing the levels of relevant markers in pregnant women's blood to determine the fetus's DS risk, or utilizing ultrasound technology to evaluate fetal growth,

development, and anatomy ^[2].

Additionally, screening for diabetes after 24 weeks of pregnancy often involves an oral glucose tolerance test (OGTT). This test evaluates a pregnant woman's glucose metabolism to identify diabetes or gestational diabetes. Early detection of gestational diabetes through OGTT enables appropriate disease management, medication, and prevention of macrosomia, thereby safeguarding the health of both mothers and children.

The utilization of ultrasound and other technologies in prenatal screening facilitates the timely detection of congenital disabilities. This enables doctors to identify fetal abnormalities promptly and initiate necessary interventions early, thereby preventing the onset of congenital disabilities ^[3]. Furthermore, it allows pregnant women and their families to prepare mentally and take proactive measures to ensure the health of their newborns. This article aims to analyze the role of prenatal screening in preventing congenital disabilities such as DS, Edwards syndrome (ES), and neural tube defects (NTD).

2. Materials and methods

2.1. General information

A total of 10,695 pregnant women at Chongyang District Maternal and Child Health and Family Planning Service Center underwent prenatal screening. The research spanned from January 2021 to December 2023, during which participants were categorized based on different age groups. The 18–30-year-old group comprised 7,068 cases, with an average age of 27.96 ± 6.55 years and gestational age ranging from 16 to 21⁺⁵ weeks, with an average of 19.03 ± 4.82 weeks. The 31–45-year-old group included 3,627 cases, with an average age of 27.98 ± 6.61 years and gestational age ranging from 16–22⁺⁴ weeks, with an average of 19.12 ± 4.77 weeks. No statistically significant differences were observed between the two groups ($P > 0.05$).

2.2. Methods

2.2.1. Prenatal screening

During prenatal screening, 3 mL of venous blood was extracted from pregnant women, and the serum was separated and stored below -20°C . Alpha-fetoprotein (AFP) and free β -human chorionic gonadotropin (β -hCG) levels in the blood were detected. Pregnant women underwent DS screening, non-invasive DNA prenatal screening, and pregnancy ultrasound screening, with relevant precautions communicated to them before the examination, such as drinking water on an empty stomach to enhance ultrasound image clarity.

2.2.2. Prenatal diagnosis methods for pregnant women at high risks of DS, ES, and NTD

Prenatal diagnosis was conducted on pregnant women identified as high risk for DS after obtaining informed consent. Amniocentesis (a method of extracting amniotic fluid for testing by puncturing the abdominal wall, uterus, and amniotic membrane, and can accurately detect chromosomal abnormalities in the fetus, including DS), chorionic villus biopsy (a method of removing and testing a small sample of villous tissue from the placenta for testing to detect placental abnormalities and chromosomal abnormalities and can also be used to screen for DS), non-invasive prenatal genetic testing (NIPT; a method that uses cell-free fetal DNA in the blood of pregnant women to detect chromosomal abnormalities such as DS at an early stage), and dual marker test (combined assessment with maternal serum DS screening and ultrasound examination results to more accurately assess the risk of DS) were employed. For those at high risk of ES, 20 mL of amniotic fluid extraction (the gold standard for prenatal screening) under ultrasound guidance was performed at 17 to 24 weeks of pregnancy for cell culture and karyotype analysis. Those identified as high risk of NTD underwent fetal system ultrasound diagnosis at 18 to 24 weeks. Upon confirmation of congenital disabilities, pregnant women were promptly

informed of the consequences, advised on labor induction, and provided with appropriate follow-up care.

2.3. Observation indicators

High-risk rates of DS, ES, and NTD were statistically analyzed among pregnant women in both age groups using DS screening software. High DS risk was determined by a comprehensive risk coefficient $\geq 1/270$; high ES risk by a comprehensive risk coefficient $\geq 1/350$; and high NTD risk by an AFP detection value ≥ 2.5 MOM^[4].

2.4. Statistical analysis

Data analysis was conducted using SPSS 27.0 software. Measurement data were expressed as mean \pm standard deviation (SD), and results were obtained through *t*-tests. Count data were presented as [*n* (%)], and results were obtained through χ^2 tests. A significance level of $P < 0.05$ indicated statistical significance.

3. Results

3.1. Comparison of the high-risk rates of DS, ES, and NTD among pregnant women between the two groups

Among 10,695 pregnant women who received prenatal screening, there were 1,588 cases found to be high-risk. The 31–45-year-old group indicated a higher rate of high congenital disease risk as compared to the 18–30-year-old group ($P < 0.05$), as shown in **Table 1**.

Table 1. Comparison of the high-risk rates of DS, ES, and NTD among pregnant women between the two groups [*n* (%)]

Group	<i>n</i>	DS high-risk rate	ES high-risk rate	NTD high-risk rate
18–30 years old	7,068	541 (7.65)	195 (2.76)	208 (2.94)
31–45 years old	3,627	350 (9.65)	150 (4.14)	144 (3.97)
χ^2	-	12.499	14.554	7.949
<i>P</i>	-	0.000	0.000	0.005

3.2. Analysis and comparison of diagnosis of three diseases among different groups of people

Table 2 shows that the high-risk group had a higher level of DS, ES, and NTD diagnosis rates ($P < 0.05$).

Table 2. Analysis and comparison of diagnosis of three diseases by different groups of people

Group	Number of examples	Number of confirmed cases	Diagnosis rate (%)
DS high risk	891	12	1.35
ES high risk	345	6	1.74
NTD high risk	352	2	0.57
Total	1,588	20	1.26

3.3. Comparison of spontaneous abortion rates and congenital disability rates among pregnant women with different risks

The high-risk group displayed a higher spontaneous abortion rate, but insignificant compared to the low-risk group ($P > 0.05$). However, the high-risk group had a higher congenital disability rate compared to the low-risk

group ($P < 0.05$). See **Table 3**.

Table 3. Comparison of spontaneous abortion rates and congenital disability rates among pregnant women with different risks [n (%)]

Group	Number of examples	Spontaneous abortion	Congenital disabilities
High risk	1,588	28 (1.76)	10 (0.63)
Low risk	9,107	0 (0.00)	18 (0.19)
χ^2	-	160.998	9.668
P	-	6.847	0.002

4. Discussion

Congenital disabilities encompass structural or functional abnormalities occurring during embryonic development, resulting in organ, systemic, or tissue anomalies at birth. These defects may stem from genetic factors, external environmental influences, or their interaction [5]. Congenital disabilities impose significant psychological, economic, and social burdens on children and families. Severe congenital disabilities may compromise a patient's quality of life, necessitating long-term or lifelong treatment. Timely prenatal screening and diagnosis are pivotal in preventing congenital disabilities.

DS arises from the presence of an additional part or a complete pair of chromosome 21, leading to individuals carrying three chromosome 21s instead of the typical two. This extra chromosome can impede both physical and mental development, with varying degrees of cognitive impairment. Prenatal screening and diagnosis play a crucial role in DS prevention [6]. High-risk pregnancies may undergo amniocentesis for further diagnostic assessment.

ES is a rare chromosomal abnormality involving an extra chromosome 18, resulting in diverse physical and intellectual developmental challenges. Children with ES may experience respiratory difficulties, skeletal deformities, kidney issues, and other complications, significantly affecting their quality of life and longevity. Early detection of ES through prenatal screening and diagnosis, particularly via amniocentesis, enables families to make informed decisions [7].

NTD represents a prevalent form of nervous system development impairment, posing significant threats to patients' health and quality of life. Physical manifestations may include spinal cord and spinal deformities, alongside neurological complications such as mental retardation and cognitive impairments. Prenatal screening methods, including ultrasound examination and amniocentesis, facilitate early NTD detection, allowing for timely treatment and intervention to reduce the incidence of congenital disabilities [8].

Presently, congenital disability prevention primarily hinges on prenatal screening. Combined with effective treatment interventions, prenatal screening can mitigate the risk of fetal congenital disabilities and reduce their incidence. However, inadequate awareness and incomplete screening procedures among pregnant women undermine congenital disability prevention efforts. Hence, it is imperative to enhance prenatal screening, raise pregnant women's examination awareness, leverage advanced medical examination technologies for fetal observation, and provide appropriate interventions for congenital disabilities. This study revealed that among 10,695 pregnant women who underwent prenatal screening, 1,588 were identified as high-risk pregnancies. High-risk rates of DS, ES, and NTD were higher among pregnant women aged 31 to 45 compared to those aged 18 to 30 ($P < 0.05$). This underscores the significant decline in physical health with advancing maternal age, contributing to fetal development restrictions and congenital disabilities, impacting both pregnant women

and their families. Prenatal screening and effective intervention play crucial roles in preventing congenital disabilities in children and reducing their incidence^[9,10]. This study further demonstrated that within the high-risk group, evaluation of DS, ES, and NTD diagnosis rates, as well as spontaneous abortion and congenital disability rates yielded higher results. This suggests a higher proportion of spontaneous abortion and congenital disabilities among high-risk groups compared to low-risk ones. Therefore, it is imperative to provide comprehensive education to high-risk pregnant women, informing them of the adverse consequences and the significance of prenatal screening and diagnosis in preventing congenital disabilities.

Prenatal screening, a non-invasive examination method, assesses fetal chromosomes, genes, biochemical markers, and other indicators in pregnant women to detect hereditary or congenital diseases. It provides families with crucial information and risk assessments, enabling informed decisions regarding further diagnostic testing, elective abortion, or childbirth preparation. While prenatal screening cannot entirely prevent congenital disabilities, it can reduce the risk of specific conditions by enabling medical teams to provide targeted interventions and management to high-risk groups. Offering various screening items, prenatal screening enhances the early detection rate of common congenital disabilities and genetic diseases, including DS, congenital heart disease, and NTD. This empowers pregnant women with more information and choices, facilitating informed decision-making during pregnancy. Prenatal screening is particularly vital for high-risk pregnant women, including older mothers and those with a family genetic history, as it mitigates disease risks and enhances newborn health. It is essential to acknowledge that while prenatal screening aids in preventing congenital disabilities to a certain extent, it cannot entirely eliminate them. Its primary role lies in providing information and risk assessments to aid families in decision-making. Moreover, prenatal screening must be evaluated comprehensively with clinical manifestations and other examination results. Thus, pregnant women should fully grasp the significance, limitations, and risks of the screening program and make rational, informed decisions.

In conclusion, prenatal screening effectively assists pregnant women in understanding their fetuses' health status, detecting potential issues early, and implementing corresponding measures to reduce the incidence of congenital disabilities.

Disclosure statement

The authors declare no conflict of interest.

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