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Analysis of the Effect of Maternal Serologic Prenatal Screening in Mid-Trimester Pregnancy

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Abstract: *Objective:* To analyze the results of maternal serological prenatal screening in the middle trimester. *Methods:* The study was conducted on 7815 middle-pregnant women who underwent prenatal screening in our hospital between January 2021 and December 2022, of which pregnant women aged 35 years and above were taken as the high-age group; those who were under 35 years old were included in the low-age group. The results of maternal serological screening in mid-pregnancy were analyzed. *Results:* The probability of detecting the disease was higher in prenatal screening results. *Conclusion:* Maternal serologic prenatal screening in the middle trimester has the advantages of economy and convenience, and can effectively reduce birth defects in newborns, which is of some value for promotion.

Keywords: Mid-trimester pregnancy; Serology; Prenatal screening

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1. Introduction

Birth defects are a major cause of infant mortality. It refers to the abnormalities of body structure, metabolic conditions, and organ functions that appear in the process of fetal development. Some examples of birth defects of body structure are congenital heart disease, cleft lip and palate, and limb abnormalities. Birth defects of metabolic conditions and organ functions can cause deafness, mental retardation, and other diseases. Therefore, birth defects not only severely impact their growth and development, but also impose a great burden on their families and society. In addressing the significant impact of birth defects, the state promotes prenatal screening. This involves testing various indicators in pregnant women to identify high-risk groups for fetal abnormal development. Through modern medical diagnostics, efforts are made to terminate undesirable pregnancies as early as possible, aiming to reduce the incidence of birth defects. Birth defects are a condition that profoundly threatens human health. However, prenatal diagnostic methods such as cord blood, amniotic fluid, and chorionic villus are invasive and cannot be used as routine diagnostic methods [1,2]. Prenatal screening serologic testing is an economical and convenient method that does not cause any harm to the patient. It plays a crucial role in the early detection of hereditary diseases and congenital defects, thereby reducing the risk of birth defects. In China, the most commonly conducted prenatal screening tests include those for Down syndrome, neural tube

deformity (ODS), and trisomy 18-trimester. These conditions have high incidence rates and pose significant threats to the health of both mothers and infants, with no definitive therapy available. In this study, we analyzed the results of serological screening conducted on 7815 women during mid-trimester pregnancy at our hospital.

2. Data and methods

2.1. Clinical data

We chose 7815 women in their mid-trimester pregnancy who underwent prenatal screening in our hospital from January 2021 to December 2022 as study subjects. Their ages ranged from 21 to 46 years, with a mean age of 30.52±1.23 years. Among them, pregnant women aged 35 years and older were classified into the high-age group, while those under 35 years were categorized into the low-age group. Pregnant women aged 35 and older were considered at high risk for prenatal screening due to their advanced age. In cases where there was a high risk of Down syndrome, cytogenetic testing using amniotic fluid was performed to detect any chromosomal abnormalities. High-risk pregnancies underwent ultrasound imaging to identify fetal structural malformations.

2.2. Methods

2.2.1. Collection of maternal information

The basic information about the mother was collected by the attending obstetrician. The study subjects have all signed an informed consent for prenatal screening. The collected information included the name of the pregnant woman, birth year and month, weight, number of fetuses, gestational week and judgment criteria, telephone number, sampling date, informed consent signature of the pregnant woman, and signatures of the doctor and blood collection nurse.

2.2.2. Specimen collection

On the day of blood collection, 5 ml of fasting venous blood was taken and stored at 22℃ for 30 minutes. The blood sample was then centrifuged at 3000r/min for 7 minutes. Subsequently, the serum was separated and stored in the refrigerator at -20℃ for future use.

2.2.3. Detection method

Time-resolved fluorescence immunoassay was conducted using the corresponding reagents and the standardized procedures were followed rigorously to ensure quality during screening. The likelihood of Down syndrome, neural tube malformation, and 18-trisomy syndrome was assessed, with different intermediate values selected for various gestational weeks. For Down syndrome, a risk cutoff of 1:270 was used, above which it was considered high risk; for 18-trisomy, the cutoff was 1:350, also considered high risk. For open neural tube defects, an alpha-fetoprotein (AFP) multiple of the median (MoM) = 2.5 was used as the risk cutoff. In cases of high-risk pregnancies for Down syndrome, cytogenetic testing could be performed on amniotic fluid to detect any chromosomal abnormalities. Additionally, high-risk pregnancies could undergo ultrasound imaging to rule out fetal structural malformations.

2.3. Statistical processing

Statistical software SPSS20.0 was used to process and analyze the data. The measurement data was expressed as the number of cases and percentage (%) and analyzed with a χ^2 test. The count data were expressed as mean ± standard deviation and analyzed using a *t*-test. $P < 0.05$ indicated statistical significance.

3. Results

The results of the screening are shown in **Tables 1 and 2**.

Table 1. Screening results in 2021

Groups	Number of cases	Number of NIPT inspections	Number of amniocentesis	Normal live births	Child with birth defects	Other adverse pregnancy outcomes
High risk	333	139	102	322	0	11
Threshold risk due to high age	254	138	47	250	0	4
High age-related risk	383	-	-	378	1	4
Threshold risk	464	-	-	455	1	8
Low risk	2863	-	-	2823	0	40

Table 2. Screening results in 2022

Groups	Number of cases	Number of NIPT inspections	Number of amniocentesis	Normal live births	Child with birth defects	Other adverse pregnancy outcomes
High risk	314	187	103	303	0	11
Threshold risk due to high age	265	190	53	262	0	3
High age-related risk	373	-	-	369	0	4
Threshold risk	407	-	-	405	0	2
Low risk	2578	-	-	2555	2	21

4. Discussion

With the implementation of the “two children” and “three children” national policies, China’s birth rate has been increasing year by year. About 5% of newborns have congenital defects, and the proportion has been increasing due to various reasons. This situation not only places significant pressure on families, affecting their livelihoods, finances, and mental well-being but also poses challenges to China’s economic and social development. To enhance the overall quality of China’s population and reduce the occurrence of birth defects, the government has enacted laws such as the Population and Family Planning Law and the Maternal and Child Health Law. These laws provide a framework for preventing and controlling birth defects and have led to the development of relevant management measures and strategies. In this context, the national health authorities have continuously strengthened their efforts in preventing and treating birth defects. With the rapid advancement of modern medicine, prenatal screening technology has become increasingly important in detecting chromosomal abnormalities and fetal anomalies. This technology has greatly reduced the uncertainty associated with pregnancy tests, enabling early intervention for newborns [5,6]. During pregnancy, women undergo screening tests for Down syndrome, trisomy 18, and open neural tube defects. The triple screening involves assessing various factors: decreased serum levels of alpha-fetoprotein and free estriol, along with increased levels of free β-human chorionic gonadotropin, indicate Down syndrome; elevated serum alpha-fetoprotein levels higher than 2.5 MoM suggest neural tube defects; and decreased levels of alpha-fetoprotein, free estriol, and free β-human chorionic gonadotropin are characteristic of trisomy 18 [7,8]. Alpha-fetoprotein is

a glycoprotein, mainly derived from vitellogenin and fetal liver. It is a glycoprotein that enters the blood and amniotic fluid of pregnant women. Its level rises linearly at 14–20 weeks of pregnancy and decreases slowly thereafter. Neural tube malformation refers to an abnormality or lesion of the chorionic villus, which leads to an increase in its permeability, resulting in the entry of fetal plasma and cerebrospinal fluid into the amniotic fluid, which leads to an elevation of alpha-fetoprotein in the serum of the pregnant woman; and 18-trisomy is a type of embryonic dysplasia. Human chorionic gonadotropin is a glycoprotein, secreted by placental trophoblasts, which consists of an alpha subunit and a beta subunit. Among them, the β-subunit is a special amino acid sequence that exhibits immunological properties that are different than other hormones, which can reduce cross-reactivity and comprehensively reflect the state of the fetus and the function of the placenta. The serum level of human chorionic gonadotropin β is about 1% during pregnancy, peaking at 8 weeks of gestation and leveling off at 18–20 weeks [9,10]. Serum levels of free-hCG gonadotropins are significantly higher in pregnant women with Down syndrome compared to healthy individuals; serum levels of free-hCG gonadotropins are significantly lower in pregnant women with trisomy 18-trimester syndrome. Free estriol, primarily synthesized by the fetal adrenal cortex and liver, is produced in the placenta and released in a free form [11,12]. In women, it becomes detectable after 8 weeks of pregnancy. Throughout gestation, estrogen levels in women rise, responding to fetal and placental function. A decrease in free estrogen levels correlates with the progression of fetal development and is observed in cases of Down syndrome and trisomy 18 [13,14].

5. Conclusion

This study highlights the effectiveness of prenatal screening in detecting chromosomal abnormalities in fetuses, alleviating the burden of prenatal diagnosis on pregnant women, and demonstrating significant clinical value. However, it is essential to recognize that prenatal screening, while straightforward, is not the definitive diagnostic standard and has inherent limitations and risks. To enhance the detection rate of prenatal screening, better communication and collaboration with clinical professionals are necessary.

Disclosure statement

The authors declare no conflict of interest.

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