

## Analysis on the indications of invasive prenatal diagnosis in 1624 cases correlated to fetal chromosomal abnormality

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### Objective

To investigate the correlation between the detection rates of fetal chromosomal abnormalities and the indications of invasive prenatal diagnosis and to study the association between chromosomal polymorphism and fetal growth.

### Methods

A total of 1626 singleton pregnancies with indications for invasive prenatal diagnosis were selected, in which the chorionic villi (CVS), amniotic fluid (AC) or umbilical cord blood (FBS) were collected respectively from 216, 974 and 436 cases under ultrasound guidance.

### Results

The success rates of CVS and cell culture were 100% (216/216) and 99.07% (214/216) respectively. AC and umbilical cord sampling and cell culture success rates were 100%. The loss rate after invasive procedure was 0.18% (3/1626). Ninety cases were diagnosed with abnormal karyotypes, giving a detection rate of 5.54% (90/1624). The highest detection rate of abnormal karyotypes was the one from the first trimester combined test for Down's syndrome (DS), accounting for 14.85% (30/202), followed by fetal structural abnormality group 8.54% (41/480), advanced maternal age group 3.23% (4/124), parental chromosome abnormality group 3.23% (1/31) and soft marker group 2.93% (6/205). The second trimester serum screening for DS high-risk group was 1.86% (8/430) and the other groups had no abnormal karyotype detected. The commonest abnormal karyotype was trisomy-21 accounting for 1.66% (27/1624), followed by Trisomy-18, accounting for 0.68% (11/1624). Abnormal karyotypes were mainly due to aneuploidies classified as high risk by the combined test or because of the presence of fetal structural abnormalities, giving an overall detection rate for aneuploidies of 89.5% (51/57). The chromosomal number abnormalities in the high-risk of first trimester combinational test for DS and fetal structural abnormalities, such as Trisomy-21 and Trisomy-18, with the detection rate of aneuploidy 89.47% (51/57) in total. When the abnormal karyotypes were referred as chromosomal structure abnormalities they were mainly found in the advanced maternal ages, parental chromosome abnormalities, soft markers and high-risk group of the mid-trimester serological screening for DS. Totally 180 cases were diagnosed as chromosomal polymorphism, accounting for 11.08% (180/1624), including 46, XN, Yqh + 74 cases, 46, XN, inv (9) 37 cases, 46, XN, 1qh + 23 cases and 46, XN, Yqh-16 cases. Fifteen cases of serious fetal malformations were found in these patients, giving an incidence rate of fetal malformations of 8.33% (15/180) in this group. These karyotypes were mainly 46, XYqh +, 46, XN, inv (9), 46, XN, 1qh+, and 46, XYqh-.

### Conclusion

High-risk results from the first-trimester combined test for DS, fetal abnormalities and soft markers found by ultrasound, advanced maternal age, parental chromosomal abnormality and high-risk from the mid-trimester serum screening for DS were the main indicators to offer invasive diagnosis.