REFERRAL REASON

Sample was referred for antenatal screening to test for fetal aneuploidy.

RESULT AND INTERPRETATION

Testing of placental cell-free DNA from this sample showed no evidence of the chromosomal abnormalities in autosomes and sex chromosomes.

Sample type	Estimated Fetal Fraction	DNA Quality	Sequencing Quality	Unique reads (M)
Maternal plasma (cfDNA)	13.48	Pass	Pass	14.31

Common Chromosomal Aneuploidies	Not detected
Sex Chromosome Aneuploidies	Not detected
Chromosomal Aneuploidies for other chromosomes	Not detected

COMMON CHROMOSOMAL ANEUPLOIDIES

Syndrome	Result	Z score
Trisomy 21-Down Syndrome	low risk	-2.553
Trisomy 18-Edwards Syndrome	low risk	0.919
Trisomy 13-Patau Syndrome	low risk	-2.831

SEX CHROMOSOMAL ANEUPLOIDIES

Syndrome	Result	Z score
Turner syndrome (45, X)	low risk	0.090
Klinefelter syndrome (XXY)	low risk	0.090
XYY syndrome	low risk	0.090
XXX syndrome	low risk	0.090

PATIENT DETAILS

Z score reference range is between -3 to +3.

Understanding the test Results: NIPT analysis can lead to one of the three following outcomes

A- **No Aneuploidy Detected:** Low probability of the fetus developing the specific chromosomal aneuploidy

B- **Aneuploidy Detected:** High probability of the fetus developing the specific chromosome aneuploidy.

Confirmatory testing via amniocentesis/CVS is recommended.

C- **No Results:** The maternal sample could not generate test results due to unavoidable reasons, therefore repeat sampling is recommended. Invasive testing is recommended if NO TEST RESULT is obtained subsequent to the repeat sampling.

OTHER CHROMOSOMAL ANEUPLOIDIES

Syndrome	Result	Z score
chromosome 1	Low risk	1.052
chromosome 2	Low risk	0.464
chromosome 3	Low risk	1.517
chromosome 4	Low risk	-0.887
chromosome 5	Low risk	-1.200
chromosome 6	Low risk	-0.905
chromosome 7	Low risk	-0.329
chromosome 8	Low risk	-1.602
chromosome 9	Low risk	0.451
chromosome 10	Low risk	-0.415
chromosome 11	Low risk	-1.004
chromosome 12	Low risk	-0.640
chromosome 14	Low risk	-0.013
chromosome 15	Low risk	-0.426
chromosome 16	Low risk	-1.122
chromosome 17	Low risk	1.667
chromosome 19	Low risk	1.812
chromosome 20	Low risk	-0.074
chromosome 22	Low risk	0.985

ABOUT THE TEST

The cell-free DNA that was extracted from the peripheral blood of the mother was subjected to whole genome sequencing. The Next Generation Sequencing is performed using MGI platform and analyzed through the JINKE NIPT technology.

The test can detect aneuploidy across the entire fetal genome (23 pairs of chromosomes) and can interpret the results for Trisomy 13, Trisomy 18, and Trisomy 21, as well as sex chromosomes. This test has a detection accuracy of up to 99 percent for fetal chromosomal aneuploidy.

RECOMMENDATIONS

1. The above results need to be interpreted in the context of all clinical findings.
2. **Patients with negative NIPT results still require follow-up ultrasound examination and diagnostic testing is recommended incase of any scan anomalies**

TEST LIMITATIONS

*If the fetal fraction is lower than 3.5%, the accuracy of the test may be reduced. To ensure the accuracy of the results, we would recommend a re-sampling of the maternal blood one or two weeks later. If the fetal fraction is less than 7%, the detection power of fetal microdeletion/microduplication syndrome (< 5 Mb) is limited. We would recommend a more careful clinical observation or a higher coverage whole genome sequencing re-test.

1. The results of this test are for reference only, not for the final diagnosis. If the test result is high risk, genetic counseling and invasive prenatal diagnosis are needed. If a high risk of microdeletion/microduplication syndrome is detected, prenatal diagnosis is recommended to be combined with maternal chromosome analysis to exclude maternal influence. If the test result is low risk, the fetus has a low risk of developing the target disease of this screening. However, the possibility of other abnormalities cannot be excluded, and systematic ultrasound examinations and other prenatal examinations should be conducted
2. The results of this test include sex chromosome aneuploidies and other chromosomal aneuploidies. Compared with trisomy 21, trisomy 18 and trisomy 13, the incidence of these aneuploidies is lower in the population, and the data available is limited. Therefore, the possibility of false positive or false negative cannot be ruled out.
3. This method is not suitable for testing: Gestational age <12+0 weeks; Pregnant women with twins or multiple pregnancies; One partner has a definite chromosomal abnormality; Received allogeneic blood transfusion, transplantation and allogeneic cell therapy within 1 year; Fetal ultrasound result suggested that there were structural abnormalities and prenatal diagnosis was needed; A family history of genetic diseases or a high risk of genetic diseases in the fetus; Pregnancy with malignant tumor; Other conditions that the doctor considers may affect the accuracy of the results.
4. Abnormalities caused by the following factors cannot be detected in this test: structural abnormalities such as chimera and translocation in chromosomes; Chromosomal polyploidies (triploid, tetraploid, etc.); Balanced translocation, inversion and ring of chromosomes; Uniparental disomy (UPD); Single/polygenic diseases; Chromosomal regions with high duplication and fixation, such as the chromosomal abnormalities of the proximal centromere and telomere.
5. The sample tested is the cell free DNA in peripheral blood of pregnant women, which mainly from placental trophoblastic cells rather than direct fetal cells. Given the limitations of the current technical level of the medical tests and the differences among pregnant women, there may still be false positives or false negatives in rare cases, even if the testing staff has fulfilled job responsibilities and operational procedures.

6. The detection accuracy may be reduced to some extent for severely obese pregnant women (BMI >40) and conception through in-vitro fertilization-embryo transfer, and the test results are only for reference.
7. The patient should provide complete, accurate and detailed personal information. The center shall not be responsible for the interruption of testing services and inaccurate results caused by inaccurate information or other misleading factors provided by the patient.
8. The test results in this report are only responsible for the samples submitted for inspection.
9. The NIPT is a screening test; a low risk does not exclude the above evaluated disorders. It is not intended, either validated for diagnosis nor for use in pregnancies with more than two fetuses, mosaicism, partial chromosomal aneuploidy, translocations or maternal aneuploidy.
10. The NIPT is a screening test and the positive predictive value is not 100%. Hence confirmation of high-risk results is recommended by invasive testing

Important: On doing PNDT test, the undersigned hereby confirms that no sex chromosome information has been passed on to anyone in whatsoever manner.

REFERENCE

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