

Patient Name :
Age / Gender :
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EmiratesID/PassportNo :

Sample UID No. :
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Non-Invasive Prenatal Test (NIPT- Advanced)

Test results:

1) Common Trisomies

Syndrome	Result	Z-score	Low risk range
Trisomy 21	Low risk	0.104	-3< Z-score <3
Trisomy 18	Low risk	-0.999	-3< Z-score <3
Trisomy 13	Low risk	-0.242	-3< Z-score <3

2) Fetal gender determination

Y chromosome detection	
Y Chromosome	Not detected

Condition	Sensitivity (95% CI)
Fetal Sex	98.6% (92.19%-99.96%)

3) 23 types of fetal chromosomal aneuploidies

Syndrome	Result	Z-score	Low risk range
Trisomy 1	Low risk	0.231	-3< Z-score <3
Trisomy 2	Low risk	-0.495	-3< Z-score <3
Trisomy 3	Low risk	0.542	-3< Z-score <3
Trisomy 4	Low risk	0.364	-3< Z-score <3
Trisomy 5	Low risk	0.043	-3< Z-score <3
Trisomy 6	Low risk	0.455	-3< Z-score <3
Trisomy 7	Low risk	0.023	-3< Z-score <3
Trisomy 8	Low risk	-0.661	-3< Z-score <3
Trisomy 9	Low risk	-0.030	-3< Z-score <3

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Trisomy 10	Low risk	-0.947	-3< Z-score <3
Trisomy 11	Low risk	0.409	-3< Z-score <3
Trisomy 12	Low risk	0.483	-3< Z-score <3
Trisomy 14	Low risk	0.424	-3< Z-score <3
Trisomy 15	Low risk	0.247	-3< Z-score <3
Trisomy 16	Low risk	-0.356	-3< Z-score <3
Trisomy 17	Low risk	0.671	-3< Z-score <3
Trisomy 19	Low risk	2.115	-3< Z-score <3
Trisomy 20	Low risk	-0.590	-3< Z-score <3
Trisomy 22	Low risk	-1.130	-3< Z-score <3
Turner syndrome (45,X)	Low risk	1.903	-3< Z-score <3
Klinefelter syndrome (XXY)	Low risk	1.903	-3< Z-score <3
XXX syndrome	Low risk	1.903	-3< Z-score <3
XXX syndrome	Low risk	1.903	-3< Z-score <3

4) 66 types of large segment deletion/duplication syndromes (> 10 Mb)

Syndrome	Result	Abnormal proportion	Low risk range
Partial monosomy 1p	Low risk	0.45%	<5 %
Monosomy 1q21→q32	Low risk	2.22%	<5 %
Monosomy 1q42→qter	Low risk	0.94%	<5 %
Trisomy 1q23→qter	Low risk	0.68%	<5 %
Partial monosomy 2p	Low risk	0.08%	<5 %
Partial trisomy 2p	Low risk	0.08%	<5 %
Partial monosomy 2q	Low risk	0.39%	<5 %
Partial trisomy 2q	Low risk	0.39%	<5 %
Monosomy 3p11→p21	Low risk	0.02%	<5 %

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Monosomy 3p25→pter	Low risk	0.22%	<5 %
Partial trisomy 3p	Low risk	0.00%	<5 %
Partial monosomy 3q	Low risk	0.66%	<5 %
Partial trisomy 3q	Low risk	0.66%	<5 %
Partial monosomy 3p and partial trisomy 3q	Low risk	0.00%	<5 %
Partial monosomy 4p	Low risk	0.43%	<5 %
Partial trisomy 4p	Low risk	0.43%	<5 %
Monosomy 4q21→q31	Low risk	0.40%	<5 %
Monosomy 4q31→qter	Low risk	0.11%	<5 %
Partial trisomy 4q	Low risk	0.21%	<5 %
Partial monosomy 4p and partial trisomy 4q	Low risk	0.43%	<5 %
Partial trisomy 5p	Low risk	0.14%	<5 %
Partial monosomy 5q	Low risk	0.05%	<5 %
Partial trisomy 5q	Low risk	0.05%	<5 %
Partial trisomy 6p	Low risk	0.34%	<5 %
Partial monosomy 6q	Low risk	0.23%	<5 %
Partial trisomy 6q	Low risk	0.23%	<5 %
Partial monosomy 7p	Low risk	0.18%	<5 %
Partial trisomy 7p	Low risk	0.18%	<5 %
monosomy 7q32→qter	Low risk	0.42%	<5 %
Trisomy 7q21→q31	Low risk	0.51%	<5 %
Trisomy 7q32→qter	Low risk	0.42%	<5 %
Partial trisomy 8q	Low risk	0.39%	<5 %
Partial monosomy 9p	Low risk	0.34%	<5 %
Partial trisomy 9p	Low risk	0.34%	<5 %
Partial monosomy 9q	Low risk	0.17%	<5 %

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Partial trisomy 9q	Low risk	0.17%	<5 %
Partial monosomy 10p	Low risk	0.41%	<5 %
Partial trisomy 10p	Low risk	0.41%	<5 %
Partial monosomy 10q	Low risk	0.40%	<5 %
Partial trisomy 10q	Low risk	0.40%	<5 %
Partial trisomy 11p	Low risk	0.16%	<5 %
Partial monosomy 11q	Low risk	0.44%	<5 %
Partial trisomy 11q	Low risk	0.44%	<5 %
Partial monosomy 12p	Low risk	0.11%	<5 %
Partial trisomy 12p	Low risk	0.11%	<5 %
Partial trisomy 12q	Low risk	0.39%	<5 %
Monosomy 13q14	Low risk	0.66%	<5 %
Monosomy 13q21→qter	Low risk	0.72%	<5 %
Trisomy 13cen→q14	Low risk	0.26%	<5 %
Trisomy 13q21→qter	Low risk	0.72%	<5 %
Partial trisomy 14	Low risk	0.32%	<5 %
Trisomy 14q24-qter	Low risk	0.11%	<5 %
supernumerary inv dup (15)	Low risk	2.85%	<5 %
Trisomy 15q22→qter	Low risk	0.14%	<5 %
Partial trisomy 16p	Low risk	0.37%	<5 %
Partial monosomy 16q	Low risk	0.84%	<5 %
Partial trisomy 16q	Low risk	0.84%	<5 %
Trisomy 17pter→q21	Low risk	0.57%	<5 %
Partial monosomy 18p	Low risk	0.19%	<5 %
Partial tetrasomy 18p	Low risk	0.19%	<5 %
Trisomy 18pter→q12	Low risk	0.38%	<5 %

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Monosomy 18q21→qter	Low risk	0.87%	<5 %
Trisomy 18q12→qter	Low risk	0.56%	<5 %
Partial monosomy 20p	Low risk	1.19%	<5 %
Trisomy 20p	Low risk	1.19%	<5 %
Partial monosomy 21	Low risk	0.14%	<5 %

- 5) 6 types of microdeletion/microduplication syndromes located at the positions of chromosome segments associated with specific syndromes (> 5 Mb)

Syndrome	Result	Abnormal proportion	Low risk range
1p36 deletion syndrome	Low risk	1.47%	<7 %
Cri-du-chat syndrome	Low risk	0.83%	<7 %
Angelman syndrome	Low risk	0.89%	<7 %
Prader-Willi syndrome	Low risk	0.89%	<7 %
10p14-p13 deletion Syndrome (DiGeorge 2 syndrome)	Low risk	1.74%	<7 %
22q11.2 deletion Syndrome (DiGeorge syndrome)	Low risk	0.89%	<7 %

- 6) Sample quality result

Sample information	Test value	Reference range	Result
Unique reads (M)	65.53	>15	Passed
GC content of unique reads (%)	40.63	37~ 43	Passed
Fetal Fraction (%)	15.81		

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Test Information and Interpretation Guidelines

- 1) Test Description: The NIPT test is a screening test and is not diagnostic. It works by isolating the cfDNA (including both maternal and fetal DNA) from a maternal blood sample and performing low coverage whole genome sequencing using Next Generation Sequencing technology. The unique reads of each chromosome are calculated and compared to an optimal reference control sample. Data is analyzed using bioinformatics algorithms and an assessment is produced for the conditions tested only. The test must not be used as the sole basis for diagnosis or other pregnancy management decisions.
- 2) Tests should always be ordered by a qualified healthcare professional and results reviewed with the patient. 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.
- 3) The results of this test include 3 types of common trisomies, 23 types of fetal chromosomal aneuploidies, 66 types of large segment deletion/duplication syndromes (> 10 Mb), and 6 types of microdeletion/microduplication syndromes located at the positions of chromosome segments associated with specific syndromes (> 5 Mb). Compared with trisomy 21, trisomy 18 and trisomy 13, the incidence of deletion/duplication syndrome is lower in the population, and the data available is limited. Therefore, the possibility of false positive or false negative cannot be ruled out. In this report, the deletion/duplication test is only used for the syndromes shown in the tables. For other conditions not tested in this report, we cannot exclude the possibility of chromosomal structures or gene fragments abnormalities.
- 4) This method is not suitable if: Gestational age <10+0 weeks; Pregnancy with triplets or more fetuses; One partner has a definite chromosomal abnormality; Received allogeneic blood transfusion, transplantation and allogeneic cell therapy within 1 year; Pregnancy with malignant tumor; Other conditions that the doctor considers may affect the accuracy of the results.
- 5) 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.
- 6) 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.
- 7) The sample tested is the cell free DNA in peripheral blood of pregnant women, which comes 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.
- 8) 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.
- 9) 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.
- 10) For fetal sex identification, this is a screening test and is not diagnostic. The Y chromosome detection uses the method of counting the molecular representation of chr Y fragments from WGS-based cell free DNA sequencing of maternal plasma. Potential sources of an inaccurate test result may include but not limited to: maternal, fetal and/or placental mosaicism, low fetal fraction, blood transfusion, transplant surgery, stem cell therapy, vanished twins, other biological reasons and so on.
- 11) The test results in this report are only responsible for the samples submitted for testing.

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12) Performance specification:

Condition	Sensitivity	Specificity
T21	99.17%	99.96%
T18	98.83%	99.95%
T13	100%	99.96%
SCA	99.6%	99.8%

Note: The data in the table is based on historical literature and internal data, and only reflects past detection, not the actual condition of the tested sample nor the promised value.

Reference:

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