Sample ID:	**11009458745**
Sample Type:	Whole Blood
Patient's Name:	***Ms. XYZ***
Patient's Age:	28 Years
Fetus number:	Singleton
Gestational age:	15^{+5} days

CliSeq
Non-Invasive Prenatal Testing

Data uploaded on:	**DD/MM/YYYY**	**Report Generated on:**	**DD/MM/YYYY**

Test Details: NIPT Advanced (Test items: 3 types of common trisomies, 22 types of fetal chromosomal aneuploidies

Test Results:-

I. 3 types of common trisomies

Syndrome	Result	Z-score	Low risk range
Trisomy 21	Low risk	0.115	-3< Z-score <3
Trisomy 18	Low risk	0.046	-3< Z-score <3
Trisomy 13	Low risk	-1.523	-3< Z-score <3

II. 22 types of fetal chromosomal aneuploidies

Syndrome	Result	Z-score	Low risk range
Trisomy 1	Low risk	1.295	-3< Z-score <3
Trisomy 2	Low risk	0.876	-3< Z-score <3
Trisomy 3	Low risk	0.115	-3< Z-score <3
Trisomy 4	Low risk	0.134	-3< Z-score <3
Trisomy 5	Low risk	0.874	-3< Z-score <3
Trisomy 6	Low risk	0.795	-3< Z-score <3
Trisomy 7	Low risk	-0.186	-3< Z-score <3
Trisomy 8	Low risk	0.674	-3< Z-score <3
Trisomy 9	Low risk	-1.769	-3< Z-score <3
Trisomy 10	Low risk	0.860	-3< Z-score <3
Trisomy 11	Low risk	0.556	-3< Z-score <3
Trisomy 12	Low risk	1.051	-3< Z-score <3
Trisomy 14	Low risk	-0.689	-3< Z-score <3
Trisomy 15	Low risk	2.117	-3< Z-score <3
Trisomy 16	Low risk	-2.196	-3< Z-score <3
Trisomy 17	Low risk	-1.560	-3< Z-score <3
Trisomy 19	Low risk	-1.569	-3< Z-score <3

Trisomy 20	Low risk	-0.488	-3< Z-score <3
Trisomy 22	Low risk	1.071	-3< Z-score <3
Turner syndrome (45,X)	Low risk	-2.057	-3< Z-score <3
Klinefelter syndrome (XXY)	Low risk	-2.057	-3< Z-score <3
XYY syndrome	Low risk	-2.057	-3< Z-score <3
XXX syndrome	Low risk	-2.057	-3< Z-score <3

III. Sample quality test

Sample information	Test value	Reference range	Result
Unique reads (M)	25.00	>15	Passed
GC content of unique reads (%)	40.10	37~43	Passed
Fetal Fraction (%)*	12.36		

*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.

Disclaimer:

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. This method is not suitable for testing: Gestational age <10 weeks;
   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.
3. 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.
4. 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.
5. 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.
6. 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.
7. The test results in this report are only responsible for the samples uploaded on Cliseq Interpreter.

References:

1. Shen S, Qi H, Yuan X, et al; The performance evaluation of NIPT for fetal chromosome microdeletion/microduplication detection: a retrospective analysis of 68,588 Chinese cases. Front Genet. 2024 Jun 7;15:1390539. PMID: 38911296.
2. Gabrielli F, Papa FT, Di Pietro F, et al; New Bioinformatic Pipeline to Detect Fetal Aneuploidies and Rearrangements Using Next-Generation Sequencing. Int J Genomics. 2024 Jun 13;2024:8859058. PMID: 38962150.
3. Bianchi DW, Platt LD, Goldberg JD, et al; MatErnal BLood IS Source to Accurately diagnose fetal aneuploidy (MELISSA) Study Group. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstet Gynecol. 2012 May;119(5):890-901. PMID: 22362253.
4. Alberry MS, Aziz E, Ahmed SR, Abdel-Fattah S. Non invasive prenatal testing (NIPT) for common aneuploidies and beyond. Eur J Obstet Gynecol Reprod Biol. 2021 Mar;258:424-429. PMID: 33550217.
5. Hui L, Bianchi DW. Fetal fraction and noninvasive prenatal testing: What clinicians need to know. Prenat Diagn. 2020 Jan;40(2):155-163. PMID: 31821597.

***For any further technical queries please contact at contact@genes2me.com**