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Excellence In Health Care

DEPARTMENT OF MOLECULAR GENETICS AND GENOMICS

Patient Name	Mrs. GULSANOBAR MOHSIN	**Ref. Doctor**	--
Age/Gender	33Y 0 M 0 D /Female	**Ref. Hospital**	

TEST REPORT FOR NIPT FIVE CHROMOSOMES

INDICATION: The antenatal scan indicative of ossified nasal bone, sample was referred for antenatal screening to test for fetal aneuploidy.

Pregnancy Type	Singleton
Gestational age	13.6weeks
Sample Quality	Pass
Fetal fraction	10.83%

RESULT AND INTERPRETATION: No Aneuploidy Detected

No chromosomal aneuploidy was detected across tested 5 chromosome pairs, suggesting a low risk for the conditions screened. As this is a non-invasive screening test, confirmatory diagnostic testing may be considered based on clinical findings.

Syndrome	Result	Z score
Trisomy 21-Down Syndrome	low risk	-1.192
Trisomy 18-Edwards Syndrome	low risk	0.198
Trisomy 13-Patau Syndrome	low risk	-1.974
Sex chr abnormalities	Low risk for, XO, XXX, XXY& XYY	**

***Zscore reference range is between -3 to +3.**Zscore is not applicable for sex chromosomal abnormalities. N/A Not Applicable*

RECOMMENDATIONS

- The above results need to be interpreted in the context of all clinical findings.
- Further follow up with your health provider is recommended.

EXPECTED TEST RESULTS

DRL-NIPT analysis can yield any of the following three results:

- **No Aneuploidy Detected:** The likelihood of the fetus having the particular chromosomalaneuploidy is low, based on the test results showing no evidence of aneuploidy.
- **Aneuploidy Detected:** A high likelihood of the fetus being affected with the specific chromosomalaneuploidy has been observed. Therefore, it is recommended to undergo confirmatory testing viaamniocentesis/CVS.
- **No Results:** A result could not be obtained from the maternal sample provided for testing due tounforeseen circumstances. It is recommended to collect another sample for testing. If a result is not obtained again, it is recommended to consider invasive testing.

TEST PERFORMANCE

Prenatal Chromosomal Aneuploidy Results for Chromosomes 13, 18, 21 & sex chromosomes		
Chromosome	**Risk**	**Sensitivity**
Chromosome 13	Low	99.99%
Chromosome 18	Low	99.99%
Chromosome 21	Low	99.99%
XO	Low	90.32%
XXY	Low	93.00%
XXX	Low	93.00%

TEST INFORMATION

Principle

The test is capable of genome-wide aneuploidy detection over the whole fetal genome (23 pairs of chromosomes) and offers an interpretation of the results for Trisomy 13, Trisomy 18, Trisomy 21, sex chromosomes and other autosomal chromosomes; as well as common microdeletions including such as Wolf-Hirschhorn syndrome (4p), DiGeorge (22q11.2), Angelman (15q11.2), Prader-Willi (15q11.2), cri-du-chat (5p), 1p36 deletion. The accuracy of the test for detecting fetal chromosome abnormalities is up to 99%.

Methodology

The test utilizes a non-invasive method to extract cell-free fetal DNA from the mother's Peripheral whole blood sample, followed by high-throughput sequencing of the extractedDNA using the Illumina platform. The molecular mass of fetal DNA is calculated for all chromosomes using this approach. The sequencing data is then analyzed using the CHROME analysis pipeline version 2.1.2.

TEST LIMITATIONS

- The NIPT-EXTENDED (23 PAIRS OF CHROMOSOMES + MICRODELETIONS) by NGS examines all 23 chromosomes for aneuploidy and Microdeletions in singleton and twin pregnancies. These tests are performed for gestational ages of at least 9 weeks.
- It's important to understand that the Non-Invasive Prenatal Test (NIPT) is a screening test, which meansthat a low-risk result does not completely rule out the possibility of the evaluated disorders. Please note that this test is not intended or validated for diagnostic purposes, and it should not be used for pregnancies with more than two fetuses, mosaicism, partial chromosomal aneuploidy, translocations, ormaternal aneuploidy.
- NIPT is a screening test, and a positive result does not confirm the presence of evaluated disorders. Invasive testing is recommended for confirmation of high-risk results due to the test's imperfect positivepredictive value.
- A low-risk NIPT result decreases the likelihood of fetal aneuploidy but does not guarantee an unaffected fetus. Sub-chromosomal abnormalities, gene defects, and birth defects are still possible. Invasive testingmay be necessary later in the pregnancy.
- NIPT accuracy may be impacted by factors such as maternal mosaicism, neoplasms, vanishing twin, low fetal fraction, and confined placental mosaicism. False positive and negative results are possible.
- In singleton pregnancies, NIPT can detect fetal DNA at a fraction of 2% or greater, while in twin pregnancies, detection is possible at a fraction of 4% or greater. Sensitivity may be reduced in twin pregnancies with a fetal fraction of 2-4%.
- NIPT can only provide results for certain types of multiple pregnancies, but it cannot identify individual fetuses

REFERENCES

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- Committee Opinion No. 545: Noninvasive prenatal testing for fetal aneuploidy. Obstet Gynecol. 2012;120(6):1532-4
- ACMG statement on noninvasive prenatal screening for fetal aneuploidy Anthony R. Gregg, S.J. Gross, R.G. Best, K.G. Monaghan, K. Bajaj, B.G. Skotko, B.H. Thompson and M.S. Watson. Genetics in Medicine 2013.

Important: The undersigned confirms that no sex chromosome information has been disclosed to anyone in any way as a result ofundergoing the PNDT test.

----------End Of Report ----------

Dr. Radhika A
Head of Department
Genetics & Molecular Testing

DRL's responsibility is limited to the test's analytical portion. Samples from non-DRL sites are assumed to belong to the patient identified on the labels and have been verified by the collector.


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