GENESYS Diagnostics INC.

prenatal screening report

REPORT RELEASED
Date: 11/23/21 Time: 12:44 PM

PROVIDER INFORMATION
Attn: Jane Doctor, MD
123 Fake Street
Springfield, NY 13531
Phone: (123) 456-7890
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SECOND RECIPIENT

PATIENT INFORMATION
Jane Patient
DOB: 05/01/1981
GA: 10 weeks
Indication: AMA
Medical record/patient ID: 123456789

SAMPLE INFORMATION
Client Sample ID:
Order ID: 742352
Date of Draw: 04/17/18
Date Received: 04/18/18
Pregnancy Type: Singleton

Electronically signed and dated on 11/23/2021 12:44:08 PM:
Lab director's e-signature is required.

RESULTS SUMMARY:

CHROMOSOME	RESULTS	PPV (%)
Chromosome 21	**NEGATIVE: No aneuploidy detected** Results consistent with two copies of chromosome 21	
Chromosome 18	**NEGATIVE: No aneuploidy detected** Results consistent with two copies of chromosome 18	
Chromosome 13	**NEGATIVE: No aneuploidy detected** Results consistent with two copies of chromosome 13	
Sex Chromosomes	**NEGATIVE: No aneuploidy detected** Results consistent with two sex chromosomes (XY)	

CLINICAL COMMENTS: This is a screening test; therefore, false positive and false negative results can occur. Clinical correlation is indicated. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary. The fetal fraction (FF) is estimated to be 8%. FF estimation is one component of this algorithm and is combined with other quality metrics to determine the confidence in the results. The FF estimate is not used in isolation to exclude samples.

PERFORMANCE AND LIMITATIONS

LIMITATIONS OF THE TEST: The Verifi™ Prenatal Test is validated for aneuploidy (both monosomies and trisomies) of all chromosomes, including 21,13, 18, X, and copy number variants (7Mb or greater) in singleton pregnancies, with a gestational age of at least 10 weeks 0 days. This is a screening test that looks only for specific chromosomal abnormalities. A normal result does not eliminate the possibility that the pregnancy is associated with other chromosomal or subchromosomal abnormalities, birth defects, genetic conditions, or other conditions, such as open neural tube defects or autism.

There is a small possibility that the test results might not reflect the chromosomes of the fetus but may reflect chromosomal changes of the placenta (confined placental mosaicism or CPM) or of you (maternal chromosomal abnormalities). Examples are, but are not all inclusive, maternal XXX, sex chromosome status, or benign and malignant maternal neoplasm. Some CPM cases have been associated with a higher chance for pregnancy complications or for uniparental disomy (UPD) depending on the chromosome in question, which may affect the growth and development of the fetus. Some of these rare chromosomal aneuploidies have been found to occur only in mosaic form. Clinical consequences depend on the chromosome(s) involved and cannot be predicted during the pregnancy. Copy number variants (CNVs) are structural changes that have been identified in all human chromosomes and can vary in size. This screening assay detects CNVs that are 7Mb or larger. Depending on the size and location of the CNV, it may correlate with a clinical consequence/effect. This test, like many tests, have limitations, including false negative and false positive results. A negative test result does not guarantee the pregnancy is unaffected.

PERFORMANCE METRICS:†

Chromosome	N	Sensitivity	95% CI	Specificity	95% CI	Accuracy	95% CI
21	500	99.9% (90/90)	96.0-100.0	99.8% (409/410)	98.7 - 100.0	—	—
18	501	97.4% (37/38)	86.2-99.9	99.6% (461/463)	98.5 - 100.0	—	—
13	501	87.5% (14/16)	61.7-98.5	99.9% (485/485)	99.2 - 100.0	—	—
Chromosome	N	Sensitivity	95% CI	Specificity	95% CI	Accuracy	95% CI
Monosomy X	508	95% (19/20)	75.1-99.9	99.0% (483/488)	97.6-99.7	—	—
XX	508	97.6% (243/249)	94.8-99.1	99.2% (257/259)	97.2-99.9	98.4%	96.9-99.3
XY	508	99.1% (227/229)	96.9-99.9	98.9% (276/279)	96.9-99.8	99.0%	97.7-99.7
XXX/XXY/XYY	Other sex aneuploidies will be reported if detected. (Limited data of these more rare aneuploidies preclude performance calculations.)						
Microdeletions, Copy Number Variants, & other autosomal aneuploidies	Microdeletions, Copy Number Variants (CNVs) and other autosomal aneuploidies if requested and detected will be reported. (Limited data of these more rare abnormalities preclude performance calculations.)						

† Data on file at Illumina, Inc. regarding Performance and Method Comparison studies.

TEST METHOD: Nucleic Acid extraction, DNA sequencing, and analysis of sequencing results to determine fetal aneuploidy.

DISCLAIMER: The manner in which this information is used to guide patient care is the responsibility of the health care provider, including advising for the need for genetic counseling or diagnostic testing. Any test should be interpreted in the context of all available clinical findings.

DISCLOSURE: This prenatal test was developed by, and its performance characteristics were determined by Verinata Health, Inc. a wholly owned subsidiary of Illumina, Inc. The VHI laboratory is CAP-accredited and certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing. This prenatal test has not been cleared or approved by the U.S. Food and Drug Administration.

This prenatal test is performed by Verinata Health, Inc., a wholly owned subsidiary of Illumina, Inc.
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State of New York Laboratory Director: Eileen S. de Feo, Ph.D.

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