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Verifi™ Plus Prenatal Test

Screen for a broader range of chromosomal aneuploidies

The all-chromosome option of the Verifi Plus Prenatal Test provides information about trisomies for all chromosomes in singleton pregnancies, giving you and your patients a broad range of information. This option within the Verifi Plus Prenatal Test allows screening for rare autosomal trisomies, which when present in an individual may lead to varying degrees of structural defects, and/or developmental and intellectual disabilities.[1]

When ultrasound anomalies are detected, guidelines recommend invasive diagnostic follow-up testing for those patients.[2] However, for those who decline invasive diagnostic follow-up testing, the all-chromosome screening test could be an option. Recent literature also suggests that rare autosomal trisomy detected on non-invasive prenatal screening is associated with increased risks of adverse pregnancy outcomes, including intrauterine fetal demise, fetal growth restriction, and preterm birth.[3] Follow up pregnancy management may be modified based on these results.[3]

Chromosomal aneuploidies identified with this test may be representative of the chromosomal make up of every fetal cell (full fetal aneuploidy), some fetal cells (fetal mosaicism) or placental cells only (confined placental mosaicism). The clinical significance of rare chromosome aneuploidies is variable and depends on the specific finding and which cells are involved. Patients with a positive Verifi Plus Prenatal Test result should be offered further genetic counseling and invasive diagnostic testing such as chorionic villus sampling or amniocentesis.[4]

Extensive options for more personalized screening

The Verifi Plus Prenatal Test offers the following testing options:

- Trisomy 21, 18, and 13 is always included.
- Sex chromosome aneuploidies (monosomy X, XXX, XXY, and XYY) are included if requested; fetal sex (XX or XY) will be reported if no sex chromosome aneuploidy is detected.
- Trisomy of all chromosomes, including sex chromosome aneuploidies, if requested.
- Select microdeletions such as 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome),15q11.2 (Prader-Willi syndrome/Angelman syndrome), and 22q11.2 deletion (DiGeorge) are included if requested.

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Singleton gestations at ≥ 10 weeks gestation*

Trisomies 21, 18, and 13 with the following options:

- Sex chromosome aneuploidy
- All chromosome trisomies (including sex chromosome aneuploidies)
- Select microdeletion syndromes

3–5 business day turnaround from sample receipt to report to partner lab

*Not available in multiple gestations.

Clear, concise results

Results from the Verifi Plus Prenatal Test are reported as “Positive: Aneuploidy Detected” or “Negative: No Aneuploidy Detected.” Results for chromosomes 21, 18, 13, X, and Y will continue to be reported individually. Results for the remaining chromosomes are reported collectively. A specific chromosomal aneuploidy will be reported in the event of a “POSITIVE: Aneuploidy Detected” result. Results for a positive microdeletion syndrome will be reported as “POSITIVE: Abnormality detected.”

Verifi™ Plus prenatal test report

Verifi™ Plus Prenatal Test

REPORT RELEASED
Date: 09/29/22 Time: 10:29 AM

PROVIDER INFORMATION
Attn: Jane Doctor, MD
123 Fake Street
Downtown City, CA 10231
Phone: (123) 456-7890
Fax: (123) 456-7899

SECOND RECIPIENT
Dr. Mary Smith
Address: 1234 State Street
Downtown City, CA 10231
Fax: (800) 555-1213

PATIENT INFORMATION
Jane Patient
DOB: 11/08/1994
GA: 11 weeks
Indication: AMA
Medical record/patient ID: 123456789

SAMPLE INFORMATION
Client Sample ID:
Order ID: 742352
Date of Draw: 05/18/22
Date Received: 05/20/22
Pregnancy Type: Singleton

ANEUPLOIDY DETECTED

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	
All Other Autosomal Chromosomes	**POSITIVE: Aneuploidy detected** Results consistent with pregnancy at increased risk for trisomy 7	NA*
Sex Chromosomes	**NEGATIVE: No aneuploidy detected** Results consistent with two sex chromosomes (XY)	
Microdeletions (1p36 deletion, 4p16.3, 5p15.2, 15q11.2, 22q11.2 deletion)	**NEGATIVE: No abnormality detected** Results consistent with no microdeletions detected in the regions of 1p36, 4p16.3, 5p15.2, 15q11.2, 22q11.2	

Analytical validation for all chromosomes

	Sensitivity	95% CI	Specificity	95% CI
All autosomes	98.7%	96.1%–99.6%	99.95%	99.62%->99.99%

The cohort was composed of samples for which high coverage sequencing data identified them as either affected by autosomal aneuploidy (N=189) or otherwise exhibiting normal diploidy (N=1330). The data for these samples were then analyzed at the normal level of sequencing coverage to establish the sensitivity and specificity of the improved algorithm.

References

1. Gardner RJM, Sutherland GR, Shaffer LG. Chromosome Abnormalities and Genetic Counseling. New York, NY: Oxford University Press; 2012.
2. Practice Bulletin No. 162 Summary: Prenatal Diagnostic Testing for Genetic Disorders. Obstet Gynecol. 2016 May;127(5):976-978. doi: 10.1097/AOG.0000000000001438. PMID: 27101119.
3. Mossfield T, Soster E, Menezes M, Agenbag G, Dubois M-L, Gekas J, Hardy T, Jurkowska M, Kleinfinger P, Loggenberg K, Marchili P and Sirica R (2022), Multisite assessment of the impact of cell-free DNA-based screening for rare autosomal aneuploidies on pregnancy management and outcomes. Front. Genet. 13:975987. doi: 10.3389/fgene.2022.975987
4. Practice Bulletin No. 226: Screening for Fetal Chromosomal Abnormalities. Obstet Gynecol. 2020;136(4): e48-e69.

Limitations of the test

Verifi™ Plus based on cell-free DNA analysis from maternal or pregnant patients' blood is a screening test. False positive and false negative results do occur. Test results must not be used as the sole basis for diagnosis. Further genetic counseling and confirmatory diagnostic testing is necessary with a positive test result. Test results might not reflect the chromosomal status of the fetus but may reflect chromosomal changes of the placenta (CPM) or of the patient, which may or may not have clinical significance. CPM may be associated with a higher chance for pregnancy complications or for uniparental disomy (UPD), which may affect the growth and development of the fetus. Some of these rare chromosomal aneuploidies may only occur in mosaic form. Clinical consequences depend on the chromosome involved and cannot be predicted prenatally. A negative test result does not eliminate the possibility of chromosomal abnormalities for the tested chromosomes or microdeletions. In addition, microdeletion conditions caused by other molecular mechanisms cannot be detected with this assy. This test does not screen for polyploidy (e.g. triploidy), birth defects such as open neural tube defects, single gene disorders, or other conditions, such as autism.

Verifi™ and Verifi™ Plus were developed by, and their performance characteristics were determined by Verinata Health, Inc. (VHI) 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. They have not been cleared or approved by the U.S. Food and Drug Administration.

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