

# Verifi™ Plus Prenatal Test

## Screen for a broader range of chromosomal aneuploidies

The new all-chromosome option of the Verifi Plus Prenatal Test will provide information about trisomies for all chromosomes, giving you and your patients a broader range of information than the standard Verifi Prenatal Test.

This option within the Verifi Plus Prenatal Test, allows screening for rare autosomal trisomies that may occur, especially in the presence of an abnormal ultrasound. Guidelines recommend invasive diagnostic follow-up for those patients with ultrasound anomalies; however, for those who decline invasive diagnostic follow-up, the all-chromosome screening test could be an option. Chromosomal aneuploidies, in general, may lead to varying degrees of structural defects, and developmental and intellectual disabilities.<sup>1</sup>

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), placental cells only (confined placental mosaicism), or some maternal cells (maternal 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 NIPT result should be offered further detailed counseling and diagnostic testing such as chorionic villus sampling or amniocentesis.<sup>2</sup>

### Extensive options for more personalized screening

The Verifi Plus Prenatal Test offers the following testing options:

- Aneuploidy of chromosomes 21, 18, and 13 (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.
- Aneuploidy (trisomy) of all chromosomes, including sex chromosome aneuploidies.
- 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.

#### Verifi Plus Prenatal Test\*

Singleton gestations at ≥ 10 weeks gestation

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

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

3–5 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 “Results consistent with a microdeletion in a certain genomic region.”

## Report example

### Verifi Plus prenatal screening report

## Verifi™ Plus Prenatal Test

**REPORT RELEASED**

Date: 09/22/15 Time: 08:45 AM

PROVIDER INFORMATION  
**University Medical Center**  
Attn: Dr. John Smith-Gonzalez  
1234 Doctor Street, Suite 541  
Downtown City, CA 10231  
Phone: (800) 555-1212  
Fax: (800) 555-1212

SECOND RECIPIENT  
**Medical Center**  
Attn: Dr. Mary Smith  
Address: 1234 State Street,  
Downtown City, CA 10231  
Phone: (800) 555-1212  
Fax: (800) 555-1212

PATIENT INFORMATION  
**Name: Jane Doe**  
DOB: 09/07/1971  
GA: XXXXXXXX  
Indication: XXXXXXXX  
Medical record/patient ID:  
XXXXXXXX

SAMPLE INFORMATION  
**Client Sample ID:  
1234567**  
**Order ID: 1234567**  
Date of Draw: 09/07/15  
Date Received: 09/22/15  
Pregnancy Type:  
Singleton

#### ANEUPLOIDY DETECTED

#### RESULTS SUMMARY:

CHROMOSOME:	RESULTS:
Chromosome 21	<b>NEGATIVE: No aneuploidy detected</b> Results consistent with two copies of chromosome 21.
Chromosome 18	<b>NEGATIVE: No aneuploidy detected</b> Results consistent with two copies of chromosome 18.
Chromosome 13	<b>NEGATIVE: No aneuploidy detected</b> Results consistent with two copies of chromosome 13.
All Other Autosomal Chromosomes	<b>POSITIVE: Aneuploidy detected</b> Results consistent with pregnancy at increased risk for trisomy 6.
Sex Chromosomes	<b>NEGATIVE: No aneuploidy detected</b> Results consistent with two sex chromosomes (XY).
Microdeletions (1p36, 4p16.3, 5p15.2, 15q11.2, 22q11.2)	<b>NEGATIVE: No microdeletions detected</b> Results consistent with no microdeletions detected in the regions of interest (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.

## Limitations of the test

Noninvasive prenatal testing (NIPT) based on cell-free DNA analysis from maternal blood is a screening test; it is not diagnostic. Test results must not be used as the sole basis for diagnosis. Further genetic counseling and confirmatory diagnostic testing is necessary prior to making any irreversible pregnancy decision. Limited data exist regarding the clinical outcome of pregnancies with rare autosomal trisomies, especially for those detected through NIPT. Health care providers should be aware of the limitations of the test and understand that limited outcome data may confound pregnancy counseling and management.

## References

- Gardner RJM, Sutherland GR, Shaffer LG. *Chromosome Abnormalities and Genetic Counseling*. New York, NY: Oxford University Press; 2012.
- Practice bulletin no. 163: Screening for fetal aneuploidy. *Obstet Gynecol*. 2016;127(5):e123-137.

The Verifi™ and Verifi™ Plus Prenatal Test 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.