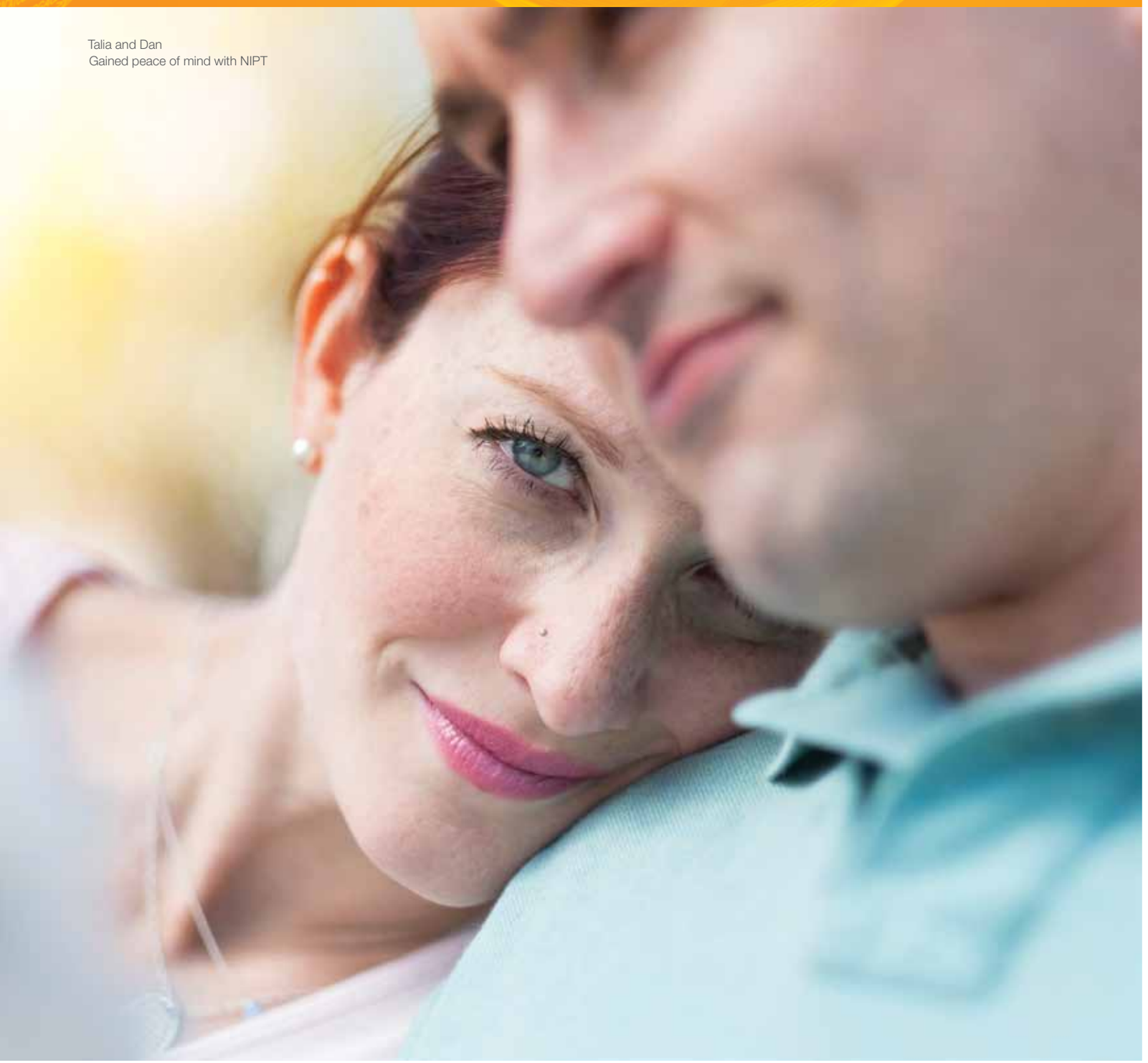
illumina®

verifi® Prenatal Test

A reliable, easy, fast noninvasive prenatal test



Talia and Dan
Gained peace of mind with NIPT

Why choose verifi?

- Proven superiority to traditional screening methods for the screening of common fetal aneuploidies, with reduced false positive rates (increased specificity) and increased positive predictive values (PPV)[1,2]
- Comprehensive portfolio with Expanded Panel available
- Fast turnaround time[7]
- Lowest published failure rate in the industry—0.1%[7-9]

0.1%
verifi® Prenatal Test[7]

1.3%
MateriT21 PLUS[8]

3.0%
Harmony Prenatal Test[6]

3.8%
Panorama Prenatal Screen[9]

FAILURE RATE

The verifi Prenatal Test has shown excellent performance in a real-life clinical population of over 86,000 patient samples.[7]

Total Cases	86,658
Average Turnaround Time (business days)	3.3
Technical Cancellations	0.1%
Aneuploidy Detected	2.2%
Aneuploidy Suspected	0.3%
Total Aneuploidy Detected or Suspected	2.5%
Observed False Positives*	0.12%
Observed False Negatives*	0.02%

*FP calculation is based on known outcome data.

The most accurate way to screen

Noninvasive prenatal testing (NIPT) is a cutting-edge, aneuploidy screening option. Professional societies, including the American College of Obstetrics and Gynecology (ACOG), have recommended NIPT as an option for all pregnant women, regardless of age or risk.[1,2] This screening option has a higher level of sensitivity and specificity than traditional serum screening.[1-3]

NIPT vs. traditional serum screening method:

- Offers the highest reported detection rate for Down syndrome[1]
- Offers the lowest reported false positive rate for Down syndrome[1]
- Offers the broadest screening window (performed as early as 10 weeks gestation until term)[1-3]

verifi® Prenatal Test

While there are different methods for performing NIPT, next-generation sequencing (NGS) is the most-published method.[4] It has demonstrated excellent detection rates and very low false positive rates.[5]

The verifi Prenatal Test from Illumina uses whole-genome next-generation sequencing (WGS) to screen for common fetal aneuploidies, with higher detection rates and significantly fewer false positives than traditional screening methods.[1-3,6]

Fast, reliable information about about common fetal aneuploidies

The verifi Prenatal Test safely and noninvasively screens for the most common chromosomal aneuploidies as early as 10 weeks gestation using a single maternal blood draw and offering the lowest test failure rate in the industry.

Test options

Basic offering:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Fetal sex

Optional add-on offerings

Sex chromosome aneuploidies:

- Monosomy X (MX; Turner syndrome)
- XXX (Triple X)
- XXY (Klinefelter syndrome)
- XYY (Jacobs syndrome)

Expanded autosomal trisomies:

- Trisomy 9 and Trisomy 16

Microdeletion syndromes:

- 1p36 deletion
- 4p- (Wolf-Hirschhorn syndrome)
- 5p- (cri-du-chat syndrome)
- 15q11 (Prader-Willi syndrome/Angelman syndrome)
- 22q11 deletion (DiGeorge)

Women whose results are not reported, indeterminate, or uninterpretable (a “no call” test result) from cell-free DNA screening should receive further genetic counseling and be offered comprehensive ultrasound evaluation and diagnostic testing because of an increased risk of aneuploidy.[1]

Reliable test results

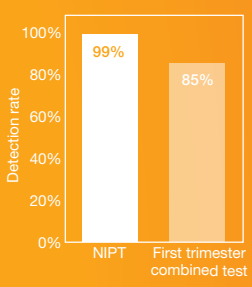
Test performance metrics are important for:

- Deciding which aneuploidy screening options to offer your patients
- Determining which laboratory to use
- Pre-test counseling about benefits and limitations of aneuploidy screening options (detection rates, false positive and false negative rates)
- Post-test counseling for patients with positive results about the likelihood of a true positive (positive predictive value)

Trisomy 21 Detection Rate: NIPT vs. First Trimester Combined Screening (FTS)[10]

NIPT detects more trisomy 21–affected pregnancies than other screening tests (eg, FTS)

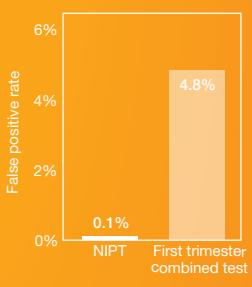
- *Detection Rate (Sensitivity)*—The proportion of affected pregnancies that will receive a positive screen result
- *False Negative Rate (1 – Sensitivity)*—The proportion of affected pregnancies that will receive a negative screen result. (ie, for trisomy 21, 1 in 100 affected pregnancies will not be detected with NIPT compared to 15 in 100 with FTS)



Trisomy 21 False Positive Rate: NIPT[1] vs. First Trimester Combined Screening[10]

NIPT has far fewer false positive results for trisomy 21 than other screening tests (eg, FTS)

- *False Positive Rate (FPR)*—The proportion of unaffected pregnancies that will receive a positive screening result
- *Specificity (1 – False Positive Rate)*—This indicates the proportion of unaffected pregnancies that will receive a negative screen result. (ie, 1 in 1000 unaffected pregnancies will screen positive for trisomy 21 with NIPT compared to 48 in 1000 with FTS)

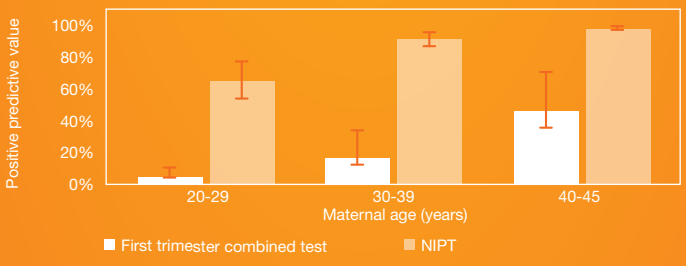


Trisomy 21 Positive Predictive Values: NIPT vs. First Trimester Combined Screening

Regardless of maternal age, NIPT has a higher PPV for trisomy 21 than other screening tests (eg, FTS)

Positive Predictive Value (PPV)

- Important test metric to consider after a patient has had aneuploidy screening
- The likelihood that a positive screen result is a true positive
- Depends on the test sensitivity, specificity, and the prevalence of the condition



PPVs calculated using sensitivity and specificity data from a NIPT meta-analysis[5] and a large study of first trimester combined screening[10] and published estimates of trisomy 21 prevalence at 10 weeks' gestation for different maternal ages.[11] PPV ranges, illustrated by error bars, were calculated using the 95% confidence intervals (CI) for sensitivity and specificity.

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 confirmatory testing is necessary prior to making any irreversible pregnancy decision.

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4. Data calculation on file. Illumina, Inc. 2016.
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8. McCullough RM, Almasri EA, Guan X, et al. Non-invasive prenatal chromosomal aneuploidy testing--clinical experience: 100,000 clinical samples. *PLoS One*. 2014;9(10):e109173.
9. Ryan A, Hunkapiller N, Banjevic M, et al. Validation of an enhanced version of a single-nucleotide polymorphism-based noninvasive prenatal test for detection of fetal aneuploidies. *Fetal Diagn Ther*. 2016;40(3):219-223.
10. Malone FD, Canick JA, Ball RH, et al. First-trimester or second-trimester screening, or both, for Down's syndrome. *N Engl J Med*. 2005;353(19):2001-2011.
11. Gardner RJM, Sutherland GR, Schaffer LG. *Chromosome Abnormalities and Genetic Counseling*. 4th ed. New York, NY: Oxford University Press; 2012.

The verifi® Prenatal Test was developed by, and its 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. It has not been cleared or approved by the U.S. Food and Drug Administration.

For more information visit illumina.com/verifi

A global genomics leader, Illumina provides comprehensive next-generation sequencing workflow solutions to the basic and translational research communities. Illumina technology is responsible for generating more than 90% of the world's sequencing data.* Through collaborative innovation, Illumina is fueling groundbreaking advancements in the fields of oncology, reproductive health, genetic disease, microbiology, agriculture, and forensic science.

*Data calculations on file. Illumina, Inc., 2015.

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