

illumina[®]

IT'S TIME FOR
NIPT

GET ACCURATE PRENATAL INSIGHTS
AS EARLY AS WEEK 10.¹

ACCURATE

EARLY

NONINVASIVE



Accurate insights. Available to all.

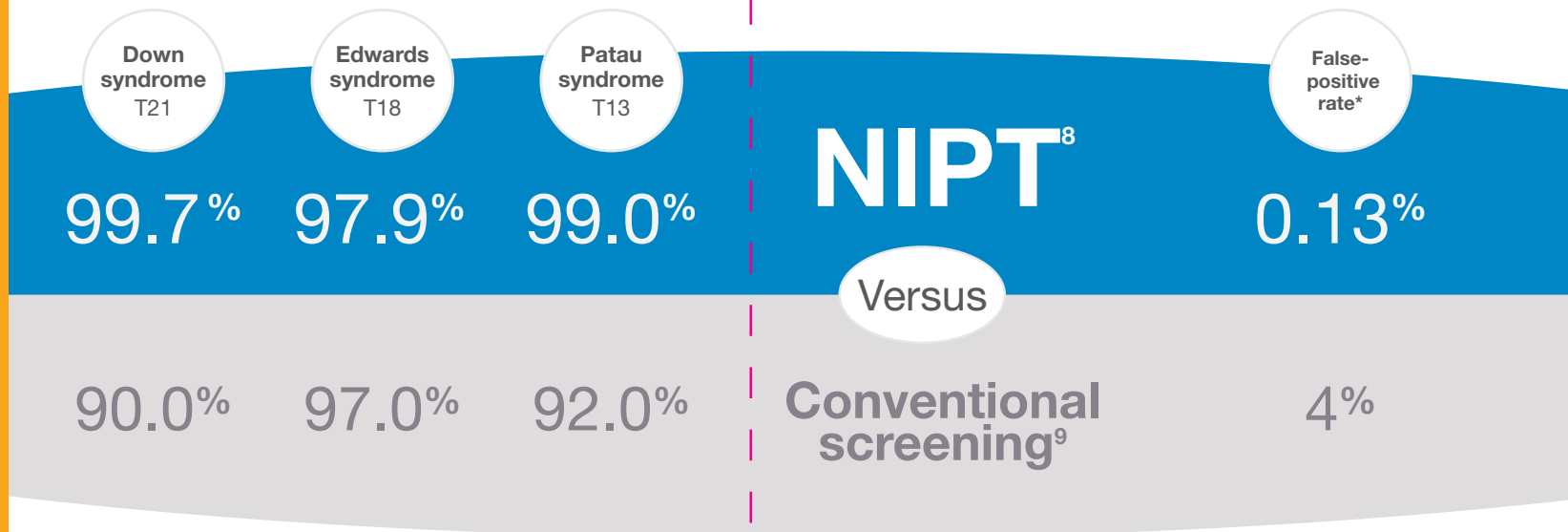
NIPT is a more accurate prenatal aneuploidy screening option than conventional prenatal serum screening, and is available for all pregnant women^{1,2,5-7}

NIPT data from a meta-analysis of the performance of NIPT screening for aneuploidies. Thirty-five studies conducted from January 2011 through December 2016 were included. The meta-analysis included peer-reviewed studies reporting on clinical validation or implementation of NIPT aneuploidy screening, in which data on pregnancy outcome were provided for >85% of the study population. These studies reported NIPT results in relation to fetal karyotype from invasive testing or clinical outcomes.⁸

Serum screening data from a prospective validation study screening for trisomies 21, 18, and 13 in 108,982 singleton pregnancies undergoing routine care in three hospitals. Subjects were screened using a combination of maternal age, fetal nuchal translucency, fetal heart rate, serum-free β-human chorionic gonadotropin, and pregnancy-associated plasma protein-A between 11 weeks 0 days and 13 weeks 6 days gestation. The detection rate and false-positive rate at estimated risk cut-offs from 1 in 2 to 1 in 1000 were determined. Rates shown are for risk cut-off of 1 in 100. The proportions of trisomies detected were compared to their expected values in different risk groups.⁹

HIGHER DETECTION RATES

LOWER FALSE-POSITIVE RATES



ACCURATE

EARLY

NONINVASIVE

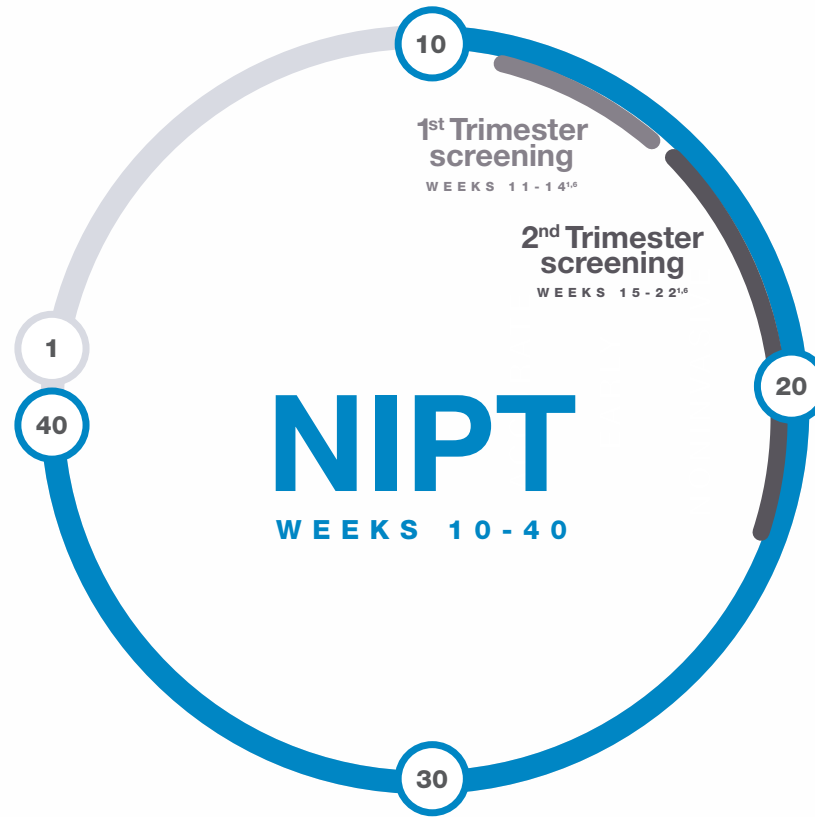
*False-positive rate shown is a combined rate for trisomies 21, 18, and 13.

NIPT can be used at any time in pregnancy, beginning as early as week 10^{1,6}



Insights earlier than ever before.

NIPT has the broadest screening window of any prenatal aneuploidy screening test^{1,6}



EARLY

NONINVASIVE

The high sensitivity and specificity of NIPT enable a reduction in confirmatory invasive procedures, their sequelae, and costs^{2,7,8,10-12}



Fewer invasive tests mean less maternal and fetal risk.

NIPT reduces the number of invasive confirmatory procedures performed in unaffected pregnancies^{2,7,8,10,11}

NUMBER OF UNNECESSARY INVASIVE PROCEDURES FOR T21, T18, AND T13 OUT OF 1000 PREGNANCIES

NIPT

False-positive rate: 0.13%⁸

~ 1

UNNECESSARY INVASIVE PROCEDURE



Conventional screening

False-positive rate: 4%⁹

40

UNNECESSARY INVASIVE PROCEDURES



Figures shown derived for a hypothetical population of 1000 pregnant women who would receive a false-positive result with each respective test, necessitating confirmatory diagnostic testing.

NONINVASIVE

IT'S TIME FOR NIPT



Screen for the presence of T21, T18, and T13 with the most accurate prenatal aneuploidy screening test available^{1,2,5-7}



Gain insights into prenatal genetic health risks as early as week 10¹



Reduce the number of invasive procedures in unaffected pregnancies^{2,7,8,10,11}

Limitations of Test

NIPT (noninvasive prenatal testing) based on cell-free DNA analysis from maternal blood is a screening test; it is not diagnostic. False-positive and false-negative results do occur. Test results must not be used as the sole basis for diagnosis. Further confirmatory testing is necessary prior to making any irreversible pregnancy decision. A negative result does not eliminate the possibility that the pregnancy has a chromosomal or subchromosomal abnormality. This test does not screen for birth defects such as open neural tube defects, or other conditions, such as autism. Some NIPT tests do not screen for polyploidy (eg, triploidy) or single-gene disorders. There is a small possibility that the test results might not reflect the chromosomal status of the fetus, but may instead reflect chromosomal changes in the placenta (ie, confined placental mosaicism [CPM]) or in the mother that may or may not have clinical significance.

References: 1. American College of Obstetricians and Gynecologists. Screening for fetal aneuploidy. Practice bulletin no. 163. *Obstet Gynecol.* 2016;127(5):e123-e137. 2. Bianchi DW, Parker RL, Wentworth J, et al; for CARE Study Group. DNA sequencing versus standard prenatal aneuploidy screening. *N Engl J Med.* 2014;370(9):799-808. 3. Farrell RM, Mercer MB, Agatisa PK, Smith MB, Philipson E. It's more than a blood test: patients' perspectives on noninvasive prenatal testing. *J Clin Med.* 2014;3(2):614-631. 4. Lewis C, Hill M, Chitty LS. Women's experiences and preferences for service delivery of non-invasive prenatal testing for aneuploidy in a public health setting: a mixed methods study. *PLoS One.* 2016;11(4):e0153147. doi: 10.1371/journal.pone.0153147. 5. Benn P, Borrell A, Chiu RWK, et al. Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn.* 2015;35(8):725-734. 6. Gil MM, Accurti V, Santacruz B, Plana MN, and Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol.* 2017;50:302-314. 7. Chudova DI, Sehnert AJ, Bianchi DW. Copy-number variation and false positive prenatal screening results. *N Engl J Med.* 2016;375(1):97-98. 8. Gil MM, Quezada MS, Revello R, Akolekar R, Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for fetal aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol.* 2015;45(3):249-266. 9. Santorum M, Wright D, Synglaki A, Karagiorgi N, Nicolaides KH. Accuracy of first-trimester combined test in screening for trisomies 21, 18 and 13. *Ultrasound Obstet Gynecol.* 2017;49(6):714-720. 10. Platt LD, Janicki MB, Prosen T, et al. Impact of noninvasive prenatal testing in regionally dispersed medical centers in the United States. *Am J Obstet Gynecol.* 2014;211(4):368.e1-368.e7. 11. Larion S, Warsof SL, Romary L, Mlynarczyk M, Peleg D, Abuhamad AZ. Association of combined first-trimester screen and noninvasive prenatal testing on diagnostic procedures. *Obstet Gynecol.* 2014;123(6):1303-1310. 12. Taneja PA, Snyder HL, de Feo E, et al. Noninvasive prenatal testing in the general obstetric population: clinical performance and counseling considerations in over 85,000 cases. *Prenat Diagn.* 2016;36(3):237-243. doi: 10.1002/pd.4766.

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