PREMIER GENOMICS

NON-INVASIVE PRENATAL TESTING

Get accurate answers about fetal chromosomal health, sooner and help avoid the risks associated with amniocentesis or CVS

10 WEEKS

Performed as early as 10 weeks gestation, Non-Invasive Prenatal Testing (NIPT) is an early adjunctive screening option for pregnant women.

It demonstrates superb sensitivity and specificity for the most prevalent aneuploidies compared to conventional maternal serum screening tests [1,2] which may help limit the need for amniocentesis or chronic villus sampling (CVS).

Using cell-free fetal DNA circulating in maternal blood, and performing whole genome massively parallel sequencing with a proprietary algorithm, this test has the lowest failure rate (0.1%) compared to other non-invasive prenatal tests.[1] It has been validated in high-risk and low-risk patient populations.[1-4]

NIPT SCREENING SPECIFICATIONS

Aneuploidy	Observed Sensitivity[1]	Observed Specificity[1]
Trisomy 21 (Down Syndrome)	99.5%	99.8%
Trisomy 18 (Edwards Syndrome)	97.2%	99.7%
Trisomy 13 (Patau Syndrome)	98.0%	99.8%

OPTION TO TEST FOR THE MOST COMMON SEX ANEUPLOIDIES

Karyotype	Observed Sensitivity[5]	Observed Specificity[5]
Monosomy X (Turner Syndrome)	95.0%	99.0%
XX (Normal Female)	97.6%	99.2%
XY (Normal Male)	99.1%	98.9%

ADDITIONAL SCREENING OPTIONS

Trisomy 9, Trisomy 16

Microdeletion Syndromes:

22q11.2 syndrome
1p36 deletion syndrome
Angelman/Prader-Willi Syndrome
Cri-du-chat Syndrome
Wolf-Hirschhorn Syndrome

NIPT SCREENING INDICATIONS

The American College of Obstetricians and Gynecologists recognizes the importance of offering aneuploidy screening or diagnostic testing to all women early in pregnancy[6]. The International Society of Prenatal Diagnosis specifically recognizes NIPT as prenatal screening option for all pregnant women[7] consistent with a large health insurance carrier coverage benefits[8]. This testing is recognized as an important advancement in prenatal screening methodology[9]and may be considered in pregnancies at increased risk for aneuploidy.[10,11]

Indication	Single	Twins
Consider in average risk	✓	
Advanced Maternal Age	≥35	≥32
Positive Serum Screen	✓	✓
Abnormal Ultrasound	✓	✓
Personal/Family history suggestive of Trisomy 21, 18 or 13	✓	✓
Personal/family history of sex chromosome aneuploidy	✓	Only valid for Y chromosome detection

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TEST RESULTS AND MEDICAL MANAGEMENT

	Trisomy 21,18,13	X, XXX, XXY, XYY	Interpretation	Implication[10-12]
No Aneuploidy Detected			Screening for Aneuploidy is negative	Very small possibility of a false negative. Discuss false negative with patient.
Aneuploidy Detected			Screening for Aneuploidy is positive	Possibility of a false positive (variable based on a priori risks) **OFFER CONFIRMATORY TESTING BY DIAGNOSTIC AMNIOCENTESIS/CVS**
Aneuploidy Suspected		Not Reported	Screening for Aneuploidy is suspected	**OFFER CONFIRMATORY TESTING BY DIAGNOSTIC AMNIOCENTESIS/CVS**

- If no aneuploidy detected, and fetal anomalies present on ultrasound, consider invasive testing, as this test does not test for all chromosome anomalies[10] or neural tube anomalies. Consider biochemical testing (AFP)/ultrasound screening in addition to NIPT.[10] If NIPT analysis results in test failure, ACOG recommends considering diagnostic testing.[10]
- The positive predictive value (PPV) (chances that if the NIPT is aneuploidy detected or suspected the diagnostic test will be concordant) varies depending on the risk of aneuploidy prior to undergoing NIPT. If performed in a high-risk population, the PPV is high.[12] If performed in the average risk population, the PPV is lower.[3] In a study of mostly high risk population, the PPV for trisomy 21 was 91%[12] and a study in an average risk population the PPV for trisomy 21 was 45.5%. This is 10 times higher than serum screening.[3]

NIPT TESTING PROCESS

THE PROCESS	A blood sample (7-10ml) is collected using the provided Streck™ BCT tube. Testing is not constrained by BMI, ethnicity, paternal sample or assisted reproductive technology. Can be performed in pregnancies with egg donors and twins.
THE TURN-AROUND TIME	3-5 days
THE REPORT	A comprehensive report is generated and uploaded to a secure portal with dedicated physician log-in and downloading capabilities. Physician-to-physician consultation is also available with our Medical Director, or with our Genetic Counselor.

Talk to your representative or visit our web site for our full catalog of genetic testing solutions.

Personalized Genetic Medicine // Inherited Genetic Disorders // Women's Genetic Health

ABOUT PREMIER GENOMICS

Premier Genomics is committed to advancing the field of personalized genetic medicine by offering cutting-edge genetic screening services to help practitioners and their patients in pursuit of tailored treatment and optimized, personalized health care. We work together with patients and their insurance providers to help ensure that access to these important genetic tests does not cause patients financial hardship.

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CLIA-accredited, CAP-certified clinical laboratory

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