

The MaterniT21 PLUS test provides you with clear, powerful test results.

### AN INCREASED RISK FOR A CHROMOSOMAL ABNORMALITY.

Anyone can have a pregnancy with a chromosomal abnormality—healthy women, mothers of all ages and all ethnicities can be at-risk. There is nothing a parent can do to cause a chromosomal abnormality and there is nothing a parent can do to prevent it. Certain factors can increase the chance to have a pregnancy with chromosomal abnormalities, including:

-  Advanced maternal age
-  Personal or family history of chromosomal abnormalities
-  Fetal ultrasound abnormality suggestive of chromosomal abnormalities
-  Abnormal serum screening test

The MaterniT21 PLUS test uses cutting edge, state-of-the-art science. Results are reported simply, with a positive or negative result. Most results are available 7 business days after the sample is received in the laboratory.

It is important to keep in mind that although the discussion of risks to the pregnancy may be necessary, most pregnancies have perfectly normal outcomes. Sometimes more information about the pregnancy can provide you clarity. The MaterniT21 PLUS test does just that—provides you and your health care provider with clear, powerful test results.

No test is perfect. While results of this testing are highly accurate, false positive and false negative results may occur in rare cases. A negative result does not ensure an unaffected pregnancy. The results of this testing, including the benefits and limitations, should be discussed with your health care provider.

Sequenom Laboratories, a wholly-owned subsidiary of Sequenom, Inc., is a CAP-accredited and CLIA-certified molecular diagnostics laboratory dedicated to improving patient outcomes by offering revolutionary laboratory-developed tests for a variety of prenatal and eye conditions. Sequenom Laboratories pioneered NIPT for fetal aneuploidies with the launch of its MaterniT21™ PLUS test, and offers a full menu of prenatal tests.

The MaterniT21™ PLUS test is a laboratory-developed test that was developed, validated and is performed exclusively by Sequenom Laboratories in the USA.

### REFERENCES

1. Palomaki GE, Kloza EM, Lambert-Messerlian GM, Haddow JE, et al. DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. *Genet Med.* 2011;13(11):913-920.
2. Palomaki GE, Deciu C, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, Ehrich M, van den Boom D, Bombard AT, Grody WW, Nelson SF, Canick JA. DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13, as well as Down syndrome: An international collaborative study. *Genet Med.* 2012;14(3):296-305.
3. Mazloom AR, Dzakula Z, Oeth P, Wang H, et al. Noninvasive prenatal detection of sex chromosomal aneuploidies by sequencing circulating cell-free DNA from maternal plasma. *Prenat Diagn.* 2013;33(6):591-597.
4. Sheets KB, Crissman BG, Feist CD, Sell SL, et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of down syndrome: Recommendations of the National Society of Genetic Counselors. *J Genet Couns.* 2011;20(5):432-441.

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MaterniT21™  
PLUS

## Noninvasive prenatal testing for fetal chromosomal abnormalities

  
**THE SCIENCE OF RESULTS YOU CAN TRUST**



MaterniT21™  
PLUS

## NONINVASIVE PRENATAL TESTING FOR FETAL CHROMOSOMAL ABNORMALITIES

### KNOWLEDGE IS EMPOWERING.

Being told by your health care provider that your pregnancy may have an “increased risk” is not an easy thing to hear. Not for the mother who is 35 years or older. Not for the couple with abnormal screening test results. Not for the family whose fetal ultrasound showed something different.

For many years, prenatal testing choices were limited. Either screening tests with little risk but limited accuracy were available, or “invasive” procedures with great accuracy but risks to the pregnancy could be chosen. Now, important complementary testing is available.

The MaterniT21 PLUS test is a “noninvasive” approach to provide a high degree of accuracy for select chromosome information, without the risk of miscarriage associated with an invasive procedure. Using premier technology, information is unlocked with the ease of a blood draw.

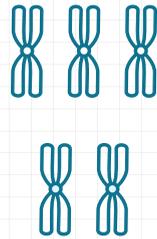
The MaterniT21 PLUS test offers a powerful glimpse into the pregnancy by evaluating the relative amount of chromosomal material. Analysis of chromosomes 21, 18, 13, X, Y, or other additional information can be requested by your health care provider.<sup>1,2,3</sup>

## UNDERSTANDING CHROMOSOMES.

Chromosomes are structures inside cells. They contain genes that instruct the body how to grow and develop. You inherit genes from your father and your mother.

Most people have 23 pairs of chromosomes, which each carry thousands of genes. The first 22 pairs are called autosomes, and are the same in males and females. The 23rd pair are chromosomes related to gender—the X and Y chromosomes. Females usually have two Xs, and males have one X and one Y.

Some people are born with an extra or missing chromosome. Having three copies of a chromosome instead of two is known as having a trisomy (tri: three, some: chromosomes). The most common fetal trisomies are trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome). People can also be born with an extra or missing gender chromosomes. An irregular number of chromosomes is a serious health concern for the baby.



MaterniT21<sup>™</sup>  
PLUS

10  
WEEKS'  
GESTATION

Important,  
timely information



Without the  
risks of prenatal  
diagnosis procedures

7  
BUSINESS  
DAY  
TURNAROUND

Clear, powerful  
test results

## CLEAR RESULTS.

The MaterniT21 PLUS tests measures the relative amount of chromosome material in maternal blood. This can be done as early as 10 weeks' gestation and without the risks associated with an invasive procedure. Some of the things the test looks for include:

### TRISOMY 21 (DOWN SYNDROME)

**21** Down syndrome is a condition caused by an extra copy of chromosome 21. Children with Down syndrome have delays in both intelligence and development. Babies with Down syndrome also have higher chances for certain health problems. The chance to have a child with Down syndrome increases with the woman's age, but women of all ages and ethnicities can have a child with Down syndrome.<sup>4</sup>

### TRISOMY 18 (EDWARDS SYNDROME)

**18** Trisomy 18, also known as Edwards syndrome, is caused by an extra copy of chromosome 18. Babies with trisomy 18 often have multiple birth defects, and many don't survive the first few months of life.

### TRISOMY 13 (PATAU SYNDROME)

**13** Trisomy 13, is caused by an extra copy of chromosome 13. Like babies with trisomy 18, these babies have multiple birth defects and often don't survive the first few months of life.

## TURNER SYNDROME (X)

Turner syndrome is caused when a girl only has one copy of the X chromosome. Many of these pregnancies face high risks of miscarriage. Girls with Turner syndrome are usually shorter than average, have delayed or absent puberty and may be infertile. Most have normal intelligence, but some have learning difficulties. Children with Turner syndrome may also have heart or kidney defects.

## KLINEFELTER SYNDROME (XXY)

A boy with Klinefelter syndrome has two X chromosomes and one Y. These boys tend to be taller than average, may have delayed or absent puberty and are often infertile. Most have normal intelligence, but some may have learning or psychological difficulties.

## TRIPLE X (XXX) AND XYY SYNDROME

Children with these conditions may be taller than average and usually have normal intelligence. A few may have learning or psychological issues. These conditions are not associated with birth defects and may go undiagnosed. People with these conditions may have normal fertility.

These are some of the more common conditions the MaterniT21 PLUS test can highlight. Other more rare conditions can also be identified and can be discussed further with your doctor. It is important to remember that there is no perfect test to detect all birth defects. The results of the MaterniT21 PLUS test, though highly accurate, are not considered diagnostic. Speaking with your health care provider about the benefits and limitations of this test can be an important conversation.

## ADDITIONAL RESOURCES

**Global Down Syndrome Foundation**  
3300 East First Avenue, Suite 390, Denver, CO 80206  
T 303.468.6667 [www.globaldownsyndrome.org](http://www.globaldownsyndrome.org)

**Klinefelter Syndrome & Associated Disorders (KS&A)**  
P.O. Box 872, Pine, CO 80470-0872  
T 888.999.9428 F 303.838.0753 [www.genetic.org](http://www.genetic.org)

**March of Dimes**  
1275 Mamaroneck Avenue, White Plains, NY 10605  
T 914.997.4488 [www.marchofdimes.com](http://www.marchofdimes.com)

**National Down Syndrome Congress**  
30 Mansell Court, Suite 108, Roswell, GA 30076  
T 800.232.NDSC (6372), 770.604.9500 [www.ndsccenter.org](http://www.ndsccenter.org)

**National Down Syndrome Society**  
666 Broadway, 8th Floor, New York, NY 10012  
T 800.221.4602 [www.ndss.org](http://www.ndss.org)

**National Society of Genetic Counselors**  
401 N. Michigan Avenue, 22nd Floor, Chicago, IL 60611  
T 312.321.6834 [www.nsgc.org](http://www.nsgc.org)

**SOFT USA (Society for Trisomy 13, 18 and Related Disorders)**  
2982 South Union Street, Rochester, NY 14624  
T 800.716.SOFT (7638), 585.594.4621 [www.trisomy.org](http://www.trisomy.org)

**Trisomy 18 Foundation**  
4491 Cheshire Station Plaza, Suite 157, Dale City, VA 22193  
T 810.867.4211 (Mon–Fri 9am–5pm ET) [www.trisomy18.org](http://www.trisomy18.org)

**Turner Syndrome Society**  
11250 West Road, Suite #G, Houston, TX 77065  
T 800.365.9944, 832.912.6006 F 832.912.6446  
[www.turnersyndrome.org](http://www.turnersyndrome.org)



For more information, ask  
your health care provider or visit  
[sequenom.com/laboratories](http://sequenom.com/laboratories)