Noninvasive prenatal testing for fetal chromosomal abnormalities

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

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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, soma: chromosome). 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.

CLEAR RESULTS.

The MaterniT21 PLUS test 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)

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.

TRISOMY 18 (EDWARDS SYNDROME)

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)

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 (0)

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 XY 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 300, Denver, CO 80206
T 720.408.0067 www.globaldownsindrome.org

Klinefelter Syndrome & Associated Disorders (KS&A)
P.O. Box 872, P.O. Box 89470-0872
T 888.390.0438 F 703.838.0763 www.ksa.org

March of Dimes
1275 Merionover Avenue, White Plains, NY 10605
T 914.297.4488 www.marchofdimes.com

National Down Syndrome Congress
30 Merrick Court, Suite 108, Roswell, GA 30076
T 1.800.232.1065 (6317), 770.634.0500 www.ndsc.org

National Down Syndrome Society
665 Broadway, 8th Floor, New York, NY 10012
T 1.800.221.4602 www.ndss.org

National Society of Genetic Counselors
401 N. Michigan Avenue, 22nd Floor, Chicago, IL 60611
T 312.321.6834 www.nsgc.org

SOFT USA (Society for Trisomy 13, 18 and Related Disorders)
2982 South Union Street, Rochester, NY 14624
T 1.800.799.2244 (9624), 585.594.4627 www.soft.org

Turner Syndrome Society
3250 West Road, Suite #G, Houston, TX 77065
T 832.644.6446 www.turnersyndrome.org

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