



Genetic Testing - What is Trisomy 21, 18 and 13?

Genes are the part of the cell that contain the biological information that control the growth and development of cells. Genes, alone or in combination, determine many of the genetic traits that a person inherits, including risks of developing certain diseases or conditions. Genes are grouped into larger structures within the cells called chromosomes. The human body has 23 distinct chromosome pairs or copies. Trisomy 21, 18 or 13 means there are three copies of that particular chromosome instead of two.

Down syndrome is a condition in which an extra copy of chromosome 21 causes delays in the way a child develops, both mentally and physically. Not everyone with Down syndrome is affected in the same way, and there is no way to determine before birth how a child may be affected. There are three different types of Down syndrome with the most common referred to as trisomy 21. The other two types are called Robertsonian translocation and Mosaicism and these occur in about 4-6% of babies born with Down syndrome. Down Syndrome affects about 1 in every 700 babies. The risk to have a child with Down syndrome does increase with the mother's age, but mothers of all ages can have a child with Down syndrome and it can occur in people of all races.

Trisomy 18, also known as Edwards syndrome, is caused by an extra copy of chromosome 18. Trisomy 18 occurs in about 1 in every 5,000 babies born. Trisomy 13, also known as Patau syndrome, is caused by an extra copy of chromosome 13. The condition is much less common and occurs in about 1 in 16,000 babies born. The MaterniT21 PLUS laboratory developed test is a noninvasive blood test that is available for women with increased risk indicators for fetal chromosome variations at 10 weeks of pregnancy and beyond. This test detects an increased amount of chromosomal 21, 18, 13 material that is circulating in maternal blood. The increased risk factors include one or more of the following:

Advanced maternal age, Positive serum screening test, Fetal Ultrasound abnormality suggestive aneuploidy, and/or Personal/family history of chromosomal abnormalities.

Sequenom offers MaterniT21 and will process billing with your commercial insurance.

Because insurance coverage (deductibles, coinsurance, copays, etc.) varies from patient to patient, it is important to learn about your financial obligations before your insurance is billed.

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Sequenom can process your verification of benefits before or after you have your blood drawn. **If you decide to call after the blood is drawn, please call Sequenom within 2 days for verification of benefits.** Should your financial obligation for commercial insurance be significant, then Sequenom will give you the opportunity to use its uninsured Patient Assistance Program. This option is dependent on Family Size and Income Level. If you decide against using your insurance or the Patient Assistance Program, then you may cancel your test and you or your insurance will not be billed. Sequenom will alert your provider that your test has been canceled. Medicaid is accepted with zero patient responsibility and uninsured patients may utilize the Patient Assistance Program. **All MaterniT21 PLUS billing matters are to be directed to Sequenom at 1-877-821-7266 option #3.**

Prenatal testing for trisomy 21, 18 and 13 can help determine whether your baby has a genetic abnormality. Knowing the risk for trisomy 21, 18 and 13 can help you, your family and your health care provider make informed decisions about your pregnancy. With your health care provider, you can prepare medically, emotionally and financially for the birth of a child with special needs, such as arranging for delivery in a medically appropriate setting. Please discuss your testing options with your health care provider.

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