MaterniT21 Testing

What is the MaterniT21 test?

This test is a new blood test conducted on the mother. It analyzes the amount of chromosome 21 in fetal DNA picked up from the maternal blood sample. It can detect an increased amount of chromosome 21 material which is associated with trisomy 21 (Down syndrome). It is completely non-invasive as it only involves a blood sample from the mother.

Who should be tested?

This test was developed and tested for pregnant women with one or more of the following:

1. Advanced maternal age (35 or older)
2. Fetal ultrasound abnormality suggestive of chromosomal abnormality
3. Positive 1st or 2nd trimester screening test
4. Personal or family history of Down syndrome.

When can I have this test?

This test can be done as early as 10 weeks gestation. Results are available in about two weeks. Genetic counseling is recommended to further discuss the risks, benefits and alternatives of the various prenatal screening methods in this group of high risk women for whom this testing may be considered.

How are the test results given?

The test results are given as positive or negative. The test has a sensitivity of 99.1% and a specificity of 99.9%.

What if I have a positive result?

A diagnostic test (CVS or amniocentesis) is recommended to confirm this blood test.

How do I interpret a positive California NT Screening test and a negative MaterniT21?

Multiply the result by 72. If you have a 1 in 80 risk of Trisomy 21 with the California Prenatal screening test, the new risk becomes a 1 in 5760. With a risk of 1 in 80 (1.25% chance of Down syndrome), there is a 98.75% chance the baby does not have Trisomy 21. With the addition of a negative MaterniT21 the risk decreases to .01%.

Does the MaterniT21 test for other conditions?

Right now, this blood test is only approved to test for Chromosome 21. It is also currently being developed to test for trisomy 13 and 18. If abnormal amounts of these chromosomes are found, the company will report these findings as well. This test does not detect all of the types of chromosome problems that genetic amniocentesis or CVS can detect.

How much does it cost?

With PPO insurance, the cost is $235 out of pocket.