

# FIRST TRIMESTER GENETIC SCREENING



Genetic screening allows us to identify if your baby is at higher risk for Down Syndrome (Trisomy 21), Trisomy 18 and Trisomy 13. Although results are **not** a definite yes or no, first trimester risk assessment detects nearly 90% of babies with Down Syndrome and about 97% of babies with Trisomy 18 or 13.

Each person has a genetic code that serves as a map for how their body will develop. In these disorders three pieces of genetic information are located in spots on the map where only two pieces of information should be. In Trisomy 21, this can be associated with birth defects, mild to severe mental retardation, and other medical problems. Trisomy 13 and 18 are much more severe disorders where the babies have profound mental retardation and birth defects that seriously effect the life expectancy of the baby.

## Some important facts about first trimester genetic testing:

- Anyone can have a baby with these genetic abnormalities but the chance of doing so increases with the mother's age
- First trimester genetic screening is not designed to provide information on genetic conditions other than Trisomy 21, 18, and 13
- For statistical reasons, we do not recommend routine second trimester genetic screening when the first trimester result is reassuring.
- The test has two parts: an ultrasound to measure the thickness of skin behind the baby's neck and the measurement of two pregnancy hormone levels in the mother's blood
- The blood test is a simple finger stick
- The combined information from the blood test and ultrasound will produce your baby's calculated risk for Trisomy 21, 18 or 13

## Timing is important!

- The test must be completed between 11 and 13 weeks of your pregnancy
- Please discuss this test with your family and inform your health care provider as soon as possible if you wish to have first trimester genetic testing.

## The results:

- Your healthcare provider will discuss the results of your genetic testing with you.

## If your test results are not reassuring:

- A more detailed ultrasound may be performed
- We may consult a high risk pregnancy specialist to assist us with your care
- We may discuss further testing options with you that can give a more definite result

As always,  
we are  
here to  
discuss  
any of your  
concerns!

**697-2035**