OB/GYN Clinic

Genetic Screening Information

Do I have to have these tests?

Although screening tests for birth defects are offered to all pregnant women, it is your choice whether to have them done. Knowing whether your baby is at risk of or has a birth defect beforehand allows you to prepare for having a child with a particular disorder and to organize the medical care that your child may need. You also may have the option of not continuing the pregnancy.

If you decide to have screening tests, there is a possibility of **false-positive and false-negative results.** A test result that shows there is a problem when one does not exist is called a false-positive result. A test result that shows there is not a problem when one does exist is called a false-negative result.

> A false-positive result can cause anxiety and may lead to unnecessary testing or treatment.
>
> A false-negative result can mean that you do not get the recommended counseling or preparation for having a child who has a medical condition or disability.

Types of screening tests:

The Integrated and Quad Screenings are the 2 most common tests offered to pregnant women. Neither tests gives a "Positive, YES" or "Negative, NO" type of answer. Instead, it gives you a calculated rate or possibility.

> For example: Your baby has a 1 in 50 chance of having the "ABC" genetic disorder. This means out of every 50 babies born, 1 baby will most likely have the disorder.

Also know that these tests are not 100% accurate. Instead, they are only about 81% right. This percentage right can be more or less depending on other things like your age, and which genetic disorder is being tested.

What factors may increase my risk of passing on a genetic disorder?

Most babies with birth defects are born to couples without risk factors. However, the risk of birth defects is higher when certain factors are present. You are at increased risk if

- ✓ You have a genetic disorder
- ✓ You already have a child who has a genetic disorder
- ✓ You are of advanced maternal age
- ✓ There is a family history of a genetic disorder
- ✓ You belong to an ethnic group that has a high risk of certain genetic disorders

INTEGRATED SCREENING: Detection rate is 82% and higher

There are 2 different times you have to go to the lab for this test. Also, you will not get results until at least 1 week after you complete the 2nd part of the test.

There are (2) different test rules to follow for Part 1 of this test. It is based on where you are in your pregnancy.

- If you are 13 weeks pregnant or more, you will only be sent to the lab.
- If you are less than 13 weeks pregnant, you can have something called a Nuchal Translucency (NT) screening. This is a type of ultrasound that looks at a very specific part of your baby.

Part 1 A: You are between: 10 weeks + 4 days pregnant and 12 weeks + 6 days pregnant
- ✓ Complete a NT screening
- ✓ Go to the lab the same day, AFTER you have your NT screening to have your blood drawn (you cannot do the lab first or on a different day!)

OR

Part 1 B: You are between: 13 weeks + 0 days pregnant and 13 weeks + 6 days pregnant
- ✓ Complete a lab draw order (only)

AND

Part 2: You will go to the lab to have your blood drawn when you are between: 15 weeks + 0 days pregnant and 21 weeks + 0 days pregnant
(Recommended best time for Part 2 of this test is between 16 -18 weeks)

If you are 14 weeks + 0 days or more pregnant this test cannot be completed, and will not be an option for you to choose.

QUAD SCREENING: Detection rate is 81% and higher for the things they check for

This test is a one-time lab test. You will go to the lab to have your blood drawn when you are between: 15 weeks +0 days and 22 weeks + 0 days of pregnancy

***You will need a paper with the lab orders on it from your doctor to have either of these test done! The lab will not accept you if you do not have the paper form, completed by your doctor.

***They will be in your Family Medicine 12 and 16 week Routine OB Patient Packets, which will be reviewed and given to you if you need it at your 12 and 16 week appointments.