Optional Prenatal Screening Tests

Prenatal screening tests can detect an increased risk of having a baby with an extra chromosome (trisomy), certain birth defects or other genetic disorders.

You will be offered full genetic counseling at Maternal Fetal Medicine if you:

- Are going to be 35 years or older by your due date.
- Have had a previous child with certain birth defects.

They will review all your genetic testing options and risk factors at that visit.

Screening tests do not tell you *for sure* if your baby has one of these issues. Instead, a positive screening test tells you that your baby *might* have an issue. A negative screening test tells you that your baby *probably does not* have an issue. If you have a positive screening test, other testing will be discussed with you.

Please watch the video at the link below before your appointment. In this video, our genetic counselor explains genetic testing and what your options are in pregnancy.

https://youtu.be/acL5iJi9ZnM

Most common genetic disorders:

- **Down syndrome (Trisomy 21).** Down syndrome is a chromosomal disorder that causes lifelong disability, developmental delays and, in some people, health problems.
- **Trisomy 18 and Trisomy 13.** These are chromosomal disorders that cause severe developmental delays and defects in the structure of the body. These are often fatal by age 1.
- **Neural tube defects:**
    - Spina bifida is a birth defect that occurs when a portion of the neural tube fails to develop or close properly. This causes defects in the spinal cord and in the bones of the spine.
    - Anencephaly is when a baby is born with an underdeveloped brain and an incomplete skull.

Screening Tests Include:

Sequential Screen: This test screens for increased risk of Down Syndrome, Trisomy 18, Trisomy 13 and neural tube defects. It includes two blood tests, the first done between 10 and 13 weeks and the second between 15 and 21 weeks. It also includes an ultrasound to measure a fold of skin on the baby's neck, called the nuchal fold. This ultrasound is done on the same day as the first blood draw. This test will detect 9 out of 10 babies with Down Syndrome and 8 out of 10 babies with neural tube defects.

Quad Screen: This test screens for increased risk of Down Syndrome, Trisomy 18, Trisomy 13 and neural tube defects. It includes a single blood draw between 15 and 21 weeks and does not include an ultrasound. This test will detect 8 out of 10 babies with Down Syndrome. The quad screen detects 92-95 percent of spina bifida and 100 percent of anencephaly.

MaterniT 21: This test involves a single blood draw that can be done any time in the pregnancy after 9 weeks gestation. It tests for Down Syndrome, Trisomy 18 and Trisomy 13. This test can tell you the sex of the baby. This test will detect 99 percent of babies with Down's syndrome, Trisomy 18 and Trisomy 13. This test **does not** screen for neural tube defects.

Ultrasound: You will have a routine ultrasound at 20 weeks gestation to check your baby's health and growth. This ultrasound will include a general survey of your baby including the brain, heart, kidneys, bladder, face, arms and legs. It can usually determine your baby's sex. This ultrasound can detect severe birth defects and growth issues. The ultrasound has a 95-98 percent detection rate for neural tube defects but only a 60 percent detection rate for Down's syndrome. The detection rate of Trisomy 18 and Trisomy 13 is more than 95 percent with ultrasound.

Carrier Testing: This test involves a blood draw and can be done at any time in your pregnancy or prior to becoming pregnant. The test will determine if you carry one of the following genes: Cystic Fibrosis (CF), Fragile X syndrome or Spinal Muscular Atrophy (SMA). A baby must get a gene from both parents. If your test shows that you do carry one of the genes, then we can order a test for the baby's father. If both parents carry the gene of one of these conditions, the baby will have a 25% chance (1 out of 4) of having that specific disorder. You will be given more information regarding carrier screening at your pre-admit appointment.

How to decide whether to have genetic testing:

There is not one right answer for all pregnant women. These tests are offered to all pregnant women. It is your choice if you have any of these tests done. Some things to think about when making the decision are:

- If you feel your risk of having a baby with a problem is very low. You may not want the tests.

- You may feel you will only worry more if a test comes back abnormal. You would rather not have any extra tests done.

- If you want to be prepared emotionally or gather information if your baby has a genetic problem. You may want to do the tests.

- If there is a serious problem with your baby, early screening tests can be done to help you decide whether to continue the pregnancy.

Insurance Coverage: It is a good idea to check with your insurance regarding coverage for prenatal screening tests. You will need to know the charge codes when you talk to your insurance company.

Quad Screen

CPT code - 81511

Sequential Screening

Part 1: 10-13 weeks gestation
CPT Codes - 76801, 76813, 84163, 84702

Part 2: 15-21 weeks gestation
CPT Codes - 82105, 82677, 84702, 86336

MaterniT 21

- You can go to the company's website and enter your insurance information to see how much you might need to pay **– https://integratedgenetics.com/billing.**
- Or you can also call 269-341-7723 and select option 1 to talk to someone about how much you might have to pay.
  CPT Code - 81420

Carrier Test

CPT code – 81265, 81220

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