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Obstetrics & Gynecology

Genetic Screening Options

At this current moment in obstetrical practice, we have a variety of tools to help us answer the common question from our patients- - "Is my baby going to be healthy?" While there is no test or set of tests that can tell us a fetus is going to be 100% "normal", there are tests which can test for the most common abnormalities seen in a developing pregnancy. This includes screening for aneuploidies, carrier status, open neural tube defects, and anatomic abnormalities (further described below). Ultimately, deciding which testing you want is up to your preferences. Some patients will decide not to do any testing, others may choose to do what is best covered by their insurance, and others may choose to do any and all tests offered regardless of insurance coverage. We hope to be your guide as you consider your options and encourage you to make the decision that feels best for your growing family.

Aneuploidy Screening:

When the genetic material of an egg and a sperm come together, there should be 46 chromosomes which make up 23 pairs. The most common chromosomal abnormality that can occur is when an extra chromosome is present, making a "trisomy" or a set of three of a chromosome. Trisomy is a type of aneuploidy.

There are a few different options for genetic screening for chromosomal problems. Your provider will discuss these with you at your first appointment, however it is a good idea to start thinking about your options. Understand that these are SCREENING tests only. Currently, the type of testing recommended for individuals is based on their age. Some patients will further decide which testing they want based on how accurate the test result is. Confirmatory or diagnostic testing (e.g., chorionic villus sampling or amniocentesis) is not provided in our office but we can refer you to Maternal Fetal Medicine for genetic counselling and testing.

Please contact your insurance company to inquire if these are covered services (See below CPT codes). Please notify our office if your insurance requires further follow-up.

Aneuploidy screening options recommended for those less than 35 years of age:

Sequential Screen (CPT codes 84163, 84702, 82105, 82677, 86336, 76801, 76813)

Screens for Trisomy 21, 18 and open neural tube defects (like Spina Bifida) in two parts

Part one: Bloodwork and ultrasound between 11weeks 0days and 13weeks 6days gestation

Part two: Bloodwork drawn at your ~ 16-week appointment.

ATTENTION: We may not be able to schedule the ultrasound for the sequential screen as scheduling access is currently limited. If you would like to do this test, please let us know as soon as possible and we can start the scheduling process. If you decide at your first OB visit that you don't want to go forward with the sequential screen, there is usually enough time to cancel.

Integrated Screen (84163, 84702, 82105, 82677, 86336)

Screens for Trisomy 21, 18 and open neural tube defects (like Spina Bifida) in two parts

- **Part one**: Bloodwork between 11weeks 0days and 13weeks 6days gestation
- **Part two**: Bloodwork drawn at your ~ 16-week appointment.

Quad screen (CPT code82105,82677,84702,86336)

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Screens for Trisomy 21, 18 and open neural tube defects (like Spina Bifida)
Bloodwork between 15weeks 0days and 22weeks and 6days gestation

- https://womenshealth.labcorp.com/patients/pregnancy/serum-screening

Test accuracy: All three of the above tests will pick up the majority of trisomy 21, 18 and open neural tube defects (like Spina Bifida). The Sequential screen is a little bit more accurate than the Integrated screen. The Integrated screen is a little bit more accurate than the Quad Screen.

***Aneuploidy screening options recommended for those greater than 35 years of age*:**

Qnatal cell-free fetal DNA (CPT code 81420)

Screens for Trisomy 21, 18, 13. If you choose, it can also test for abnormalities in the number of sex chromosomes

*Bloodwork any time after 10 weeks 0 days
*Special considerations:
-Designed for women 35 years and older because they are most at risks for trisomies Can result in false positives in patients who are younger than 35. However, of all available screening options for aneuploidy, it is most sensitive for picking up chromosomal abnormalities.
- Qnatal is an expensive test that is sometimes covered by insurance, we would recommend completing the Quest Diagnostics Cost Estimator form at www.myNIPTcost.com prior to having test done

- *For more information on Qnatal testing, you can visit*
  - *https://www.questwomenshealth.com/pregnancy-and-fertility/pregnant-patients/noninvasive-prenatal-screening*

Open Neural Tube Defects:

Serum Alpha fetoprotein or AFP (CPT code: 82105)

Open neural tube defects (ONTD) are problems with the way the fetus' skull, brain, spinal cord, and/or spine develop. Less than 1 pregnancy per 1,000 is affected. Serum AFP is a blood test drawn in our clinic between 15 and 18 weeks that screens for ONTD. If you are getting the Sequential Screen, Integrated Screen or Quad Screen, the serum AFP testing is already included. Patients that opted out of aneuploidy screening or who are getting the qnatal test will need to consider this test. If one wants screening done for ONTD as soon as available, the serum AFP is recommended. Otherwise, the anatomy ultrasound that every patient has around 20 weeks also screens for this anomaly.

Carrier Screening:

Cystic fibrosis (CPT code: 81220)
Spinal muscular atrophy (CPT code: 81329)

Carrier screening is performed once in a parent's life. Ideally it is done before conceiving or early in a first pregnancy. Carrier screening is done with a blood sample that looks to see if you carry "silent" genes for the most

common heritable conditions that could affect your baby. These most common heritable conditions in the United States are cystic fibrosis (CF) and spinal muscular atrophy (SMA). The chance that you carry the gene for CF or SMA is 1 in 25 to 1 in 114 (depending on your ethnicity). There may be other heritable conditions that we recommend screening for depending on your family history and/or ethnicity. If you test positive for a gene associated with one of the tested for conditions, we will likely want to test your partner next. If your partner is positive for the same gene, there is a chance that your baby will be affected by that condition. At any point during this process, we are happy to refer you to a genetic specialist to learn more. *If you have already had carrier screening performed prior to this pregnancy, it is not necessary to have it done again.*

Common Questions:

What is the most accurate test to make sure there is nothing wrong with the chromosomes of my baby?

The only way to look at all of the chromosomes of the pregnancy and surely diagnose that there is a correct number is to do an invasive test called an amniocentesis or chorionic villus sampling. These are procedures that use a needle to sample some of the cells from the pregnancy to exactly determine the chromosomal make-up for the fetus(es). Because these procedures have risk (albeit a small amount), it is usually only recommended to those that are found to have an abnormality found on ultrasound or aneuploidy screening test or for those at highest risk for chromosomal abnormalities in their pregnancies (women older than 35). This procedure is done by our high-risk OB specialists called Maternal Fetal Medicine physicians.

This is all very confusing.

All this information can be very overwhelming. You and your physician will be going over this all at your first obstetrical appointment. Another option is to be referred to a genetics counselor to discuss these options, as well as any patient specific questions.

What will my insurance cover?

This is your responsibility to determine. Please call your insurance company to verify what they will cover. Your insurance company will ask for the CPT codes, these are listed above.

Please feel free to reach out to our office with any other questions or concerns,
Monday through Friday 8:00 A.M. to 4:30 P.M.
at (207)874-2445