Obstetrics & Gynecology Genetic Screening Options

We have a variety of tools to help us answer the common question we hear from patients, "is my baby going to be healthy?" While there is no individual test or set of tests that can tell us a fetus is going to be 100% "healthy", there are screening tests for the most common abnormalities seen in pregnancy. This includes screening for aneuploidies (fetal chromosomal abnormalities), parent carrier status, open neural tube defects, and anatomic abnormalities, further described below.

Ultimately, deciding which test(s) you want is up to you. 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 all tests offered regardless of insurance coverage. We will support you as you consider these options and encourage you to make the decision that feels best for your growing family.

Aneuploidy Screening

When the genetic materials of an egg and 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 abnormalities. 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**,** meaning they do not confirm a diagnosis but raise a red flag that an abnormality may be present.

While increasing age is associated with a higher risk of aneuploidy, the American College of Obstetricians and Gynecologists (Practice Bulletin 226) recommends offering all types of genetic testing options (see below) to pregnant people of all ages, regardless of risk for chromosomal abnormality.

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 diagnostic testing. Diagnostic testing is the only way to confirm a genetic abnormality in a developing pregnancy.

Please contact your insurance company to check on coverage (See below CPT codes). Please notify our office if your insurance requires further follow-up.

Serum Analyte Testing:

Screening for aneuploidy using blood markers associated with chromosomal abnormalities.

Purpose	Screening Option	CPT Codes	Process
Screens for Trisomy 21, 18 and includes the AFP screen for open neural tube defects (i.e.Spina Bifida)	Integrated Screen	84163, 84702, 82105, 82677, 86336	Two parts: 1. Bloodwork between 11 weeks, 0 days and 13 weeks, 6 days gestation. 2. Bloodwork drawn at your ~ 16-week appointment.
	Quad Screen	82105, 82677, 84702, 86336	Bloodwork between 15 weeks, 0 days and 22 weeks, 6 days gestation.

Considerations:

- The above tests screen for trisomy 21, 18 and open neural tube defects, such as Spina Bifida. The Integrated screen is a little bit more accurate than the Quad Screen.
- For more info: https://womenshealth.labcorp.com/patients/pregnancy/serum-screening
- You may consider including a nuchal translucency ultrasound with these testing options for more information, see below.

Cell-free fetal DNA testing (cffDNA):

Screening for aneuploidy using fetal DNA fragments in a pregnant person's blood.

Purpose	Screening Option	CPT Codes	Process
Screens for Trisomy 21, 18, 13. The test can also test for sex chromosome abnormalities (optional).	QNatal cell-free fetal DNA	81420	Bloodwork any time after 10 weeks, 0 days.

Considerations:

- This screening can result in false positives in patients who are younger than 35. However, of all available screening options for aneuploidy, it is the most accurate screening test for trisomy 21 .
- QNatal is an expensive test that may be covered by insurance. For questions regarding coverage and costs, please contact Quest's Patient Navigator team via email at patientnavigators@questdiagnostics.com, or by calling 1-888-445-5011.
- For more information on QNatal testing: https://www.questwomenshealth.com/pregnancy-and-fertility/pregnant-patients/noninvasive-prenatal-screening.

Nuchal Translucency Ultrasound

- Formal ultrasound scheduled between 12w0d and 13w6d of pregnancy
- A thickened nuchal translucency has been associated with trisomy 21
- Provides an opportunity to detect early fetal anomalies

Open Neural Tube Defects

Open neural tube defects (ONTD) are problems with the way the fetal skull, brain, spinal cord, and/or spine develop.

Purpose	Screening Option	CPT Code	Process
Screens for Open neural tube defects (ONTD).	Serum Alpha fetoprotein or "AFP"	82105	Blood test drawn in our clinic between 15 and 18 weeks gestation.

Considerations:

- If you are getting the Integrated Screen or Quad Screen, the serum AFP testing is already included. Patients who opted out of aneuploidy screening or who are getting the QNatal test will need to consider this test.
- The anatomy ultrasound that every patient has around 20 weeks also screens for open neural tube defects.

Carrier & Hemoglobinopathy Screening

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

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).

Hemoglobinopathy is a group of genetic blood disorders that are inherited and can vary in their symptoms and severity. More common types of hemoglobinopathies include Sickle Cell Disease and Thalassemia. As 1 in 66 people in the United States have a hemoglobinopathy trait, we offer this testing to all persons planning pregnancy or currently pregnant.

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 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.

Screening Option	CPT Codes	Process
Cystic fibrosis	81220	Blood test drawn in our clinic any time in pregnancy or prior to pregnancy. They can be added into the lab panel drawn between 10 and 12 weeks gestation.
Spinal muscular atrophy	81329	
Hemoglobinopathy Evaluation	83020, 85014, 85018, 85041	This testing includes Hemoglobin A, Hemoglobin F, Hemoglobin A2 (Quant), Hemoglobin S, Hemoglobin C, Hemoglobin E, and any hemoglobin variants.

Expanded Carrier Screening

There are a variety of options for expanded carrier screening to test your carrier status of many other genes. These expanded panels are optional and may be considered by people planning a pregnancy by sperm donor or of certain ethnicities.

Screening Option	CPT Codes	Process
QHerit	81443, 81243 (Fragile X), 81329 (SMA)	Quest offers a variety of expanded carrier screening options ranging from 24 genes to 612 genes. These include common hemoglobinopathies. Please see their website for more details on the variety of tests offered and the possible cost associated with the test: https://www.questwomenshealth.com/pregnancy-and-fertility/for-your-pregnant-patients/qherit-carrier-screening

Common Questions:

Q: What is the most accurate test to make sure there is nothing wrong with the chromosomes of my baby?
A: The only way to look at all 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). These procedures do carry some risk to the pregnancy and are done by our high-risk OB specialists called Maternal Fetal Medicine physicians. If you are interested in diagnostic testing, please let your provider know at your first appointment.

Q: This is all very confusing, how do I decide what to do?
A: 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.

Q: What will my insurance cover?
A: 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.