Fetal Genetic Screening Information and Codes

What is the 20-week anatomy scan ultrasound? All women will receive the anatomy ultrasound that looks at the baby's body parts. If you would like to find out the sex of your baby, we can tell on this ultrasound. The ultrasound will take about 20-50 minutes. You are not able to take video or photos during the exam, although we will print you some photos.

Does the 20-week anatomy ultrasound detect issues? The ultrasound will look for body part issues on the baby and not at genetics. When there is an issue, the anatomy scan will only find it 70% of the time. Given the high rate of missing something at this ultrasound, we offer additional fetal genetic screening.

What does the additional fetal genetic screening look for? These fetal genetic screening tests look for specific chromosome problems and open neural tube defects (spina bifida or anencephaly). There is no test that will screen for all issues or tell us if baby is "healthy".

Are fetal genetic screening tests harmful to my baby? No, these tests are blood draws for you as well as an ultrasound. Ultrasound is considered safe during pregnancy.

I will not do anything different if my baby has genetic issues. Should I still consider fetal genetic testing? Yes. If we know about an issue prior to birth, it could change what we need to do during your pregnancy care and at delivery.

What if my screening test is positive? When a screening test is positive, we need to do additional testing to see if this is a true positive or a false positive. Screening tests will identify all pregnant women whose baby may have a genetic issue and some pregnant women whose baby does not have any issues. This is known as a false positive. A diagnostic test (amniocentesis) would determine if this is a false positive.

What are my chances of having a baby with a genetic issue? Your risk level is determined mostly by age. High risk is age 35 or older at time of delivery. Additional risk factors include having a prior pregnancy complicated by a genetic issue such as Trisomy 21 (Downs Syndrome), Trisomy 18 (Edwards Syndrome), Trisomy 13 (Patau Syndrome), Sex Chromosomal or other Chromosomal issues.

	Trisomy 21 (Downs)	Trisomy 18 (Edwards)	Trisomy 13 (Patau)	Sex Chromosomal Issues	Rare Chromosomal Issues	All Chromosomal Issues
Age 20	1 in 1,250	1 in 5,000	1 in 10,000	1 in 294	1 in 270	1 in 122
Age 25	1 in 1,000	1 in 5,000	1 in 10,000	1 in 294	1 in 270	1 in 119
Age 30	1 in 714	1 in 2,500	1 in 5,000	1 in 294	1 in 270	1 in 110
Age 35	1 in 294	1 in 1,111	1 in 2,500	1 in 285	1 in 270	1 in 84
Age 40	1 in 86	1 in 333	1 in 714	1 in 196	1 in 270	1 in 40

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What are my options for fetal genetic screening? The most common fetal genetic screening test is Cell Free DNA. This test will look at baby's chromosomes that are in your blood. This test can also determine the baby's sex. Cell Free DNA tests have a high detection rate with a low false positive rate and can be offered to both low risk and high-risk women. Low risk women may be required by their insurance to do a different test (Full Integrated, Quad test, etc.) which we can offer you. Commonly the company that performs Cell Free DNA (Arisoa, Natera or Sequenom) can offer a discount directly to the patient.

We also recommend doing a Nuchal Translucency ultrasound and AFP with the Cell Free DNA. These are 3 separate tests that are done together to check different issues.

Test	What it tests for	Timing and Results Available	Insurance CPT Code	Performing Lab
Nuchal Translucency (NT)	Neural tube defects, some heart and anatomy issues, some genetic issues	Ultrasound performed at 10wk – 13w6d Results within 24 hours of Ultrasound	76813	Alliance OB/GYN
Alpha-Fetoprotein (AFP)	Neural tube defects	16-22 weeks Results within a few days after the blood draw	82105	Sparrow 517-364-6000
Cell Free DNA ClariTest Core, Panorama, Materni21	Down Syndrome (Trisomy 21), Trisomy 18, Trisomy 13, sex chromosome issues and fetal sex	10 weeks or later Results within 7-10 days after blood draw	81420 81507 (ClariTest Core only)	ClariTest Core: GenPath Womens Health 1-800-633-4522 Panorama: Natera 1-650-249-9090 Materni21: Sequenom Laboratory 1-877-821-7266

What is my cost for this test? It is important to talk with your insurance about coverage prior to having the test done. These diagnosis codes (below) and CPT codes (above) will help in the conversation with your insurance company. Pricing for the test is determined by the performing lab and is subject to change. If you are low risk, your insurance may require a prior authorization for Cell Free DNA – we request that you contact the company (Ariosa, Natera or Sequenom) directly for lower cost test options.

Diagnosis Codes - use the code that applies to you	
Z34.01	Low risk woman in her first pregnancy
Z34.81	Low risk woman in her 2nd (or more) pregnancy
O09.511	Advanced maternal age (>35yo at time of delivery) in first pregnancy
O09.521	Advanced maternal age (>35yo at time of delivery) in 2nd (or more) pregnancy
Z36.82	Testing for Nuchal Translucency Ultrasound
Z13.79	Other screening for genetic or chromosomal issues for high-risk women