Prenatal Testing for Abnormal Fetal Chromosomes

Prenatal testing is available to all women who are currently pregnant. Most babies with birth defects are born to people without risk factors. Some birth defects are caused by inherited abnormal genes; others are caused by abnormal chromosomes. Deciding to have prenatal testing is a personal choice. Some women prefer not to test. Others feel having the information allows time to prepare for caring for a child with a particular disorder or consider the option to interrupt the pregnancy.

Please ask your insurance company about coverage prior to proceeding with testing

First Trimester

Non-invasive prenatal testing (NIPT) is a screening test performed any time after 10 weeks 0 days and identifies women whose fetuses are at risk of having an abnormal number of chromosomes (known as aneuploidy). Two common aneuploidies are having an extra chromosome ("trisomy") or a missing chromosome ("monosomy"). There are three common trisomy conditions: trisomy 21 (Down Syndrome), trisomy 13 (Patau Syndrome) and trisomy 18 (Edwards Syndrome). A common monosomy is monosomy X (Turner Syndrome). The risk of having a pregnancy affected by an aneuploidy increases with age (see chart below). The NIPT North Clinic administers is called "informaSeq Prenatal Test". InformaSeq detects up to 99% of trisomy 13, 18 and 21 with false positive rate as low as 0.1%. Please note, if your insurance company does not cover this test, you can refer to the informaSeq brochure for more information about alternative payment options at www.intregratgedgenetics.com

Risk of aneuploidy and age at time of delivery

Age*	Down Syndrome	All**	Age*	Down Syndrome	All**
20	1 in 1477		35	1 in 353	1 in 179
21	1 in 1461		36	1 in 267	1 in 149
22	1 in 1441		37	1 in 199	1 in 123
23	1 in 1415		38	1 in 148	1 in 105
24	1 in 1382		39	1 in 111	1 in 81
25	1 in 1340	1 in 476	40	1 in 85	1 in 63
26	1 in 1287	1 in 476	41	1 in 67	1 in 49
27	1 in 1221	1 in 455	42	1 in 54	1 in 39
28	1 in 1141	1 in 435	43	1 in 45	1 in 39
29	1 in 1047	1 in 417	44	1 in 39	1 in 31
30	1 in 939	1 in 385	45	1 in 35	1 in 24
31	1 in 821	1 in 385			
32	1 in 696	1 in 323			
33	1 in 572	1 in 286			
34	1 in 456	1 in 244			

*Maternal age at time of delivery, **Includes risk for trisomy 13, 18, 21

The **first trimester screen** (FTS) is a prenatal test that is performed between 11 weeks + 6 days and 13 weeks + 6 days of pregnancy and screens for the risk of trisomy 13, 18 and 21. FTS combines maternal age with two blood markers (PAPP-A and Beta-HCG) and an ultrasound measurement of the thickness of the back of the fetal neck (nuchal translucency). The FTS results provide a revised risk for trisomy 13, 18

and 21 that may be higher or lower than the pre-screen, age-based risk. Learn more at www.ntdlabs.com

Follow-up testing: If you have an abnormal **informaSeq** or **first trimester screen**, you will be referred to a genetic counselor and offered diagnostic testing. The decision to pursue diagnostic testing is a personal decision. The advantage of screening early in your pregnancy is obtaining earlier information about your pregnancy which may allow for more options regarding the pregnancy as well as more time to make those decisions. A genetic counselor will review what your options are for diagnostic testing. If you opt for informaSeq or FTS, you will be offered separate screening for neural tube defects in the second trimester. We perform informaSeq in our office but refer patients to Maternal Fetal Medicinefor first trimester screening and diagnostic testing.

Second Trimester

Quad Screen

The Quad Screen is a prenatal test performed between 15 weeks and 21 weeks + 6 days of pregnancy. Similar to the first trimester screen, the quad screen identifies women whose fetuses are at risk of having an aneuploidy (specifically trisomy 13, 18 and 21); it also identifies women whose fetuses are at risk of having open neural tube defects. The quad screen is a blood test that combines four blood markers (MSAFP, beta-HCG, estriol and inhibin A) and maternal age. The quad screen results provide a revised risk for aneuploidies that may be higher or lower than the pre-screen, age-based risk. The decision to pursue additional diagnostic testing is a personal decision. If the risk for aneuploidy or neural tube defect is increased, you will be referred to Maternal Fetal Medicine specialist and a genetic counselor to review your options for diagnostic testing.

Ultrasound

An ultrasound to evaluate fetal anatomy is a part of routine prenatal care and is performed around 20 weeks. At North Clinic we perform “Level 1” ultrasounds; this is a screening ultrasound for anatomic and placental abnormalities and abnormalities that increase the risk for aneuploidy. It is not possible to detect every possible abnormality by ultrasound; however, we strive to provide the highest quality by following national guidelines. When a patient has an abnormality on a “Level 1” ultrasound or has a medical condition associated with increased risk for birth defects, a “Level 2” ultrasound is performed. We refer patients to Maternal Fetal Medicine for a Level 2 ultrasound.

Diagnostic Testing

All women can opt for diagnostic testing instead of screening. The decision to pursue invasive diagnostic testing (such as chorionic villus sampling or amniocentesis) is personal and each individual will weigh risks and benefits of testing differently. Chorionic villus sampling (CVS is performed between 10 to 14 weeks) and amniocentesis (performed after 15 weeks) are performed by Maternal Fetal Medicine specialists. Both are 99.9% accurate in detecting aneuploidies and carry a 1 in 500 risk of miscarriage.

Insurance Codes:

Please ask your insurance company if these tests are covered prior to moving forward with testing:

- First Trimester Screen with Nuchal Translucency: 84163, 84702, 86336
- Quad Screen: 82105, 82677, 86336, 84702
- AFP: 82105
- informaSeq®: 81420