Prenatal Screening for Abnormal Fetal Chromosomes

Prenatal screening is available to anyone who is currently pregnant. There is no risk to the pregnancy with any of these screening tests. Chromosomal abnormalities occur in approximately 1 in 150 live births; however, the chance increases as a pregnant person ages. The most common chromosomal abnormalities are trisomy 21 (Down syndrome), trisomy 18 (Edward syndrome), trisomy 13 (Patau syndrome). Sex chromosome (X and Y) abnormalities can also occur. Screening tests can tell you if your pregnancy has an increased chance of a chromosomal abnormality. Screening tests do not confirm a diagnosis of a disorder. Deciding to have prenatal screening is a personal choice. Some women prefer not to screen. Others feel having the information allows time to prepare for caring for a child with a particular disorder or consider the option to terminate the pregnancy.

<u>Please ask your insurance company about coverage prior to proceeding with testing</u>

Risk of chromosomal abnormality 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

Screening tests offered during pregnancy:

1. **Cell-free DNA screening (MaterniT21)**: cell-free DNA is a small amount of DNA that is released from the placenta into a pregnant woman's bloodstream. This cell-free DNA can be used to screen for chromosomal abnormalities in the pregnancy. This blood test is performed any time after 9 weeks 0 day of pregnancy. The accuracy (false positive and false negative rate) depends on the chromosome being evaluated, but the detection rate can be as high as 99% with a false positive rate as low as 0.1%.
   The following are screened using MaterniT21:

Trisomy 21 (Down Syndrome)
Trisomy 18 (Edward Syndrome)
Trisomy 13 (Patau Syndrome)
Sex chromosomes (X and Y)
Trisomy 16
Trisomy 22
1p36 Deletion Syndrome
22q11 deletion (DiGeorge Syndrome)
15q11 deletion (Angelman Syndrome)
11q23 deletion (Jacobsen Syndrome)
8q24 deletion (Langer-Giedion Syndrome)
5q15 deletion (Cri-due-Chat Syndrome)
4p16 deletion (Wolf-Hirschhorn Syndrome)

2. The **First Trimester Screen** is a prenatal screening test that uses an ultrasound and blood testing performed between 10 and 14 weeks of pregnancy. The ultrasound measures the nuchal translucency (area of thickness on the back of the fetal neck) and the blood testing screens for blood markers (PAPP-A and Beta-HCG) with a detection rate of approximately 85%. Learn more at www.ntdlabs.com
   The following are screened using the First Trimester Screen:

   Trisomy 21 (Down Syndrome)
   Trisomy 18 (Edward Syndrome)
   Trisomy 13 (Patau Syndrome)

3. The **Quad Screen (Second Trimester Screen)** is a blood test performed between 15 and 23 weeks of pregnancy. Four blood markers are measured (MSAFP, beta-HCG, estriol and inhibin A). Similar to the first trimester screen, the Quad Screen identifies women whose pregnancy is at higher risk of having trisomy 18 and 21; it also identifies women whose fetuses are at risk of having open neural tube defects (such as spina bifida). The detection rate is approximately 80%. The following are screened using the Quad Screen:

   Trisomy 21 (Down Syndrome)
   Trisomy 18 (Edward Syndrome)
   Neural tube defects

4. Single **Alpha Fetal Protein** (AFP) is a blood test performed between 15 and 18 weeks of pregnancy and screens for increased risk of neural tube defects (such as spina bifida). This screening test alone does not evaluate for chromosomal abnormalities. The detection rate ranges from 65-95% depending on the abnormality with a false positive rate of 1-3%.

Follow-up testing: If you have an abnormal **MaterniT21, First Trimester Screen or Quad Screen,** you will be referred to a genetic counselor and Maternal Fetal Medicine. An abnormality on one of the screening tests suggests an increased risk of chromosomal or neural tube defect (depending on screening test performed), but does not confirm a disorder in the pregnancy. Therefore, the option of diagnostic testing will be discussed with the genetic counselor.

Ultrasound: An ultrasound to evaluate fetal anatomy is a part of routine prenatal care and is performed around 18- 22 weeks. This screening test evaluates for major physical abnormalities that can be associated with or independent of chromosomal abnormalities. It is not possible to detect every possible abnormality by ultrasound, however ultrasound routinely detects 80-90% of structural abnormalities. If

an abnormality is noted on ultrasound, patients are referred for further evaluation with Maternal Fetal Medicine.

Insurance Codes:

Please ask your insurance company if the screening test is covered prior to proceeding with the testing. The following are CPT codes for the screening tests.

- MaterniT21 Plus: 81420
- First Trimester Screen with Nuchal Translucency: 84163, 84702, 86336
- Quad Screen: 82105, 82677, 86336, 84702
- AFP: 82105