

Prenatal Genetic Screening

What is prenatal genetic screening?

Also referred to as aneuploidy screening, this group of tests are used to determine the risk that your baby has a chromosomal abnormality, such as Down Syndrome (Trisomy 21), Trisomy 13 or 18 or a neural tube defect.

*A SCREENING tests determines if a patient is at higher or lower risk, based on age and health factors, than someone else just like them. For instance, a result might return as having a 1 in 10,000 risk or it could come back as a 1 in 43 chance of a chromosomal abnormality. This is different from a DIAGNOSTIC test which gives a specific result.

What is Trisomy 21?

Down syndrome results from an extra chromosome 21. Individuals with Down syndrome have different levels of intellectual disability and they may have other birth defects. Down syndrome occurs in 1 in 700 live births but the chance increases with the age of the mother.

What is Trisomy 18?

Like Down syndrome, Trisomy 18 has an extra chromosome. These individuals have severe intellectual and physical disabilities. Most babies are stillborn or do not survive past infancy. Trisomy 18 occurs in about 1 in 4000 live births and the chance increases with the mother's age.

What are neural tube defects?

This type of birth defect impacts the development of the brain and spinal cord. The most common type is spina bifida. People with spina bifida may have problems with walking and bowel or bladder control. Neural tubes defects occur in 1 in 1500 live births.

Is screening right for me?

While this may seem like a routine test, this is not a decision you should take lightly. Most results are reassuring but it is important to think about how you might feel if you have a positive result.

*Are you prepared to deal with the worry if you have a positive screening result?

*Would you consider pregnancy termination if you find out that your baby has a chromosomal or neural tube defect?

*If you decided to continue the pregnancy, would knowing that your baby has one of the conditions help you prepare for your baby's birth?

*Would you rather find out after your baby's birth if they have one of these conditions?

What screening tests are available (all have approximately a 5% false positive rate)?

*"Sensitivity" is the term used to describe the proportion of patients with a chromosomal abnormality who will have a positive test.

*"False positive rate" is the term used to describe the proportion of patients without a chromosomal abnormality who have a positive test.

1. Ultrascreen (**87% sensitive**):
 - *Late 1st trimester (11-13 weeks), with results prior to 15 weeks gestation.
 - *An ultrasound of the fetal neck and a finger-prick blood test is performed at University of Vermont Medical Center Fetal Diagnostic Center.
 - *Does not give risk of neural tube defects.
 - *False positive rate for all tests higher than 5% if over age 30.
2. Sequential Screening (**95% sensitive**):
 - *After you get the results of the Ultrascreen, a second test will modify the results.
3. Integrated Screening (**95% sensitive**):
 - *Ultrascreen followed by a second blood test. You will only receive results after the second part of the test.
4. Quad Marker Screening (QMS) (**85% Sensitive- lowest detection rate**):
 - *Second trimester (after 15 weeks).
 - *Maternal blood test can be performed at Porter.
 - *It also provides risk of neural tube defects.
 - *Results return in approximately 2 weeks.
5. Non-invasive prenatal testing (NIPT) (**99% sensitive for T21 and 97% for T18**)
 - *A blood test that looks for baby's chromosomes in your blood.
 - *Performed any time after 10 weeks.
 - *Also reports gender.
 - *Only covered by insurance if you are over 35, have a personal or family history of chromosomal problems or if one of the other tests is abnormal.

What is the next step if the screening test indicates a high risk of a chromosomal abnormality?

Diagnostic testing is available in several forms.

1. Non-invasive prenatal testing (see above)
2. Chorionic Villus Sampling
 - *Performed between 10-14 weeks with a needle through the vagina or abdomen to sample placental tissue.
 - *Risk of miscarriage following the procedure is 1:100-1:150 secondary to rupture of amniotic membranes, infection, or injury to the placenta or fetus.
 - *Preliminary results return in approximately 2 days. Final chromosome analysis takes approximately 10 days to be completed.
3. Amniocentesis:
 - *Performed after 15 weeks with a needle through the abdominal wall into the uterus to obtain amniotic fluid.
 - *Risk of miscarriage following the procedure is 1:500 (see above).
 - *Preliminary results return in approximately 2 days. Final chromosome analysis takes approximately 10 days to be completed.

Cystic Fibrosis Screening

- *There are over 1700 different genetic mutations that can lead to cystic fibrosis, though there are approximately 23 specific mutations found in MOST patients with cystic fibrosis. These 23 mutations are the ones tested for when screening.
- *A patient's risk of being a carrier also depends on her family history and her ethnic background.
- *Even if the patient is a carrier, her partner may not be. If the partner is not a carrier, the risk of the child having CF is extremely low.
- *The only way to definitively determine prenatally if a baby will have cystic fibrosis is by amniocentesis.
- *If neither a patient nor her partner has a personal or family history of cystic fibrosis, this testing is not the most cost-effective. Vermont includes Cystic Fibrosis in its newborn screening panel -- blood will be drawn from baby in the birthing center.

