Chromosomal Testing

Risk Table for Chromosomal Abnormalities by Maternal Age

Maternal Age (yrs)	Risk of Trisomy 21 by Age:		Risk for Any Chromosomal Abnormality:
	At 12 weeks	At birth	At birth
20	1 in 1070	1 in 1480	1 in 525
25	1 in 950	1 in 1340	1 in 475
30	1 in 630	1 in 940	1 in 384
32	1 in 460	1 in 700	1 in 322
34	1 in 310	1 in 456	1 in 243
35	1 in 250	1 in 353	1 in 178
36	1 in 200	1 in 267	1 in 148
38	1 in 120	1 in 148	1 in 104
40	1 in 70	1 in 85	1 in 62
42	1 in 40	1 in 54	1 in 38
44	1 in 20	1 in 39	1 in 23

Screening for Downs syndrome (Trisomy 21) and other chromosomal abnormalities:
The only way to be certain whether the fetus has Downs syndrome or not is by doing an invasive diagnostic test – an amniocentesis or chorionic villus sampling (CVS). Both tests provide a sample that contains tissue that has the same genetic make-up as the baby which allows the baby's chromosomes to be examined. Because of the increased risk of miscarriage associated with these two tests, they are not generally recommended unless the fetus is at increased risk. Traditionally, this is a mother over 35 years old. Many patients elect to do the nuchal translucency screening test to determine their individual risk of Trisomy 13 and 18. Genetic counseling is recommended for women over 35 and those with a nuchal screen showing 1/1000 chance of having a baby with Trisomy 13 or 18.

Amniocentesis

What is an amniocentesis?
An amniocentesis is a procedure where a small amount of amniotic fluid (fluid surrounding the developing baby) is removed from the uterus through a thin needle, using ultrasound guidance. This procedure is typically performed during 16 to 20 weeks of pregnancy. It can be done as early as 12 to 14 weeks and as late as near term. Some women say amniocentesis does not hurt, while others say they feel pressure or a cramp.

What tests can be performed on amniotic fluid specimen?
Different tests can be done on amniotic fluid; the most common tests are listed below.

- Chromosome analysis to detect chromosome abnormalities such as Down syndrome or Trisomy 18.
- AFP (alpha-fetoprotein) and AChE (acetylcholinesterase) measurements to detect neural tube defects such as spina bifida and anencephaly. In spina bifida there is an opening in the back/spinal cord, usually requiring multiple surgeries, and may be associated with physical disabilities. In anencephaly the brain development is incomplete, usually resulting in death.

- Genetic diseases that can be diagnosed prenatally, including Cystic fibrosis, Fragile X syndrome, Hemophilia, Sickle cell disease, Thalassemia, Tay-Sachs disease, Canavan disease and Gaucher's disease.

Who should consider having an amniocentesis?

- Women who will be 35 years or older at the time of delivery. The risk of having a child with Down syndrome or other chromosome abnormalities increases with increasing maternal age.
- Women with an abnormal nuchal translucency screening test.
- Either parent can be a carrier of a chromosome rearrangement. Some individuals have chromosome rearrangements, in which some of the genetic materials on a chromosome may be moved from their normal location. These individuals are healthy, but they may have a child with a chromosome imbalance that can be associated with developmental and physical defects.
- Previous child with chromosome abnormality. These couples have an increased risk of having another child with a chromosome abnormality.
- Parents are carriers of a prenatally diagnosable genetic disorder. These couples have an increased risk of having a child with the genetic disorder. If diagnosis for the disorder is available, amniocentesis can be performed for this purpose. Carrier screening is available for a number of disorders. Ask your doctor for more information.
- Women with abnormal ultrasound findings. When ultrasound examination shows abnormalities, amniocentesis for diagnostic testing of the amniotic fluid may be recommended.
- Women with abnormal Expanded AFP screening test. This may indicate an increased risk for chromosome abnormalities or neural tube defects.
- Family history of neural tube defects. The risk of having a child with a neural tube defect, such as spina bifida, is increased when a close relative has the disorder.
- Certain seizure medications may increase the risk for neural defects and amniocentesis should be considered.

Chorionic Villus Sampling (CVS)

What is a CVS?

A CVS is performed between 10 and 12 weeks and involves taking a small amount of tissue from the placenta. Although methods can vary, the procedure involves inserting a small tube called a catheter through the cervix into the uterine cavity. It may be performed after an abnormal nuchal thickness to evaluate the chromosomes of the fetus for abnormalities. CVS chromosomal results are available earlier in pregnancy than amniocentesis results. To complete the testing, AFP only and Level II ultrasound are still recommended. This test is performed by a perinatologist.

Scheduling Genetic Counseling and Genetic Testing

How do I schedule chromosomal testing?

You must call a perinatology office (high-risk obstetrician) to schedule and appointment for genetic counseling, nuchal screening, CVS or amniocentesis. If you would like to schedule nuchal thickness testing or CVS, you should call prior to 11 weeks of pregnancy. If you want an amniocentesis this is usually performed with a Level II ultrasound between 16 and 18 weeks of pregnancy. If you elect not to have the amniocentesis, you should still consider genetic counseling and a Level II ultrasound. You may also have the Expanded AFP test performed if you did not do a nuchal screening test and are not planning an amniocentesis. The AFP test does not have the same accuracy as the amniocentesis. If you are undecided about testing, schedule genetic counseling during the first trimester of your pregnancy.

Who should I call?

Please check with your insurance carrier to determine which physician and facility is contracted with your insurance. Locally, we recommend the Obstetrix Medical Group at (408) 371-7111, or Stanford perinatology department at (650) 725-7030.