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 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.

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 perinatology group at (408) 371-7111, or Stanford perinatology department at (650) 725-7030.