TESTING FOR GENETIC PROBLEMS AFFECTING PREGNANCY

Christie Clinic Ob/Gyn

SCREENING FOR DISORDER THAT COULD PASS ON TO ANY FUTURE PREGNANCY

Cystic Fibrosis

- Blood test can be taken before or during pregnancy.
- Patient without any symptoms may be carrier of one abnormal gene copy. Patient with two abnormal gene copies will have the disease.
- Disease affects breathing and digestion.
- About 1 in 27 patients without the disease will be carrier of an abnormal copy. If both mom and dad have an abnormal copy, the baby has 1 in 25% chance of having the disease.
- This test may or may not be covered by insurance.

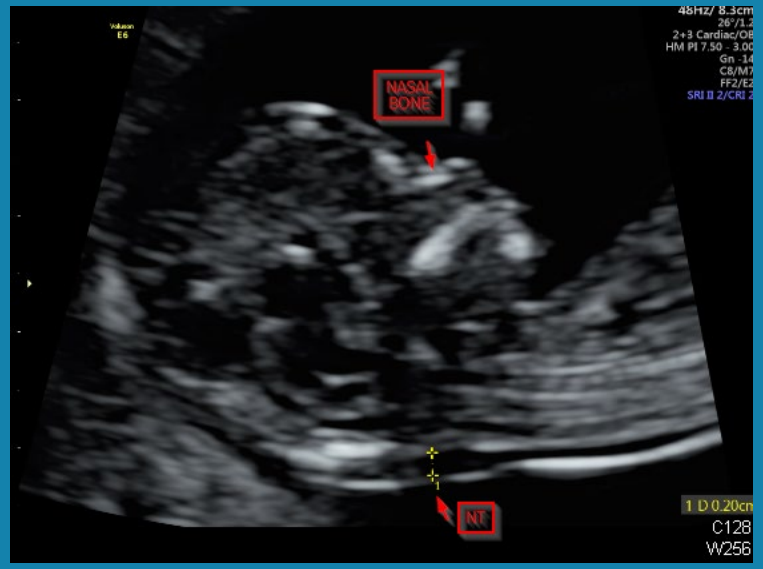
Spinal Muscular Atrophy

- Blood test can be taken before or during pregnancy.
- Patient without any symptoms may be carrier of one abnormal gene copy. Patient with two abnormal gene copies will have the disease.
- Disease causes muscle weakness and even paralysis.
- About 1 in 50 patients without the disease will be carrier of an abnormal copy. If both mom and dad have an abnormal copy, the baby has 1 in 25% chance of having the disease.
- This test may or may not be covered by insurance.

FIRST TRIMESTER SCREENING FOR DISORDER AFFECTING THIS SPECIFIC PREGNANCY

Nuchal Translucency

- Ultrasound performed between 12 and 13.5 weeks of pregnancy.
- Measures thickness of a clear space at the back of the baby's neck. This space can be abnormal in babies with chromosome problems like Down Syndrome and other birth defects including heart defects.



Serum Screen

- Blood test can be taken within 5 days of ultrasound.
- Test measures two proteins in the mother's blood: PAPP-A and HCG which can be affected by chromosome problems like Down Syndrome.
- The results will be combined with the ultrasound information to provide a risk assessment of this baby being affected with chromosome problems like Down Syndrome.
- This test may or may not be covered by insurance.

SECOND TRIMESTER SCREENING FOR DISORDER AFFECTING THIS SPECIFIC PREGNANCY

Quad Screen

- Blood test can be taken between 15 and 22.5 weeks of pregnancy.
- Test measures four proteins in the mother's blood: AFP, HCG, Estriol, Inhibin.
- The results will provide a risk assessment of this baby being affected with chromosome problems like Down Syndrome or structural problems like spina bifida.
- This test may or may not be covered by insurance.

AFP Screen

- Blood test taken between 15 and 22.5 weeks of pregnancy in patients who did first trimester screening for genetics.
- Test measures AFP protein in mother's blood.
- The results will be combined with the ultrasound information to provide a risk assessment of this baby being affected with structural problems like spina bifida.
- This test may or may not be covered by insurance.

SCREENING FOR DISORDER AFFECTING THIS SPECIFIC PREGNANCY IN HIGH RISK PATIENTS

Claritest

- Blood test can be taken after 10 weeks of pregnancy.
- Offered to women who are high risk (those who are >34 years old, have previous baby affected with chromosome problem, or who have abnormal nuchal translucency ultrasound.
- Test measures baby's DNA fragments found in the mother's blood which can be affected by chromosome problems like Down Syndrome.
- The results will provide a risk assessment of this baby being affected with chromosome problems like Down Syndrome.
- This test may or may not be covered by insurance.

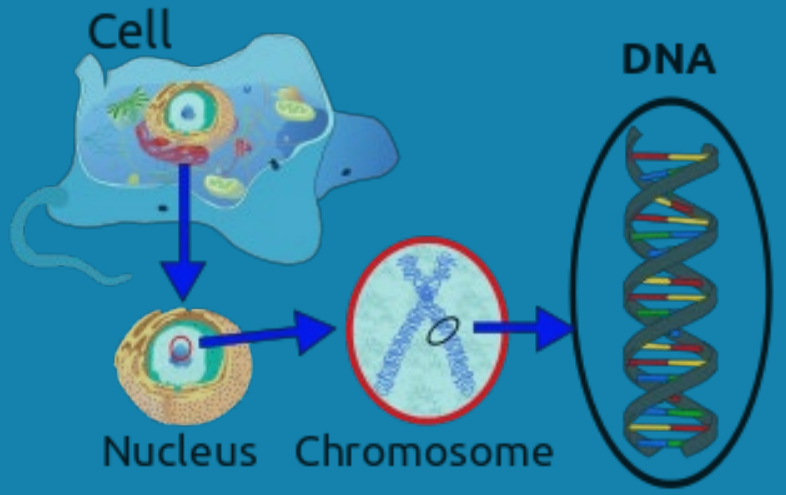


Photo credit to Wikipedia

SCREENING FOR DISORDER AFFECTING THIS SPECIFIC PREGNANCY IN HIGH RISK PATIENTS

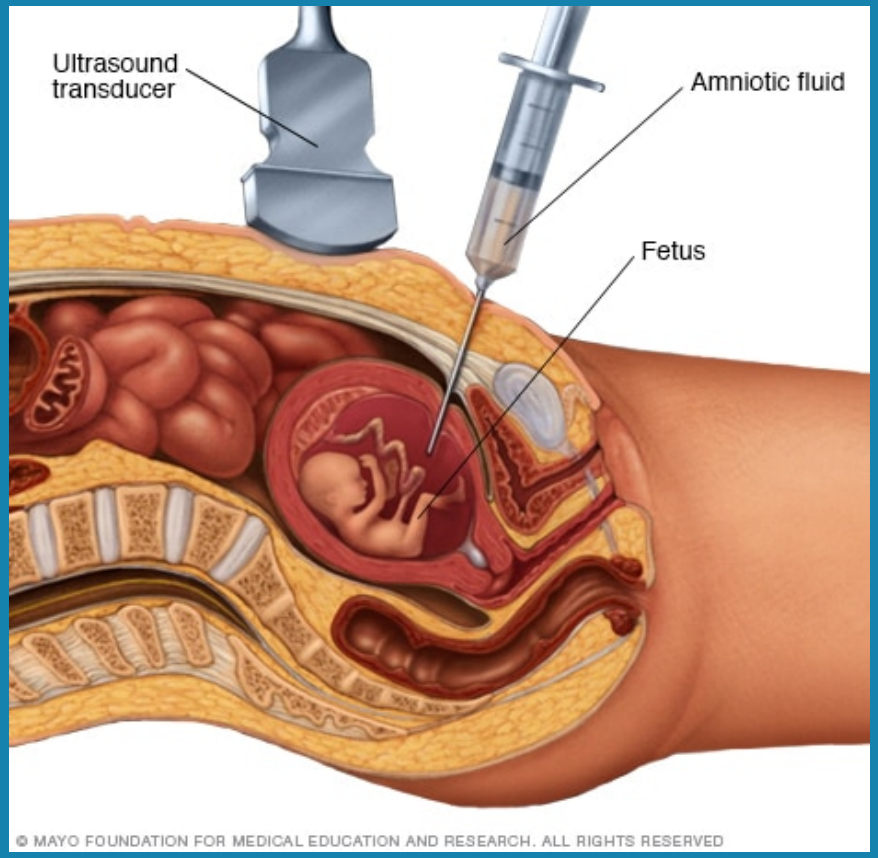


Photo credit to Mayo Clinic

Amniocentesis

- Test requires taking a sample of the baby's amniotic fluid using a needle through the mother's abdomen.
- Offered to women who are high risk (those who are >34 years old, have previous baby affected with chromosome problem, or who have abnormal genetic screening test result.
- Test evaluates baby's DNA found cells that have shed into the amniotic fluid.
- The results will provide a risk assessment of this baby being affected with chromosome problems like Down Syndrome.
- There are risks to this procedure including pain, infection, breaking the bag of waters, and loss of the pregnancy which can be discussed in more detail with the physician.
- This test may or may not be covered by insurance.