Prenatal Screening and Diagnostic Counseling

Screening Tests

A screening test tell us the likelihood that a baby has a condition, NOT if the baby has that diagnosis or condition. The conditions that these tests screen for are Down syndrome, Trisomy 18, Trisomy 13, or an open neural tube or abdominal defect.

Timing by Gestational Age	Type of Test	Advantages	Disadvantages
First trimester 10-13 weeks, 6 days	Nuchal translucency ultrasound + pregnancy associated plasma protein-A test	Screens for genetic disorders including anencephaly, neural tube defects, and cardiac defects.	Requires specifically trained ultrasound technicians. False positive rate ~5%.
Anytime after 10 weeks old	Cell free fetal DNA blood test	Potential for earlier detection. False positive rate 0.5%.	Requires dating ultrasound for accurate pregnancy dating.

A ***positive*** screening test means that the fetus is at a higher risk of having the disorder compared with the general population. It does not mean that the fetus has the disorder.

A ***negative*** result means that the fetus is at lower risk of having the disorder compared with the general population. It does not rule out the possibility that the fetus has the disorder.

Notes:

Obstetrics and Gynecology
(907) 729-3100
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Diagnostic Testing

Prenatal genetic diagnostic tests uses cells directly from the pregnancy to tell whether the baby has a specific condition like Down syndrome, Trisomy 18, or specific inherited disorders. If you have a positive screening test, follow-up services can include genetic counseling, an ultrasound exam, and possible diagnostic testing.

Timing by Gestational Age	Type of Test	Advantages	Disadvantages
First trimester 12-13 weeks 6 days	Chorionic villus sampling	Potential for earlier detection than amniocentesis	Takes a needle to sample villus tissue. Very rare, 1 in 455 (0.22%) chance of pregnancy loss.
Second trimester 16-20+ weeks	Amniocentesis		Takes a needle to sample the fluid around your baby. Very rare, 1 in 900 (0.11%) chance of pregnancy loss.

Notes:

Glossary:

Amniocentesis– The sampling of amniotic fluid usign a hollow needle inserted into the uterus to screen for developmental abnormalities

Anencephaly – A birth defect in which a baby is born without parts of the brain and skull.

Cell free fetal DNA blood test – A screening to determine if the fetus has a higher chance of Down Syndrome, Trisomy 18 and Trisomy 13.

Chorionic Villus Sampling – A diagonostic test that involves taking a sample of tissue from the placenta to test for certain other genetic problems.

Chromosome – A long DNA molecule with part or all of the genetic material for an organism (baby).

False Positive – An outcome where the screening incorrectly predicts a positive result. The result of the test is positive but the baby does not have the condition.

Nuchal Translucency Ultrasound – A screening performed in the first trimester to help determine your baby's risk of congenital conditions.

Open Neural Tube Defect – Problems with the way the brain, spinal cord, or spine forms while the baby is growing in the womb.

Pregnancy Associated Plasma Protein-A – A protein produced by the placenta that is needed for the implantation process and to maintain a healthy placenta.

Trisomy 13 – A disorder in which babies are born with three copies of chromosome 13 instead of two.

Trisomy 18 – A disorder in which babies are born with three copies of chromosome 18 instead of two.

Down Syndrome or Trisomy 21 – A disorder in which babies are born with three copies of chromosome 21 instead of two.

Villus Tissue – Thin membrane of placental tissue that contains the same genetic material as the fetus.

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