The American College of Obstetricians and Gynecologists

FAQ

FREQUENTLY ASKED QUESTIONS
FAQ165
PREGNANCY

Screening for Birth Defects

What is a birth defect?

A birth defect is a physical problem that is present at birth. A birth defect may affect how the body looks, functions, or both. Many birth defects are mild, but some can be severe. Babies with birth defects may need surgery or medical treatment.

What is a screening test?

A screening test can give information about a pregnant woman's risk of having a baby with certain birth defects. Not all birth defects can be detected with screening tests before birth.

What are some of the birth defects for which there are screening tests?

- Neural tube defect—Incomplete closure of the fetal spine that can result in spina bifida or ***anencephaly***.
- Abdominal wall defects—One type of defect occurs when the muscle and skin that cover the wall of the abdomen are missing and the bowel sticks out through a hole in the abdominal wall (gastroschisis). Another type is when the tissue around the umbilical cord is weak and allows organs to protrude into this area (omphalocele).
- Heart defect—The chambers or pathways through the heart are not properly developed.
- Down syndrome—Mental retardation, abnormal features of the face, and medical problems such as heart defects occur as a result of an extra chromosome 21 (trisomy 21).
- Trisomy 18—There is an extra chromosome 18, which causes severe mental retardation and birth defects and sometimes death.

Who should receive screening tests?

Screening tests are offered to all pregnant women to assess their risk of having a baby with a birth defect or genetic disorder. If a screening test shows an increased risk of having an affected baby, further tests may be used to diagnose the problem (see the FAQ Diagnosing Birth Defects). An abnormal screening test result, while alarming, only signals a possible problem. In most cases, the baby is healthy even if there is an abnormal screening test result. Likewise, a birth defect can occur even if the test result does not show a problem.

What screening tests are done in the first trimester?

First trimester screening tests include blood tests and an ***ultrasound exam***. This screening can be done as a single combined test or as part of a step-by-step process. Some women may not need further testing. First trimester screening is

done between 11 weeks and 14 weeks of pregnancy to detect the risk of Down syndrome and trisomy 18. The blood tests measure the level of two substances in the mother's blood:

1. Pregnancy-associated plasma protein-A (PAPP-A)
2. ***Human chorionic gonadotropin (hCG)***

An ultrasound exam, called ***nuchal translucency screening***, is used to measure the thickness at the back of the neck of the fetus. An increase in this space may be a sign of Down syndrome, trisomy 18, or other chromosomal problems.

How is the risk for birth defects assessed in first trimester screening?

In first trimester screening, the results of the nuchal translucency screening are combined with those of the blood tests and the mother's age to assess the risk for the fetus. In the first trimester, this combined test detects Down syndrome in most but not all cases (82–87%). When the nuchal translucency thickness is increased, the fetus may have a heart defect or other genetic condition. In this case, your health care provider may suggest a more detailed ultrasound exam around 20 weeks of pregnancy.

What screening tests are done in the second trimester?

In the second trimester, a test called "multiple marker screening" is offered to screen for Down syndrome, trisomy 18, and neural tube defects. This test measures the level of three or four of the following substances in your blood:

- Alpha-fetoprotein (AFP)—A substance made by a growing fetus, which is found in amniotic fluid, fetal blood, and, in smaller amounts, in the mother's blood.
- Estriol—A hormone made by the ***placenta*** and the liver of the fetus.
- Human chorionic gonadotropin—A hormone made by the placenta.
- Inhibin-A—A hormone produced by the placenta.

The test using the first three of these substances is called a triple screen. When the fourth substance (inhibin-A) is added, the test is called a quadruple screen. The triple screen test detects Down syndrome in 69% of the cases. The quadruple screen detects Down syndrome in 81% of the cases. The AFP test detects neural tube defects in 80% of the cases. These tests usually are done around 15–20 weeks of pregnancy. The stage of pregnancy at the time of the test is important because levels of the substances measured change during pregnancy.

What is combined screening?

The results from both first- and second-trimester tests can be combined to increase their ability to detect Down syndrome. When both the first- and second-trimester tests are used, about 90–95% of Down syndrome cases can be detected. With this type of testing, the final result may not be available until all tests are completed.

What happens when the results from screening tests are a cause for concern?

If the results of a screening test or other factors raise concerns about your pregnancy, diagnostic tests can be done to provide more information. These tests include the following:

- Detailed ultrasound exam—A type of ultrasound exam that can help explain abnormal results and provide more detailed information about the growth and development of the fetus.
- Amniocentesis—A procedure in which a small amount of amniotic fluid and cells are withdrawn from the sac surrounding the fetus and tested.
- Chorionic villus sampling (CVS)—A procedure in which a small sample of cells from the placenta is tested.

Glossary

Anencephaly: A type of neural tube defect that occurs when the fetus's head and brain do not develop normally.

Human Chorionic Gonadotropin (hCG): A hormone produced during pregnancy; its detection is the basis for most pregnancy tests.

Nuchal Translucency Screening: A special ultrasound test of the fetus to screen for the risk of Down syndrome and other birth defects.

Placenta: Tissue that provides nourishment to and takes waste away from the fetus.

Ultrasound Exam: A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.

If you have further questions, contact your obstetrician–gynecologist.