CONDITION

Birth defects testing

Learn how prenatal and newborn tests screen for and diagnose birth defects, what results mean, and when follow-up care is needed.

WHAT IT IS
Testing for conditions developing before birth

BODY SYSTEM
Multiple body systems

MAIN SYMPTOMS
No noticeable prenatal changes, abnormal fetal growth, unusual amniotic fluid, structural differences on ultrasound

WHAT CAUSES IT
Genetic, environmental, infection-related, or unknown

HOW IT IS DIAGNOSED
CVS or amniocentesis with laboratory testing

MAIN TREATMENT
Condition-specific specialist care

CAN IT BE CURED?
Sometimes - depends on the specific condition

IS IT AN EMERGENCY?
Sometimes - heavy bleeding after CVS or amniocentesis

LONG-TERM OUTLOOK
Varies by condition, with severe illness or shortened life possible

Overview

Birth defects testing looks for health conditions that develop before birth. Birth defects may affect the structure or function of the body. Examples include heart defects, differences in brain or spine development, and genetic conditions caused by changes in chromosomes or individual genes.

Testing can take place before pregnancy, during pregnancy, or shortly after birth. The best approach depends on the stage of pregnancy, medical and family history, and what information the patient wants.

There are two main types of prenatal testing:

Professional guidance recommends that prenatal screening and diagnostic options be

discussed with every pregnant patient, regardless of age or known risk factors. Choosing whether to have testing is personal. Some people want as much information as possible, while others decide that certain results would not change their plans.

Birth defects testing matters because an accurate result may guide pregnancy care, delivery planning, treatment after birth, or decisions about the pregnancy. It can also give families time to meet specialists and arrange practical or emotional support.

- Screening tests estimate the chance that a fetus has a particular condition. They do not provide a definite diagnosis.
- Diagnostic tests examine fetal cells or tissue and can confirm or rule out many chromosome or genetic conditions. They still cannot detect every possible birth defect.

Symptoms

Birth defects testing is not usually prompted by symptoms in the pregnant person. Many fetal conditions do not cause noticeable changes during pregnancy. A concern may first appear during a routine ultrasound, blood test, or review of family history.

Possible prenatal findings include:

After birth, some conditions are found during the newborn physical examination. Signs may include unusual facial or body features, a heart murmur, difficulty feeding, low muscle tone, or problems with movement. Other conditions are detected through newborn blood-spot testing, hearing screening, or pulse oximetry, which measures oxygen in the blood.

Many birth defects are not obvious at birth. Developmental, hearing, vision, heart, or metabolic conditions may become apparent later. A normal prenatal screen or ultrasound cannot guarantee that a baby has no medical condition.

- A structural difference seen on ultrasound
- Fetal growth that is smaller or larger than expected
- An unusual amount of amniotic fluid
- A screening result showing an increased chance of a chromosome condition
- A known genetic condition in one or both biological parents
- A previous pregnancy or child affected by a birth defect

Causes and risk factors

Birth defects have many possible causes. Some result from an extra, missing, or rearranged chromosome. Down syndrome, for example, is usually caused by an extra copy of chromosome 21. Other conditions result from a change in one gene.

Some birth defects involve several influences rather than one clear cause. Genes, health conditions, environmental exposures, and chance may all contribute. In many cases, the cause is never identified. A birth defect is not usually the result of something a parent intentionally did or did not do.

Factors that may affect the chance of certain conditions include:

A risk factor does not mean that a fetus has a birth defect. Many people with risk factors have unaffected pregnancies, and birth defects can occur when no risk factor is known.

People planning a pregnancy can review medications, health conditions, vaccinations, family history, and possible exposures with a clinician. Medications should not be stopped without medical advice because untreated illness may also affect pregnancy.

- A personal or family history of a genetic condition or birth defect
- A previous pregnancy affected by a chromosome or structural condition
- Increasing age of the pregnant person, which raises the chance of some chromosome conditions
- A genetic variant carried by one or both biological parents
- Certain infections during pregnancy
- Poorly controlled health conditions, such as diabetes
- Exposure to alcohol, tobacco, some drugs, or certain medications
- Abnormal prenatal screening or ultrasound findings

Diagnosis

Testing may begin before conception with carrier screening . A carrier has a genetic change that usually does not cause symptoms in that person but may be passed to a child. Carrier screening does not test the fetus directly. If a person is found to be a carrier, testing the other biological parent may clarify the chance of an inherited condition.

During pregnancy, screening options include blood tests and ultrasound.

Cell-free DNA screening , also called noninvasive prenatal screening, examines small DNA fragments from the placenta that circulate in the pregnant person's blood. It is the most sensitive screening test for common chromosome conditions, including trisomy 21, but it is not

diagnostic. Placental DNA does not always match fetal DNA, so false-positive and false-negative results can occur.

Other screening methods combine a blood test with ultrasound measurements during the first trimester. Second-trimester blood screening may estimate the chance of certain chromosome conditions and open neural tube defects, such as spina bifida.

A detailed anatomy ultrasound is commonly performed in the second trimester. It examines the brain, spine, heart, limbs, kidneys, and other structures. Ultrasound can identify many structural differences, but detection depends on the condition, fetal position, stage of pregnancy, equipment, and the examiner's experience.

When screening or ultrasound raises concern, diagnostic testing may be offered:

Laboratory testing of these samples may include a chromosome analysis, chromosomal microarray, or testing for a specific gene condition. A microarray can find small missing or extra chromosome segments that may not appear on a standard chromosome analysis.

CVS and amniocentesis are invasive procedures and carry a small risk of pregnancy loss and other complications. A clinician can explain the expected benefits and procedure-related risks for the individual situation.

Some results are uncertain. A variant of uncertain significance is a genetic change whose health effect is not yet understood. It should not be treated as a confirmed diagnosis. A genetics specialist can explain what is known and whether testing relatives or reviewing the result later may help.

After birth, diagnosis may involve a physical examination, imaging, blood testing, chromosome or gene testing, hearing screening, and pulse oximetry. Newborn blood-spot screening looks for specific serious conditions that may benefit from early treatment. The conditions included vary by location.

- Chorionic villus sampling , or CVS, removes a small sample of placental tissue. It is generally performed earlier in pregnancy.
- Amniocentesis removes a small amount of the fluid surrounding the fetus. It is usually performed from the second trimester onward.

Treatment and management

Testing itself does not treat a birth defect. Management depends on the condition, the certainty of the result, the expected health effects, and the family's goals.

After an increased-chance screening result, patients are usually offered genetic counseling, a

detailed ultrasound, and the option of diagnostic testing. Major medical or pregnancy decisions generally should not be based on a screening result alone.

If a diagnosis is confirmed, care may involve a maternal-fetal medicine specialist, genetic counselor, pediatric specialist, or fetal treatment center. These professionals can discuss how certain the diagnosis is, the range of possible outcomes, and whether more testing would change care.

Some structural conditions can be monitored with additional ultrasound or fetal heart imaging. A few may be treated before birth at specialized centers. More often, prenatal diagnosis helps clinicians choose an appropriate delivery location and prepare for treatment soon after birth.

Depending on the condition and local laws, pregnancy options may include continuing the pregnancy with planned medical care, arranging adoption, or ending the pregnancy. Counseling should be respectful, medically accurate, and based on the patient's values rather than pressure from others.

If a family continues the pregnancy, a written delivery and newborn-care plan may be helpful. It can cover where delivery should occur, which specialists should be present, what tests the baby may need, and whether immediate treatment is expected. Supportive or palliative care may also be discussed for conditions expected to cause severe illness or a shortened life.

Complications

Screening can produce an incorrect or unclear result. A false-positive result suggests increased risk when the fetus is unaffected. A false-negative result does not identify a condition that is present. These possibilities are why screening and diagnosis must be clearly distinguished.

CVS and amniocentesis may cause cramping, bleeding, infection, fluid leakage, or pregnancy loss. Serious complications are uncommon, but the exact risk depends on the procedure and clinical circumstances.

Genetic testing can also reveal unexpected information, such as a chromosome change in a biological parent or a finding unrelated to the original reason for testing. Results may create anxiety or affect other family members who could carry the same genetic change.

After CVS or amniocentesis, contact the care team promptly for fluid leaking from the vagina, significant bleeding, fever, worsening abdominal pain, regular contractions, or reduced fetal movement when movement is normally felt. Heavy bleeding, severe pain, fainting, or major breathing difficulty requires emergency care.

For a newborn, blue or gray coloring, trouble breathing, unusual unresponsiveness, or a seizure

requires emergency evaluation.

Follow up

Routine follow-up depends on the type of result. A low-chance screening result usually leads back to standard prenatal care, including recommended ultrasound examinations. It does not rule out every genetic or structural condition.

An increased-chance or unclear result should be reviewed with the prenatal clinician or a genetic counselor. Follow-up may include diagnostic testing, specialist ultrasound, fetal heart imaging, or testing of biological parents. Patients can ask for a copy of the laboratory report because the exact test and wording matter.

A confirmed diagnosis may require repeated imaging, coordinated delivery planning, and consultation with pediatric specialists. After birth, the baby may need examination or testing even when prenatal results were reassuring.

Some genetic results may be reclassified as medical knowledge improves. Families with an uncertain result can ask whether it should be reviewed again in the future.

Routine appointments are appropriate for discussing test choices, reviewing stable results, and planning further evaluation. Symptoms suggesting a complication after an invasive procedure should not wait for the next scheduled visit.

Sources

- American College of Obstetricians and Gynecologists. "Screening for Fetal Chromosomal Abnormalities." ACOG Practice Bulletin No. 226 . Obstetrics & Gynecology . 2020;136(4):e48-e69.
- American College of Obstetricians and Gynecologists. "Prenatal Diagnostic Testing for Genetic Disorders." ACOG Practice Bulletin No. 162 . Obstetrics & Gynecology . 2016;127(5):e108-e122.
- American College of Obstetricians and Gynecologists. "Carrier Screening for Genetic Conditions." Committee Opinion No. 691 . Obstetrics & Gynecology . 2017;129(3):e41-e55.
- Dungan JS, Klugman S, Darilek S, et al. "Noninvasive Prenatal Screening for Fetal Chromosome Abnormalities in a General-Risk Population: An Evidence-Based Clinical Guideline of the American College of Medical Genetics and Genomics." Genetics in Medicine . 2023;25(11):100874.

- Centers for Disease Control and Prevention. "Diagnosing Birth Defects." National Center on Birth Defects and Developmental Disabilities.
- Health Resources and Services Administration. "Newborn Screening." Maternal and Child Health Bureau. U.S. Department of Health and Human Services.