

A **soft marker** is something your healthcare provider might see during a routine pregnancy mid-trimester **ultrasound** exam. If a soft marker is found, it does not necessarily indicate an abnormality in the development or growth of the baby's organs. In many cases, it may simply represent a *normal variant*—a harmless difference in the baby's anatomy. Follow-up ultrasounds or additional testing may or may not be needed, depending on the specific circumstance. However, if other risk factors are present, finding a soft marker can mean that the fetus has an increased risk of a **genetic disorder** known as **aneuploidy**.

What is aneuploidy?

Aneuploidy is a genetic disorder where there are missing or extra **chromosomes**. The most common aneuploidy is **Down syndrome**. Instead of the normal two copies of chromosome 21, there is an extra copy. Having an extra chromosome is called **trisomy**. Other common aneuploidies include trisomy 13 and trisomy 18. Prenatal testing for these disorders is offered to all pregnant people.

What prenatal tests are available for aneuploidy?

Two different types of tests are available to detect certain genetic disorders, including aneuploidy: **prenatal genetic screening tests** and **prenatal genetic diagnostic tests**. Screening tests can tell you if the fetus has a high or low risk of having a genetic disorder. Diagnostic tests can tell you with a high degree of certainty if the fetus has a genetic disorder.

Prenatal Genetic Screening Tests

Prenatal genetic screening tests include **cell-free (cf) DNA screening** (also called **noninvasive prenatal testing**, or **NIPT**) and serum screening. Serum screening measures the amount of certain substances in your blood to assess the risk of aneuploidy. These tests are not as commonly performed as cfDNA.

Cell-free (cf) DNA screening (also called **noninvasive prenatal testing**, or **NIPT**) is a blood test that can be done starting at 10 weeks of pregnancy. This test looks for chromosome abnormalities in small fragments of genetic material (DNA) that come from the **placenta**. cfDNA screening can assess the risk of Down syndrome, trisomy 13, trisomy 18, and some sex chromosome disorders in the fetus.

cfDNA test results are reported as “low-risk” or “high-risk”:

- A low-risk cfDNA screening test result means that there is a lower chance that the fetus has a chromosome disorder compared with the general population. There is still a small chance, though, that the fetus could be affected.
- A high-risk cfDNA screening test result means that there is a higher chance that the fetus has a chromosome disorder compared with the general population. Diagnostic testing is needed to know with certainty whether the fetus is affected.

Prenatal Diagnostic Genetic Tests

Diagnostic tests include a detailed ultrasound exam, **amniocentesis**, and **chorionic villus sampling**. Prenatal genetic diagnostic testing is the most accurate way to find out whether the fetus has an aneuploidy. Diagnostic testing is offered to all pregnant people as a first genetic test (instead of a screening test), regardless of age or risk factors.

What are the risk factors for having a fetus with an aneuploidy?

The following risk factors have been linked to an increased risk of aneuploidy:

- Maternal age (usually older than age 35)
- Results of first or second-trimester genetic screening tests that show a high risk for aneuploidy
- First or second-trimester ultrasound exam findings linked to a high risk for aneuploidy


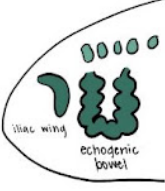
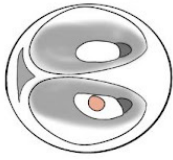





When are soft markers found?

Most soft markers are found during the second-trimester ultrasound exam, sometimes called the “**anatomy scan**.”

This exam is done between 18 and 22 weeks of pregnancy to check the development of the fetal organs and body parts. Some structural anomalies can be found during this exam.

What are the different types of soft markers?

Common soft markers found on an ultrasound are listed in the [table below](#).

Illustration	Soft marker	Description	Extra care needed during pregnancy
	Echogenic intracardiac focus (EIF)	A small area in the heart that appears as bright as bone	None
	Echogenic bowel	A brighter-than-normal area in the digestive tract	This marker has been linked to cystic fibrosis , poor fetal growth, cytomegalovirus infection (CMV) , and bleeding. Depending on your medical history, an ultrasound exam to check fetal growth in the third trimester, genetic testing for cystic fibrosis, and tests for CMV infection may be recommended.
	Choroid plexus cyst (CPC)	A small area of fluid in the part of the brain called the choroid plexus	None
	Single umbilical artery (SUA)	An umbilical cord that has only 1 instead of the normal 2 arteries	The risk of low birth weight (fetal growth restriction) can be higher in fetuses with an SUA. Growth evaluation may be recommended in the third trimester.
	Urinary tract dilatation (UTD)	Enlargement of an area of the kidney	A repeat ultrasound in the third trimester may be recommended to check the kidneys.
	Shortened humerus, femur, or both	Shortened bones of the upper arm or thigh	When found in the second trimester, a repeat ultrasound in the third trimester may be recommended to check fetal growth and limbs.
	Thickened nuchal fold	Thickened skin of the fetal neck; can also be detected during the first trimester	None
	Absent or hypoplastic nasal bone	The nasal bone is absent or smaller than expected	None

What can I expect if my ultrasound exam shows a soft marker, I have no other risk factors for aneuploidy, and my pregnancy is otherwise healthy?

If a soft marker is found on the routine anatomy scan, you will be offered a detailed ultrasound exam to check whether the marker is isolated. An isolated soft marker occurs in the absence of another anomaly, poor fetal growth, or any other soft marker.

If the exam confirms that the soft marker is isolated, the next steps depend on your cfDNA test results. If your cfDNA test result is low-risk, additional testing is usually not recommended. If you haven't had a cfDNA test, genetic counseling to assess your genetic risk factors and genetic testing may be offered. Some soft markers also prompt your doctor to look for an infection or other genetic syndromes, such as cystic fibrosis, that cfDNA or serum screening.

You have a choice in deciding whether to have a detailed ultrasound exam. SMFM recommends discussing this exam with your healthcare provider so that you can make an informed decision.

What can I expect if my detailed ultrasound exam shows multiple soft markers or if I have other genetic risk factors for aneuploidy?

Diagnostic testing with amniocentesis may be offered depending on the type of soft marker, your prior cfDNA test results and detailed ultrasound exam results, and the health of your pregnancy. An amniocentesis can tell with a high degree of accuracy whether the fetus has an aneuploidy. It can also identify other genetic disorders that you and your healthcare provider can request.

Your preferences and beliefs about what you would do if amniocentesis results show a disorder play a role in whether you want to have this testing. Your healthcare provider can explain what is known about the soft marker, review your risk factors, and discuss all of your options to help you and your family make an informed decision.

Will I need special care during pregnancy if a soft marker is found?

Some isolated soft markers are linked to pregnancy problems unrelated to aneuploidy. For this reason, your healthcare provider may recommend extra tests and further ultrasounds for some soft markers. The table explains the recommended tests and why they might be needed.

Quick Facts

- A soft marker is a minor finding seen during a pregnancy ultrasound exam.
- A soft marker is usually a variation of normal fetal development. But when other risk factors are present or there is more than 1 soft marker, it can mean that the fetus is at increased risk of a genetic disorder known as aneuploidy (an abnormal number of chromosomes).
- A detailed ultrasound exam is offered if a soft marker is found.
- If the detailed ultrasound exam confirms that the soft marker is isolated, and you have had a low-risk prenatal screening test (e.g., cell-free DNA test) result, you can be reassured that the chance of aneuploidy in the fetus is very low. No other genetic testing is needed in this case.
- Some isolated soft markers are linked to pregnancy problems unrelated to aneuploidy. For this reason, your healthcare provider may recommend extra tests and imaging for some soft markers.
- If the soft marker is not isolated or if there are other risk factors for aneuploidy, an amniocentesis may be offered depending on the type of soft marker, your prior cell-free DNA test and detailed ultrasound exam results, and the health of your pregnancy.

Glossary

Amniocentesis: A procedure in which a sample of amniotic fluid is removed from the uterus during pregnancy and tested to look for genetic problems in the fetus.

Anatomy Scan: A routine ultrasound exam done between 18 and 22 weeks of pregnancy to check the development of the fetus's body parts and organs.

Aneuploidy: A genetic disorder in which there are missing or extra chromosomes.

Cell-free (cf) DNA Screening Test: A prenatal screening test that looks for certain chromosomal disorders in the fetus. It uses small pieces of DNA (genetic material) from the placenta that circulate in the blood of a pregnant person. Also called noninvasive prenatal testing (NIPT).

Chorionic Villus Sampling (CVS): A procedure in which a small sample of the villi, a part of the placenta, is removed and tested to look for genetic problems in the fetus.

Chromosomes: The structures inside cells that carry genes, the pieces of hereditary material passed down from parents to offspring.

Cystic Fibrosis: An inherited disorder caused by the production of thick, sticky mucus, which can build up in organs such as the lungs and pancreas and lead to damage, blockages, and infections. It is the result of a gene passed down in families.

Cytomegalovirus (CMV): A virus that can be passed from a pregnant person to the developing fetus. Babies born with CMV can have problems such as jaundice, an enlarged liver and spleen, and a skin rash. Babies without symptoms at birth can develop deafness, cognitive difficulties, eye problems, and seizures.

Detailed Ultrasound Exam: An ultrasound exam that provides a comprehensive assessment of fetal body parts and organs.

Fetus: The stage of development during pregnancy from 9 weeks to birth.

Genetic Disorder: Any disorder caused by a genetic change, an abnormal number or structure of chromosomes, or a combination of genetic and other factors.

Noninvasive Prenatal Testing (NIPT): Prenatal blood tests that measure certain substances in the mother's blood or analyze fragments of placental DNA to screen for aneuploidy.

Placenta: A special organ that develops during pregnancy. It allows the transfer of nutrients, antibodies, and oxygen to the fetus. It also makes hormones that sustain the pregnancy.

Prenatal Genetic Diagnostic Tests: Tests done during pregnancy that determine with a high degree of accuracy whether a genetic disease or disorder is present in the fetus.

Prenatal Genetic Screening Tests: Tests done during pregnancy that assess the chance that a genetic disorder is present in the fetus.

Sex Chromosome Disorder: A genetic disorder in which there is an abnormal number or structure of the sex chromosomes.

Sickle Cell Disease: An inherited disorder that causes blood cells to become crescent (sickle)-shaped, making it difficult for them to pass through small blood vessels.

Soft Marker: A finding on ultrasound that is usually a normal variation but can be associated with a chromosome disorder.

Trimester: The three-month periods in which pregnancy is divided. The first trimester is months 1 to 3 (weeks 1 to 12); second trimester is months 4 to 6 (weeks 13 to 27); and the third trimester is months 7 to 9 (weeks 28 to 40).

Ultrasound: Use of sound waves to create images of internal organs or the fetus during pregnancy.

To find a maternal-fetal medicine subspecialist in your area, go to [Find an MFM - Society for Maternal-Fetal Medicine](#). The Society for Maternal-Fetal Medicine's Patient Education Series reflects the content of current, published SMFM practice guidelines. Each series document has undergone extensive internal review prior to publication. Patient Education documents should not be used as a substitute for the advice and care of a medical professional.

Last updated: June 2026