

Testing for Fetal Aneuploidy

What is aneuploidy?

Chromosomes contain large amounts of a baby's genetic material, which is what makes us who we are. Aneuploidy (AN-yuh-ploy-dee) happens when a baby has too few or too many chromosomes. This can cause a chromosome disorder like Down syndrome.

What are aneuploidy screening tests?

Screening tests give information about your baby's risk of a chromosome disorder. This testing is optional, and not all pregnant women have it. Some of these tests also screen for problems with your baby's brain or spine. Your doctor will help you decide if these tests are right for you.

How does screening work?

Screening tests usually are done at 10 weeks of pregnancy or later. Some screening tests are done in the first trimester, some in the second trimester, and some in both. It depends on your baby's risk and the type of testing you choose.

There are several ways to do the screening. One type of screening is an ultrasound. This uses sound waves to

measure the thickness of the liquid under the skin on your baby's neck. If the thickness is not normal, your baby may not have the normal number of chromosomes. Your doctor may also do a blood test to check your baby's risk of having a chromosome disorder.

Screening tests only determine whether your baby is at risk of a chromosome disorder. They can't tell for sure.

How do I know if my baby has aneuploidy?

If you choose to have a screening test and it shows that your baby is at risk, your doctor may talk to you about more testing. Two tests can tell for sure whether your baby has a chromosome disorder. They both have a small risk of causing a miscarriage. They are done by a specialist.

One test is called amniocentesis (AM-nee-oh-sen-TEE-sis). The doctor uses a needle to take a small amount of fluid from the bag of water around the baby.

The other test is called chorionic villus (CORE-ee-AH-nick VIL-us) sampling. The doctor uses a needle to take a small piece of the placenta. This test is done earlier in the pregnancy than amniocentesis.

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This handout is provided to you by your family doctor and the American Academy of Family Physicians. Other health-related information is available from the AAFP online at <https://familydoctor.org>.

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Fetal Aneuploidy *(continued)*

Where can I get more information?

Your doctor

AAFP's Patient Information Resource

<https://familydoctor.org/screening-in-pregnancy>

American College of Obstetricians and Gynecologists

<https://www.acog.org/Patients/FAQsGenetic-Disorders>

<https://www.acog.org/Patients/FAQs/Prenatal-Genetic-Testing-Chart-Infographic>

<https://www.acog.org/Patients/FAQs/Cell-free-DNA-Prenatal-Screening-Test-Infographic>

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