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Advanced Aneuploidy Screening with Cell-Free DNA

A guide to prenatal testing

We have written this handout to help you decide if you want to have an advanced aneuploidy screening test. Having this test is up to you. Some people do not find this test helpful. You may refuse testing at any time. This handout explains the screening test and some medical terms. Talk with your healthcare provider to learn more.

What is advanced aneuploidy screening with cell-free DNA?

Having this screening test is up to you.

Advanced aneuploidy screening with cell-free DNA is done using blood drawn from your arm. It can be done starting at 10 weeks of pregnancy. The test screens for specific disorders in your baby that are related to *chromosomes*. Chromosomes are the "packages" of DNA contained in our cells.

DNA is *deoxyribonucleic acid*, a molecule that carries genetic information from one generation to the next. Most of our DNA is stored inside the cells of our body. *Cell-free DNA* is not contained within a cell. Everyone has some cell-free DNA in their blood. When you are pregnant, most of the cell-free DNA in your blood is from you, but some is from your fetus.

In this test, the lab measures the total amount of cell-free DNA from chromosomes 21, 18, 13, and X in your blood. If there is an abnormal amount of DNA from one of these chromosomes in your blood, there is a high chance that your baby has aneuploidy for that chromosome.

What is aneuploidy?

Most people have 2 copies each of 23 chromosomes, for a total of 46. *Aneuploidy* is when a person has extra copies or missing copies of certain chromosomes. There are different types of aneuploidy.

Trisomy is when there are 3 copies of a certain chromosome in all of the cells in the body. *Monosomy* is when there is only 1 copy of a certain chromosome in all of the cells in the body.

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- **Trisomy 21** is when there are 3 copies of the chromosome number 21 in all cells. It is the most common cause of *Down syndrome*.
- **Trisomy 18** is when there are 3 copies of chromosome 18 in all cells. This condition is also called *Edward syndrome*.
- **Trisomy 13** is when there are 3 copies of chromosome 13 in all cells. This condition is also called *Patau syndrome*.
- **Monosomy X** is when there is only 1 copy of the X chromosome. It is the most common cause of *Turner syndrome*.

What are the benefits of this test?

- It is the most accurate screening test for aneuploidy we have today.
- Taking this test does **not** pose any risk to your fetus.
- A **normal result** means that the chances of your baby having aneuploidy are very low. Getting a normal result may be reassuring and help lower anxiety you may have about your baby's health.

What are the limitations of this test?

- Advanced aneuploidy screening detects **almost** all cases of Down syndrome, trisomy 18, trisomy 13, and monosomy X. **But, it will not tell you for sure whether or not your baby has any of these disorders.**
- Rarely, the result will be abnormal, even though the fetus **does not** have aneuploidy.
- Rarely, the result will be normal, even though the fetus **does** have aneuploidy.
- About 1% of blood samples (1 out of 100) cannot be interpreted. If this happens, you will not get a result.
- Advanced aneuploidy screening does not test for any other chromosome disorders, other birth defects, or other genetic conditions.

Questions?

Your questions are important. Call your doctor or health care provider if you have questions or concerns.

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If the test is abnormal, how can I confirm the results?

If you want to confirm that your baby has aneuploidy:

- Before you deliver, you can have a *chorionic villus sampling* (CVS) or an *amniocentesis* test. Please ask for handouts about these tests.
- After delivery, a small blood sample can be taken from your baby to test your baby's chromosomes.

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