UW Medicine
UNIVERSITY OF WASHINGTON MEDICAL CENTER

Advanced Prenatal Screening with Cell-Free DNA

At UW Maternal Fetal Medicine in Yakima

At UW Maternal Fetal Medicine in Yakima, we partner with our patients and families in making decisions about their healthcare. This handout gives information to help you decide if you want to have cell-free DNA screening.

It is your decision whether or not to have this test. Talk with your pregnancy care provider to learn more.

What is advanced prenatal screening with cell-free DNA?

Advanced screening with cell-free DNA is a blood test done during pregnancy. *Cell-free DNA* is genetic material from the pregnancy. It is normal for a pregnant woman to have some of this material in her blood.

This test tells us the risk of your baby having health conditions such as *Down syndrome, trisomy 18,* or *trisomy 13.* It can also tell the risk of your baby having an *X chromosome issue*. The test does **not** diagnose these conditions, but it can tell us if your baby might have them.

Who can have this test?

This test can be done after the 10th week of pregnancy. It is usually done for women who have a higher risk of having babies with these conditions. Women are at higher risk if they:

- Will be **35 years or older** at their expected due date
- Have a personal or family history of certain **inherited (*genetic*) conditions**
- Had a **fetal ultrasound** that showed a possible problem
- Had a positive ***serum screening*** blood test

Talk with your pregnancy care provider if you have any questions about prenatal screening.

How is a cell-free DNA screen done?

A cell-free DNA screen has 2 parts. The pregnant woman will:

- Meet with a genetic counselor
- Have 1 blood draw from her arm

Meeting with a Genetic Counselor

Talking with a genetic counselor can help you decide what tests, if any, are right for you. This visit is usually on the same day as your blood draw. It usually lasts about 30 minutes and is at the UW Maternal Fetal Medicine clinic in Yakima.

At this visit, a genetic counselor will:

- Ask you about your pregnancy and family history
- Explain Down syndrome, trisomy 18, trisomy 13, and X chromosome issues
- Explain how the cell-free DNA test works and how you will get results

Blood Draw

The blood draw is usually done in our clinic right after you meet with the genetic counselor. Blood is taken from your arm for this test. The blood sample will be sent to the lab you and the genetic counselor have chosen.

The blood test checks your blood for cell-free DNA from certain *chromosomes* (the parts of a cell that contain genetic information). The amount of this DNA is usually different if a baby has Down syndrome, trisomy 18, trisomy 13, or an X chromosome issue.

What will I learn from this screening?

Most people have 23 pairs of chromosomes, for a total of 46. The screening will check your baby for these disorders:

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

- **XXY**, when there are 2 copies of the X chromosomes and 1 copy of the Y chromosome in all the baby's cells. XXY causes a condition called Klinefelter syndrome.

How accurate is cell-free DNA testing?

Cell-free DNA testing detects **nearly** all cases of Down syndrome, trisomy 18, trisomy 13, and X chromosome issue. **But, it will not tell you for sure if your baby has any of these conditions.**

What are the benefits of this test?

- A cell-free DNA test gives you information about your baby's health without any risk to your pregnancy. It is the best test we have for checking an unborn baby's chromosomes.
- Because the results tell us more about your baby's health, they may also help us give you better care during your pregnancy.
- The test does not pose any risk to you or your baby.
- Normal results may help reassure you about your baby's health.

What are the limitations of this test?

- An *abnormal* result can cause worry or stress for the parents and family.
- This test does not give information about *spina bifida* or other birth defects or genetic conditions. Another blood test may be done between 15 weeks and 22 weeks of pregnancy to check for spina bifida.

What can I expect after the test?

Most women receive their results within 2 weeks after their blood draw. Results can either be *normal* or *abnormal*:

Normal Results

A normal result means that you have a **low risk** of having a baby with Down syndrome, trisomy 18, trisomy 13, or a sex chromosome issue. But, your baby may still have one of these conditions. It is rare, but sometimes the test result is normal even when the baby **does** have one of these conditions.

Abnormal Results

An abnormal result means that your baby **may** have Down syndrome, trisomy 18, trisomy 13, or an X chromosome issue. But, it does not tell us for sure.

It is rare, but sometimes the screening result is abnormal even when the baby **does not** have one of these conditions. Other tests can be done to confirm that the baby has one these conditions.

If the test is abnormal, what can I do next?

If your test results are abnormal, you will be scheduled to meet with a genetic counselor. At this visit, you will:

- Review your results
- Talk about possible outcomes for your pregnancy and your baby
- Ask any questions you have
- Find out about more tests you can take

If you want to confirm that your baby has one of these genetic conditions:

- **Before you deliver:** You can have a *chorionic villus sampling* (CVS) or an *amniocentesis test*. Please ask for handouts that explain these tests.
- **After you deliver:** We can take a small blood sample from your baby to test your baby's chromosomes.

Questions?

Your questions are important. If you have any more questions about cell-free DNA, talk with your provider before you sign any consent forms.

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