my body + my baby

prenatal genetic screening

What is prenatal genetic screening?

This test is to see if your pregnancy is at risk for having the following conditions:

- **Down syndrome, also called Trisomy 21**, is a condition that causes intellectual disability, heart defects, and other health issues. Down syndrome is caused by an extra chromosome in the developing fetus. Although the chances of Down syndrome increase with the age of the pregnant person, it can happen at any age.
- **Trisomy 18 and Trisomy 13** are both caused by an extra chromosome as well. These are very severe conditions that are often life-threatening.
- **Open neural tube defects**, which are problems with the spinal cord, such as spina bifida.

Prenatal genetic screening is an option.

Whether you get genetic screening is up to you. There are both benefits and risks to it. Sometimes babies born with genetic problems can require extra care at birth, so it can be helpful for your care team to be prepared. And, if you choose, screening will allow you to make a more informed decision about continuing your pregnancy. However, a positive or abnormal result could lead to more testing, including higher-risk testing. Positive or abnormal results can also happen when in reality everything is fine. This can lead to unnecessary stress. To make the decision whether to not to proceed with prenatal genetic screening, talk to your midwife, your family, and your friends.

What are my screening options?

Integrated screening

- This test uses measurements from two blood tests and a first trimester ultrasound. This information is combined to tell you more about your pregnancy. Medical insurance typically covers this for all patients.
- The blood tests measure proteins that the pregnancy makes as it develops. The first blood test is taken between 11 and 13 weeks of your pregnancy, and the second is taken ideally between 15 and 18 weeks.
- Between 11 and 13 weeks, you will have an ultrasound. This will measure the length of the fetus and the fluid-filled space at the back of the neck.
- Results will be available several days after your second blood test at 16-18 weeks.

Cell Free DNA screening

- Cell free DNA testing is a single blood test performed at ten weeks or later. The laboratory looks for the fetus's chromosomes that are in cells circulating within your blood. If there is a higher than expected amount of chromosomes present in your blood, this suggests that there may be a genetic condition.
- This testing is NOT always covered by medical insurance. It is slightly more accurate than Integrated Screening, and provides results earlier in pregnancy, but is more expensive. Sometimes insurance will cover it if a pregnant person is over age 35 or if they have a history of having a baby with a genetic condition.
- Results typically take approximately two weeks from the time of the blood draw.

What do my genetic screening results mean?

Regardless of which type of testing you choose:

If your result is positive or abnormal:

- There is still more testing needed to determine if your fetus has any genetic conditions. Other factors may affect the test results. Your midwife may suggest further testing such as a detailed ultrasound or an amniocentesis to get more information.

If your result is negative or normal:

- You are considered at low-risk to give birth to a baby with Down syndrome, trisomy 18 or 13, or an open neural tube defect.

What are the side effects of genetic screening?

The screening options on the previous page include drawing your blood and/or performing an ultrasound, and pose no risk to the fetus. You may have bruising or soreness in your arm at the site of the blood draw.

Source: https://www.uwmedicine.org/sites/stevie/files/2018-11/Guide-Prenatal-Testing-English.pdf