



# *First Trimester Screening for Down Syndrome*

## **Introduction**

First trimester screening is an optional test done between weeks 10 and 14 of pregnancy. The result may show whether you have an increased risk of giving birth to a baby with Down syndrome.

Every woman regardless of age has a risk of having a child with Down syndrome. The risk increases as a woman gets older.

This material provides information about Down syndrome and the first trimester screening test.

## **What is Down syndrome?**

Down syndrome is a genetic disorder that causes varying degrees of mental retardation and can lead to other problems such as heart defects, poor vision and hearing loss. It is not possible to know before birth how Down syndrome will affect babies. Most babies born with Down syndrome live through their first year, and almost half reach 60 years of age.

Though Down syndrome is a serious disorder, early intervention may help those who have the condition lead fulfilling lives. Intervention typically begins in infancy. Medical specialists such as physical therapists, speech pathologists and occupational therapists may help children with Down syndrome develop their skills as fully as possible. Many children who have Down syndrome live with their families, attend school and learn to read and write. Many adults with Down syndrome have jobs.

## **What is first trimester screening for Down syndrome?**

First trimester screening is used to determine your risk of having a baby with Down syndrome. The screening is not harmful and has two parts, usually done the same day:

- A blood sample is drawn from your arm to measure certain protein and hormone levels.
- An ultrasound, also called sonography, is a procedure that uses high-frequency sound waves to produce an image of your developing pregnancy. The ultrasound is used to measure the size of a specific region on the back of a developing neck. When Down syndrome is present, more fluid than usual may collect in this tissue.

First trimester screening cannot determine if you will give birth to a baby with Down syndrome, but the information from the blood test and ultrasound, along with your age, may help determine whether the risk is increased. If the screening result shows a higher risk, further tests (diagnostic tests) can be done to determine if your baby could be born with Down syndrome.

First trimester screening is optional. The decision to have it is yours. Mayo Clinic Laboratories offers a test, Mayo ID: 1STT / First Trimester Maternal Screen, and can be ordered by your physician. If you want the test, ask your health care provider to help you set up an appointment. If you have the screening, you may first meet with a genetic counselor to discuss the screening and your questions.

Preparation for a first trimester screening may require that you follow certain instructions such as coming to the ultrasound with a full bladder. If your appointment for the screening is in a few days and you have not received preparation instructions, talk to your health care provider. It is important to follow instructions for getting ready.

### **What does the result mean?**

The result usually is ready within three working days after the screening and is sent to your health care provider. Ask when and how you will receive the screening result. Discuss your result with your health care provider. The result is reported as either screen negative or screen positive.

**What does a screen negative result mean?** Screen negative means the risk of Down syndrome is low.

**What does a screen positive result mean?** Screen positive means the risk of Down syndrome is increased above a risk cut-off determined by the laboratory. Your health care provider can provide you with more information about the specific risk cut-off. If you have a screen positive result, you are offered the opportunity to have a diagnostic test to find out whether you could have a baby with Down syndrome. About 1 out of 20 women screened has a positive screen result; however, most women with positive screen results do not have a baby with Down syndrome.

### **What are the limitations of first trimester screening?**

First trimester screening identifies most, but not all, pregnancies with Down syndrome. Rarely, a woman with a screen negative result gives birth to a baby with Down syndrome. Approximately 8 out of 10 Down syndrome pregnancies are identified as being screen positive through first trimester screening. Talk with your health care provider if you would like more information.

### **Can first trimester screening find other abnormalities?**

The screening may also show the possibility of a chromosome abnormality called trisomy 18. Trisomy 18, also known as Edward syndrome, is caused by an extra copy of chromosome 18 and can be fatal. Infants born with trisomy 18 typically have small birth size, mental retardation and many developmental abnormalities. If you have a screen positive result for trisomy 18, you may choose to have a diagnostic test that may determine if your baby could be born with trisomy 18.

First trimester screening does not check for a group of serious birth defects called neural tube defects. These defects are caused by improper development of the brain or spinal cord. Spina bifida is one example. Between weeks 15 and 18 of pregnancy, you may have a blood test that measures a substance called alphafetoprotein (AFP). This test may help diagnose neural tube defects. In addition, an ultrasound done at this time may also detect neural tube defects.

## **What happens if Down syndrome is diagnosed?**

You may meet with a genetic counselor and your health care provider to discuss the diagnosis and your options.

## **Contacting your health care provider**

If you have questions about the first trimester screening or this information, discuss them with your health care provider.

*This material is for your education and information only. This content does not replace medical advice, diagnosis or treatment. New medical research may change this information. If you have questions about a medical condition, always talk with your health care provider.*

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