



Quest
Diagnostics®



**Sequential Integrated
Screening**

Prenatal Screening



You just want your baby to be healthy.

Your pregnancy can be a busy time, yet you and your doctor need to understand how your pregnancy is developing.

Prenatal screening (testing before birth) can tell you if your baby is at greater risk of certain birth defects.

The following questions and answers will help you understand the Sequential Integrated Prenatal Screening test and what your next steps might be if your results show the baby is at increased risk.



Is prenatal screening for you?

Who should have prenatal screening?

The American College of Obstetricians and Gynecologists recommends that prenatal screening for certain types of genetic disorders—including Down syndrome, trisomy 18, and neural tube defects—be offered to all pregnant women, regardless of their age.

Do I have to have prenatal screening?

You should discuss your options with your doctor, but **the decision about whether or not to have prenatal screening is up to you.** The following information will help you decide.



Screening for the following disorders

What is Down syndrome?

Down syndrome (trisomy 21) is caused by an extra copy of chromosome number 21. Babies with Down syndrome have a distinct physical appearance and mild to moderate intellectual disability. They also have an increased chance of having other health problems, such as heart defects.



About one in every 700 babies is born with Down syndrome.

What is trisomy 18?

Edwards syndrome (trisomy 18) is caused by an extra copy of chromosome number 18. Babies with Edwards syndrome have severe intellectual disabilities, often with life-threatening birth defects. Only 5% to 10% of babies with Edwards syndrome survive to their first birthday.



About one in every 6,000 babies is born with Edwards syndrome.

What is a neural tube defect?

The neural tube develops into the spine and brain of the baby very early in pregnancy. If the neural tube doesn't develop properly, the result can be an open defect in the baby's spine and/or skull. Neural tube defects are not associated with the mother's age.



One to two out of every 1,000 babies are born with neural tube defects.

Screening

What is the Sequential Integrated Screen?

It is a test that provides information about the baby's chance of having Down syndrome, trisomy 18, or a neural tube defect.

It can detect:

- About 92% of babies with Down syndrome
- About 90% of those with trisomy 18
- About 80% of babies with a neural tube defect

How is the Sequential Integrated Screen performed?

The test has two steps:

- 1** In the first step, a small sample of blood will be taken during the first trimester of pregnancy. This sample is used to measure the level of two substances in your blood.
- 2** Additionally, an ultrasound is done to take a measurement at the back of the baby's neck. This measurement is called nuchal translucency.

The results from step 1 are used to identify pregnancies with a high risk for Down syndrome or trisomy 18.

If the results from step 1 indicate the baby is at high risk, your doctor may suggest a second step: diagnostic testing. In step 2, the level of four substances is measured with a blood test in the second trimester.

If the results from step 1 *do not* indicate a high risk, no result is reported until the second blood sample is taken.

The measurements from step 1 are combined with the measurements from step 2 to determine the baby's risk.



Results

What if my result is “in process”?

After step 1, “in process” means that so far, the baby is not known to be at high risk of Down syndrome or trisomy 18. You will proceed to step 2.

After step 2, “screen negative” means that the baby likely does not have Down syndrome, trisomy 18, or a neural tube defect. But a negative screen does not guarantee the birth of a baby without these conditions.

What if my result is “screen positive”?

If your result is “screen positive” after step 1, it means that the baby has a higher risk of having Down syndrome or trisomy 18. It *does not* mean that the baby is definitely affected. You will not proceed to step 2. Instead, you need to consider diagnostic testing. Your doctor can help you understand the risk and explain the additional tests that may be recommended.

If your result is “screen positive” after step 2, it means there is an increased chance your baby may have Down syndrome, trisomy 18 or a neural tube defect. It *does not* mean that your baby definitely has one of these disorders.



Your doctor can help you understand the risk and explain the additional tests that may be recommended.

What additional tests might my doctor recommend?

If your test result is “screen positive” after step 1, your doctor may recommend chorionic villus sampling (CVS) or amniocentesis.

If your test result is “screen positive” after step 2, your doctor might order a detailed ultrasound and/or recommend amniocentesis.

Since there is a small risk of miscarriage associated with amniocentesis, you should discuss the risks and benefits of each procedure with your doctor.

What if the additional test results are negative?

If the additional test results are negative, your baby is likely to be healthy. However, negative test results do not guarantee that your baby will not have some form of birth defect.

What if the additional test results are positive?

If an additional test result is positive, genetic counseling will help you learn more about your baby's condition. This will help you make decisions in the best interests of you and your family.



Summary

The Sequential Integrated Screening:

- 1 Helps you find out your chances of having a baby with Down syndrome, trisomy 18, or a neural tube defect.
- 2 Is performed in two steps, one during the first trimester and the other during the second trimester of pregnancy.
- 3 Allows an earlier diagnosis if you are “screen positive” after step 1.
- 4 Detects about 92% of babies with Down syndrome, 90% with trisomy 18, and 80% with an open neural tube defect.
- 5 Does not guarantee a healthy baby if the result is “screen negative.”
- 6 Is not diagnostic. A “screen positive” result does not necessarily mean your baby has a problem. Additional diagnostic testing is needed.



For more information, visit [QuestDiagnostics.com](https://www.questdiagnostics.com) and talk with your healthcare provider.

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