Screening for Down Syndrome During Pregnancy: What You Need to Know

Highlights

Down syndrome is a common genetic \(chromosomal\) disorder that leads to intellectual disability.
You can have a screening test to find out if your baby is likely to be born with Down syndrome.
Screening tests are usually done during the first trimester of pregnancy.
Talk to your doctor or a genetic counselor to decide if screening is right for you.

In This Article

Questions for Your Midwife or Doctor
Tools and Assistance

Down Syndrome Overview

Screening for Down Syndrome

Reasons for Screening for Down Syndrome

Planning for medical care and support after birth.
Making decisions about additional diagnostic testing.
Monitoring the pregnancy more closely if needed.

Preparing for Screening for Down Syndrome

Methods of Screening for Down Syndrome

Combined First Trimester Screening: This is the most common screening method and involves: A blood test between week 10 and week 13 of pregnancy. A nuchal translucency ultrasound scan between week 11 and week 14 of pregnancy. The blood test measures two substances: Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta. Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone. These results, combined with the nuchal translucency measurement, along with maternal age and weight, estimate the likelihood of the baby having Down syndrome. This test detects about 85% to 90% of cases.
A blood test between week 10 and week 13 of pregnancy.
A nuchal translucency ultrasound scan between week 11 and week 14 of pregnancy.
The blood test measures two substances: Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta. Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone.
Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta.
Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone.
These results, combined with the nuchal translucency measurement, along with maternal age and weight, estimate the likelihood of the baby having Down syndrome. This test detects about 85% to 90% of cases.
Non-Invasive Prenatal Testing \(NIPT\): NIPT is a more advanced screening

method that involves a blood test taken after 10 weeks of pregnancy. It analyzes fetal DNA circulating in the mother’s blood and is 99% accurate in detecting Down syndrome. This test does not require an ultrasound but can be expensive.
This test does not require an ultrasound but can be expensive.
Second Trimester Serum Screening: If the first-trimester screening is missed, an alternative blood test can be taken between weeks 14 and 20 of pregnancy. This test measures certain proteins and hormones in the mother’s blood and can detect about 75% of Down syndrome cases.
This test measures certain proteins and hormones in the mother’s blood and can detect about 75% of Down syndrome cases.

A blood test between week 10 and week 13 of pregnancy.
A nuchal translucency ultrasound scan between week 11 and week 14 of pregnancy.
The blood test measures two substances: Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta. Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone.
Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta.
Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone.
These results, combined with the nuchal translucency measurement, along with maternal age and weight, estimate the likelihood of the baby having Down syndrome. This test detects about 85% to 90% of cases.

Pregnancy-associated plasma protein A \(PAPP-A\), which is made in the placenta.
Free beta human chorionic gonadotropin \(free ˛†-hCG\), a pregnancy-related hormone.

This test does not require an ultrasound but can be expensive.

This test measures certain proteins and hormones in the mother’s blood and can detect about 75% of Down syndrome cases.

Cost of Screening for Down Syndrome

Understanding Screening Test Results

Low-risk result: Indicates that the baby is unlikely to have Down syndrome.

High-risk result: Suggests a higher probability that the baby has Down syndrome, and further diagnostic testing may be recommended.

Diagnostic Tests for Down Syndrome

Chorionic Villus Sampling \(CVS\): A test performed between weeks 10 and 13 that examines placental tissue for chromosomal abnormalities.
Amniocentesis: A test conducted after week 15 that analyzes amniotic fluid surrounding the baby.

Optional Nature of Down Syndrome Screening

Questions for Your Midwife or Doctor

Why are you offering me this test?
What does the test involve?
Do I need to prepare in any way for the test?
When will I receive the results?
Who will discuss the results with me?
What are my options if the test results show a high probability of Down syndrome?

Tools and Assistance

Information on Down syndrome and what to expect.
Parental guidance and counseling.
Resources for early intervention and medical care.