



What happens if I have a high risk (screen positive) result?

If the result of the screening test shows the chance of your baby having Down's syndrome is greater than the national cut off risk, we will offer you a diagnostic test. Overall about one in 33 (3%) women screened has a high risk (screen positive) result and are offered a diagnostic test.

You can decide not to have a diagnostic test

This carries no risk to your pregnancy but may leave some uncertainty until after your baby is born.

You can decide to have a diagnostic test

This will give you a definite result but carries a small risk of miscarriage.

You need to think about what you would do if you found yourself in this position. If you would not be happy with either of the above options, you need to consider carefully whether the screening process is the right choice for you.

Diagnostic procedures for Down's syndrome

There are two diagnostic procedures offered for Down's syndrome:

- Chorionic Villus Sampling
- Amniocentesis

Are these procedures safe?

These procedures do carry some risk, and this is why we don't offer them to everybody.

1. Risk of miscarriage = For every 1000 women who have amniocentesis, one will miscarry. And for every 1000 women who have CVS, one or two will miscarry.
2. Risk of infection = For 1 in 1000 women undergoing these procedure.
3. Risk of sample culture failure = For 1 in 10,000 women undergoing these procedure. If the sample of amniotic fluid is contaminated with maternal blood or if fetal cells are not in adequate amount in sample so as to get fetal DNA in adequate amount or fetal cell culture failed to grow then we need to repeat the procedure.

What are the possible results from diagnostic tests?

- Your baby does not have Down's syndrome; this is the most common result.
- No or unclear result
Very rarely a CVS or amniocentesis does not produce a result because the cells do not grow, or the results are not clear. If this happens, further tests may be offered.
- Your baby has Down's syndrome
A very small number of women will get this result. It is not possible to tell how affected your baby will be.

What happens if my baby has Down's syndrome?

There are then three options – it is entirely your decision which one you choose:

- Some parents will decide to continue with the pregnancy, make plans and prepare for any extra challenges they might face bringing up a child with Down's syndrome
- Some parents may feel that they are unable to bring up their child themselves and may want to consider adoption as an option
- Some parents decide they do not want to continue with the pregnancy and will choose to have a termination. You have the opportunity to discuss your results with our fetal medicine specialist. You will have time to decide what you are going to do and will be supported by us.



Contact Us

-  Bhandari Hospital and Research Center (BHRC),
Opposite Meghdoot Garden, Vijay Nagar, Indore (MP) 452010
-  Helpline: 78699 14699, 98930 34666, 0731-4733666
-  aayushyaindore@gmail.com
-  www.fetalmedicineindore.com



First Trimester Screening

Helpful screening modality for common genetic abnormalities

What causes Down's Syndrome?

Inside the cells of our bodies there are tiny thread like structures called chromosomes. These chromosomes carry the genes that determine how we develop. Most people have 23 pairs of chromosomes in each of their cells. When our bodies produce the cells needed to make babies, the chromosome pairs divide and rearrange themselves. Sometimes these pairs of chromosomes accidentally do not divide correctly, and this can cause the baby's cells to have an extra copy of chromosome 21. This causes Down's syndrome (also called Trisomy 21). The extra chromosome cannot be removed from the cells, so there is no cure for the condition.

Should I have the test for Down's syndrome?

Only you can decide that. Some women want to find out if their baby has Down's syndrome, and some do not. Information about the tests and how they work can help you make up your mind. However we recommend this test for all pregnant women.

Will the tests tell me for certain if my baby has Down's syndrome?

We begin by offering all women a test that carries no risk of miscarriage. This type of test is called a screening test. Screening tests do not give a definite answer, but they do help us to tell which babies have an increased risk of having Down's syndrome. We then offer diagnostic tests to the women with an increased risk. These are tests that give definite information but they do carry a small risk of miscarriage. This is why we only offer a diagnostic test to women who have been found to have an increased risk.

Combined first trimester screening test?

At Aayushya Center For Fetal Medicine you can choose to have a screening test for Down's syndrome that involves a scan and blood test. This test can be taken from 11 weeks and 2 days up to 14 weeks of pregnancy (depending upon the length of the fetus).

All babies have a very thin film of fluid under the skin at the back of their neck (nuchal area) at this stage of pregnancy. Provided your baby is lying in a suitable position, we can measure this as part of the scan and this is known as the nuchal translucency measurement. If the nuchal translucency measurement is above 3.5mm, it is considered abnormal and will be discussed with you at the time of the scan, even if you choose not to have Down's syndrome screening.

A blood sample is taken from the mother's arm during the same appointment as the scan (providing you are between 11 weeks and 2 days and 14 weeks and 1 day pregnant). The blood test measures the amount of some hormones that are found naturally in the mother's blood. These have passed to the mother from the baby. If a baby has Down's syndrome, different amounts of these hormones can sometimes be found in the mother's blood. This is the combined test and the result is given as a risk figure. A risk can also be calculated for twin pregnancies also.

Second trimester screening

If you wish to have Down's syndrome screening and your pregnancy is 14 weeks and over when you come to the first trimester screening clinic, you will be offered second trimester screening. This is a blood test and can be carried out between 15 weeks – 20 weeks of pregnancy. The result is given as a risk figure.

What is a high risk (screen positive) result?

Both the screening tests for Down's syndrome give results in the form of, 'one in ...'; for example 'one in 100' (1%) or 'one in 1000' (0.1%). These numbers tell us how likely it is that the baby has Down's syndrome. For example, the result 'one in 100' means that there is one chance in 100 that the baby has Down's syndrome. The

result 'one in 1000' means that there is one chance in 1000 that the baby has the condition. It is important to understand that as the second number in the result gets bigger, Down's syndrome gets less likely. **If your screening result is above the national cut-off figure (>1:20 to 1:200) we will offer you further tests. This is known as a high risk (screen positive) result.**

What is a low risk (screen negative) result?

If the screening test shows the risk of your baby having Down's syndrome is lower than the national cut-off figure, (>1:1000) this is known as having a low risk (screen negative) result and we will not offer you further tests. Most screening test results fall into this category. It is important to understand that a low risk (screen negative) result means exactly that. It does not mean that there is no risk at all that your baby has Down's syndrome, just that it is unlikely. There is still a small risk because some babies with Down's syndrome are not detected by screening tests. Overall, about a quarter of babies with Down's syndrome are not detected by screening tests.

It is very important to remember that screening tests fit into a two-stage process, so if you get a high risk (screen positive) result, it means we will offer you more information and tests. It does not mean that your baby definitely has Down's syndrome.

How long does it take to get the result of my screening test?

First Trimester Screening

The combined screening test result takes 2-3 working days.

Second Trimester Screening

The second trimester screen result takes between 3-5 days.

