

What does a Low-risk result mean?

One in three hundred is the dividing line between high and low risk. Low-risk means a chance of less than 1 in 300 of a foetus with Down syndrome. A low-risk result does not rule out the possibility of Down's, but the risk for this group is small. Only one woman in over ~3,000 screened as low-risk will have a Down syndrome foetus.

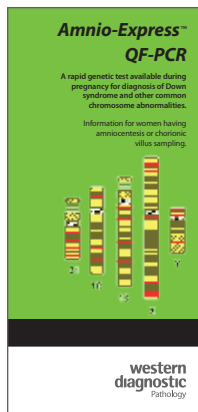
What does a High-risk result mean?

It means a chance for Down syndrome in the pregnancy of 1 in 300 or more. A high-risk result does not mean that the pregnancy is definitely abnormal. It only means that further tests should be offered to determine whether there is an abnormality or not. Approximately 1 in 20 women will have an increased risk result.

Further testing is offered to determine which women in the high-risk group really have an abnormality. This testing will usually involve amniocentesis or chorionic villus sampling, which take a small amount of fluid or tissue from around the baby for chromosomal analysis.

Both tests have a small risk of miscarriage. Final results from the chromosomal analysis may take 2-3 weeks. Sometimes an interim result may be available in a few days if specialised 'AmnioExpress QF-PCR'* testing is requested.

**see our pamphlet on Amnio Express - QF-PCR*



Conclusion

The First Trimester Screening test is, as its name implies, only a screening test. However it does detect significantly more Down pregnancies than the older triple test/maternal serum screen and it is able to achieve this at a much earlier stage of pregnancy. This test will classify patients into high and low risk groups. Patients at higher risk are offered further investigations to determine whether abnormalities are present or not. Those at low risk can be reassured earlier that these adverse outcomes are unlikely.

Why will I receive an account?

There will be an out-of-pocket charge (For details of the exact cost, see our Billing Guide for Out Patients, **My Pathology Test - What will it cost?**) for this test for all patients. You will receive an account. Medicare will reimburse a percentage of this and you will be responsible for the out-of-pocket expense. In line with our normal billing policy, current Health Care Cardholders, Pension Cardholders, Veterans with Gold Cards and residents of Nursing Homes will be exempt from this out-of-pocket charge.

The information in this pamphlet is designed to help you answer questions regarding screening for Down Syndrome. Should you have further questions talk to your doctor. Further information can be provided by your obstetrician, your ultrasonologist, the Western Australia Health Department, Genetic Services of Western Australia and King Edward Memorial Hospital.

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Understanding First Trimester Pregnancy Screening

Information for Patients



**western
diagnostic**
Pathology

74 McCoy Street, Myaree
Telephone 08 9317 0999

**western
diagnostic**
Pathology

The First Trimester Screening (FTS) test has important implications for you and your family. This brochure explains how First Trimester Screening is performed and what the results mean. Before undergoing this test it is very important you have discussed this testing procedure with your doctor.



What is First Trimester Screening?

First Trimester Screening is a test to detect women at risk for Down syndrome and some other problems during the early part of pregnancy. It involves two tests, a blood test and an ultrasound.

Down's screening tests do not provide a diagnosis, but instead group women as low risk or high risk. Women at high risk are offered further tests. It is important to remember that most women will have normal babies, even if they are grouped as high risk.

Down Syndrome

Down syndrome occurs in about 1 in every 700 pregnancies and is a chromosomal abnormality which is an important cause of congenital intellectual disability. Chromosomes contain the genetic information we require to grow and develop normally. Each cell contains 23 pairs of chromosomes. Down syndrome usually occurs by having 3 copies of chromosome 21. What causes this replication is still unknown, no racial, geographical, social, economic or environmental factors have been identified.

Chances of having a baby with Down syndrome increase as a woman gets older, but a child with Down Syndrome can be born to a mother of any age.

The First Trimester Screening Test

First Trimester Screening requires a blood test and an ultrasound scan which are performed early in pregnancy. A specialised ultrasound measurement is taken of the thickness (translucency) of the baby's neck and this reading is combined with the blood test results and the woman's age to give an assessment of the risk of Down Syndrome.

The First Trimester Screening Detection Rate

The FTS detects between 85%-90% of Down (Trisomy 21) pregnancies. As it is only a screening test and not a diagnostic test, a small proportion of fetuses with Down syndrome (approximately 10%) will not be detected as "increased risk".

When is the Test Performed?

Blood Test - We recommend women have the blood test at week 10 of pregnancy. However, the blood test can be performed up until 13 weeks and 6 days of pregnancy.

Ultrasound - We recommend the ultrasound is performed at week 12 of pregnancy. However, the ultrasound can be performed from 11 weeks through to 13 weeks and 6 days of pregnancy.

How do I arrange the testing?

1. *Make an appointment with your doctor and discuss the test fully. You need two referrals, one for the blood test (Western Diagnostic Pathology) and one for the ultrasound.*
2. *Have your blood test at week 10 of pregnancy*
3. *Book your ultrasound for week 12 of your pregnancy*
4. *The report of your risk assessment may be given to you at the time of the ultrasound or mailed to your doctor.*

What does the lab test?

The laboratory will measure PAPP-A (Pregnancy Associated Plasma Protein-A) and f β hCG (free beta hCG) hormones. We will send a copy of these results to your ultrasound clinic to include in your risk assessment and a copy to your doctor so that they know the blood test component of the test has been done.