## What are some examples of prenatal diagnostic tests?

Examples of diagnostic tests include chorionic villus sampling (CVS) and amniocentesis.

## How are these performed?

Both amniocentesis and CVS are done under guidance of an ultrasound. During **amniocentesis**, a small amount of amniotic fluid surrounding the baby is drawn through a small needle that is inserted through your abdomen. This is typically done after 16 weeks of pregnancy. **CVS**, on the other hand, involves collecting small sample of tissue from the placenta by inserting a needle through the abdomen. CVS is typically done between 10 to 14 weeks of pregnancy.

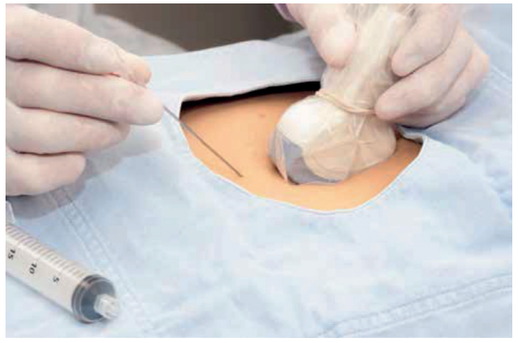

Figure 1 Picture showing amniocentesis procedure done under ultrasound guidance.

Various laboratory tests can be ordered on the sample of placental tissue or amniotic fluid, but the basic test is karyotype analysis that gives you a picture of the chromosomes. This covers the majority of chromosome problems including Down syndrome, Trisomy 18 and Trisomy 13.

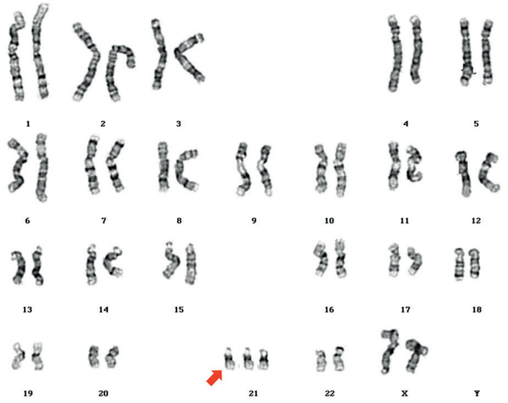

Figure 2 Karyotype analysis demonstrating an extra third copy of chromosome 21 (Down syndrome).

## Are there any risks associated with CVS and amniocentesis?

The risk of miscarriage associated with CVS and amniocentesis at KKH is 0.3% (1 in 300).

## Besides the major chromosome problems, can CVS and amniocentesis be done to test for other diseases as well?

Yes, the sample obtained from CVS and amniocentesis can be used to test for other conditions in the pregnancy. For example, very small deletions / duplications within the chromosomes, genetic diseases such as thalassaemia and fetal infection can also be tested for. Such extra testing will need to be specially ordered, so please let your doctor and the AMC counsellor know if this is something that concerns you.

For more information on screening options or how to proceed with arrangement of testing, please contact us at 6394-1288, Antenatal Monitoring Clinic (AMC), Women's Tower, Level 1.

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# Introduction to Down syndrome Testing

PATIENTS. AT THE HEART OF ALL WE DO.®

Congratulations on your pregnancy! Thank you for choosing KKH for your prenatal care. Some pregnant women may be concerned about the health of the baby. Fortunately, there are various of tests that can be done during pregnancy, some of which help test for birth defects, chromosomal or genetic conditions.

## Why should I consider prenatal testing?

The vast majority of babies are born normal. However, a small percentage of them may have a major birth defect, intellectual disability or health problem that could be associated with chromosomal/genetic abnormalities. Babies can be born with these conditions even if the parents are healthy. Hence, some women choose to pursue testing during pregnancy.

There are various prenatal tests available for your consideration depending on the gestational age of your pregnancy.

## I hear that testing for chromosome problems such as Down syndrome is important for pregnant women who are older than 35 years old. I am younger than 35 years old, are these tests still relevant to me?

You might be surprised to know that the majority of babies with Down syndrome are born to young women. This is because younger women have more babies than women who are older. Although the risk of having a baby with Down syndrome increases with a woman's age, all pregnancies are at risk for Down syndrome.

## Types of testing available

**Screening tests** can be done to determine if a pregnancy is at increased risk for a condition. However, it does not give a definitive answer to whether or not the pregnancy is affected. Nevertheless, understanding the risks may help you decide about further diagnostic testing.

**Diagnostic tests** can identify whether the pregnancy is affected by a certain condition. Examples of prenatal diagnostic tests include chorionic villus sampling (CVS) or amniocentesis. These procedures however are invasive in nature and are associated with a possibility of miscarriage.

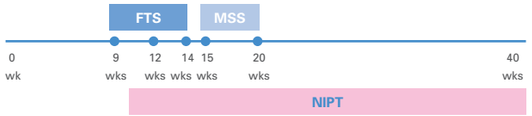
## What are some examples of prenatal screening tests for Down syndrome?

Examples of screening tests for Down syndrome include the First Trimester Screen (FTS), Non-invasive Prenatal Testing (NIPT) and the Maternal Serum Screen (MSS).

## Comparing FTS, NIPT and MSS

| | FTS | NIPT | MSS |
|---|---|---|---|
| **What condition the test screen for** | •Down syndrome<br>• Trisomy 18<br>• Trisomy 13<br>• Birth defects | • Down syndrome<br>• Trisomy 18<br>• Trisomy 13 | • Down syndrome<br>• Trisomy 18 |
| **How it is done** | Ultrasound and blood test | Blood test | Blood test |
| **When the test is done (gestational age)** | Between 9 weeks to 13 weeks + 6 days | After 10 weeks | Between 15 to 20 weeks |
| **Detection rate of DS** | 90% | >99% | 60% to 70% |

**Down syndrome screening timeline during pregnancy (weeks)**




**First Trimester Screen (FTS)** comprises an ultrasound scan and a blood test. The ultrasound scan is done between 11 weeks and 13 weeks + 6 days gestation. This involves taking measurements of the nuchal translucency (a fluid-filled space behind your baby's neck) and accessing the visibility of the nose bone of your baby. Fetus with an increased nuchal translucency measurement could also be at risk for other conditions. Furthermore, the ultrasound scan done at FTS will also screen for major structural defects such as heart defects that may already be obvious in the first trimester. Knowing these information early in your pregnancy is important for pregnancy management.

The ultrasound scan result will be combined with a blood test that measures biochemical markers (hCG and PAPP-A) to provide risk estimations for Down syndrome, Trisomy 18 and Trisomy 13. The risk is considered to be increased if it is higher than 1 in 300. This blood test is done between 9 weeks and 13 weeks + 6 days gestation.

**Non-invasive Prenatal Testing (NIPT)** is a blood test that analyses DNA from the placenta that exists in the mother's blood. The DNA from the placenta is usually the same as that of the baby's. Like FTS, NIPT provides risk estimations for Down syndrome, Trisomy 18 and Trisomy 13. However, some patients are not able to obtain a result despite testing due to various reasons. Your doctor will be able to advise alternative testing in this situation.

**Maternal Serum Screen (MSS)** involves a blood test that measures three biochemical markers (hCG, uE3, AFP) to provide risk estimations for Down syndrome and Trisomy 18. The risk is considered to be increased if it is higher than 1 in 250.

## The detection rate for Down syndrome appears to be higher in NIPT than FTS. Will I be missing out on anything if I choose to have NIPT done and not FTS?

NIPT is a blood test, and does not screen for structural defects such as heart defects in the baby. In fact, heart defects are seven times more common than Down syndrome. For this reason, KKH recommends the FTS for its ability to detect structural defects during the early stages in pregnancy, as well as for the relatively high detection rate for Down syndrome. FTS still remains as the gold standard for Down syndrome testing at KKH.

## Can I do both FTS and NIPT simultaneously?

Yes, you may. In fact, some patients who have had NIPT done may not receive a result after testing due to certain reasons. Hence, we recommend FTS to be done regardless of your decision on NIPT.