KK Women's and Children's Hospital
SingHealth

Down Syndrome Testing During Pregnancy

PATIENTS. AT THE HEART OF ALL WE DO.®

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Introduction

Congratulations on your pregnancy! Thank you for choosing KKH for your prenatal care. The Antenatal Diagnostic Clinic (ADC) and Antenatal Monitoring Clinic (AMC) can help provide you with comprehensive prenatal genetic counselling and prenatal testing.

The vast majority of babies are born normal. However, a small percentage of them (2%) have a major physical defect or intellectual problem. This may be due to chromosomal abnormalities, genetic diseases or structural birth defects. Babies can be born with these conditions even if the parents are healthy, which is why it is important to screen for such conditions in all pregnancies.

There are various tests available for your consideration, depending on how far along the pregnancy you are. These tests may be screening or diagnostic in nature.

A screening test shows if a pregnancy is at increased risk for a condition, and does not give a definitive answer to whether or not the pregnancy is affected. However, understanding the risks can help you decide if you want to pursue further diagnostic testing.

A diagnostic test can identify if the pregnancy is or is not affected by a certain condition. A diagnostic test however involves invasive testing either through chorionic villus sampling (CVS) or amniocentesis. These procedures are associated with a possibility of miscarriage, and hence are not routinely offered to all pregnant women.

It is important to note that these tests are unable to assess all birth defects / genetic conditions that may be present in a pregnancy. Based on your specific family and pregnancy history, your healthcare provider may recommend additional testing options. Please let your healthcare provider know about your concerns.

Screening Tests

First Trimester Screen (FTS)

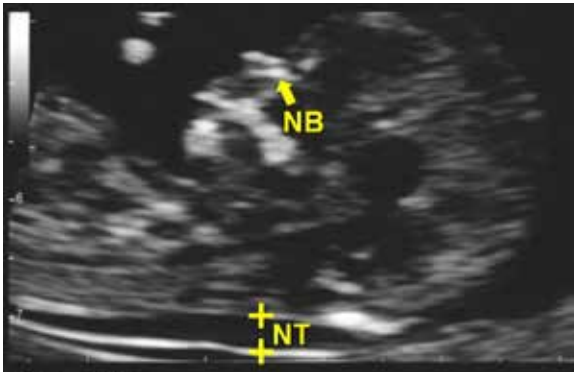
What is it?

The FTS comprises an ultrasound and a blood test.

The FTS ultrasound involves taking measurements of the nuchal translucency and assessing the visibility of the nasal bone (image shown below). If the nuchal translucency measurement is found to be increased or if the nasal bone is not visible, this may increase your risks for Down syndrome, Trisomy 18 and Trisomy 13. The nuchal translucency measurement is particularly important because if it is found to be substantially increased, the baby could also be at risk for other conditions. For this reason, testing beyond Down syndrome, Trisomy 18 and Trisomy 13 may be indicated.

Furthermore, the ultrasound done at FTS will also screen for major structural defects such as heart defects that may already be obvious in the first trimester. Knowing this information early in your pregnancy is important for pregnancy management.

The ultrasound results will then 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.



FTS ultrasound

When is it done?

The first trimester ultrasound is ideally done between 11 weeks to 13 weeks 6 days, and the blood needs to be drawn between 9 weeks to 13 weeks 6 days.

What do results look like?

Risk estimations for Down syndrome, Trisomy 18 and Trisomy 13 will be calculated, and you may find yourself at increased or decreased risk for the diseases tested for. The risk is considered to be increased if it is higher than 1 in 300.

Screening Tests

Noninvasive Prenatal Testing (NIPT)

What is it?

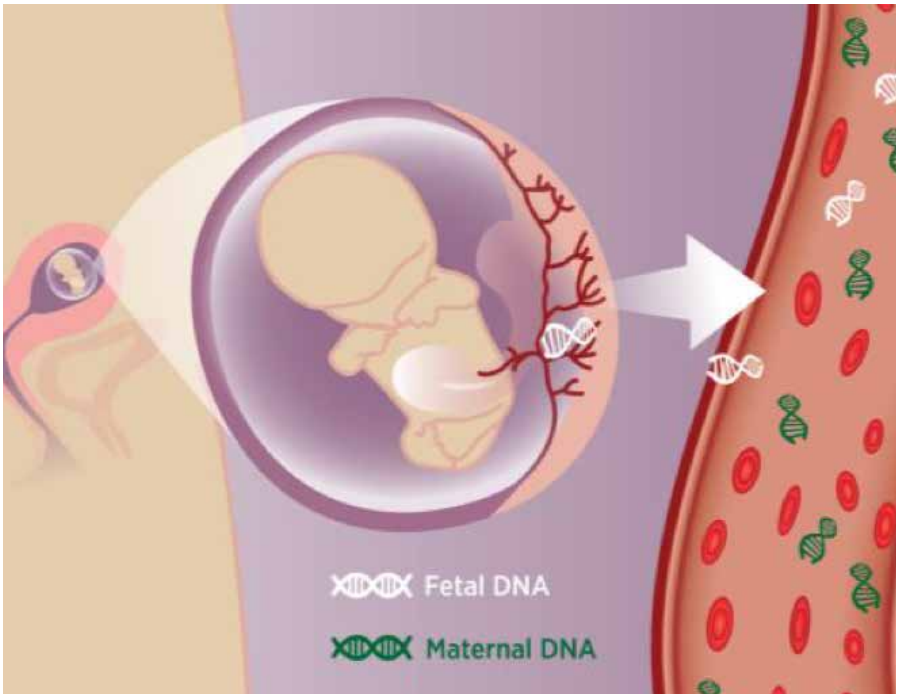
NIPT is a blood test that analyses DNA from the placenta that exists in the maternal blood. The DNA from the placenta is usually the same as that of the baby's. Like the FTS, NIPT provides risk estimations for Down syndrome, Trisomy 18 and Trisomy 13.

When is it done?

After 10 weeks of pregnancy to term.

What do results look like?

Risk estimations for Down syndrome, Trisomy 18 and Trisomy 13 will be calculated, and you may find yourself at increased or decreased risk for the diseases tested for. Some patients however, are not able to obtain a result on NIPT despite testing.



Screening Tests

Maternal Serum Screen (MSS)

What is it?

A blood test that measures three biochemical markers (hCG, uE3, AFP) to provide risk estimations for Down syndrome and Trisomy 18.

When is it done?

Between 15 weeks to 20 weeks of pregnancy.

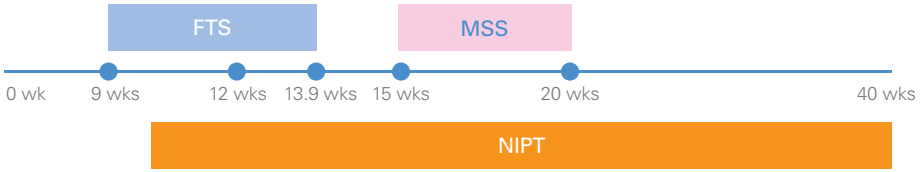
What do results look like?

Risk estimations for Down syndrome and Trisomy 18 will be calculated, and you may find yourself at increased or decreased risk for the diseases tested for. The risk is considered to be increased if it is higher than 1 in 250.

Comparing FTS, NIPT and MSS

	Diseases	How It Is Done	Time	Detection Rate Of DS
FTS	• Birth defects and Down syndrome • Trisomy 18 • Trisomy 13	Ultrasound and Blood test	11 weeks to 13 weeks 6 days and 9 weeks to 13 weeks 6 days	90%
NIPT	• Down syndrome • Trisomy 18 • Trisomy 13	Blood test	After 10 weeks	>99%
MSS	• Down syndrome • Trisomy 18	Blood test	Between 15 to 20 weeks	60 to 70%

Down syndrome screening timeline during pregnancy (weeks)



Frequently Asked Questions (FAQs)

Will NIPT tell me if the baby is affected by Down Syndrome?

NIPT, like the FTS, is only a screening test and gives you risk estimations for an affected pregnancy. False positives and false negatives can still occur in NIPT, although the chance of which is lower than that of FTS.

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.

Diagnostic Tests

Amniocentesis

What is it?

Under the guidance of an ultrasound, a small amount of the amniotic fluid surrounding the baby is drawn through a small needle that is inserted through your abdomen.

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

When is it done?

After 16 weeks of pregnancy.

What do results look like?

This depends on the laboratory test that is ordered. Please speak to your doctor or the AMC counsellor for further information.

Diagnostic Tests

Chorionic villus sampling (CVS)

What is it?

Under the guidance of an ultrasound, a small sample of tissue from the placenta is taken by inserting a needle through the abdomen.

Various laboratory tests can be ordered on the villus sample, but the basic test is the karyotype analysis that gives you a picture of the placental chromosomes (which is usually the same as that of the baby's). This covers the majority of chromosome problems, including Down syndrome, Trisomy 18 and Trisomy 13.

When is it done?

CVS is typically done between 10 to 14 weeks of pregnancy. The procedure can also be done after 14 weeks of pregnancy, but this is determined on a case-by-case basis and is dependent on the laboratory test that is requested.

What do results look like?

This depends on the laboratory test that is ordered. Please speak to your doctor or the AMC counsellor for further information.

Frequently Asked Questions (FAQs)

Are there any risks associated with CVS and amniocentesis?

The risk of miscarriage associated with CVS and amniocentesis at KKH is 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.

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Patient's label

Patient Acknowledgement

I acknowledge that this Down syndrome screening booklet has been given to me on DD / MM / YYYY at _____ (location) by _____ (staff's name).

I have been informed about the various Down Syndrome test options available to me and will review the details in the booklet for further information.

Patient's signature: _____

Date: _____

TEAR ALONG THIS LINE.

Reg No 198904227G OAMCdstp0317

Useful telephone number

Appointments / Specialist Outpatient Clinics Enquiries Hotline	6294-4050

Note: For more information on the test options or how to proceed with the tests, please call 6394-1288. The KK Antenatal Monitoring Clinic (AMC) is located at Women's Specialist Clinics, Women's Tower, Level 1.

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