



NUH Women's Centre
Is My Answer

Diagnostic Tests For Chromosomal Abnormalities

A member of the NUHS

How long does it take to obtain the result?

A rapid result can be obtained for Down syndrome within 2 days. The detailed result of the chromosome test takes about 10-12 days. For thalassaemias, the result is available within 3 days.

Will a repeat of the procedure be required?

In approximately 1% of the cases, a second invasive test (an amniocentesis) will need to be performed because the results are inconclusive.

What happens if the tests indicate abnormality in the fetus?

You will be notified by the doctor about the results. The abnormality and options will be discussed to allow you to make your own decision about the pregnancy.

Fetal Blood Sampling (FBS)

Fetal blood sampling (FBS) is a procedure to remove a small amount of blood from the fetus during pregnancy.

What types of disorder can be detected by FBS?

1. Chromosomal disorders – FBS is done to rapidly verify the presence of Down syndrome or other fetal chromosomal abnormality, especially when a physical abnormality is found in the fetus via an ultrasound scan.
2. Certain inherited disorders (e.g. thalassaemia, haemophilia, cystic fibrosis, etc)
3. Certain infections which can affect the fetus.
4. Anaemia or low platelet levels in the fetus (through performing a blood count).

When is it done?

It is usually carried out between 18 and 23 weeks of pregnancy.

How is it performed?

An ultrasound is performed before FBS to confirm the gestational age of the fetus. The doctor guides a needle from the patient's abdomen into an umbilical or fetal liver blood vessel under ultrasound guidance. Once an adequate amount of specimen is obtained, the needle is withdrawn.

How much discomfort is involved during FBS?

Most women feel mild pain when FBS is performed but the pain goes away soon after the procedure.

Is local anaesthesia used?

Yes, local anaesthesia is used to make the area numb to reduce the discomfort.

Is it necessary to rest after the procedure?

Patients are advised to rest at home for 2 days after the procedure. Complete bed rest is not required.

What are the risks involved?

The risk of miscarriage is around 1 - 1.5%. Please contact your doctor if you experience persistent abdominal pain, vaginal bleeding or leaking.

How long does it take to obtain result?

Around five days.

What happens if the tests indicate abnormality in the fetus?

You will be notified by the doctor about the results. The abnormality and options will be discussed to allow you to make your own decision about the pregnancy.

Contact Us

Fetal Care Centre (FCC)
Kent Ridge Wing, Level 3

Fetal Care Centre Annex
Main Building, Level 1

Tel/Appointment : 6772 5180
Fax : 6772 4547
Email : FCC@nuhs.edu.sg
Website : www.nuhgynaecology.com.sg

Opening Hours : 8.30 am - 6.00 pm (Mondays – Thursdays)
8.30 am - 5.30 pm (Fridays)

Location



National University Hospital
5 Lower Kent Ridge Road, Singapore 119074
Tel: (65)6779 5555 Fax: (65)6779 5678
Website : www.nuh.com.sg
Co. Reg. No. 198500843R

The information provided in this publication is meant purely for educational purposes and may not be used as a substitute for medical diagnosis or treatment. You should seek the advice of your doctor or a qualified healthcare provider before starting any treatment or if you have any questions related to your health, physical fitness or medical conditions.

Information is correct at time of printing (Jan 2014) and subject to revision without prior notice.

Copyright (2014). National University Hospital
All Rights reserved. No part of this publication may be reproduced without permission in writing from the National University Hospital.

Diagnostic Tests For Chromosomal Abnormalities

During your pregnancy, a number of procedures (diagnostic tests) can be done to confirm the presence or absence of chromosomal abnormalities in the fetus. The type of procedure depends on the stage of your pregnancy.

Amniocentesis

This is a procedure by which the fluid in the sac surrounding the baby is withdrawn. Known as the amniotic fluid, it contains cells shed by the developing baby. A variety of tests can be carried out on an amniotic fluid sample, including chromosomal analysis.

What types of disorder can be detected by amniocentesis?



1. Nearly all chromosomal abnormalities, which may be caused by the lack or addition of an extra chromosome, such as Down syndrome.
2. Certain inherited disorders such as thalassaemia, haemophilia, cystic fibrosis.
3. Certain fetal infections.

When is it done?



It is usually carried out between 15 and 20 weeks of pregnancy.

Who should consider amniocentesis?

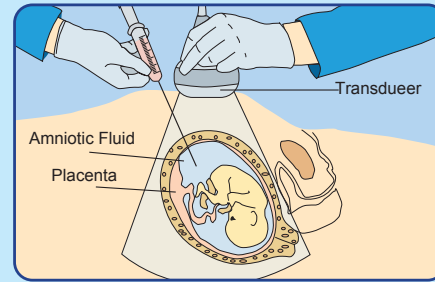


1. Women who are 35 years and older, and wish to know for certain whether the baby is affected by chromosomal disorders.
2. Women who had a chromosomal abnormal child previously.
3. Women who are aware that they and their partner are carriers of certain disorders that can be passed down to the next generation.
4. Women with positive results from a prior screening test e.g. Nuchal Translucency screen or Triple Test (refer to our brochure on "Screening for Chromosomal Abnormalities".)

How is it performed?



The doctor performs an ultrasound scan to determine the position of the placenta and the safest location to perform the test. The doctor cleans your abdomen with antiseptic solution and inserts a very thin needle into the uterus through the abdomen under ultrasound guidance. About 20ml of the fluid is drawn.



How much discomfort is involved during amniocentesis?



Most women feel a slight pressure or pain when amniocentesis is performed. It is described as being similar to taking a blood test. Very rarely will patients experience severe pain.

Is local anaesthesia used?



No, the needle used for anaesthesia usually causes more discomfort than the needle used for amniocentesis.

Will the amniotic fluid continue to seep out after the needle is removed?



Most of the time it does not, as a very fine needle is used and the tissues around the tract of the needle will seal the path after removal of the needle and prevent seepage. Consult your doctor if it does.

Is it necessary to rest after the procedure?



The patient is advised to rest at home for 2 days after the procedure. Complete bed rest is not required.

Will amniocentesis hurt the baby?



Due to continual use of the ultrasound during the procedure, injury to the baby is rare. Care is taken to avoid placing the needle near the baby.

What are the risks involved?



The risk of a miscarriage following an amniocentesis is about 0.5% (1 in 200 procedures). Please consult your doctor if you experience persistent abdominal pain, vaginal bleeding or leaking after the procedure.

How long does it take to obtain the result?



If chromosomal culture (karyotype) is being done, the result is available in about 10-12 days. Rapid tests called FISH (fluorescence in situ hybridisation) or PCR (polymerase chain reaction) are available for chromosomes 13, 18, 21, X and Y. These additional tests can be done if the patient wishes to. The results are available by the next working day.

What happens if the tests indicate abnormality in the fetus?



You will be notified by the doctor about the results. In the case of an abnormal result, the nature of the abnormality and options will be discussed to allow you to make your own decision about the pregnancy.

Chorionic Villus Sampling (CVS)

Chorionic villus sampling (CVS) is a prenatal test that involves taking a tiny placental tissue sample from outside the sac where the fetus develops. The tissue is tested to diagnose or rule out certain birth defects. Both the baby and placenta (afterbirth) originate from the same cell, so the chromosomes present in the cells of the placenta are the same as those of the baby.

What types of disorder can be detected by chorionic villus sampling?



1. Chromosomal abnormalities such as Down syndrome.
2. Certain inherited disorders such as thalassaemia, haemophilia, cystic fibrosis.

When is it done?

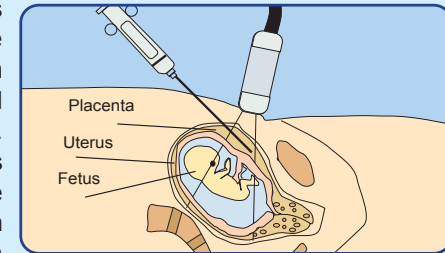


It is usually carried out between 11 and 13 weeks of pregnancy.

How is it performed?



An ultrasound is performed before CVS to confirm the gestational age of the fetus. The doctor guides a needle from the patient's abdomen into the placenta under ultrasound guidance. Once an adequate amount of specimen is obtained, the needle is withdrawn.



How much discomfort is involved during CVS?



Most women feel mild pain when CVS is performed. The pain goes away soon after the procedure.

Is local anaesthesia used?



Yes, local anaesthesia is used to make the area numb to reduce the discomfort.

Is it necessary to rest after the procedure? What should I expect following a CVS?



Patients are advised to rest at home for 2 days after the procedure. Complete bed rest is not required.

For the first couple of days, you may experience some abdominal discomfort, period-like pain or a little bleeding, which is relatively common. In majority of the cases, the pregnancy continues without any problems. You may find it helpful to take painkillers like paracetamol.

What are the risks involved?



The risk of miscarriage following CVS is around 1%. Please contact your doctor if you experience persistent abdominal pain, vaginal bleeding or leaking.