MEDIHEAL GROUP OF HOSPITALS

Transforming Health Care In Africa

Prenatal Screening *for*

Genetic Conditions

Concern about your baby's health?
Prenatal genetic screening opens:

Monday to Saturday; 9:00am to 6.00pm

PRENATAL GENETIC TESTING

Globally, 2-3% of newborns have a congenital malformation or genetic defects identified at birth. Chromosomal abnormalities are present in about 0.9% of newborns and include abnormalities of chromosome number and structure. They can involve one or more autosomes (1-22), sex chromosomes, or both simultaneously. The most common aneuploidy detected at birth is Down's syndrome (trisomy 21), which occurs in approximately 1/700 live births. The chance of having a baby with down's syndrome increases with the maternal age. However, about 50% of affected babies are born to mothers who are under 35, because more young women are having babies. Therefore, there should be universal screening irrespective of age, race, nationality, religion or socioeconomic status

What is prenatal genetic testing?

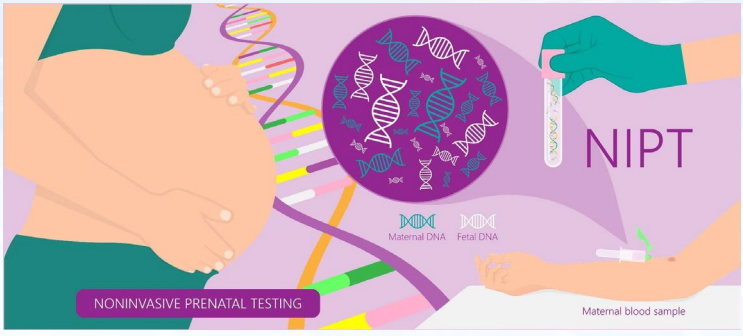
There are two types of tests done during pregnancy to determine if your baby has Down syndrome or other chromosomal defects.

- **Screening tests**

  A screening test can tell you the chance or risk score that the baby could have a chromosomal abnormality. It can sometimes give an abnormal result even when there is nothing wrong with the baby. It does not provide specific diagnosis. Prenatal screening usually involves maternal blood testing (dual marker test/ quadruple test/ NIPT) and / or ultrasound examination (Nuchal translucency/ first trimester scan or second trimester anatomy scan).

- **Diagnostic tests**

  A diagnostic test can tell you for sure that your baby has genetic defect or not. It is usually offered when your screening test shows abnormal results (positive or high risk on serum marker screening or abnormal ultrasound findings or previous affected pregnancy). It includes testing of placental tissue or fetal cells obtained by various methods like chorionic villous sampling or amniocentesis respectively.

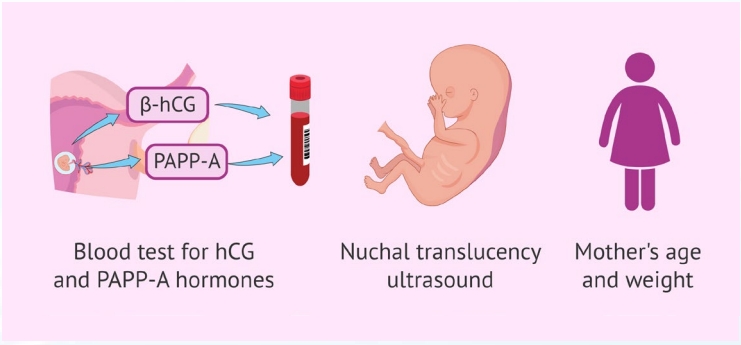


When can prenatal screening be done or offered?

- First trimester screening (11-13+6 weeks)
- Second trimester screening (15-20 weeks)
- Non-invasive prenatal screening/ testing (NIPS/NIPT) (10 weeks onwards)

What is first trimester screening?

It comprises all tests which are done between 9 and 13+6 weeks to look for certain birth defects related to the fetal cardiac or genetic defects, such as Down syndrome (trisomy 21) and Edward's syndrome (trisomy 18) primarily as well as trisomy 13 in some laboratories. It includes maternal blood test (serum beta- HCG and PAPP-A) and an ultrasound for nuchal translucency measurement. These tests are usually combined with maternal age to provide a patient specific risk. Women with positive screening test are counselled regarding options for further testing by CVS. Benefits of identifying aneuploidy in first trimester is that it affords patients information earlier in pregnancy so that decisions regarding termination may be made representing less maternal risk.



What are the different options available for first trimester screening?

- **Double marker/ dual marker test**

  It is one of the methods of maternal serum marker screening for aneuploidy done between 10-13+6 weeks. It measures the levels of two proteins, human chorionic gonadotropin and pregnancy associated plasma protein A. These markers are abnormally high or low in cases of genetic abnormalities. The test gives you a risk score (e.g. 1 in 100) for having a baby with Down's syndrome or other chromosome conditions.

- **First trimester nuchal translucency scan**

  It is a special ultrasound done during first trimester for nuchal translucency measurement (amount of fluid at the back of baby's

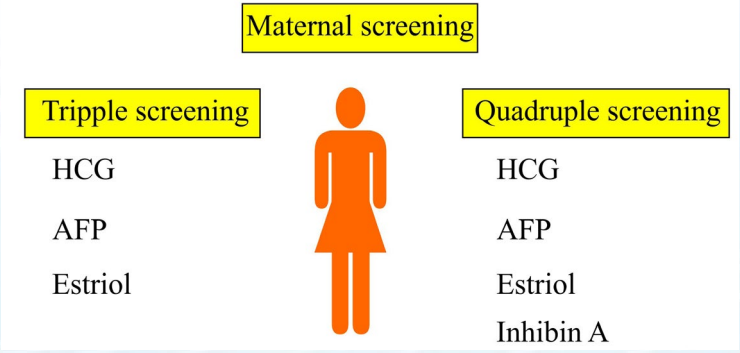
neck during early pregnancy i.e. 11-13+6 wks). It is often increased in fetuses with Down's syndrome (75%), trisomy 13, trisomy 18, turner syndrome, triploidy and structural birth defects, particularly Congenital heart defects. The risk of aneuploidy increases as nuchal translucency thickness increases. Other markers can be included to improve detection rate and to rule out major anomaly. E.g. absent nasal bone in 60-70% fetuses with Down's syndrome)

- Combined first trimester screen (Dual marker test + NT scan)
- NIPS (cell free DNA screening)

This test is based on identification of fetal cell free DNA in maternal blood circulation which is derived from trophoblast. It is considered as screening test for Down's syndrome and reserved for high-risk women (i.e. advanced maternal age >35yrs at the time of delivery, abnormal ultrasound findings, abnormal serum marker screening result, previous baby or a close family member with down's syndrome). It can be done any time in pregnancy after 10 weeks up to delivery. All positive results need to be confirmed by diagnostic tests. It involves screening for trisomy 21,18,13 with detection rates of >99%, 96%, 92% respectively.

What is second trimester screening?

Second trimester screening is done between 15-20 weeks to rule out certain birth defects which includes a maternal serum screen (quad screen) and a comprehensive ultrasound evaluation of the baby for presence of structural anomalies.



What are the different options available for second trimester screening?

- **Quadruple marker/quad test**

It is one of the methods of maternal serum marker screening done between 15-20 weeks. This test involves measurement of 4 proteins (alpha feto protein, human chorionic gonadotropin, estriol and inhibin A). The test gives you a risk score (e.g. 1 in 100)

for having a baby with Down's syndrome or other chromosome conditions and neural tube defects.

- **Second trimester anomaly scan**

  A detailed anatomic survey in the second trimester (18-22weeks) is an important tool for chromosomal defects screening. Approximately one third of fetuses with Down's syndrome have an identifiable sonographic major or minor structural abnormality.

- **Fetal Echocardiogram**

  A fetal echocardiogram is a test done by Fetal Medicine expert/ Paediatric cardiologist that uses ultrasound to evaluate the baby's heart for congenital heart defects before birth.

Method of screening	Detection rate (%)
Maternal age (MA)	**30**
MA + Quad test at 15-18 weeks	**50-70**
MA + Fetal NT at 11-13+6weeks	**70-80**
MA + Fetal NT + Dual marker test (β-HCG + PAPP-A) at 11-13+6weeks	**85-90**
MA + Fetal NT + Fetal Nasal Bone at 11 - 13 + 6weeks	**90**
MA + Fetal NT + Fetal Nasal Bone + Dual marker test at 11 - 13 + 6weeks	**95**

Combined first and second trimester risk assessment

- Integrated testing has the highest Down's syndrome detection rate (90%) with the lowest false positive rate (2%) compared to other screening methods. This test includes combined first trimester screen (Dual marker test + NT scan) and second trimester screen to provide a single risk calculation.

- Stepwise sequential screening uses same combination of tests but preliminary results are disclosed to patients in first trimester. If patient shows high risk for aneuploidy, diagnostic testing is offered.

Prenatal diagnostic testing / invasive prenatal diagnosis

Chorionic villous sampling and amniocentesis are both routinely

used for prenatal diagnostic testing and can provide fetal cells/ tissue for tests like FISH, Karyotyping, chromosomal microarray and DNA based tests. Pregnancy loss rate due to invasive procedures is estimated to be between 1/500-1/1000.

- **Chorionic villous sampling (CVS)**
  It is an invasive diagnostic test done by fetal medicine expert between 11-14 weeks where he/she collects a tiny piece of the placenta, called chorionic villous, sent for genetic testing to rule out certain chromosomal abnormalities. It is offered to all women who show positive / abnormal screening result or at high risk for certain birth defects.

- **Amniocentesis**
  It is an invasive diagnostic test done by fetal medicine expert between 15-20 weeks where he/she collects a small amount of amniotic fluid around the fetus, sent for genetic testing to rule out certain chromosomal abnormalities. It is offered to all women who show positive / abnormal screening result or at high risk for certain birth defects.

WHAT WE SCREEN FOR?

Trisomy 21 (Down Syndrome)

What is Down syndrome in children?

Down syndrome is a genetic disorder. It is also called trisomy 21. This includes certain birth defects, learning problems, and facial features. A child with Down syndrome also may have heart defects and problems with vision and hearing and how severe or mild these problems are varies from child to child. It affects about 1 in 800 babies.

What causes Down syndrome in a child?

When a baby is conceived, a normal egg cell and normal sperm cell start with 46 chromosomes. The egg and sperm cells then divide in half. The egg and sperm cells then have 23 chromosomes each. When a sperm with 23 chromosomes fertilizes an egg with 23 chromosomes, the baby will then have a complete set of 46 chromosomes. Half are from the father and half are from the mother.

But sometimes an error occurs when the 46 chromosomes are being divided in half. An egg or sperm cell may keep both copies of chromosome number 21, instead of just 1 copy. If this egg or sperm is

fertilized, then the baby will have 3 copies of chromosome number 21. This is called trisomy 21.

Sometimes the extra number 21 chromosome or part of it is attached to another chromosome in the egg or sperm. This may cause translocation Down syndrome. This is the only form of Down syndrome that may be inherited from a parent.

A rare form is called mosaic trisomy 21. This is when an error in cell division happens after the egg is fertilized. People with this syndrome have both normal cells and some cells with an extra chromosome number 21.

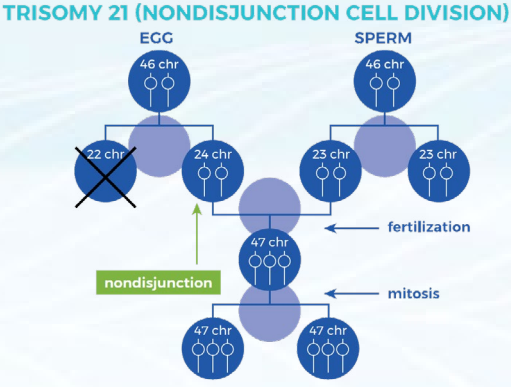
Symptoms

- Eyes that slant upward
- Small ears that may fold over slightly at the top
- Small mouth that makes the tongue appear large
- Small nose with a flattened bridge
- Short neck
- Small hands with short fingers
- 2 instead of 3 palm creases, including one across the palm and one around the base of the thumb
- Short height
- Loose joints

How is Down syndrome treated in a child?

There is no cure for Down syndrome. But a child with Down syndrome may need treatment for problems such as:

- Heart defects. .
- Intestinal problems.
- Vision problems.
- Hearing loss.
- Other health problems.
- Learning problems.



Trisomy 18 (Edward Syndrome)

Edwards' syndrome, also known as trisomy 18, is a rare but serious condition. Sadly, most babies will die before or shortly after being born. A small number (about 13 in 100) babies born alive with Edwards' syndrome will live past their 1st birthday.

Symptoms of Edwards' syndrome

Your healthcare provider will look for signs of Edwards syndrome (trisomy 18) during a prenatal ultrasound, including:

- Very little fetal activity.
- A single artery in your umbilical cord.
- A small placenta.
- Birth defects.
- Your fetus is surrounded by too much amniotic fluid

What causes Edwards syndrome (trisomy 18)?

Having three copies of chromosome 18 instead of the typical two causes Edwards syndrome (trisomy 18).

When cells form, they start in reproductive organs as one fertilized cell (sperm in males and eggs in females). Cells divide (meiosis) to create pairs by copying themselves. The cell's copy contains half the amount of DNA as the original cell, 23 chromosomes from 46, and each chromosome pair has a number.

Characteristics of Edwards syndrome (trisomy 18) after birth

After your baby is born, your child likely has physical characteristics of Edwards syndrome (trisomy 18), including:

- Decreased muscle tone (hypotonia).
- Low-set ears.
- Internal organs forming or functioning differently (heart and lungs).
- Issues with cognitive development (intellectual disabilities), which are typically severe.
- Overlapping fingers and/or clubfeet.
- Small physical size (head, mouth and jaw).
- Weak cry and minimal response to sound.

What tests diagnose Edwards syndrome (trisomy 18)?

During pregnancy, your healthcare provider will offer different tests to determine the diagnosis of your child if they show symptoms of Edwards syndrome (trisomy 18). These tests include:

- **Amniocentesis**: Between 15 and 20 weeks of pregnancy, your healthcare provider will take a small sample of amniotic fluid to identify potential health conditions in your baby.
- **Chorionic villus sampling (CVS):** Between 10 and 13 weeks of

pregnancy, your healthcare provider will take a small sample of cells from your placenta to look for genetic conditions.

- **Screenings:** After 10 weeks of pregnancy, your healthcare provider will examine a sample of your blood to assess whether or not your child has common extra chromosome conditions, including trisomy 18.

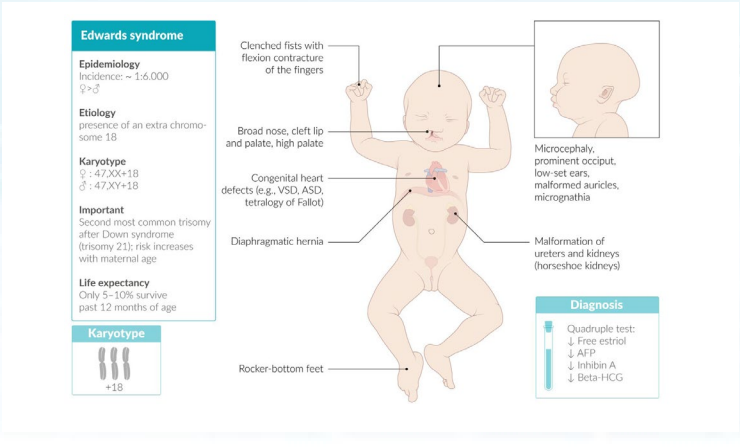
After your child is born, child's heart will be examined via ultrasound to identify and treat any heart-related conditions as a result of their diagnosis.

How is Edwards syndrome (trisomy 18) treated?

Often, the condition is so severe that babies who survive being born are treated with comfort care. But treatment for Edwards syndrome (trisomy 18) is unique for each child, based on the severity of their diagnosis. There's no cure for Edwards syndrome (trisomy 18).

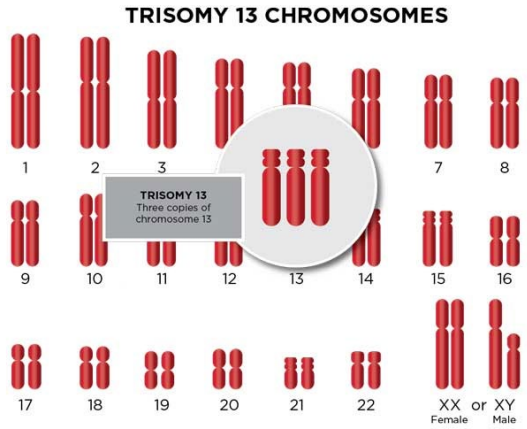
Treatment for Edwards syndrome (trisomy 18) might include:

- **Cardiac treatment:** Heart problems affect nearly all cases of Edwards syndrome (trisomy 18). Not all babies with cardiac problems due to Edwards syndrome (trisomy 18) are eligible for surgery, but some might be.
- **Assisted feeding:** Children diagnosed with Edwards syndrome (trisomy 18) can have problems eating normally due to delayed physical growth. A feeding tube might be necessary to address early feeding problems after your child is born.
- **Orthopaedic treatment:** Children diagnosed with Edwards syndrome (trisomy 18) may have spinal problems like scoliosis, which could impact how your child moves. Orthopaedic treatment could involve bracing or surgery.
- **Psychosocial support:** Support is available to you, your family and your child diagnosed with Edwards syndrome (trisomy 18), especially to help cope with the loss of your child or help you navigate your child's complex diagnosis.



Trisomy 13 (Patau Syndrome)

Patau's syndrome is a serious rare genetic disorder caused by having an additional copy of chromosome 13 in some or all of the body's cells. It's also called trisomy 13. Each cell normally contains 23 pairs of chromosomes, which carry the genes you inherit from your parents.



Causes trisomy 13 (patau syndrome)

They occur randomly during conception, when the sperm and egg combine and the foetus starts to develop. An error occurs when the cells divide, resulting in an additional copy, or part of a copy, of chromosome 13, which severely affects the baby's development in the womb.

Symptoms trisomy 13 (patau syndrome)

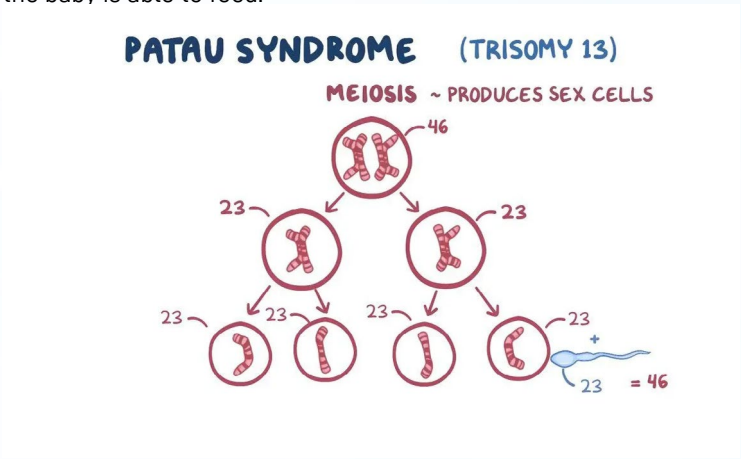
- Cleft lip and palate.
- An abnormally small eye or eyes (microphthalmia)
- Absence of 1 or both eyes (anophthalmia)
- Reduced distance between the eyes (hypotelorism)
- Problems with the development of the nasal passages.

Types of Trisomy 13

- **Full Trisomy 13:** The existence of a third copy of chromosome 13 in all of the cells. About 95% of cases of Trisomy 13 are this type.
- **Mosaic Trisomy 13:** The existence of a third copy of chromosome 13 in some of the cells. About 5% of cases of Trisomy 13 are this type.
- **Partial Trisomy 13**: The existence of a part of a third copy of chromosome 13 in the cells. Less than 1% of cases of Trisomy 13 are this type.

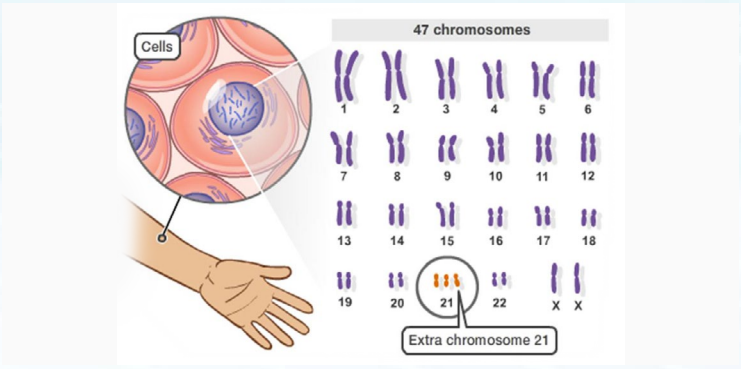
Treatment

There's no specific treatment for Patau's syndrome. As a result of the severe health problems a newborn baby with the syndrome will have, doctors usually focus on minimising discomfort and ensuring the baby is able to feed.



BOTTOMLINE

Every woman has a risk that her fetus / baby has a chromosomal defect. All these tests are completely optional. At our center, we offer maternal-fetal medicine care which includes genetic counselling, prenatal screening, high resolution ultrasound services including 3D scanning, nuchal scan, detailed anomaly scan, fetal doppler, fetal echocardiography and fetal neurosonogram. We also conduct invasive fetal procedures like amniocentesis, cordocentesis, CVS for prenatal genetic diagnosis. If you have any query about any of these screening tests, you can consult our maternal-fetal medicine expert or open the link given below to get more details on our website to understand about the risks, benefits and limitations of these tests.



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OUR JOURNEY

"Healthy Africa
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