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OBSTETRICS AND GYNAECOLOGY
ULTRASOUND
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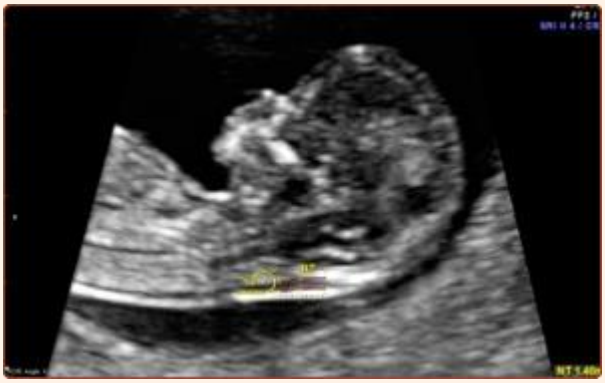
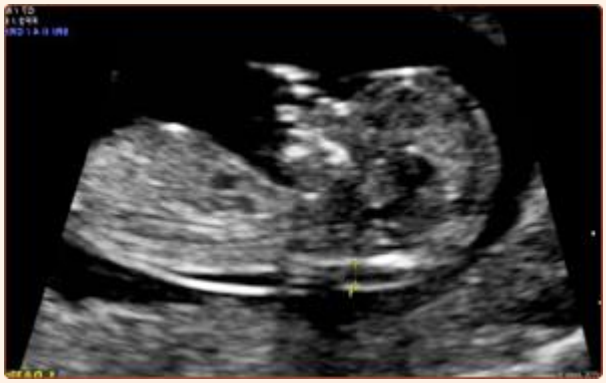
First Trimester Screening (Nuchal Translucency scan)

At the end of the first trimester, normally there is a small amount of fluid seen beneath the skin of fetal neck and head. This fluid space is called as nuchal translucency. In an ultrasound image this is identified as the echo free space at the back of the fetal neck and can be accurately measured to within a tenth of a millimeter. All fetuses have a measurable nuchal translucency.

When there is extra fluid, ie when the nuchal translucency measurement is thicker than normal, there is an association with chromosomal anomalies such as Down Syndrome and some structural abnormalities in the fetus.





What is involved in first trimester screening test?

There are two separate components to first trimester screening.

- A blood test (ideally done at 10 weeks)
- Nuchal translucency scan (ideally done at 12-13 weeks)

The results of the blood test, scan findings and maternal details are fed into a computer package (developed by Fetal Medicine Foundation, London) which then calculates the individual risk for chromosome abnormalities. This will either be ‘low risk’ (risk is less than 1in 300) or a ‘high risk’ (risk is greater than 1in 300).

What are chromosomal disorders?

Chromosomes are structures that hold our genes. The body is made up of individual units called cells. In the center of most cells is a structure called the nucleus and this is where chromosomes can be found. The typical number of chromosomes in humans is 46 (two pairs of 23) holding an estimated 25,000 genes. One set of 23 chromosomes is inherited from the egg and the other set is inherited from the sperm. Each individual’s chromosomal makeup is determined at the time of conception and cannot be altered

A chromosomal disorder reflects an abnormality of chromosomal number or structure. There are many types of chromosome abnormalities. It means that occasionally a fetus may receive too many or too few chromosomes or there is rearrangement of portions of the chromosomes.

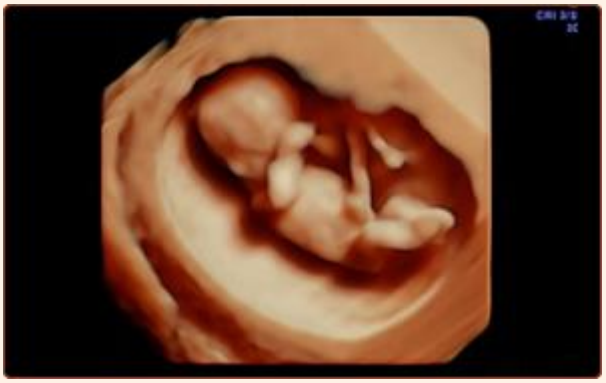
The most common chromosome abnormalities are:

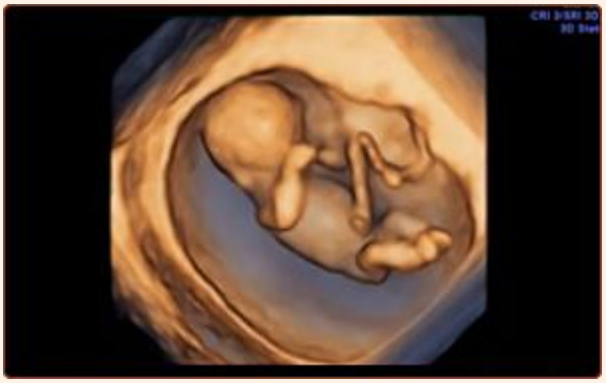
- Downs Syndrome (Trisomy 21)- extra number 21 chromosome
- Edward Syndrome (Trisomy 18)- extra number 18 chromosome
- Patau Syndrome (Trisomy13)- extra number 13 chromosome
- If an abnormality is found at the time of an ultrasound scan, an amniocentesis is offered in most cases.
- Turner syndrome (Monosomy X)- one missing sex chromosome

What information is obtained from blood test?

Beta human chorionic gonadotropin (Free- BhCG) and pregnancy associated plasma protein- A (PAPP-A) levels are assessed from the blood test. Though this test is best done at 10weeks, it can be done just before or on the same day as the scan. In fetuses affected by chromosomal abnormalities the levels of these hormones are frequently abnormal.

What is the additional information obtained from the scan?





The 12 weeks scan helps to provide the following additional information:

- More accurately date the pregnancy
- Diagnose multiple pregnancy
- Detect early pregnancy failure
- Diagnose some major fetal structural abnormalities .

When will I get the results?

If you have already had your blood test and results are available at the time of your scan, we will be able to give you the results immediately after your scan. If you haven't had the blood test, we will need to wait until the blood results become available to assess your risk.

What happens if my test result is 'high risk'?

If you want to have a definitive result to 100% exclude a chromosome abnormality, you should then consider having prenatal diagnostic testing either with CVS or Amniocentesis.

How accurate is the test?

The combined First trimester Screening is the most sensitive and specific screening test for Down syndrome. It detects 90% of chromosomal abnormalities. This means that no matter how low the risk is calculated to be, a chromosomal abnormality like Down syndrome is not completely excluded.

What are the limitations of this test?

A low risk screening result does not guarantee your baby does not have a chromosomal disorder. It is only a screening test and does not actually tell you whether your baby does or does not have a chromosomal disorder. The test can miss about 10% of chromosomally abnormal fetuses.

In addition, a low risk result does not mean that your baby will be born healthy, though majority of babies are healthy at birth.

First trimester screening is offered to all women during their pregnancy. It is a voluntary test where you can 'opt out' if you think it is not an appropriate test for you. You should discuss with your partner and the doctor before deciding to proceed with the testing.