What is antenatal screening?

Antenatal screening is a way of assessing whether your unborn baby is at risk of developing or has developed an abnormality during pregnancy,like Down syndrome, Neural tube defects NTD (spina bifida or anencephaly) and other birth defects.

What is Down syndrome?

Down syndrome is one of the most common genetic birth defects, affecting approximately one in 800 to 1,000 babies. Down syndrome is a disorder that includes a combination of birth defects; among them, some degree of mental retardation, characteristic facial features and, often, heart defects, increased infections, problems with vision and hearing, and other health problems. The severity of all of these problems varies greatly among affected individuals.

What are First & Second trimester screening tests during pregnancy?

	FIRST TRIMESTER	SECOND TRIMESTER
WEEKS	10-13+6 weeks	15-22 weeks
SCREENING TESTS	Nuchal translucency scan Free b-hCG PAPP-A **(dual test)**	Detailed Ultrasound **Triple Test** (AFP, hCG & UE3) **Quadruple test** (triple test+inhibinA)
DIAGNOSTIC TESTS	Chorionic villus sampling	Amniocentesis
SUSPECTED ABNORMALITIES	Down Syndrome Heart Defects Trisomy 18	Down Syndrome Neural tube defects Trisomy 18

What is Triple Screen Test?

A triple test is a screening test that is performed on patient's blood during pregnancy, usually in the second trimester (15-20 weeks). It measures the following protein levels associated with pregnancy in the maternal serum:

1. Alphafetoprotein (AFP)
2. Human chorionic gonadotropin (hCG)
3. Unconjugated estriol (UE3)

Why is it called a "Screen" test?

Screening tests have been designed to identify women who are at increased risk of having a baby with a birth defect. These tests have no risks of miscarriage, but cannot determine with certainty whether a fetus is affected.

What if a screening test is positive?

If you have positive results on a screening test, you can discuss what this means with a genetic counselor or concerned doctor. Options for further diagnostic testing will be explained. The decision as to whether to have invasive genetic testing is up to you.

Who should be offered screening?

All pregnant women should be offered screening for Down syndrome. They should understand that it is their choice after informed discussion whether or not to proceed with the screening programme.

What is the timing of screening?

First trimester-Screening should be performed by the end of the first trimester (11 to 13 weeks 6days)

Second trimester- 15-20weeks (for women booking later in pregnancy)

Who has the greatest risk of having a baby with Down syndrome?

Parents who have already had a baby with Down syndrome, mothers or fathers who have a rearrangement-involving chromosome 21, and mothers over 35 years old are at greatest risk. The risk of Down syndrome increases with age, from about 1 in 1,383 for a woman at age 25, to 1 in 1,000 at age 30, 1 in 350 at age 35, and 1 in 84 at age 40. However, about 80 percent of babies with Down syndrome are born to women who are under age 35, as younger women have far more babies.

What are Diagnostic tests?

Diagnostic tests are extremely accurate at identifying certain abnormalities in the fetus. Diagnostic tests include Amniocentesis, which involves removing amniotic fluid, Chorionic villus sampling (CVS), which involves removing some tissue from the placenta.

Amniocentesis is an invasive test done at 16 weeks to 20 weeks to look for the fetal chromosomes. For Amniotic cell studies about 20 ml of amniotic fluid is removed at 16 weeks of pregnancy by putting a needle through the abdominal wall. This is simple and safe procedure.

Diagnosis of Down syndrome is made by growing the amniotic fluid cells and counting the number of chromosomes in the cells. If the fetus has Down syndrome, the choice is left to the parents to decide whether they would like to get the pregnancy terminated or not. Medical termination of pregnancy can be arranged in a Hospital before 20 weeks of pregnancy by

safe techniques. Risk of fetal loss with amniocentesis is 0.5-1%.

Chorionic Villi Sample is collected at 10- 12 weeks of pregnancy by using a special catheter under ultrasound guidance through the vagina or the sample can also be collected through the abdominal route. This must be done by an experienced obstetrician. The Chorionic Villi cells are examined and the number and structure of chromosomes in the cells is determined. There is a risk of about 3-5% of fetal loss after this procedure.

For Further Information

Contact

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Antenatal Screening

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