Dear Parents to be,

Congratulations! You are stepping into the one of the most rewarding journey of your life, the destination of which will be blissful parenthood. It is indeed our pleasure to support you through this journey as the Fetal Medicine Unit. The endeavor of Fetal Medicine is to safeguard the health and wellbeing of your unborn baby. We specifically follow the growth and development of the fetus and look for any problems and suggest the corrective treatment. Another important aspect in fetal wellbeing is to look for chromosomal or genetic problems that may be inherent and that can profoundly affect the quality of life after birth. In this context, we want to educate you about screening for Down Syndrome.

What is Down Syndrome?

Down Syndrome (DS) otherwise called Trisomy 21 is a chromosomal disorder in which the affected individual has one extra chromosome. Every human being has 46 chromosomes arranged in 23 pairs. In people with Down Syndrome, the chromosome pair number 21 has three copies instead of two so that the total number of chromosomes is 47 instead of the normal 46. Usually it is not inherited and so a baby can be affected even if there is no history of Down's syndrome in the family

What are the characteristics of Down's syndrome?

Children with Down Syndrome will all have some degree of learning difficulty. Many may be able to go to ordinary schools and lead semi-independent lives, but others will need full-time care with a normal life expectancy.

They also have certain physical characteristics, such as eyes that slant upwards and outwards, a single crease running across the palm of the hand, a big gap between the great toe and the 2nd toe, low-set ears and small hands.

Certain medical conditions are more likely to occur in these children:

- Heart defects, which occur in one in three children with Down's
- Digestive tract defects
- Problems with sight
- Problems with hearing

Why should we test for Down's syndrome?

It is the commonest cause of global developmental delay in children and can have a major social, financial and emotional impact on a family.

Are you at risk of having a baby with Down's syndrome?

Every mother is at risk of having a child with Down Syndrome which increases with the mother's age, especially over the age of 35. However, majority of babies with Down's syndrome are born to the relatively younger mothers because there are more number of pregnancies in women below the age of 35.

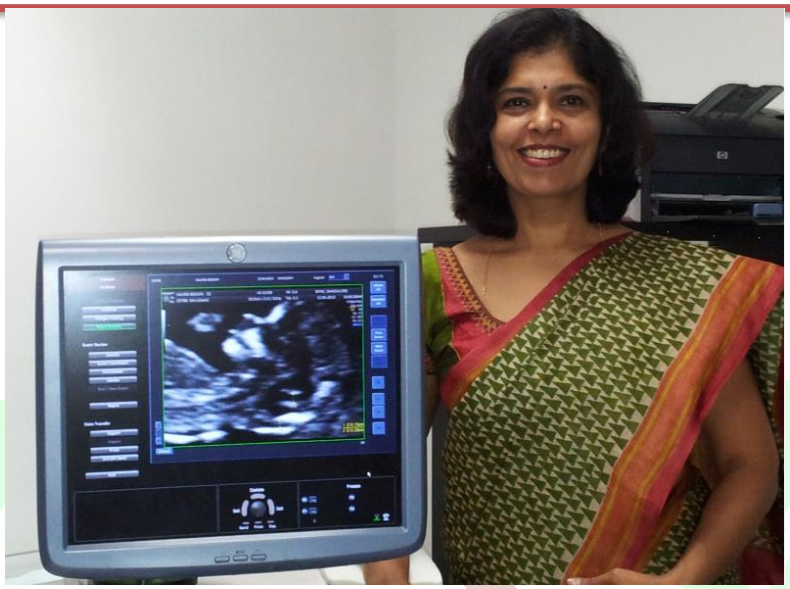
What can you do to find out whether or not you baby has Down's syndrome?

You can undergo certain tests. There are two types of tests available – screening tests and diagnostic tests.

A screening test only tells you whether or not you are at particularly high risk of having a baby with Down's Syndrome. This does not confirm the chromosome status of your baby.

A diagnostic test can give you a definite diagnosis, but also carries a small risk of miscarriage.

What are the screening tests available?



These are ultrasound tests, blood tests and a combination of the two.

The ultrasound tests include the nuchal translucency scan between 11- 14 weeks of pregnancy and the anomaly scan between 18-20 weeks. In these scans, apart from the general growth and development, we look for markers for Down Syndrome in the fetus and depending on the presence or absence of these markers, we assess the risk for Down Syndrome

The blood tests include the first trimester test

which measures two/four hormones in the mother – beta HCG, PAPP-A(Double marker test) or beta HCG, PAPP-A,PLGF, AFP(first trimester quadruple marker test) between 10 weeks to 13+6 weeks of pregnancy or the second trimester Quadruple marker test in which four hormones (beta HCG, AFP,uE3,Inhibin- A) are measured in the maternal blood between 15 weeks to 20+5 weeks.

If both ultrasound and blood tests are done, then the risk assessment obtained from both can be combined to get a more accurate final risk assessment.

A cut-off point of 1 in 300 is usually used at BFMC. If your risk is less than that, you will be described as "screen negative" which means that you are

unlikely to be carrying a baby with Down's (but it's not impossible). If it is > 1 in 300, then you are screen positive meaning you are at high risk of having a baby with Down's syndrome.

What are the diagnostic tests available?

The diagnostic test is either a CVS (Chorionic Villus Sampling) or Amniocentesis which will confirm the chromosomal status of the fetus and tell you definitely whether or not your baby has Down Syndrome. However, these are invasive tests and they carry a risk of miscarriage. Hence, they are generally offered only if the screening tests are suggestive of a screen positive result.

WE HAVE ADDITIONAL INFORMATION LEAFLETS DETAILING FIRST AND SECOND TRIMESTER SCREENING TESTS.