PRENATAL SCREENING

NON INVASIVE PRENATAL TESTING

- The NIPT is a DNA test on maternal blood to safely and reliably screen pregnancies for the most common aneuploidies like Trisomies 21 (downs syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13(Patau syndrome).
- At least 20 ml blood in specific tubes is required from mother which can be done from 10 weeks onwards.
- DNA isolated from maternal blood is sequenced using high throughput next-generation sequencer.
- Turn around time is 2 weeks.
- In case of a high-risk result on NIPT, we have to perform diagnostic test (CVS or Amniocentesis) to confirm the result.
- It is highly accurate screening test till date with detection rate (DR) for T21, T18, and T14 as 99.7%, 98.2%, and 99% respectively.

ADVANTAGES OF COMBINED FIRST TRIMESTER SCREENING:

- Allows earlier detection of an increased risk of Down's syndrome and other chromosomal abnormalities during pregnancy.
- Allows more time for counseling and decision making by the couple.
- The Combined FTS detection rate is approximately 90 to 95% (for a false positive rate of 5%) when the test includes NT along with biochemical analysis for free hCG and PAPP-A.

FOR CHARGES / BOOKING CONTACT RECEPTION.

Services

- Pre Conception Counselling
- Infertility Workup
- IUI or Intra Uterine Insemination
- IVF or In Vitro Fertilization
- ICSI or Intra Cytoplasmic Sperm Injection
- Oocyte Donation and Egg Sharing Programmes
- Surrogacy
- Embyro Freezing and Oocyte Freezing
- Cyro Preserved Semen Bank
- Andrology Clinic
- TESA and PESA
- Basic and Advanced Endoscopic Surgery
- Hormone Analysis
- Genetic Counseling
- Pre Implantation Genetic Diagnosis (PGD)
- Pre Implantation Genetic Screening (PGS)
- Recurrent Miscarriage Clinic
- Nutrition Clinic
- Ultrasound and Fetal Machine

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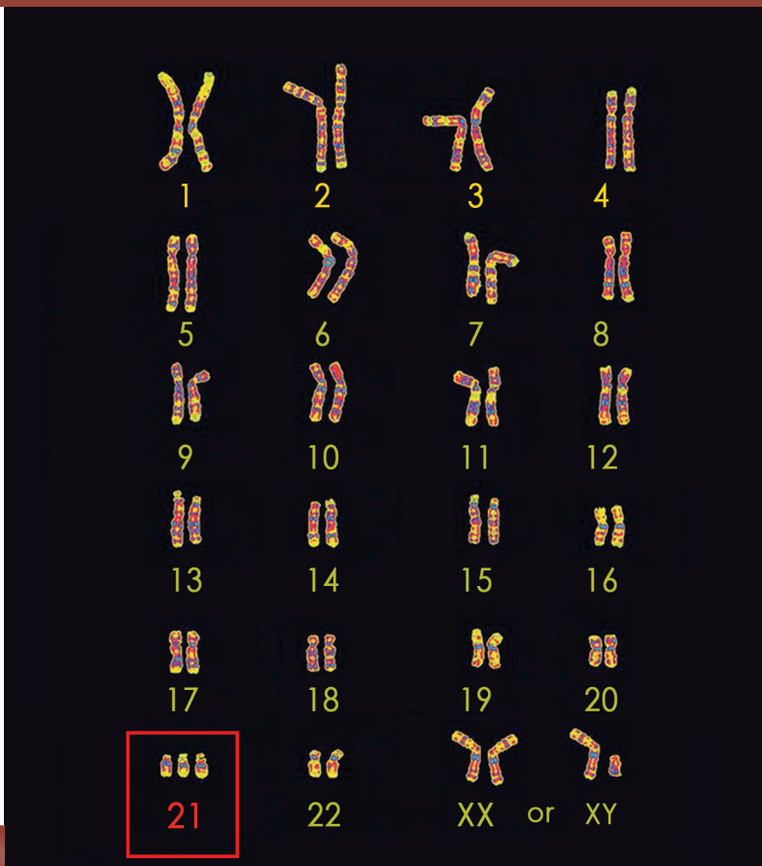

TRISOMY 21 / Downs Syndrome

DETECT BEFORE ITS TOO LATE.

Care IVF

Empowering women's health

Care IVF

DOWN'S SYNDROME

Down's syndrome (DS) is the commonest single cause of significant learning disability in children of school age. Children with Downs Syndrome carry three instead of two copies of the chromosome 21. This is usually a completely random happening, though it is more common in older mothers.

This manifests in children with varying degrees of mental retardation, congenital heart defects, gastrointestinal abnormalities, low muscle tone and abnormal palmar crease.

Throughout the world, the frequency of DS is about 3 per 2000 births.

DIAGNOSTIC TESTING FOR DOWN'S SYNDROME/TRISOMY 18/TRISOMY 13

Amniocentesis : Down syndrome can be diagnosed early in pregnancy (at about 15 to 16 weeks) by amniocentesis. This involves a very fine needle being passed into the womb, under guidance by ultrasound, and sampling of the (amniotic) fluid around the baby. It is done under local anaesthetic, and most women don't find it too uncomfortable. There is a risk, however, of about 1 in 100 to 200 of a spontaneous miscarriage after the procedure. Because of this miscarriage amniocentesis is only offered to women deemed to be at high risk of having baby with Down's Syndrome (Trisomy 21) / Trisomy 18 / Trisomy 13

WHO IS AT HIGH RISK THEN?

Until recently, the only factor used to identify women at high risk for DS was their age. At age 35, for example, the chance of having a baby with DS is about 1 in 360 as compared to 1 in 1350 in a woman at age 25. This has led to many hospitals offering amniocentesis to women over a certain age, usually 35 or 37. The problem with this is that it only will identify 15-30% of all cases of DS, since ALTHOUGH THE RISK IS HIGHER IN WOMEN OF HIGHER AGE, BECAUSE OF HIGHER FERTILITY 80% OF DOWNS SYNDROME CASES OCCUR IN YOUNGER WOMAN.

WHAT IS PRENATAL SCREENING

Prenatal Screening is used to identify those pregnancies in which there is a relatively greater risk of fetal disorder that will cause the child, if it survives to term, to be born disabled. The anomalies most commonly associated with screening programs are Trisomies (such as Down's syndrome, which could be missed by regular ultrasound alone) and open neural tube defects (ONTD's)

Combined First Trimester Screening (FTS) offers a way out as a means of finding those individuals on whom the use of a diagnostic test is warranted. Prenatal Screening Tests only requires the mothers blood sample and fetal parameters from USG scan reports to filter out those pregnancies which are at increased risk for a particular disease from general population.

GLOBAL TRENDS IN PRENATAL SCREENING

American College of Obstetricians and Gynaecologists (ACOG), The Society of Obstetricians and Gynaecologists of Canada (SOGC), The National Health Service, UK states as policy that screening for Down's syndrome should be offered to all pregnant women presenting for maternity care, before 20 weeks of gestation.

HOW DO WE SCREEN FOR DOWN'S SYNDROME WITHOUT DOING INVASIVE TESTS?

Various strategies for Down's syndrome screening exist. The Screening might take place in the first trimester, second trimester or both trimesters and might involve from one to six markers in addition to maternal age. Some of the methods used in different screening programs are summarized in the following table.

Screening Strategy	Analytes and times of tests		Accuracy
FIRST TRMESTER SCREENING			90% - 95%
Combined First Trimester Screening	NT	at 11 weeks, 6 days	
	free HCG & PAPP-A	at 11-13 weeks, 6 days	
SECOND TRIMESTER SCREENING			60% - 65%
Triple test	AFP, hCG (OR FREE hCG) and uE3	at 14-20 Weeks	
Quadruple test	AFP, hCG (OR FREE hCG) and uE3 and inhibin A	at 14-20 Weeks	67% - 71%

In an advanced IVF center like Care IVF, apart from the above markers we would also check the fetus for other markers of aneuploidy like Nasal bone, Tricuspid-regurgitation, ductus venosus, Fronto maxillary Facial angle (FGF angle) which has the potential to further improve the rate to 95%

CAUTION:

However, in order to derive maximum benefit, the program should be conducted as per the guidelines laid down by the Fetal Medicine Foundation (FMF) UK. This means, it is important to perform both, nuchal translucency (NT) as well as biochemical evaluation and get risk assessments based on both. It is also important that the NT is performed by a clinician certified by the FMF. It is equally important that the biochemistry is run on systems that have been tested and accredited by the FMF utilizing chemistry recommended by the foundation and the risk assessments are done on validated software that utilizes local ethnic data, multiples-of-the-median (Moms) as reference.