Center of Medical Genetics

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Genetic Alert for Obstetricians

1. **Genetic Counseling Services:** We have four highly trained Medical Geneticists who provide upto date counseling for risks to the fetus of Chromosomal disease, Genetic disorders and Teratogenic effects of infections, Drugs & Radiation etc. The group has special expertise in Genetic causes of Recurrent Spontaneous Abortions and their Management.

2. **Fetal Medicine and Ultrasound:** Our centre provides fetal intervention techniques such as Amniocentesis, Chorionic villus sampling (CVS), Cordocentesis, Amnio-infusion, and Intra-uterine Transfusions for Rh-incompatibility or Fetal anaemia, in addition to Ist trimester scan, level 2 Ultrasounds and Doppler studies.

3. **Biochemical screening for chromosomal disease** during pregnancy **(First & 2nd trimester)**
   OSCAR (One Stop Clinic for Assessment of Risk in Foetal Anomalies)

These tests are carried out by Delfia Xpress of Perkin Elmer, which is an advanced automated system standardized internationally. It uses Elipse software, considered the best in the world, because it calculates the risk based on ultrasound findings as well as other variables such as ethnic origin, IVF pregnancy, which are not available in the commonly used softwares.
FIRST trimester test (PAPP-A, Free β hCG, NT): 10 week to 13week +6 days
SECOND trimester triple test (AFP+Free β hCG +uE3) & Quadruple test (beta hCG, AFP,Estriol, Inhibin A): 15 to 21 weeks

4. **Amniotic fluid cells – F.I.S.H. studies and Culture** (most economical in Delhi)
   F.I.S.H. for 5 chromosomes aneuploidy (Chr 21/18/13/X/Y)
   F.I.S.H. + Karyotype by Culture

5. **Fetal infections:** Comprehensive management, Avidity testing, PCR in AF/CVS/Cord blood for Rubella, Toxoplasmosis, CMV, Herpes, Parvo, Chickenpox & Enteroviruses.

6. **Non-Invasive Prenatal Diagnosis by Examination of Maternal Blood;**
   Fetal trisomies (21,13,18) (NIFTY), Rh status of foetus by test on maternal blood.

7. **Fetal Autopsy:** Done by experts in Dysmorphology & Perinatal Pathology

8. **Prenatal Diagnosis by Molecular techniques:** Thalassemia, Muscular dystrophy, Mental retardation syndromes (Fragile X etc), Short-limbed dwarfism & many other diseases. maternal cell contamination checked routinely - Enquire for other disorders

- **Chorionic villus sampling** best between **10 – 12 weeks.**
- **Amniocentesis -** best done **after 16 weeks.**
- We provide **F.I.S.H results in 3-4 days.** FISH study should include **5 commonly** involved chromosomes **– 13, 18, 21 X and Y,** and not only chromosome 21

- **Fetal Genetic Evaluation (Fetal Autopsy),** (in cases of *Obstetric Mishaps (IUD, Still Birth, IUGR, abortions Malformed child)*
- ***In short-limbed dwarfism*** *always get a* ***radiograph*** *of baby for proper diagnosis****, For fetus with malformations -*** *record the abnormalities; Collect blood in* ***EDTA for DNA studies, in HEPARIN for chromosomal studies, in PLAIN TUBE for antibodies.*** *If blood is unobtainable collect about 50 MG OF PLACENTA in sterile saline.*

For Further information Contact: Dr I C Verma, FRCP, FAMS, FAAP Dr Ratna D Puri, M.D, D.M
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Recommended Testing Protocol During Early Pregnancy

At the Time of First Contact with Pregnant Woman

- **Hemogram** in place of **only Hb, to exclude thalassemia carrier status**
  Minimum tests Hb, RBC count, MCV, MCH.
  Exclude thalassemia at first contact, as North India has 5 % Carrier rate

- **ABO and Rh typing, Urine – Routine and microscopic**

- **Hepatitis BsAg, HIV, VDRL, Blood Sugar**

- **TORCH – IgG and IgM both, not only IgM.** In early pregnancy IgG must be done to establish immune status, and to provide a base line for judging rise of antibody levels.

Use Avidity test for the antibodies to establish whether Recent or Old infection

Do not recommend abortion based on Maternal TORCH antibodies, Test for infection in the fetus by PCR on Amniotic fluid, or CVS, or Cord blood

- **Thyroid test. Do TSH and Free T4, Not only TSH (High T.S.H.) and Low Free T4** (in the lowest 10^{th} centile) is also a risk factor for lowering of IQ of baby.

- **Biochemical Screening for Down syndrome is required for every woman, and not only for older women (older than 35 years.)**

- The current **recommendation** is to go **for FIRST TRIMESTER SCREENING (PAPP A & Free Beta HCG), along with Nuchal translucency measurement.**
  - Best time for this is **10 to 13 weeks of gestation (based on ultrasound)**
  - **Software should calculate on MOMS, and should include NT in calculations.**
  - If risk of Down syndrome is > 1:50 do CVS, If between 1:50 to 1:1500 do triple test later, If > 1:1500 triple test is not required

- For **TRIPLE TEST, best period of gestation is 16 to 20 weeks. AFP, Unconjugated Estriol, and Free B-HCG, Software should use MOMS of Indian women norms.**
- **Ultrasound 1^{st} Trimester – 11 – 13 weeks, 2^{nd} Trimester - 18-20 weeks (Level 2)**

- **During pregnancy always ask for history of genetic disease - thalassemia, mental retardation, muscular dystrophy.**
- **The gestational period for Chorionic villus sampling is earliest after 10 weeks. For all prenatal diagnosis requiring DNA techniques chorionic villus sampling (CVS) is preferred.**
- **Amniocentesis is best done after 16 weeks.**
- **We provide F.I.S.H results in 3-4 days. FISH study should include 5 commonly involved chromosomes – 13, 18, 21 X and Y, and not only chromosome 21**