SIR GANGA RAM HOSPITAL: NEW DELHI

Center of Medical Genetics

PROFESSIONAL SERVICES

The Center of Medical Genetics offers comprehensive services for the diagnosis, counselling, prenatal diagnosis and management of patients with genetic disorders.

1. **Clinical Diagnosis & Genetics Counselling-** The center has one of the best diagnostic group for Dysmorphology , metabolic diseases, genetic neurological disorders and genetic cancers.

   What is Genetic counselling?
   Genetic counselling is given to the patient and the family explaining the disorder, implications of the test results, the options available, prognosis of the disorder, and its treatment. Specially designed charts and diagrams are used for genetic counselling, to explain in simple terms.
   Genetic counselling is available for:
   - A) Malformations - Single and multiple
   - B) Intellectual Disability - Comprehensive evaluation
   - C) Autism - Protocol driven evaluation
   - D) Neuromuscular disorders - Clinical, Immunohistochemical, and Molecular studies.
   - E) Neurogenetic disorders - Clinical, Biochemical & Molecular studies
   - F) Metabolic disorders –Clinical, Biochemical & Molecular tests special dietary therapies TMS. GC/MS.
   - G) Short stature - Syndrome diagnosis, Hormonal, Cytogenetic & Molecular studies.
   - H) Prenatal diagnosis and Counseling - The most comprehensive program in India.
   - I) Triple, Quadruple and First trimester testing - Using DELFIA Express and Life Express software.
   - J) Cancer genetics - Molecular Testing for all familial cancers
   - K) Thalassemia - The most experienced lab for prenatal diagnosis of thalassemia in India
   - L) Hemophilia - Complete works - inversion 1/22, point mutation detection, prenatal diagnosis, carrier screening.
   - M) Recurrent abortions - Comprehensive diagnosis & management, Lymphocyte Immune therapy
   - N) TORCH infection & Antibody & Avidity testing, PCR for intrauterine infections.

   Who needs Genetic Counselling?
   - A) Couple with previous child with malformations, intellectual disability, developmental delay, autism, neuroregression, dysmorphism, failure to thrive.
   - B) Couple with previous termination of pregnancy in view of ultrasound abnormalities, intrauterine demise, still births, and intrauterine growth retardation.
   - C) Family History of genetic diseases like Thalassemia, Hemophilia, Muscular Dystrophy, Familial Cancers.
   - D) Couple with Recurrent Spontaneous Abortions.
   - E) Child with short stature.

2. **Report Interpretation and Counselling**
   Reports of genetic studies like Fluorescent In Situ hybridisation (FISH), Karyotype, Tandem Mass Spectrometry (TMS), Urine Gas Chromatography Mass Spectrometry (GCMS), Microarray, Next generation Sequencing (NGS).

3. **Fetal Autopsy**
   Fetal Autopsy is recommended in pregnancies terminated for antenatally diagnosed fetal malformations, skeletal dysplasia, Intrauterine Growth retardation (IUGR) and Intrauterine Demise (IUD).
   Requirements for Autopsy:
   A) Fetus should be sent in 10% Formalin (Commercially available formalin is 40%)
   B) Fetal cord blood or fetal cardiac blood in heparin vial (2ml) for karyotyping and EDTA vial (2ml) for DNA storage.
   C) Small piece of placenta from the fetal surface should be sent in Normal Saline. Rest of placenta should be sent in formalin.
   D) Detailed antenatal history & ultrasound Reports.
   E) Fetogram if possible

4. **Enzyme Therapy Infusion**
   Enzyme infusion is given at our center for Mucopolysaccaridosis Type I (Hurler), Mucopolysaccharidosis II (Hunter), Gaucher disease, Fabry disease and Pompe Disease.
   Enzyme infusion requires admission for minimum 6 hrs.

5. **Sweat Chloride Test**
   Sweat chloride test for Cystic Fibrosis
   Prior appointment is required. Test takes around 3 hours.

6. **Glycogen Storage disease stimulation test**
   Glycogen storage stimulation test for diagnosis and management of Glycogen storage disease (GSD).
   Prior consultation and appointment is required.

7. **Growth Hormone Stimulation Test**
   Prior appoinment is required. The patient has to fasting for at least 4 hrs prior to the test. We perform clonidine stimulation test.

8. **Skin Biopsy**
   Skin Biopsy for Karyotype, culture, enzyme studies, diagnosis of Epidermolysis Bullosa.
   Prior appointment is required.

9. **Muscle Biopsy**
   Muscle Biopsy for histopathology and immunostaining for diagnosis of Duchenne Muscular dystrophy, Limb Girdle Muscular Dystrophy, Merosin deficient Congenital muscular dystrophy.
   Prior consultation & appointment is required.

10. **Intravenous Immunoglobulin and intralipid infusion** for Recurrent Spontaneous Abortions.
    This procedure requires admission in hospital for 4 hrs. Prior appointment is required.

11. **Lymphocyte immune therapy**
    Prior appointment with HLA department is required.

12. **Pamidronate infusion** for Osteogenesis Imperfecta
    This requires 3 days admission with prior appointment.