

# Center of Medical Genetics Sir Ganga Ram Hospital

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## Advanced Genetic Tests For Paediatricians

### Biochemical Tests:

1. **Galactosemia** –GALT enzyme (Heparin blood), DNA Test .....
2. **Succinylacetone + Fumaryl acetoacetate for Tyrosinemia** (Heparin blood) .
3. **Organic acids (urine)+ Mass Tandem spectroscopy** Blood (Filter paper specimens)
4. **Aminoacid chromatography**, quantitative (Urine/ Plasma) .....
5. **Metabolic screening-urine (Aminoacid chromatography + chemical tests)** ....
6. **Enzyme assays** for Metachromatic leukodystrophy, Gm1 gangliosidosis, Tay Sachs disease, Pompe's, Gaucher's disease. Krabbe's, Niemann Pick's, MPS VII. Hurler's/ Hunter syndrome.

### DNA Diagnostics:

- **Beta Thalassemia**, Mutation analysis (family) / XMN polymorphism / Prenatal dx
- **Duchenne Muscular Dystrophy**, Deletions/ Prenatal diagnosis/ carrier screen
- **Cystic fibrosis. 25 mutations** including  $\Delta 508$  /  $\Delta 508$  only.....
- **Spinal muscular atrophy** - Diagnosis/Prenatal diagnosis.....
- **Hemophilia A and B**, Carrier screening / Prenatal diagnosis.....
- **Mitochondrial disorders (MELAS, MERRF, Leighs, NARP)**
- **Many other disorders** .....

*For Prenatal Diagnosis Enquire for Requirements & Cost*

### Chromosomal Studies

**Blood/ dysmorphic child/ Bone Marrow for leukemias** .....

**Amniotic cell cultures/ Chorionic villus samples** .....

**Fragile X/ Ataxia telangiectasia / Prader Willi syndrome**.....

**F.I.S.H. studies for Micro-deletion syndromes/ Quick Prenatal diagnosis** .....

**Muscle Biopsy staining with**→ Dystrophin (3 regions), Sarcoglycan antibodies ( $\beta, \alpha, \delta, \Psi$ )

**Merosin, Dysferlin and Dystroglycan antibodies**

**For Further information Contact Staff of Dept. of Genetic Medicine, in Hospital 9.30 am to 5 p.m.;**

**Telephone Nos: 25861767 / 42251382 (Dr. Verma), 42252115 (Molecular),**

**42252111 (Cytogenetics), 42252112 (Biochemical), 42252114 (HLA).**