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, Pompie’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 Δ508 / Δ508 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 (β,α,δ,Υ)
Merosin, Dysferlin and Dystroglycan antibodies

For Further information Contact Staff of Dept. of Genetic Medicine, in Hospital 9.30 am to 5 p.m.;
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42252111 (Cytogenetics), 42252112 (Biochemical), 42252114 (HLA).