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The Most Comprehensive Neurogenetic Diagnostic Program in India

DNA Diagnostics:

- Adrenoleukodystrophy – Sequencing of ABCD 1 gene
- Angelman syndrome – Methylation test
- Apo E Genotyping
- Ataxia telengiectasia – Linkage
- Canavan disease – Sequencing of gene / NAA in urine
- **CMT1** (Charcot – Marie Tooth disease) / **HMSN** - Deletion / Duplication of PMP gene
- **Craniosynostosis**, Apert syndrome, Crouzon disease – Gene studies
- **Duchenne/ Becker Muscular Dystrophy** *Deletion / Duplication test* ***of all 79 exons***/ Carrier testing / Prenatal
- **Dystonia** DYT1 gene (Early onset< 30 years, localised or generalised), common deletion
- **Dystonia** DYT5 (DOPA responsive) – GTP Cyclohydrolase 1 gene sequencing
- Fragile X syndrome (PCR screening test / Methylation test)
- **Friedreich's ataxia –** Molecular studies provide a definitive diagnosis
- Hallervorden-Spatz disease/ Carrier screening / Gene sequencing
- **Huntington disease –** Symptomatic & Predictive testing
- **Hydrocephalus –** X-linked (MASA syndrome) – Linkage studies, and sequencing
- **Krabbe's disease** – Common deletion (by DNA), Japanese encephalitis – Enzyme assay
- Leber's Hereditary Optic Nerve atrophy (LHON) - 3 mutations
- **Limb Girdle Muscular dystrophy** - Gene SEQUENCING of α, β, γ, and δ sarcoglycanopathies
- McArdle disease - R49X mutation by sequencing, Arm ischemia test, Machado-joseph disease
- **Megalencephaly, cystic** (van der Knaap type), Specific mutation in Aggarwals, For Other Ethnic groups - SEQUENCING of gene
- **Mental Retardation Panel 1** (**9 chm** regions); & **Panel 2** (**6 chm**) for *checking microdeletions*
- **Mental Retardation X panel,** for checking deletions in X chromosome (**11 genes**)
- **Merosin deficiency Muscle dystrophy:** Antibody & Gene studies / Prenatal diagnosis
- **Mitochondrial disorders**: MERRF (3 Mutations); MELAS (3 mutations), NARP (3 mutations), **L**eigh's (Two mutations), SEQUENCING of Mitochondrial genome
- **Mitochondria – Deletion studies (Kaern-Sayre** syndrome plus other deletions**)**
- **MTHFR** (Methylene tetrahydrofolate reductase gene polymorphism) **677C>T, 1298A>C**
- **Mytonic dystrophy**. Type 1 & type 2 (PCR based)
- **Neuronal Ceroid lipofuscinosis (NCL)** – Mutation studies + Enzyme assays

- Neurofibromatosis – Predictive testing, Prenatal diagnosis by linkage
- **Parkinsons disease (idiopathic)** – Gly2019 Ser mutation – by SEQUENCING of LRRK2 gene
- **Pelizaus Merzbacher** disease: Deletion / duplication of PLP gene (majority of cases)
- **Porphyria** (AIP) – Specific mutation in *HMBS* gene in Rajasthanis, SEQUENCING of the gene in others
- **Rett syndrome** – SEQUENCING, plus DELETION / DUPLICATION study
- **Spinal bulbar muscular atrophy (SBMA) - Kennedy disease ,** CAG repeats in AR gene
- **Spinal muscular atrophy**- (SMA) Diagnosis/ **Carrier screening (Dosage study) / Prenatal diagnosis**
- **Spinocerebellar ataxias** (SCA) type 1,2,3,6,7,12,17, DRPLA, others being set up
- **Subtelomeric deletions** of all 46 chromosomes
- Thrombophilia - Factor V Leiden, MTHFR (677C>T, 1298 A>C, & Prothrombin gene G20210A)
- **Tuberous Sclerosis :** Gene Sequencing of TSC 1 and TSC 2 genes
- **Von Hipple Landau disease (VHL)-** Gene SEQUENCING & Deletion/ Duplication study
- **Wilson disease**, Diagnosis by linkage/ Mutation analysis/ SEQUENCING of gene

PCR Diagnosis of Infections in C.S.F / blood:

- Cytomegalovirus , Enteroviruses , Dengue, Japanese Encephalitis
- Herpes viruses 1 / 2, Toxoplasmosis , Tuberculosis, Chikingunya virus

Pharmacogenomic tests to reduce adverse reactions

- **Anti-coagulant therapy** - Warfarin / Acetrom – Polymorphism VKROC1 and Cyp2C19 gene
- **Carbamazepine therapy** (HLAB*1502)

Biochemical Tests:

- Adrenoleukodystrophy & Peroxisomal disorders (VLCFA analysis)
- Aminoacid chromatography, and Quantification by HPLC
- Organic acids analysis
- Tandem Mass Spectrometry for aminoacid & fatty acid oxidation disorders

Enzyme assays

- GM1 gangliosidosis
- GM2 gangliosidosis (**Tay Sach disease**, Sandhoff disease))
- Krabbe's disease (Enzyme + gene studies)
- **Metachromatic leukodystrophy**
- MPS (Mucopolysacharidosis) I, II, III, IV, VI, Sly disease
- Neuronal Ceroid Lipofuscinosis (NCL) infantile, Late infantile (two enzymes)
- Niemann Pick disease, Other Sphingolipid disorders

Muscle Biopsy staining with antibodies

- Calpain (Western Blotting, Dysferlin antibody
- $\propto$-Dystroglycan antibodies (for merosin +ve congenital muscular dystrophies)
- Dystrophin – 3 regions
- Merosin, Sarcoglycan antibodies (β,α, δ, γ)

Enquire for Charges; and for Availability of other Tests

Send Samples by Courier to Reach within 48 hours. No Refrigeration required**.**

Molecular diagnosis 5 ml EDTA blood**; Biochemical Enzyme assays** 6 ml HEPARIN blood;

For Further information Contact Staff of Dept. of Genetic Medicine, FROM 9.30 AM to 5 PM;

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