



## MOLECULAR DIAGNOSTIC FACILITY

### a. Indications

- Hemoglobinopathies** including thalassemias, sickle cell anemia, hemoglobin E disease
- Musculopathies** including Duchenne and Becker Muscular Dystrophy, Spinal Muscular Atrophy, myotonic dystrophy etc
- Neurodegenerative diseases** like Spinocerebellar Ataxias, Huntington's disease etc
- Mental retardation** syndromes like Fragile-X Syndrome
- Bleeding disorders** like Hemophilia A and B
- Non-Syndromic Hearing Loss (NSHL)**
- Cystic fibrosis (CF)**
- Mitochondrial disorders** such LHON, Leigh Syndrome, MELAS etc.
- Susceptibility screening** for MTHFR, Factor V, Prothrombin, HFE, etc.

### b. Sample collection

#### (i) *Molecular analysis*

5-10 ml of blood should be collected aseptically in an **EDTA vacutainer (purple cap)**, **labelled with patient's name, age, sex and date of collection of sample**. The details of dispatch may be intimated earlier through e-mail so that proper arrangements for receiving samples are made.

#### (ii) *Prenatal Diagnosis*

A minimum of 30 to 40 mg of **properly processed** chorion villus sample (in sterile culture medium or absolute alcohol or normal saline) or 20-30 ml of amniotic fluid (in plain sterile container) or 1-2 ml fetal cord blood sample (in EDTA vacutainer) collected aseptically should be sent. It is mandatory that the DNA marker for specific disease in the proband/family be established before proceeding for prenatal diagnosis. Please contact us before referral in case of any doubt. It is further advised to inform us at least one day in advance before proceeding for prenatal diagnosis.

### c. Sample transport

The samples may be sent at room temperature via courier so as to reach us within 48 hours. All the samples should be accompanied with the properly filled Information Sheet and the consent form duly signed by the patient or his relatives. **Please ensure that any sample for prenatal analysis is accompanied by Form G as specified by the Pre-Conception and Pre-Natal Diagnostic Techniques (Regulation and Prevention of Misuse) Act, 1994 (Amended in 2003).**

### d. Contact details

Please contact us for any further information, especially regarding the choice of test for a particular family.

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**e. Charges & Reporting time (From 01.02.2009)**

<b>Disease</b>	<b>Charges per sample</b>	<b>Service Tax (10.3%)</b>	<b>Total Charges</b>	<b>Reporting Time</b>
<b>β-Thalassemia</b> Seven common mutations (IVS 1-5 G>C, IVS 1-1 G>T, 619 bp deletion, Codon 15 G>A, Codon 30 G>A, FS 8/9 +G, FS 41/42 – CTTT) Screening for specific mutation (as per the request of doctor) Detection of novel mutations (by sequencing)	Rs 2,038/- Rs 1,223/- Rs 4,075/-	Rs 209.91 Rs 125.96 Rs 419.73	Rs 2,248/- Rs 1,349/- Rs 4,495/-	2 weeks 10 days 4 weeks
<b>Sickle Cell Anemia</b> <b>Hemoglobin-E Disease</b>	Rs 1,223/- Rs 1,223/-	Rs 125.96 Rs 125.96	Rs 1,349/- Rs 1,349/-	10 days 10 days
<b>Duchenne and Becker Muscular Dystrophy</b> Deletion analysis for 24 exons <b>Carrier analysis</b> Using intragenic markers (Intron 3, 44 and 49) Q-PCR for specific exon	Rs 1,834/- Rs 4,075/- Rs 4,075/-	Rs 188.90 Rs 419.73 Rs 419.73	Rs 2,023/- Rs 4,495/- Rs 4,495/-	2 weeks 4 weeks 4 weeks
<b>Spinal Muscular Atrophy</b> Deletion analysis (exon 7 and 8 only) Carrier analysis using Q-PCR (exon 8 only)	Rs 1,531/- Rs 4,075/-	Rs 155.00 Rs 419.73	Rs 1,686/- Rs 4,495/-	2 weeks 4 weeks
<b>Fragile-X Syndrome</b> Southern Blot based analysis for FRAXA locus using EcoRI, EcoRI+EagI or PstI digestion ( <i>Please specify</i> ) <b>Myotonic Dystrophy</b> Southern Blot based analysis	Rs 2,038/- Rs 2,038/-	Rs 209.91 Rs 209.91	Rs 2,248/- Rs 2,248/-	4 weeks 4 weeks
<b>Triplet Repeat Expansion Associated Diseases</b> Huntington's Disease, Spinocerebellar Ataxias 1, 2, 3 and 6, DRPLA, Friedrich's Ataxia	Rs 1,531/- for single disease Rs 4,075/- for panel (SCA 1, 2, 3)	Rs 155.00 Rs 419.73	Rs 1,686/- Rs 4,495/-	2 weeks 4 weeks
<b>Hemophilia-A</b> Intron-22 inversion by Southern Blot analysis Carrier analysis @ intragenic markers (BclI & XbaI RFLP) <b>Hemophilia-B</b> Carrier analysis @ intragenic markers (HhaI, DdeI, TaqI)	Rs 2,038/- Rs 2,038/- Rs 2,038/-	Rs 209.91 Rs 209.91 Rs 209.91	Rs 2,248/- Rs 2,248/- Rs 2,248	4 weeks 3 weeks 3 weeks
<b>Chronic Pancreatitis</b> (N34S SPINK1 mutation)	Rs 1,223/-	Rs 125.96	Rs 1,349/-	10 days
<b>Cystic fibrosis</b> delta F508 mutation Four common mutations (dF508, G542X, G551D, R553X) 5T mutation <b>G6PD deficiency</b> Orissa, Mediterranean and Kerala-Kalyan mutations <b>Pre-coagulation profile</b> Factor V Leiden and Prothrombin (G20210A) mutation Single mutation analysis (any of the above two mutations) <b>MTHFR gene polymorphisms</b> 677T>C and 1298A>C mutations <b>Hereditary Haemochromatosis</b> H63D and C282Y mutations <b>Mitochondrial encephalopathy</b> <b>LHON</b> - 3 mutations (G3460A, G11778A, T14484C) <b>Leigh' disease</b> - 3 mutations (T12706C, A13084T, G13513A) Specific mutation (as per the request of referring doctor)	Rs 1,223/- Rs 2,038/- Rs 1,223/- Rs 2,038/- Rs 2,038/- Rs 2,038/- Rs 1,223/- Rs 2,038/- Rs 2,038/- Rs 2,038/- Rs 2,038/- Rs 1,223/-	Rs 125.96 Rs 209.91 Rs 125.96 Rs 209.91 Rs 209.91 Rs 125.96 Rs 209.91 Rs 209.91 Rs 209.91 Rs 125.96	Rs 1,349/- Rs 2,248/- Rs 1,349/- Rs 2,248/- Rs 2,248/- Rs 1,349/- Rs 2,248/- Rs 2,248/- Rs 2,248/- Rs 2,248/- Rs 2,248/- Rs 1,349/-	10 days 2 weeks 10 days 2 weeks 2 weeks 2 weeks 10 days 2 weeks 2 weeks 3 weeks 3 weeks 10 days
<b>Non-Syndromic Hearing Loss (NSHL)</b> Connexin 26 (GJB2) exon 2 screening (by sequencing)	Rs 2,038/-	Rs 209.91	Rs 2,248/-	2 weeks
<b>DNA Isolation and Storage upto two years</b>	Rs 509/-	Rs 52.42	Rs 562/-	1 week
<b>Prenatal Diagnosis</b>	Rs 4,075/-	Rs 419.72	Rs 4,495/-	3-7 days
<b>Maternal cell contamination</b>	Rs 2,546/-	Rs 262.23	Rs 2,809/-	3-7 days
<b>Prenatal Diagnosis &amp; Maternal cell contamination</b>	Rs 6,112/-	Rs 629.53	Rs 6,742/-	3-7 days

