सी सी एम बी CCMB
कोशिकीय एवं आणविक जीवविज्ञान केन्द्र
CENTRE FOR CELLULAR AND MOLECULAR BIOLOGY
उप्पल रोड़, हैदराबाद 500 007 भारत.
Uppal Road, Hyderabad 500 007 India.

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

1-2 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/her relatives. **Please ensure that any sample for prenatal analysis is accompanied by Form F and 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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Scientist and Group Leader
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Revised charges effective from 15th July 2016

Disease	Charges per sample	Service Tax @ (15%)	Total Charges	Reporting Time
β-Thalassemia				
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)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Screening for specific mutation (as per the request of doctor)	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Detection of novel mutations (by sequencing)	Rs 1,739/-	Rs 261/-	Rs 2,000/-	4 weeks
Sickle Cell Anemia	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Hemoglobin-E Disease	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Duchenne and Becker Muscular Dystrophy				
Deletion analysis for 24 exons	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Deletion analysis for 79 exons by MLPA analysis	Rs 2,174/-	Rs 326/-	Rs 2,500/-	2 weeks
Carrier analysis using MLPA	Rs 2,174/-	Rs 326/-	Rs 2,500/-	2 weeks
Spinal Muscular Atrophy				
Deletion analysis (exon 7 and 8 only)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Carrier analysis using MLPA	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
**SMN* gene sequencing (exons 1 to 8)	Rs 3,913/-	Rs 587/-	Rs 4,500/-	4 weeks
Fragile-X Syndrome				
*CGG repeat tract analysis by Amplidex Kit	Rs 3,044/-	Rs 456/-	Rs 3,500/-	10 days
Myotonic Dystrophy				
TP-PCR based analysis	Rs 1,304/-	Rs 196/-	Rs 1,500/-	4 weeks
Triplet Repeat Expansion Associated Diseases	Rs 1,304/- for single disease	Rs 196/-	Rs 1,500/-	2 weeks
Huntington's Disease, Spinocerebellar Ataxias 1, 2, 3 and 6, DRPLA, Friedrich's Ataxia	Rs 3,044/- for panel (SCA 1, 2, 3)	Rs 456/-	Rs 3,500/-	4 weeks
Hemophilia-A				
Carrier analysis @ intragenic markers (BclI & XbaI RFLP)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	3 weeks
Hemophilia-B				
Carrier analysis @ intragenic markers (HhaI, DdeI, TaqI)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	3 weeks
Chronic Pancreatitis (N34S *SPINK1* mutation)	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Cystic fibrosis				
delta F508 mutation	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Four common mutations (dF508, G542X, G551D, R553X)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
5T mutation	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Glucose 6 phosphate dehydogrenase deficiency				
Orissa, Mediterranean and Kerala-Kalyan mutations	Rs 1,739/-	Rs 261/-	Rs 2,000/-	2 weeks
Pre-coagulation profile				
Factor V Leiden and Prothrombin (G20210A) mutation	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Single mutation analysis (any of the above two mutations)	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
***MTHFR* gene polymorphisms**				
677T>C and 1298A>C mutations	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Hereditary Haemochromatosis				
H63D and C282Y mutations	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
Mitochondrial encephalopathy				
LHON - 3 mutations (G3460A, G11778A, T14484C)	Rs 1,739/-	Rs 261/-	Rs 2,000/-	3 weeks
Leigh' disease - 3 mutations (T12706C, A13084T, G13513A)	Rs 1,739/-	Rs 261/-	Rs 2,000/-	3 weeks
Specific mutation (as per the request of referring doctor)	Rs 870/-	Rs 130/-	Rs 1,000/-	10 days
Non-Syndromic Hearing Loss (NSHL)				
Connexin 26 (*GJB2*) exon 2 screening (by sequencing)	Rs 1,304/-	Rs 196/-	Rs 1,500/-	2 weeks
DNA Isolation and Storage up to two years	Rs 500/-	Rs 75/-	Rs 575/-	1 week
Prenatal Diagnosis	Rs 3,044/-	Rs 456/-	Rs 3,500/-	3-7 days
Maternal cell contamination	Rs 1,304/-	Rs 196/-	Rs 1,500/-	3-7 days
Prenatal Diagnosis & Maternal cell contamination	Rs 4,348/-	Rs 652/-	Rs 5,000/-	3-7 days