सी सी एम बी
कोशिकीय एवं आणविक जीवविज्ञान केन्द्र
(वैज्ञानिक तथा औद्योगिक अनुसंधान परिषद्)
उप्पल रोड, हैदराबाद - 500 007, भारत.

CCMB
CENTRE FOR CELLULAR AND MOLECULAR BIOLOGY
(Council of Scientific & Industrial Research)
Uppal Road, Hyderabad - 500 007, India.

MOLECULAR DIAGNOSTIC FACILITY

a. Indications

- **Hemoglobinopathies** including Thalassemias, Sickle Cell Anemia, Hemoglobin E disease.
- **Neuromuscular diseases** including Duchenne and Becker Muscular Dystrophy, Spinal Muscular Atrophy, Myotonic Dystrophy, etc.
- **Neurodegenerative diseases** like Spinocerebellar Ataxias, Huntington disease, Fragile-X Syndrome, etc.
- **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.
- **Any disorder with evidence of genetic susceptibility** for NGS based approaches like Exome Sequencing etc.

b. Sample collection

(i) Molecular analysis

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.** The dispatch details may be intimated earlier through e-mail for proper care.

(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 pathogenic variation for specific disease in the proband/family be established before proceeding for prenatal diagnosis. Please contact us before referral, in case of any doubt and 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. The samples should be accompanied with a properly filled Information Sheet and the Consent Form duly signed by the patient or his/her relatives and a demand draft/online payment receipt for appropriate amount as per the attached sheet. Online payment can be made through the link *https://www.onlinesbi.com/sbicollect/icollecthome.htm?corpID=378316* by choosing the drop down option as TSP1 MOLECULAR DIAGNOSTICS. **Please ensure that any sample for prenatal analysis MUST BE 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) and a document justifying the test (Previous test report, USG report, etc.).**

d. Contact details

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

Dr Giriraj Ratan Chandak/Dr T Karthik Bharadwaj/Dr Surya Balakrishnan
Molecular Diagnostics Division
Clinical Research Facility (CRF)
ANNEXE- II of CSIR-CCMB (Opposite GENPACT)
Uppal Road, Hyderabad 500 039.
E-Mail: nidan@ccmb.res.in, nidanccmb@gmail.com
Website: *http://www.ccmb.res.in/index.php?view=molecular_dignosis&mid=192&id=81*
Please call on **040-27195612** in case of any enquiry.

Revised test list and charges from 15th February 2020		
Disease	**Total Charges**	**Reporting Time**
Beta-Thalassemia		
Six common mutations (IVS 1-5 G>C, IVS 1-1 G>T, Codon 15 G>A, Codon 30 G>A, FS 8/9 +G, FS 41/42 - CTTT)	Rs 2,000/-	2 weeks
619 bp deletion	Rs 1,400/-	10 days
Screening for specific mutation (as per the request)	Rs 1,400/-	10 days
Detection of novel mutations (by sequencing)	Rs 2,700/-	4 weeks
Sickle Cell Anemia	Rs 1,400/-	10 days
Hemoglobin-E Disease	Rs 1,400/-	10 days
Duchenne and Becker Muscular Dystrophy		
Deletion analysis for 24 exons	Rs 2,000/-	2 weeks
Deletion analysis for 79 exons by MLPA analysis	Rs 3,300/-	2 weeks
Carrier analysis using MLPA	Rs 3,300/-	2 weeks
Spinal Muscular Atrophy		
Deletion analysis (exon 7 and 8 only)	Rs 2,000/-	2 weeks
Carrier analysis using MLPA	Rs 2,000/-	2 weeks
SMN1 gene sequencing (exons 1 to 8)	Rs 6,000/-	4 weeks
Triplet Repeat Expansion Associated Diseases		
Fragile-X Syndrome (CGG repeat tract analysis by Amplidex Kit)	Rs 4,700/-	10 days
Myotonic Dystrophy (TP-PCR based analysis)	Rs 2,000/-	2 weeks
Dentato-Rubral Pallido Luysian Atrophy (DRPLA)	Rs 2,000/-	2 weeks
Huntington Disease	Rs 2,000/-	2 weeks
Friedrich Ataxia	Rs 2,000/-	2 weeks
Spino Bulbar Muscular Atrophy (SBMA)	Rs 2,000/-	2 weeks
Spinocerebellar Ataxia- 1, 2 & 3	Rs 4,700/-	2 weeks
Spinocerebellar Ataxia- 1, 2, 3, 6 & 12	Rs 8,700/-	4 weeks
Spinocerebellar Ataxia (any one type)	Rs 2,000/-	2 weeks
Other Diseases		
Chronic Pancreatitis		
(N34S *SPINK1* mutation)	Rs 1,400/-	10 days
Cystic fibrosis		
delta F508 mutation	Rs 1,400/-	10 days
Four common mutations (dF508, G542X, G551D, R553X)	Rs 2,000/-	2 weeks
5T mutation	Rs 1,400/-	10 days
CFTR 36 mutation panel (Devyser Kit)	Rs 6,000/-	4 weeks
Glucose 6 phosphate dehydrogrenase deficiency		
Orissa, Mediterranean and Kerala-Kalyan mutations	Rs 2,700/-	2 weeks
Pro-coagulation profile		
Factor V Leiden and Prothrombin (G20210A) mutation	Rs 2,000/-	2 weeks
Single mutation analysis (any of the above two mutations)	Rs 1,400/-	10 days
***MTHFR* gene polymorphisms**		
677C>T and 1298A>C mutations	Rs 2,000/-	2 weeks
Hereditary Haemochromatosis		
H63D and C282Y mutations	Rs 2,000/-	2 weeks
Mitochondrial diseases		
LHON - 3 mutations (G3460A, G11778A, T14484C)	Rs 2,700/-	3 weeks
Leigh disease-3 mutations (T12706C, A13084T, G13513A)	Rs 2,700/-	3 weeks
Specific mutation (LHON/ Leigh) (as per the request)	Rs 1,400/-	10 days
Non-Syndromic Hearing Loss (NSHL)		
Connexin 26 (*GJB2*) exon 2 screening (by sequencing)	Rs 2,000/-	2 weeks
Miscellaneous		
JAK2 mutation analysis for Polycythaemia Vera (V617F)	Rs 2,000/-	2 weeks
FGFR3 mutation analysis for Achondroplasia (c.1138G>A/ c.1138G>C)	Rs 2,000/-	2 weeks
UGT1A1 promoter region polymorphism for Gilbert syndrome [$(TA)_7TAA$]	Rs 2,000/-	2 weeks
CAPN3 founder mutations for LGMD2A (c.2051-1G>T and c.2051-1G>T)	Rs 2,000/-	2 weeks
Targeted mutation analysis (as per the request)	Rs 4000/-	4-6 weeks
DNA Isolation and Storage up to two years	Rs 1,000/-	1 week
Prenatal Diagnosis	Rs 4,700/-	3-7 days
Maternal cell contamination	Rs 2,000/-	3-7 days
Prenatal Diagnosis & Maternal cell contamination	Rs 6,700/-	3-7 days
Next Generation Sequencing		
Exome sequencing	Rs 22,000/-	6-8 weeks
Exome/Panel - data analysis	Rs 5,000/-	4 weeks