GENETIC SERVICES UNIT

(Supported by the Government of West Bengal, Department of Biotechnology)

3rd Floor, PG Polyclinic Building, 5 Suburban Hospital Road, Kolkata 700020

National Institute of Biomedical Genomics

(An Autonomous Institution of the Government of India, Department of Biotechnology)

Email: gsu.bmgu@nibmg.ac.in

Test Request Form

(Please contact the laboratory before sending the sample.)

For a sample to be accepted for testing, it is essential to provide all information required below.

Patient Information

Name: _____ _____ _____

(BLOCK LETTERS) (FIRST) (MIDDLE) (LAST)

Sex: **Male** / **Female** Date of Birth (dd/mm/yyyy): _ _/_ _/ _ _ _ _ Age (if DoB is unavailable):_____

Father's Name: _____ Mother's Name: _____

Patient's Hospital Registration No.: _____

Clinical Information

Clinical Diagnosis:

Clinical / Laboratory / Radiological Data Supporting Clinical Diagnosis:

Test Requested: _____

Prescribing Physician's Information

Name: Dr. _____ _____ _____

(BLOCK LETTERS) (FIRST) (MIDDLE) (LAST)

Prescribing Government Hospital and Department: _____

Phone Number: _____ E-mail Address: _____

Date of sample collection: ____/___/___ **Prescribing physicians signature with date**: _____

The Genetic Services Unit of NIBMG has my consent to carry out the test(s) requested herein by my physician in a research mode and to store my DNA for any future research for the benefit of humankind without revealing my identity. I have understood that the results of the tests requested herein will only be provided to my physician and will not be provided directly to me or to anyone else.

Signature of patient / legal guardian with date: _____

Date sample received at the laboratory: ___/____/_____ Acceptable sample: Yes / No

Name of person receiving the sample: _____ Sample Number: _____

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LIST OF TESTS OFFERED BY THE GENETIC SERVICES UNIT

Sl. no	Name of the test	Method	Sl. no	Name of the test	Method
1.	Beta Thalassemia major (4 common mutations)	Multiplex ARMS-PCR	24.	Down Syndrome (Trisomy 21)	QF-PCR
2.	HbE trait	PCR-RFLP	25.	Patau Syndrome (Trisomy 13)	QF-PCR
3.	HBB (exon1-2, exon 3) sequencing	Sanger Sequencing	26.	Edwards Syndrome(Trisomy 18)	QF-PCR
4.	HBG2 XmnI polymorphism in Beta Thalassemia	PCR-RFLP	27.	Chromosomal aneuploidies (13, 18, 21, X and Y)	MLPA
5.	Sickle cell anaemia	ARMS-PCR	28.	Microdeletion Syndromes ▪ 1p36 deletion syndrome ▪ Wolf-Hirschhorn syndrome ▪ Cri-du-Chat Syndrome ▪ Sotos syndrome ▪ Saethre-Chotzen syndrome ▪ Williams-Beuren syndrome ▪ Langer-Giedion syndrome ▪ WAGR syndrome ▪ Prader-Willi syndrome ▪ Angelman syndrome ▪ Rubinstein-Taybi syndrome ▪ Miller-Dieker syndrome ▪ Lissencephaly 1 ▪ Smith-Magenis syndrome ▪ Alagille syndrome ▪ DiGeorge syndrome ▪ 22q11.2 microduplication syndrome ▪ Phelan-McDermid syndrome	MLPA
6.	Hemophilia A	Inverse-PCR			
7.	TPMT *3C (rs1142345) genotyping	ARMS-PCR			
8.	Spinal Muscular Atrophy	MLPA			
9.	Duchenne / Becker Muscular Dystrophy	MLPA			
*10.	Spinocerebellar ataxia types 1,2,3,7,12	Fluorescent PCR, TP-PCR			
*11.	Friedreich`s ataxia	Fluorescent PCR, TP-PCR			
*12.	Myotonic dystrophy	Fluorescent PCR, TP-PCR			
13.	Dystonia types 1 & 6 [sequencing of exon 5 of TOR1A gene (c.907_909delGAG mutation) and all coding exons of THAP1 gene]	Sanger Sequencing			
*14.	Huntington disease	Fluorescent PCR			
15.	Wilson disease	Sanger Sequencing			
16.	Familial hypokalaemia periodic paralysis	Sanger Sequencing			
17.	Cystic Fibrosis (Del F508 mutation in CFTR gene)	ARMS-PCR			
18.	MELAS	PCR-RFLP			
19.	Achondroplasia	PCR-RFLP			
20.	Connexin 26 (GJB2) c.71G > A(p.W24X) genotyping in pre-lingual sensorineural hearing loss	PCR-RFLP			
21.	Factor V Leiden mutation (F5 gene)	PCR-RFLP			
22.	Prothrombin G20210A mutation (F2 gene) in thrombophilia	PCR-RFLP			
*23.	Fragile X syndrome (sizing of alleles within normal range)	Fluorescent PCR			

PROCEDURE OF REFERRAL TO GSU FOR A GENETIC TEST

- Contact the Genetic Services Unit before sending a sample.
- For DNA based tests, 2-3 ml blood should be collected in a sterile tube containing EDTA. The tube should be labelled immediately before collection (patient's name, referring Government hospital registration number and date of collection).
- The tube containing the blood sample should be sent so as to reach GSU within 4 hours of blood collection at room temperature (**preferable**) or
- The tube containing the blood sample may be stored at 4 °C inside the main compartment – not the ice compartment – of a refrigerator and sent within 7 days in an container filled with ice.
- The tube must be sent together with the signed and completely filled test request form (TRF) between 10 am to 5 pm, Monday through Friday.
- All the tests done in GSU are in research mode & free of cost.

*External validation pending

Turn Around Time (TAT): Period required for reporting is variable depending on the nature of the test (expected 6-8 weeks). Report will be provided only to the referring physician and not to the patient. Report can be provided either personally or through post / email.