REQUEST FORM FOR MOLECULAR DIAGNOSTICS SERVICES

Unit of Molecular Diagnostics
Specialised Diagnostics Centre
Institute for Medical Research
National Institute of Health, MOH
Jalan Pahang, 50588 Kuala Lumpur
Tel: 03-26162783/ 2581

To The Requesting Lab / Person,
Please STAMP HERE

Patient Name :

Patient IC/ID :

Date of Birth : **Age :**

Gender : Male / Female / Unknown

Ethnicity/Nationality :

Hospital :

Ward/Clinic :

Name of Attending Doctor (Specialist) :

If this is a parental or family member sample, please state
Proband/Child's Full Name …….. **IC/ID** …… **DOB** ……

Reason for Referral:

Diagnostic test : ❒ Affected patient ❒ Possibly affected patient
Carrier testing : ❒ Father of affected patient ❒ Mother of affected patient
❒ Sibling of affected patient ❒ Other family member of affected patient (please specify) : ……
Predictive testing : ❒

Type of Specimen Sent:
❒ Whole blood ❒ Blood spot ❒ Tissue (please specify) : …… ❒ Urine ❒ Extracted DNA
❒ Others (please specify): …… Date of sample taken: ……

Please Read This Section Before You Proceed

Requirements for clients requesting molecular diagnostics services from UMD, IMR :

1. ***All cases requiring molecular diagnostics testing must be referred to any Clinical Geneticist/Neurologist and they must endorse the test before any sample submission. Samples received without referral by Clinical Geneticist/Neurologist will be rejected.***
2. *Please ensure that the patient and/or their legal guardian understands the implications of genetic testing and provide his/her consent to undertake the test.*
3. *Please send the samples according to the criteria for sample collection as outlined below.*
4. *Kindly ensure samples are sent together with both the request form and informed consent form.*

Criteria for sample collection :

1. *2.5 ml blood in EDTA (purple/lavender cap) tube, **DO NOT** use Heparin (green cap) tube. Send about 1-2 tubes in appropriate packaging at AMBIENT condition as soon as possible after collection. If more than 3 hours, keep sample chilled. Please protect from freezing.*
2. *10 – 20 ml urine in appropriate container. Urine must be refrigerated after collection.*
3. *Tissue samples must be placed inside sterile container. Please contact us for a detailed guideline on tissue sample collection, preservation and storage.*
4. *DNA, urine and tissue samples must always be kept chilled until the samples arrive at the laboratory.*

Clinical Signs and Symptoms, Age of Onset, Relevant Laboratory (eg.: biochemical testing result) and Imaging Findings :

Clinical Diagnosis : ……
Parental Consanguinity : ❒ Yes ❒ No

Pedigree (Family Tree)
(Can also be attached on a separate sheet) :

I certify that the patient specified above and/or their legal guardian has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained informed consent from the patient or his/her legal guardian for this testing.

Consultant's Name : Signature and/or Stamp : Date :

LIST OF DISORDERS/GENES TESTED IN UNIT OF MOLECULAR DIAGNOSTICS (UMD), IMR

- Please mark √ to select
- Please note that genetic testing **will only be accepted upon consultation with Clinical Geneticist/Neurologist** and/or if **biochemical testing result** or any relevant screening test result is **suggestive** of the respective disease

INHERITED METABOLIC DISORDERS (IEM)

(A) Disorders of Amino Acids & Organic Acids Metabolism

1	Argininosuccinate Lyase Deficiency *(ASL Sequence Analysis)*		10	Glutaric Aciduria Type 1 *(GCDH Sequence Analysis)*		19	Methylmalonyl-CoA Epimerase Deficiency *(MCEE Sequence Analysis)*	
2	Argininosuccinate Synthase Deficiency *(ASS1 Sequence Analysis)*		11	Hypophosphatasia *(ALPL Sequence Analysis)*		20	N-Acetylglutamate Synthase (NAGS) Deficiency *(NAGS Sequence Analysis)*	
3	Aromatic Amino Acid Decarboxylase Deficiency *(DDC Sequence Analysis)*		12	Lysinuric Protein Intolerance (LPI) *(SLC7A7 Sequence Analysis)*		21	Non Ketotic Hyperglycinemia (NKH) - Panel *(AMT / GLDC / GCSH Sequence Analysis / GLDC Deletion/Duplication Analysis)*	
4	Biotinidase Deficiency *(BTD Sequence Analysis)*		13	Maple Syrup Urine Disease (MSUD) *(DLD Sequence Analysis)*		22	Ornithine Transcarbamylase (OTC) Deficiency *(OTC Sequence Analysis)*	
5	Carbamoyl Phosphate Synthetase 1 (CPS1) Deficiency *(CPS1 Sequence Analysis)*		14	Maple Syrup Urine Disease (MSUD) - Panel *(BCKDHA / BCKDHB / DBT Sequence Analysis)*		23	Primary Hyperoxaluria Type 1 *(AGXT Sequence Analysis)*	
6	Citrin Deficiency (Type II Citrullinemia) *(SLC25A13 Sequence Analysis)*		15	Methylenetetrahydrofolate Reductase Deficiency *(MTHFR Sequence Analysis)*		24	Pyruvate Dehydrogenase Deficiency *(PDHA1 Sequence Analysis)*	
7	Classical Homocystinuria *(CBS Sequence Analysis)*		16	Methylmalonic Acidemia (MMA) - Panel *(MMUT / MMAA / MMAB Sequence Analysis)*		25	Tyrosine Hydroxylase Deficiency *(TH Sequence Analysis)*	
8	Cystinuria *(SLC3A1 Sequence Analysis)*		17	Methylmalonic Aciduria and Homocystinuria Type C *(MMACHC Sequence Analysis)*				
9	Ethylmalonic Encephalopathy *(ETHE1 Sequence Analysis)*		18	Methylmalonic Aciduria and Homocystinuria Type D *(MMADHC Sequence Analysis)*				

(B) Fatty Acids Oxidation Defects

26	Carnitine Palmitoyltransferase 1 (CPT1) Deficiency (*CPT1A Sequence Analysis*)	
27	Carnitine Palmitoyltransferase 2 (CPT2) Deficiency (*CPT2 Sequence Analysis*)	
28	Carnitine Uptake Deficiency *(OCTN2 Sequence Analysis)*	
29	Carnitine-Acylcarnitine Translocase Deficiency *(SLC25A20 Sequence Analysis)*	
30	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency (*HADHA Sequence Analysis*)	
31	Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency (*ACADM Sequence Analysis*)	
32	Mitochondrial Trifunctional Protein Deficiency *(HADHB Sequence Analysis)*	
33	Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency *(ACADS Sequence Analysis)*	
34	Short-Chain 3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency *(HADH Sequence Analysis)*	
35	Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency *(ACADVL Sequence Analysis)*	

(C) Disorders of Carbohydrate Metabolism

36	Classical Galactosemia *(GALT Sequence Analysis)*	
37	Fructose-1,6-Bisphosphatase Deficiency *(FBP1 Sequence Analysis)*	
38	Galactokinase Deficiency *(GALK1 Sequence Analysis)*	
39	Galactose Epimerase Deficiency *(GALE Sequence Analysis)*	
40	Glycogen Storage Disease Type Ia *(G6PC Sequence Analysis)*	
41	Glycogen Storage Disease Type Ib *(SLC37A4 Sequence Analysis)*	
42	Glycogen Storage Disease Type III *(AGL Sequence Analysis)*	
43	Phosphomannomutase 2 Deficiency (PMM2-CDG) *(PMM2 Sequence Analysis)*	

(D) Lysosomal Storage Diseases

44	Gaucher Disease *(GBA Sequence Analysis)*	
45	Pompe Disease (GSD II) *(GAA Sequence Analysis)*	
46	Maroteaux-Lamy Syndrome (MPS VI) *(ARSB Sequence Analysis)*	
47	Morquio A Disease (MPS IVA) *(GALNS Sequence Analysis)*	
48	Metachromatic Leukodystrophy (MLD) *(ARSA Sequence Analysis)*	
49	Fucosidosis *(FUCA1 Sequence Analysis)*	

(E) Disorders of Purine & Pyrimidine Metabolism

50	Dihydropyrimidinase (DHP) Deficiency *(DPYS Sequence Analysis)*	
51	Hereditary Orotic Aciduria *(UMPS Sequence Analysis)*	
52	Purine Nucleoside Phosphorylase Deficiency *(PNP Sequence Analysis)*	
53	Lesch-Nyhan Syndrome *(HPRT1 Sequence Analysis)*	

(F) Other Metabolic Disorders

54	Alpha 1-Antitrypsin Deficiency *(SERPINA1 Sequence Analysis)*	
55	Acute Intermittent Porphyria - Panel *(HMBS Sequence Analysis / Deletion/Duplication Analysis)*	
56	Canavan Disease *(ASPA Sequence Analysis)*	
57	Sulfite Oxidase (SUOX) Deficiency *(SUOX Sequence Analysis)*	
58	X-linked Adrenoleukodystrophy *(ABCD1 Sequence Analysis)*	

- Please mark √ to select
- Please note that genetic testing **will only be accepted upon consultation with Clinical Geneticist/Neurologist** and/or if **biochemical testing result** or any relevant screening test result is **suggestive** of the respective disease

MITOCHONDRIAL DISORDERS

59	Leber Hereditary Optic Neuropathy (LHON) - Panel *(m.3460G>A, m.11778G>A, m.14459G>A and m.14484T>C Sequence Analysis)*		66	Mitochondrial HMG-CoA Synthase Deficiency *(HMGCS2 Sequence Analysis)*		73	Multiple Respiratory Chain Deficiencies (Mitochondrial Translation Defect/ OXPHOS Deficiency) *(GFM1 Sequence Analysis)*	
60	Leigh Syndrome - 8993 Hotspot *(MT-ATP6 Sequence Analysis)*		67	Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE) *(TYMP Sequence Analysis)*		74	Myoclonic Epilepsy with Ragged-Red Fibers (MERRF) Syndrome – 8344 Hotspot *(m.8344A>G Sequence Analysis)*	
61	Leigh Syndrome – Full Panel *(MT-ATP6, MT-TL1, MT-TK, MT-TW, MT-TV, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-CO3 Sequence Analysis)*		68	Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency *(ECHS1 Sequence Analysis)*		75	Neuropathy, Ataxia and Retinitis Pigmentosa (NARP) Syndrome - 8993 Hotspot *(m.8993T>G/C Sequence Analysis)*	
62	Leigh Syndrome *(SURF1 Sequence Analysis)*		69	mtDNA Deletion Syndromes - Chronic Progressive External Ophthalmoplegia (CPEO) *(mtDNA Deletion/Duplication Analysis)*		76	POLG-Related Disorders - Panel *(POLG Sequence Analysis / Deletion/Duplication Analysis)*	
63	Mitochondrial Deletion *(mtDNA Deletion/Duplication Analysis)*		70	mtDNA Deletion Syndromes - Kearns-Sayre Syndrome (KSS) *(mtDNA Deletion/Duplication Analysis)*		77	Whole Mitochondrial DNA - mtDNA hotspots *(mtDNA Sequence Analysis)*	
64	Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome – 3243 Hotspot *(m.3243A>G Sequence Analysis)*		71	mtDNA Deletion Syndromes - Pearson Syndrome *(mtDNA Deletion/Duplication Analysis)*				
65	Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome – Full Panel *(m.3243A>G, m.3252A>G, m.3256C>T, m.3271T>C, m.3291T>C, m.3697G>A, m.4332G>A, m.12147G>A, and m.13514A>G Sequence Analysis)*		72	mtDNA Depletion Syndrome (MDS) - Panel *(ANT1 / DGUOK / POLG / RRM2B / SUCLA2 / SUCLG1 / TK2 / TWINKLE / TYMP / MPV17 Sequence Analysis)*				

NEUROGENETIC DISORDERS

78	Alexander Disease *(GFAP Sequence Analysis)*		83	Primary Dystonia - Panel *(TOR1A / THAP1 Sequence Analysis)*		88	Spinocerebellar Ataxia Type 2 (*CAG Repeat Analysis - SCA2)*	
79	Friedreich Ataxia (FRDA) (*GAA Repeat Analysis – FXN)*		84	SCN1A-Related Seizure Disorders *(SCN1A Sequence Analysis)*		89	Spinocerebellar Ataxia Type 3 (*CAG Repeat Analysis - SCA3)*	
80	Kennedy Disease (*CAG Repeat Analysis – AR)*		85	Spinal Muscular Atrophy (SMA) - Panel *(SMN1 Gene Dosage Analysis / Sequence Analysis)*		90	Spinocerebellar Ataxia Type 6 (*CAG Repeat Analysis - SCA6)*	
81	Lissencephaly - Panel *(LIS1 / DCX Sequence Analysis)*		86	Spinocerebellar Ataxia (SCA) – Full Panel (*CAG Repeat Analysis - SCA1, SCA2, SCA3, SCA6, SCA7)*		91	Spinocerebellar Ataxia Type 7 (*CAG Repeat Analysis - SCA7)*	
82	MCT8-Specific Thyroid Hormone Cell Transporter Deficiency *(SLC16A2 Sequence Analysis)*		87	Spinocerebellar Ataxia Type 1 (*CAG Repeat Analysis - SCA1)*				

GENETIC SYNDROMES

92	Alagille Syndrome - Panel *(JAG1 Sequence Analysis / Deletion/Duplication Analysis)*		96	Floating-Harbor Syndrome (FHS) *(SRCAP Sequence Analysis - Hotspots)*		100	Schinzel Giedion Syndrome *(SETBP1 Sequence Analysis)*	
93	Angelman Syndrome - Panel *(SNRPN Methylation & Gene Dosage Analysis / Uniparental Disomy & Imprinting Defect Analysis)*		97	Fragile X Syndrome (FRAXA) *(CGG Repeat Analysis – FMR1)*		101	Short Syndrome *(PIK3R1 Sequence Analysis)*	
94	Angelman Syndrome *(UBE3A Sequence Analysis)*		98	Leopard Syndrome *(PTPN11 Sequence Analysis)*		102	Prader-Willi Syndrome - Panel *(SNRPN Methylation & Gene Dosage Analysis / Uniparental Disomy & Imprinting Defect Analysis)*	
95	Barth Syndrome *(TAZ Sequence Analysis)*		99	Noonan Syndrome *(PTPN11 Sequence Analysis)*				

OTHER GENETIC DISORDERS

103	Berardinelli-Seip Congenital Lipodystrophy - Panel *(BSCL2 / AGPAT2 Sequence Analysis)*		107	FGFR3-Related Disorders *(FGFR3 Sequence Analysis)*		111	PTEN-Related Disorders - Panel *(PTEN Sequence Analysis / Deletion/Duplication Analysis)*	
104	Cartilage Hair Hypoplasia (CHH) *(RMRP Sequence Analysis)*		108	Mucopolysaccharidosis Type III B (MPS III B) *(NAGLU Sequence Analysis)*		112	Retinoblastoma - Panel *(RB1 Sequence Analysis / Deletion/Duplication Analysis)*	
105	Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) *(NOTCH3 Sequence Analysis - Hotspots)*		109	Myotonic Dystrophy Type 1 (DM1) *(CTG Repeat Analysis – DMPK)*		113	Severe Congenital Neutropenia *(ELANE Sequence Analysis)*	
106	FGFR2-Related Disorders *(FGFR2 Sequence Analysis)*		110	Pseudorheumatoid Dysplasia *(WISP3 Sequence Analysis)*		114	X-Chromosome Inactivation *(AR Fragment Analysis)*	

OTHER SERVICES

115	DNA Extraction & Storage		117	Specific Mutation Screening ……				
116	Testing of Familial Mutations/Carrier Testing ……		118	Others (*Please discuss with the Head of Unit first*) ……				

Unit of Molecular Diagnostics
Specialised Diagnostics Centre
Institute for Medical Research
National Institute of Health, MOH
Jalan Pahang, 50588 Kuala Lumpur
Tel: 03-26162783/ 2581

CONSENT FOR MOLECULAR DIAGNOSTICS SERVICES

Patient Name: _____ Patient ID:_____

The samples that I provide together with the request form are to be used for molecular genetic testing of:

_____

(Specify the disorder or disease to be tested)

- The molecular genetic testing may provide a diagnosis of or indication of risk for me or my offspring for the disorder or disease specified above.
- I understand the molecular genetic testing may not yield results for any combination of the following reasons: 1) unavailable blood or tissue samples from critical family members; 2) uninformativeness of the available genetic markers; 3) maternal contamination of prenatal samples; 4) technical reasons.
- I understand that DNA analysis may yield information on biological paternity, the results of which will not be disclosed to me unless biological paternity is relevant in counseling for the reason for which I have submitted this DNA sample. I agree to provide a family history to the best of my knowledge.
- I **AGREE/DO NOT AGREE** to have my samples or DNA extracted from my samples be used for the purpose of research and development or as quality control in diagnostics laboratory.
- Additional samples may need to be collected from me in the absence of results, or if the results are inconclusive.
- The DNA extracted from my (my child's) samples will be stored in the DNA bank at the Institute for Medical Research or its responsible delegate.
- I understand that any information identifying me (my child) will be kept confidential and that any exchange of samples or information will be coded.
- No compensation will be given to me (my child) nor will funds be forthcoming to me (my child) due to invention resulting from research and development using my (my child's) DNA.

Your signature on this form indicates that you have understood to your satisfaction the information regarding molecular genetic testing and agree to participate. In no way does this waive your legal rights nor release the investigators, sponsors, or involved institutions from their legal and professional responsibilities. If you have further questions concerning matters related to this consent, please discuss them with your medical geneticist, genetic counselor, or referring physician.

_____

(Signature of patient or legal guardian and date)

_____

(Signature of witness and date)