REQUEST FORM FOR MOLECULAR DIAGNOSTICS SERVICES

Molecular Diagnostics Unit
Specialised Diagnostics Centre
Institute for Medical Research
National Institutes of Health, MOH
Jalan Pahang, 50588 Kuala Lumpur
Tel: 03-26162783/ 2581/ 2707

To The Requesting Lab / Person,
Please STAMP HERE

Patient Name :

Patient IC/ID :

Date of Birth : **Age :**

Hospital :

Ward/Clinic :

Gender : Male / Female / Unknown

Name of Attending Doctor (Specialist) :

Ethnicity: **Nationality :**

Referral Geneticist/Neurologist :

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) : ……
Presymptomatic/ Predictive testing : ❒ At-risk relatives

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

1. **All cases requiring molecular diagnostics testing MUST HAVE:**
   a) **Endorsement by any Clinical Geneticist/Neurologist before any sample submission;**
   b) **Suggestive biochemical test result(s) or any relevant screening test result(s).**
   If these requirements are not fulfilled, request 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. Kindly ensure samples are sent together with both the request form and informed consent form. **Please send one (1) request form per test/gene.**
4. **For details on sample criteria, please refer to SMIS.** Please send the samples according to the criteria for specimen requirements as outlined below:

Type	Specimen requirements	Recommended storage condition during delivery
Blood	One EDTA (lavender top) tube containing 2-5 ml whole blood. 1-2 ml is acceptable for infants. **DO NOT** use Heparin tube.	Ambient for overnight delivery or can be refrigerated up to 7 days before shipping
Urine	Volume of ≥20 ml of an early morning sample.	Frozen
Tissue	Minimum specimen size is a cube 0.5 cm on each side or ~50 mg, frozen within minutes after collection.	Frozen
DNA	Concentration of ≥50 ng/ul AND a volume of ≥100 ul.	Chilled/frozen
Others	Please consult the laboratory	

Clinical Signs and Symptoms, Age of Onset, Relevant Laboratory (e.g. 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 MOLECULAR DIAGNOSTICS UNIT (UMD), IMR

- Please mark √ to select.
- Please note that genetic testing **will only be accepted upon consultation with Clinical Geneticist/Neurologist AND** if **biochemical test result(s)** or any relevant **screening test result(s)** are **suggestive** of the respective disease.
- When multiple tests are requested, turnaround time (TAT) for the 2nd test will begin after completion of the 1st report. The same rule applies to the subsequent test request.

INHERITED METABOLIC DISORDERS (IEM)

(A) Disorders of Amino Acids & Organic Acids Metabolism

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

(B) Fatty Acids Oxidation Defects

23	Carnitine Palmitoyltransferase 1 (CPT1) Deficiency (*CPT1A Sequence Analysis*)		29	Classical Galactosemia *(GALT Sequence Analysis)*		37	Gaucher Disease *(GBA1 Sequence Analysis)*	
24	Carnitine Palmitoyltransferase 2 (CPT2) Deficiency (*CPT2 Sequence Analysis*)		30	Galactokinase Deficiency *(GALK1 Sequence Analysis)*		38	Pompe Disease (GSD II) *(GAA Sequence Analysis)*	
25	Carnitine-Acylcarnitine Translocase Deficiency *(SLC25A20 Sequence Analysis)*		31	Galactose Epimerase Deficiency *(GALE Sequence Analysis)*		39	Maroteaux-Lamy Syndrome (MPS VI) *(ARSB Sequence Analysis)*	
26	Carnitine Uptake Deficiency *(OCTN2 Sequence Analysis)*		32	Fructose-1,6-Bisphosphatase Deficiency *(FBP1 Sequence Analysis)*		40	Morquio A Disease (MPS IVA) *(GALNS Sequence Analysis)*	
27	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency (*HADHA Sequence Analysis*)		33	Glycogen Storage Disease Type Ia *(G6PC Sequence Analysis)*		41	Metachromatic Leukodystrophy (MLD) *(ARSA Sequence Analysis)*	
28	Mitochondrial Trifunctional Protein Deficiency *(HADHB Sequence Analysis)*		34	Glycogen Storage Disease Type Ib *(SLC37A4 Sequence Analysis)*		42	Fucosidosis *(FUCA1 Sequence Analysis)*	

(C) Disorders of Carbohydrate Metabolism

			35	Glycogen Storage Disease Type III *(AGL Sequence Analysis)*				
			36	Phosphomannomutase 2 Deficiency (PMM2-CDG) *(PMM2 Sequence Analysis)*				

(D) Lysosomal Storage Diseases

(E) Disorders of Purine & Pyrimidine Metabolism

43	Dihydropyrimidinase (DHP) Deficiency *(DPYS Sequence Analysis)*		47	Canavan Disease *(ASPA Sequence Analysis)*				
44	Hereditary Orotic Aciduria *(UMPS Sequence Analysis)*		48	Multiple Acyl-CoA Dehydrogenase Deficiency (*ETFDH Sequence Analysis*)				
45	Purine Nucleoside Phosphorylase Deficiency *(PNP Sequence Analysis)*		49	NAXE-Related Progressive Encephalopathy *(NAXE Sequence Analysis)*				
46	Lesch-Nyhan Syndrome *(HPRT1 Sequence Analysis)*		50	Sulfite Oxidase (SUOX) Deficiency *(SUOX Sequence Analysis)*				
			51	X-linked Adrenoleukodystrophy *(ABCD1 Sequence Analysis)*				

(F) Other Metabolic Disorders

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MITOCHONDRIAL DISORDERS

52	Leber Hereditary Optic Neuropathy (LHON) - Panel *(m.3460G>A, m.11778G>A, m.14459G>A and m.14484T>C Sequence Analysis)*		59	Mitochondrial HMG-CoA Synthase Deficiency *(HMGCS2 Sequence Analysis)*		66	mtDNA Depletion Syndrome (MDS) - Panel *(ANT1 / DGUOK / POLG / RRM2B / SUCLA2 / SUCLG1 / TK2 / TWNK / TYMP / MPV17 Sequence Analysis)*	
53	Leigh Syndrome - 8993 Hotspot *(MT-ATP6 Sequence Analysis)*		60	Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE) *(TYMP Sequence Analysis)*		67	Multiple Respiratory Chain Deficiencies (Mitochondrial Translation Defect/ OXPHOS Deficiency) *(GFM1 Sequence Analysis)*	
54	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)*		61	Mitochondrial Nonsyndromic Hearing Loss and Deafness (mtDNA Gene Panel)		68	Myoclonic Epilepsy with Ragged-Red Fibers (MERRF) Syndrome – 8344 Hotspot *(m.8344A>G Sequence Analysis)*	
55	Leigh Syndrome *(SURF1 Sequence Analysis)*		62	Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency *(ECHS1 Sequence Analysis)*		69	Neuropathy, Ataxia and Retinitis Pigmentosa (NARP) Syndrome - 8993 Hotspot *(m.8993T>G/C Sequence Analysis)*	
56	Mitochondrial Deletion *(mtDNA Deletion/Duplication Analysis)*		63	mtDNA Deletion Syndromes - Chronic Progressive External Ophthalmoplegia (CPEO) *(mtDNA Deletion/Duplication Analysis)*		70	POLG-Related Disorders - Panel *(POLG Sequence Analysis / Deletion/Duplication Analysis)*	
57	Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome – 3243 Hotspot *(m.3243A>G Sequence Analysis)*		64	mtDNA Deletion Syndromes - Kearns-Sayre Syndrome (KSS) *(mtDNA Deletion/Duplication Analysis)*		71	Whole Mitochondrial DNA - mtDNA hotspots *(mtDNA Sequence Analysis)*	
58	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)*		65	mtDNA Deletion Syndromes - Pearson Syndrome *(mtDNA Deletion/Duplication Analysis)*				

NEUROGENETIC DISORDERS

72	Alexander Disease *(GFAP Sequence Analysis)*		77	MCT8-Specific Thyroid Hormone Cell Transporter Deficiency *(SLC16A2 Sequence Analysis)*		82	Spinocerebellar Ataxia Type 1 (SCA1) *(CAG Repeat Analysis - ATXN1)*	
73	Dentatorubral-Pallidoluysian Atrophy (DRPLA) *(CAG Repeat Analysis - ATN1)*		78	Primary Dystonia - Panel *(TOR1A / THAP1 Sequence Analysis)*		83	Spinocerebellar Ataxia Type 2 (SCA2) *(CAG Repeat Analysis - ATXN2)*	
74	Friedreich Ataxia (FRDA) *(GAA Repeat Analysis - FXN)*		79	SCN1A-Related Seizure Disorders *(SCN1A Sequence Analysis)*		84	Spinocerebellar Ataxia Type 3 (SCA3) *(CAG Repeat Analysis - ATXN3)*	
75	Kennedy Disease *(CAG Repeat Analysis - AR)*		80	Spinal Muscular Atrophy (SMA) - Panel *i) SMN1/SMN2 Gene Dosage Analysis* *ii) SMN Gene Sequence Analysis* ***(can only be requested if result of test (i) is suggestive)***		85	Spinocerebellar Ataxia Type 6 (SCA6) *(CAG Repeat Analysis - CACNA1A)*	
76	Lissencephaly - Panel *(LIS1 / DCX Sequence Analysis)*		81	Spinocerebellar Ataxia (SCA) – Full Panel *(CAG Repeat Analysis - SCA1, 2, 3, 6 & 7)*		86	Spinocerebellar Ataxia Type 7 (SCA7) *(CAG Repeat Analysis - ATXN7)*	

GENETIC SYNDROMES

87	Alagille Syndrome - Panel *(JAG1 Sequence Analysis / Deletion/Duplication Analysis)*		90	Floating-Harbor Syndrome (FHS) *(SRCAP Sequence Analysis - Hotspots)*		94	Schinzel Giedion Syndrome *(SETBP1 Sequence Analysis)*	
88	Angelman Syndrome - Panel *i) SNRPN Methylation & Gene Dosage Analysis* *ii) Uniparental Disomy & Imprinting Defect Analysis)* ***(can only be requested if result of test (i) is suggestive)*** iii) *UBE3A Sequence Analysis* ***(can only be requested if result of test (i) is suggestive)***		91	*FMR1* Disorders (Fragile X, FXTAS, FXPOI, FXAND) *(CGG Repeat Analysis – FMR1)*		95	Short Syndrome *(PIK3R1 Sequence Analysis)*	
			92	Leopard Syndrome *(PTPN11 Sequence Analysis)*		96	Prader-Willi Syndrome - Panel *i) SNRPN Methylation & Gene Dosage Analysis* *ii) Uniparental Disomy & Imprinting Defect Analysis* ***(can only be requested if result of test (i) is suggestive)***	
89	Barth Syndrome *(TAFAZZIN Sequence Analysis)*		93	Noonan Syndrome *(PTPN11 Sequence Analysis)*				

OTHER GENETIC DISORDERS

97	Berardinelli-Seip Congenital Lipodystrophy - Panel *(BSCL2 / AGPAT2 Sequence Analysis)*		101	FGFR3-Related Disorders *(FGFR3 Restriction Enzyme Analysis / FGFR3 Sequence Analysis)*		105	PTEN-Related Disorders - Panel *(PTEN Sequence Analysis / Deletion/Duplication Analysis)*	
98	Cartilage Hair Hypoplasia (CHH) *(RMRP Sequence Analysis)*		102	Mucopolysaccharidosis Type III B (MPS III B) *(NAGLU Sequence Analysis)*		106	Retinoblastoma - Panel *(RB1 Sequence Analysis / Deletion/Duplication Analysis)*	
99	Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) *(NOTCH3 Sequence Analysis - Hotspots)*		103	Myotonic Dystrophy Type 1 (DM1) *(CTG Repeat Analysis – DMPK)*		107	Severe Congenital Neutropenia *(ELANE Sequence Analysis)*	
100	FGFR2-Related Disorders *(FGFR2 Sequence Analysis)*		104	Pseudorheumatoid Dysplasia *(WISP3 Sequence Analysis)*		108	X-Chromosome Inactivation *(AR Fragment Analysis)*	

OTHER SERVICES

109	DNA Extraction & Storage		111	Specific Mutation Screening ……				
110	Testing of Familial Mutations / Carrier Testing ……		112	Others (*Please discuss with the Head of Unit first – by appointment only*) ……				