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 :
Name of Attending Doctor (Specialist) :

Gender : Male / Female / Unknown

Ethnicity: **Nationality :**

Referral Geneticist/Neurologist :
Date: **Time:**

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 MEET the following requirements, otherwise it will be REJECTED:**
   - **a) Prior endorsement by any Clinical Geneticist is mandatory** before any sample submission;
   - b) Requests for testing related to **Inherited Metabolic Disorders and Mitochondrial Disorders** may **only be initiated by a Clinical Geneticist**.
   - c) For all other conditions, molecular diagnostic testing may be requested by a **Clinical Geneticist, Neurologist or Paediatrician.** Requests from other specialties for these conditions must be referred to a Clinical Geneticist for evaluation and approval.
   - **d) Relevant supporting results** must be provided, including suggestive biochemical test result(s) and/or other appropriate screening test result(s).
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 x 2 container 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:

Note: Please refer to Human Phenotype Ontology (HPO) website for standardized vocabulary of phenotypic abnormalities associated with human disease.

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.
- 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) – REQUEST BY CLINICAL GENETICIST ONLY

(A) Disorders of Amino Acids & Organic Acids Metabolism

1	Argininosuccinate Lyase Deficiency *(ASL Sequence Analysis)*		7	Glutaric Aciduria Type 1 *(GCDH Sequence Analysis)*		13	Non Ketotic Hyperglycinemia (NKH) - Panel *(AMT / GLDC / GCSH Sequence Analysis / GLDC Deletion/Duplication Analysis)*	
2	Argininosuccinate Synthase Deficiency *(ASS1 Sequence Analysis)*		8	Hypophosphatasia *(ALPL Sequence Analysis)*		14	N-Acetylglutamate Synthase (NAGS) Deficiency *(NAGS Sequence Analysis)*	
3	Biotinidase Deficiency *(BTD Sequence Analysis)*		9	Lysinuric Protein Intolerance (LPI) *(SLC7A7 Sequence Analysis)*		15	Ornithine Transcarbamylase (OTC) Deficiency *(OTC Sequence Analysis)*	
4	Carbamoyl Phosphate Synthetase 1 (CPS1) Deficiency (*CPS1 Sequence Analysis)*		10	Maple Syrup Urine Disease (MSUD) *(DLD Sequence Analysis)*		16	Methylmalonic Acidemia (MMA) - Panel *(MMUT / MMAA / MMAB Sequence Analysis)*	
5	Citrin Deficiency (Type II Citrullinemia) *(SLC25A13 Sequence Analysis)*		11	Maple Syrup Urine Disease (MSUD) - Panel *(BCKDHA / BCKDHB / DBT Sequence Analysis)*		17	Methylmalonic Aciduria and Homocystinuria Type C *(MMACHC Sequence Analysis)*	
6	Classical Homocystinuria *(CBS Sequence Analysis)*		12	Pyruvate Dehydrogenase Deficiency *(PDHA1 Sequence Analysis)*				

(B) Fatty Acids Oxidation Defects

18	Carnitine Palmitoyltransferase 1 (CPT1) Deficiency (*CPT1A Sequence Analysis*)		24	Classical Galactosemia *(GALT Sequence Analysis)*		27	Lesch-Nyhan Syndrome *(HPRT1 Sequence Analysis)*	
19	Carnitine Palmitoyltransferase 2 (CPT2) Deficiency (*CPT2 Sequence Analysis*)		25	Fructose-1,6-Bisphosphatase Deficiency *(FBP1 Sequence Analysis)*				
20	Carnitine-Acylcarnitine Translocase Deficiency *(SLC25A20 Sequence Analysis)*		26	Glycogen Storage Disease Type Ia *(G6PC Sequence Analysis)*				
21	Carnitine Uptake Deficiency *(OCTN2 Sequence Analysis)*							
22	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency (*HADHA Sequence Analysis*)							
23	Mitochondrial Trifunctional Protein Deficiency *(HADHB Sequence Analysis)*							

(C) Disorders of Carbohydrate Metabolism

(D) Disorders of Purine & Pyrimidine Metabolism

(E) Other Metabolic Disorders

						28	Multiple Acyl-CoA Dehydrogenase Deficiency (ETFDH Sequence Analysis)	
						29	NAXE-Related Progressive Encephalopathy (NAXE Sequence Analysis)	
						30	X-linked Adrenoleukodystrophy *(ABCD1 Sequence Analysis)*	

MITOCHONDRIAL DISORDERS – REQUEST BY CLINICAL GENETICIST ONLY

31	Leber Hereditary Optic Neuropathy (LHON) - Panel *(m.3460G>A, m.11778G>A, m.14459G>A and m.14484T>C Sequence Analysis)*		36	Mitochondrial HMG-CoA Synthase Deficiency *(HMGCS2 Sequence Analysis)*		41	mtDNA Deletion Syndromes - Chronic Progressive External Ophthalmoplegia (CPEO) *(mtDNA Deletion/Duplication Analysis)*	
32	Leigh Syndrome - 8993 Hotspot *(MT-ATP6 Sequence Analysis)*		37	Myoclonic Epilepsy with Ragged-Red Fibers (MERRF) Syndrome – 8344 Hotspot *(m.8344A>G Sequence Analysis)*		42	mtDNA Deletion Syndromes - Kearns-Sayre Syndrome (KSS) *(mtDNA Deletion/Duplication Analysis)*	
33	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)*		38	Neuropathy, Ataxia and Retinitis Pigmentosa (NARP) Syndrome - 8993 Hotspot *(m.8993T>G/C Sequence Analysis)*		43	mtDNA Deletion Syndromes - Pearson Syndrome *(mtDNA Deletion/Duplication Analysis)*	
34	Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome – 3243 Hotspot *(m.3243A>G Sequence Analysis)*		39	Mitochondrial Nonsyndromic Hearing Loss and Deafness (mtDNA Gene Panel)		44	Mitochondrial Deletion *(mtDNA Deletion/Duplication Analysis)*	
35	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)*		40	POLG-Related Disorders - Panel *(POLG Sequence Analysis / Deletion/Duplication Analysis)*				

- Please mark √ to select
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NEUROGENETIC DISORDERS – REQUEST BY CLINICAL GENETICIST/NEUROLOGIST/PAEDIATRICIAN

45	Dentatorubral-PallidoluysianAtrophy (DRPLA) (CAG Repeat Analysis -ATN1)		47	Kennedy Disease (*CAG Repeat Analysis – AR)*		49	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)**	
46	Friedreich Ataxia (FRDA) (*GAA Repeat Analysis – FXN)*		48	MCT8-Specific Thyroid Hormone Cell Transporter Deficiency *(SLC16A2 Sequence Analysis)*		50	Spinocerebellar Ataxia (SCA) – Full Panel (*CAG Repeat Analysis - SCA1, SCA2, SCA3, SCA6, SCA7)*	

GENETIC SYNDROMES – REQUEST BY CLINICAL GENETICIST/NEUROLOGIST/PAEDIATRICIAN

21	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)**		52	FMR1Disorders (Fragile X, FXTAS, FXPOI, FXAND) (CGG Repeat Analysis –FMR1)		53	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)**	

OTHER GENETIC DISORDERS – REQUEST BY CLINICAL GENETICIST/NEUROLOGIST/PAEDIATRICIAN

54	FGFR2-Related Disorders *(FGFR2 Sequence Analysis)*		56	Myotonic Dystrophy Type 1 (DM1) *(CTG Repeat Analysis – DMPK)*		58	X-Chromosome Inactivation *(AR Fragment Analysis)*	
55	FGFR3-Related Disorders (*FGFR3* Restriction Enzyme Analysis / *FGFR3* Sequence Analysis)		57	Retinoblastoma - Panel *(RB1 Sequence Analysis / Deletion/Duplication Analysis)*				

OTHER SERVICES – REQUEST BY CLINICAL GENETICIST/NEUROLOGIST/PAEDIATRICIAN

59	DNA Extraction & Storage		62	Mutation analysis (up to 10 exons) (by consultation)		65	Mutation analysis (more than 40 exons) (by consultation)	
60	Testing of Familial Mutations/Carrier Testing ……		63	Mutation analysis (up to 20 exons) (by consultation)		66	Others (Please discuss with the Head of Unit) ……	
61	Specific Mutation Screening ……		64	Mutation analysis (up to 30 exons) (by consultation)				

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CONSENT FOR MOLECULAR DIAGNOSTICS SERVICES

Patient Name: _____ Patient ID:_____

Gene/ Disorder/ Disease testing: _____

Genetic Test Purpose and Specimen:
The purpose of this molecular genetic test is to determine if you or your child carry any mutation(s) predisposing to or causing a specific genetic condition. This test will entail analysis of all genes included on the test or panel that was ordered by your healthcare provider

By signing this form, I understand that:

1) Molecular genetic testing may provide a diagnosis of or indication of risk for me or my child for a disorder or disease.

2) Molecular genetic testing may not yield results for any combination of the following reasons: (i) Unavailability of samples from critical family members; (ii) Absence of appropriate genetic markers; (iii) Maternal contamination of the prenatal samples; (iv) Technical reasons.

3) 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 sample. I agree to provide a family history to the best of my knowledge.

4) My (my child's) samples or DNA extracted from my (my child's) samples may be used: (i) as quality control; (ii) for research and development in our laboratory.

5) Additional samples may need to be collected from me (my child) in the absence of results, or if the results are inconclusive.

6) DNA extracted from my (my child's) samples will be stored in the DNA bank in Institute for Medical Research.

7) Any information identifying me (my child) will be kept confidential and that any exchange of samples or information will be coded.

8) 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 counsellor or referring physician.

_____	_____
Signature of patient or legal guardian	**Signature of witness**
Name:	**Name:**
Date:	**Date:**