REQUEST FORM FOR MOLECULAR DIAGNOSTICS SERVICES

Unit of Molecular Diagnostics and Protein (UMDP)
Specialized Diagnostics Centre
Institute for Medical Research, Kuala Lumpur
Tel: 03-2616 2540/2590 Fax: 03-26162533

To The Requesting Lab / Person, Please STAMP HERE

Patient Name :	
Patient IC/ID :	**Hospital :**
Date of Birth : **Age :**	**Ward/Clinic :**
Gender : Male / Female / Unknown	**Name of Attending Doctor (Specialist) :**
Ethnic Background :	**Tel :** **Fax :**
If this is a parental or family member sample : **Proband/Child Full Name :**_____ **IC/ID :**_____ **DOB :**	**Email :**

Reason for referral:

Diagnostic test : ❒ Affected patient ❒ Possibly affected patient

Carrier test : ❒ Father of affected patient ❒ Mother of affected patient

❒ Sibling of affected patient ❒ Other family member of affected patient, specify…….....

Predictive diagnosis ❒ **DNA storage** ❒

Type of Specimen Sent :

❒ Whole blood ❒ Blood Spot ❒ Tissue, 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 UMDP, IMR :

1. ***All cases requiring molecular diagnostics testing must be referred to any Clinical Geneticist/Neurologist/Physician/ Paediatrician and they must endorse the test before any sample submission be made. Samples received without referral by Clinical Geneticist/Neurologist/Physician/ Paediatrician will be rejected.***
2. *Please ensure that the patient or their parent/guardian both understand the implications of genetic testing and provide their 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 under AMBIENT condition as soon as possible after collection. If more than 3 hours, keep sample cooled. 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 be kept chilled at all times until the sample/s arrive at the laboratory.*

Clinical Signs and Symptoms, Age of Onset, Relevant Laboratory 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 their legal guardian for this testing.

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

LIST OF DISORDERS/GENES TESTED IN MOLECULAR LAB
UNIT OF MOLECULAR DIAGNOSTICS AND PROTEIN (UMDP), IMR

Please mark √ to order

INHERITED METABOLIC DISORDERS / IEM

(a) Disorders of Amino Acids & Organic Acids Metabolism

❒ 1. Non Ketotic Hyperglycinemia (NKH):
(AMT, GCSH, GLDC Sequencing, GLDC Del/Dup Analysis)
❒ 2. N-Acetylglutamate Synthase (NAGS) Deficiency: *(NAGS)*
❒ 3. Carbamoyl Phosphate Synthetase 1 (CPS1) Deficiency: *(CPS1)*
❒ 4. Ornithine Transcarbamylase (OTC) Deficiency: *(OTC)*
❒ 5. Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria): *(ASL)*
❒ 6. Lysinuric Protein Intolerance: *(SLC7A7)*
❒ 7. Classical Homocystinuria: *(CBS)*
❒ 8. Methylenetetrahydrofolate Reductase Deficiency: *(MTHFR)*
❒ 9. Glutaric Aciduria Type 1: *(GCDH)*
❒10. Methylmalonic Aciduria and Homocystinuria Type D: *(MMADHC)*
❒11. Methylmalonyl-CoA Epimerase Deficiency: *(MCEE)*
❒12. Citrin Deficiency (Type II Citrullinemia): *(SLC25A13)*
❒13. Biotinidase Deficiency: *(BTD)*
❒14. Tyrosine Hydroxylase Deficiency: *(TH)*
❒15. Aromatic Amino Acid Decarboxylase Deficiency: *(DDC)*
❒16. Ethylmalonic Encephalopathy (ETHE): *(ETHE1)*
❒17. Hypophosphatasia: *(ALPL)*
❒18. Primary Hyperoxaluria Type 1: *(AGXT)*
❒19. Pyruvate Dehydrogenase Deficiency: *(PDHA1)*

(b) Fatty Acids Oxidation Defects

❒20. Carnitine Palmitoyltransferase 1 (CPT1) Deficiency: *(CPT1A)*
❒21. Carnitine Palmitoyltransferase 2 (CPT2) Deficiency: *(CPT2)*
❒22. Carnitine-Acylcarnitine Translocase Deficiency: *(SLC25A20)*
❒23. Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency: *(HADHA)*
❒24. Short-Chain 3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency: *(HADH)*
❒25. Carnitine Uptake Deficiency: *(OCTN2)*
❒26. Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency: *(ACADVL)*
❒27. Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency: *(ACADM)*
❒28. Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency: *(ACADS)*
❒29. Mitochondrial Trifunctional Protein Deficiency: *(HADHB)*

(c) Disorders of Carbohydrate Metabolism

❒30. Classical Galactosemia: *(GALT)*
❒31. Galactokinase Deficiency: *(GALK1)*
❒32. Galactose Epimerase Deficiency: *(GALE)*
❒33. Fructose-1,6-Bisphosphatase Deficiency: *(FBP1)*
❒34. Glycogen Storage Disease Type 1a: *(G6PC)*
❒35. Glycogen Storage Disease Type 1b: *(SLC37A4)*
❒36. Glycogen Storage Disease Type III: *(AGL)*
❒37. Phosphomannomutase 2 Deficiency (PMM2-CDG): *(PMM2)*

(d) Lysosomal Storage Diseases

❒38. Gaucher Disease: *(GBA)*
❒39. Pompe Disease (GSD II): *(GAA)*
❒40. Maroteaux-Lamy Syndrome (MPS VI): *(ARSB)*
❒41. Morquio A Disease (MPS IVA): *(GALNS)*

(e) Disorders of Purine & Pyrimidine Metabolism

❒42. Hereditary Orotic Aciduria: *(UMPS)*
❒43. Purine Nucleoside Phosphorylase Deficiency: *(PNP)*
❒44. Lesch-Nyhan Syndrome: *(HPRT1)*

(f) Other Metabolic Disorders

❒45. X-linked Adrenoleukodystrophy: *(ABCD1)*
❒46. Canavan Disease: *(ASPA)*
❒47. Alpha 1-Antitrypsin Deficiency: *(SERPINA1)*
❒48. Acute Intermittent Porphyria: *(HMBS)*

MITOCHONDRIAL DISORDERS

❒49. Leigh Syndrome 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 and SURF1)
❒50. mtDNA Deletion Syndromes [Pearson Syndrome, Kearns-Sayre Syndrome (KSS), Chronic Progressive External Ophthalmoplegia (CPEO)]: *(mtDNA Del/Dup Analysis)*
❒51. Leber Hereditary Optic Neuropathy (LHON) Panel:
(m.3460G>A, m.11778G>A, m.14484T>C)
❒52. Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome:
(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)
❒53. Myoclonic Epilepsy with Ragged-Red Fibers (MERRF) Syndrome: *(m.8344A>G)*
❒54. Neuropathy, Ataxia and Retinitis Pigmentosa (NARP) Syndrome: *(m.8993T>G/C)*
❒55. POLG-Related Disorders: *(POLG)*
❒56. mtDNA Depletion Syndrome (MDS) Panel:
(POLG, DGUOK, MPV17, ANT1, TWINKLE, RRM2B, SUCLA2, SUCLG1, TYMP)
❒57. Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE): *(TYMP)*
❒58. Multiple Respiratory Chain Deficiencies (Mitochondrial Translation Defect): *(GFM1)*
❒59. Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency: *(ECHS1)*
❒60. Mitochondrial HMG-CoA Synthase Deficiency: *(HMGCS2)*

GENETIC SYNDROMES

❒61. Fragile X Syndrome (FRAXA): *(CGG Repeat Analysis – FMR1)*
❒62. Prader-Willi Syndrome: *(SNRPN Methylation Analysis)*
❒63. Angelman Syndrome:
(SNRPN Methylation Analysis, UBE3A Sequencing)
❒64. Alagille Syndrome: *(JAG1, JAG1 Del/Dup Analysis, NOTCH2 Sequencing)*
❒65. Noonan Syndrome: *(PTPN11)*
❒66. Schinzel Giedion Syndrome: *(SETBP1)*
❒67. Leopard Syndrome: *(PTPN11)*
❒68. Floating-Harbor Syndrome (FHS): *(SRCAP-Hotspots)*

NEUROGENETIC DISORDERS

❒69. SCN1A-Related Seizure Disorders: *(SCN1A)*
❒70. Spinal Muscular Atrophy (SMA):
(SMN1 Gene Dosage Analysis, SMN Sequencing)
❒71. Primary Dystonia: DYT1 *(TOR1A)*, DYT6 *(THAP1)*
❒72. MCT8-Specific Thyroid Hormone Cell Transporter Deficiency: *(SLC16A2)*
❒73. Lissencephaly: *(LIS1, DCX)*
❒74. Alexander Disease: *(GFAP)*
❒75. Spinocerebellar Ataxia (SCA):
(Trinucleotide Repeat Analysis - SCA1, SCA2, SCA3, SCA6, SCA7)
❒76. Kennedy Disease: *(CAG Repeat Analysis – AR)*

OTHER GENETIC DISORDERS

❒77. Pseudorheumatoid Dysplasia: *(WISP3)*
❒78. Berardinelli Congenital Lipodystrophy: *(BSCL2, AGPAT2)*
❒79. Retinoblastoma: *(RB1 Sequencing, RB1 Del/Dup Analysis)*
❒80. PTEN-Associated Disease: *(PTEN)*
❒81. X-Chromosome Inactivation: *(AR Fragment Analysis)*
❒82. Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL): *(NOTCH3 - Hotspots)*
❒83. Severe Congenital Neutropenia: *(ELANE)*
❒84. Others (Please discuss with Head of Laboratory first)

OTHER SERVICES

❒85. DNA Extraction & Storage

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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)