



REQUEST FORM FOR BIOCHEMICAL GENETIC TESTS

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IMR Lab. Number

PLEASE FILL UP THE ENTIRE FORM TO ENSURE CORRECT, RELIABLE RESULT AND INTERPRETATION

Name: _____ Age: _____ DOB: _____ Sex: M/ F/ U Race: M/ C/ I/ O/ Foreigner
RN: _____ ID: *(preferably patient's IC)* _____ Hospital: _____ Ward : _____
Address : _____ Tel : _____

1. Symptoms / Signs of Current Illness:

Fever		Poor sucking / feeding	
Pallor		Respiratory problem	
Jaundice		Difficulty in breathing	
Hypothermia		Mental retardation	
Hypotonia / floppy		Developmental delay	
Cyanosed		Failure to thrive	
Lethargy		Feeding intolerance	
Easily irritable		Septicaemic-like illness	
Seizures or h/o seizures		Headache	
Drowsy		Smelly urine	
Coma		Colored urine	
Abnormal behaviour		Skin lesions	
Frequent vomiting		Eye lesions	

Other symptoms/ signs:

2. Feeding History:

Breast milk/ Formula milk/ Mixed/ NBM/
Special Diet _____
Infusion: Saline/ Dextrose/ Mannitol/ TPN/ _____

3. Family History: (please specify)

Consanguinity: No/ Yes _____
Stillbirth/ Neonatal death: No/ Yes _____
Metabolic disease: No/ Yes _____

4. Physical Examination :

Respiratory distress		Hyperreflexia	
Dysmorphic features		Nystagmus	
Hypothermia		Optical atrophy	
Cardiomyopathy		Ptosis	
Drowsy		Abnormal odour	
Coma		Abnormal hair	
Opisthotonus		Hepatomegaly	
Dystonia		Splenomegaly	
Choreoathetoid movement		Eczema / Other rashes	
Hypotonia		Others (specify)	

5. Treatment Given : (specimen should be taken before any form of treatment given or stop for 2-3 days)

Antibiotic: No / Yes _____
Steroid: No / Yes _____
Anticonvulsant: No / Yes _____
Transfusion: No/ Yes _____
Other drug: (please state) _____

6. Lab Result : (before treatment is given)

ALT : _____ U/L **Blood Glucose** : _____ mmol/L **Urine pH** : _____
AST : _____ U/L **Blood Ammonia** : _____ umol/L **Urine Ketones** : Pos/ Neg
ALP : _____ U/L **Blood Lactate** : _____ mmol/L **Urine Reducing Sugar** : Pos/ Neg

Blood Gases : Normal / Met acidosis / Met alkalosis / Resp acidosis / Resp alkalosis **Anion Gap** : _____

CT Scan/ MRI : _____

Other relevant test (specify) : _____

WORKING AND PROVISIONAL DIAGNOSIS :

(For office uses only)

7. Test Required: (Please tick ONLY appropriate test(s) required)

DRIED BLOOD SPOT (DBS)	
1	Acid Alpha-Glucosidase (POMPE)
2	Biotinidase Enzyme Activity
3	Creatine & Guanidinoacetic Acid
4	Galactosemia Screening (TG & GALT)

PLASMA AND/ OR SERUM	
♦ SEPARATE plasma/ serum from RBC IMMEDIATELY	
♦ KEEP AND SEND FROZEN	
1	Carnitine Total & Free (2ml, heparin/ plain tube)
2	Creatine & Guanidinoacetic Acid (1ml, EDTA/ heparin/ plain tube)
3	Homocysteine Total (2ml, EDTA/ plain tube)
4	Organic Acids (FORENSIC ONLY) (1ml, EDTA/ heparin/ plain tube)
5	Peroxisomal Disorder Profile (VLCFA, Phytanic & Pristanic Acid) (1ml, EDTA/ heparin/ plain tube)

URINE	
♦ KEEP AND SEND FROZEN	
1	5-HIAA, 24 H Urine (10ml 25% HCl as preservative, 2ml of 24-Hr Urine) State Total Volume : _____ L , State pH : _____
2	Carnitine, 24 H Urine (2ml of 24-Hr Urine) State Total Volume : _____ L
3	Argininosuccinic Acid (ASA) (2ml)
4	Biogenic Amines (2ml, protect from light)
5	Creatine & Guanidinoacetic Acid (2ml)
6	Cystine & Homocystine (2ml)
7	Lysine Metabolism Profile (P6C, Pipecolic Acid, AASA) (2ml)
8	Mucopolysaccharides (GAGs / HRE) (5ml)
9	Oligosaccharide (2ml)
10	Organic Acids (2ml)
11	Orotic Acid (2ml)
12	Porphyria Profile (2ml, protect from light)
13	Pterins, Urine (2ml, protect from light)
14	Sialic Acid, Total & Free (2ml)
15	Succinylacetone (2ml)
16	Sugar & Polyols (2ml)

CEREBROSPINAL FLUID (CSF)	
♦ KEEP AND SEND FROZEN	
1	Biogenic Amines, CSF (2ml, protect from light)
2	Pterins, CSF (0.5ml, protect from light, special microtube with preservative EDTA and DTE provided by the Biochemistry Unit)

VITREOUS HUMOUR	
♦ KEEP AND SEND FROZEN	
1	Organic Acids (FORENSIC ONLY) (2ml)

WHOLE BLOOD IN EDTA TUBE	
♦ DO NOT SPIN, DO NOT FREEZE	
♦ KEEP AND SEND CHILLED	
♦ Sample shall arrived IMR within 72 HOURS after collection	
BY CONSULTATION ONLY (Kindly specify the name of the individual being consulted)	
SPOKEN TO:	
1	Lysosomal Storage Disease Enzyme Assays (6ml) (ONLY up to TWO (2) diseases of enzyme per request)
	General LSD Marker - Chitotriosidase (CHITO)
	α-Mannosidosis - α-mannosidase (AMAN)
	β-Mannosidosis - Total β-Mannosidase (BMAN)
	Aspartylglucosaminuria - Total Aspartylglucosaminidase (GASP)
	Ceroid Lipofuscinosis - Palmitoyl-protein thioesterase (PPT)
	Fabry Disease - α-galactosidase (AGAL)
	Fucosidosis - α-fucosidosis (AFUC)
	Gaucher Disease - β-glucosidase (BGLU)
	GM1-Gangliosidosis - β-galactosidase (BGAL)
	Metachromatic Leukodystrophy - Aryl sulphatase A (ASA)
	Mucopolipidosis - α-mannosidase (AMANP)
	Niemann Pick A/B - Acid Sphingomyelinase (ASM)
	Krabbe Disease - Galactocerebrosidase (GALC)
	Sandhoff Disease - Total Hexosaminidase (BHEX)
	Schindler Disease - α-N-acetyl galactosamidase (ANAG)
	Tay-Sachs Disease - β-hexosaminidase A (MUGS)
2	Mucopolysaccharidoses Enzyme Assays (6ml) (ONLY up to TWO (2) diseases of enzyme per request)
	MPS Type I - α-Iduronidase (IDA)
	MPS Type II - Iduronate-2-sulphatase (IDS)
	MPS Type IIIa - Sulphaminidase (SULP)
	MPS Type IIIb - α-N-acetyl glucosaminidase (AHEX)
	MPS Type IVa - Galactose-6-sulphatase (GALSO)
	MPS Type IVb - β-galactosidase (BGAL)
	MPS Type VI - Aryl sulphatase B (ASB)
	MPS Type VII - β-glucuronidase (BGLUCU)
	Multiple Sulphatase - Aryl Sulphatase A (ASA)

OTHER TESTS (Please specify)	

Collected by :	
Collected date :	
Specialist In-Charge (Sign & Stamp) :	

**For more details information of sample requirements, please refer to IMR Test List available at IMR Website (<http://imr.nih.gov.my/>) and SMIS Website (<http://smis.imr.gov.my/>)*