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# IEM REQUEST FORM

**IMPORTANT NOTICE: To ensure correct, reliable result and interpretation given, the following must be followed:**

1. **Please fill up the entire form.**
2. **At least 1ml plasma and 5ml urine are needed. Heparinised plasma is preferred.**
3. **Separate plasma from RBC immediately. Haemolysed samples will be rejected.**
4. **ALL processed samples (plasma and urine) must be frozen immediately and transport in DRY ICE to IMR.**

Name:______________________ Age:_______ Sex: M / F / U Race: M / C / I / O_________

RN:______________ M I/C:________________ Hospital:__________ Wad:____________

House 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 | ☐ | Coloured urine | ☐ |
| Abnormal behaviour | ☐ | Skin lesions | ☐ |
| Frequent vomiting | ☐ | Eye lesions | ☐ |

Other symptoms/signs:____________________________________________

2. Feeding history: Type of milk: Breast/ Formula/ Mixed/ Solid diet:____________________

3. Family history: Consanguinity: Yes / No. If Yes please specify:____________________

| Occurrence of / in | Stillbirth | neonatal death | neonatal seizures | metabolic disease |
|---|---|---|---|---|
| Siblings | | | | |
| Maternal side | | | | |
| Paternal side | | | | |

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 | ☐ |
| Hyptonia | ☐ | Others (specify) | |

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

Drug therapy: Antibiotic: No/Yes________________ Anticonvulsant: No/Yes______________

Steroid: No/Yes__________________ Other drug:_________________________

Fluid infusion: Saline / Dextrose / Mannitol / Parenteral feeding / Others:________________

**6. Lab Result (before treatment is given)**

**LFT**: ALT:_____________mmol/l **Blood Glucose**:___________mmol/l

AST:_____________mmol/l **Blood Ammonia**___________

ALP:_____________mmol/ l **Blood Lactate**:___________pyruvate:_________

**Blood Gases**: Normal / Met. Acidosis / Met.alkalosis / Resp.acidosis / Resp.alkalosis

**Anion Gap**:______________________Other relevant test ( specify)__________________

CTscan/MRI:_____________________________________________________________

**Urine Analysis**: pH__________Ketones: Pos/ Neg Reducing Sugar: Pos/Neg

**Provisional Diagnosis**:_______________________________________________________

**7. Test required: (Please tick appropriate test/s required)**

| Test | ☐ | |
|---|---|---|
| 1. Plasma amino acid | ☐ | Specimen No: ______ |
| 2. CSF amino acid | ☐ | |
| 3. Urine Amino acid (HPLC-by appt) | ☐ | **Collected by:** |
| 4. Urine Orotic acid | ☐ | Date specimen collected ______ |
| 5. Urine Organic Acid | ☐ | |
| 6. Urine GAG / MPS (DMB & HRE) | ☐ | **Collected by:** |
| 7. Plasma Total and Free Carnitine | ☐ | (Operator ID) ______ |
| 8. Plasma Total homocysteine | ☐ | |
| 9. Blood spot: Total Galactose | ☐ | Date specimen send ______ |
| 10. Blood spot: GALT / G1PUT | ☐ | |
| 11. Blood spot: AA & Acylcarnitines (IEM screening) | ☐ | Paediatrician In-Charge: |
| 12. Blood spot for biotinidase assay | ☐ | |
| 13. Blood spot for Fabry | ☐ | |
| 14. Blood spot for Pompe | ☐ | |
| 15. Plasma VLCFA | ☐ | Sign. And chop: |
| 16. Plasma Phytanic acid | ☐ | |
| 17. Urine Succinylacetone | ☐ | |
| 18. Urine S-sulphocysteine | ☐ | : |
| 19. Urine Oligosaccharide/Tetraglucoside | ☐ | |
| 20. Urine Delta ALA | ☐ | |
| 21. Urine Phorphyrin & Phorphobilinogen | ☐ | |
| 22. Urine Cystine | ☐ | |
| 23. Urine Homocystine | ☐ | |
| 24. Urine Sialic Acid (Neuraminic acid) | ☐ | |

25. Others (Please specify):________________________________________