Annexure XII

ICMR TASK FORCE ON METABOLIC DISORDER

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HIGH RISK REQUEST FORM

Name: ______ Center ID No: ______ Ward No: ______

Sex: ______ Date of Birth: ______ Date: ______ Time: ______

Hours after feeding: ______

CLINICAL PRESENTATION

General Presentation

Acute decompensation after discharge: ______ Unexplained encephalopathy: ______

Failure to thrive: ______ Recurrent Vomiting: ______

Psychomotor retardation: ______ Poor Feeding/Lethargy: ______

Hyperventilation: ______ Tachycardia: ______

Abnormal odor: ______ Photosensitivity: ______

Neurological

Seizures: ______ Myoclonus: ______ Muscular Hypertonia: ______

Muscular Hypotonia: ______ Ataxia, Cerebellar Dysfunction: ______ Pryamisal signs: ______

Extrapyramidal signs: ______ Stroke like episodes: ______

Organ Dysfunction

Hepatopathy: ______ Hepatosplenomegaly: ______

Nephropathy: ______ Renal tubular Dysfunction: ______

Cardiomyopathy: ______ Ocular abnormalities: ______

Neuroradiological Findings

No pathological abnormalities: ______ White matter abnormalities: ______

Grey matter abnormalities: ______ Malformation/Lack of gyration: ______

Supratentorial Atrophy: ______ Infratentorial atrophy: ______

Cerebral Hematoma/Hemorrhage: ______ Others: ______

FAMILY HISTORY

Consanguinity:

Dystrophy at birth:

SIDS or unclear illness in siblings:

BIOCHEMICAL CHECKLIST

Hypoglycaemia & level: Time & concentration to normalization:

Acid base abnormality: Lactate Levels: Pyruvate Levels: Anion Gap:

Anemia: Pancytopenia:

Hyperammonemia & Level: CPK elevated & level:

AST/ALT elevated: Creatinine elevated/decreased:

Others, please specify:

Presumptive Diagnosis

Previous metabolic screening results (if any)

What diet was the patient on when this specimen was collected?

- [ ] Fasting
- [ ] Normal milk diet
- [ ] Medium chain triglycerides
- [ ] Cooking medium- Mustard oil

Is this a pre-treatment sample?

If not, what treatment (e.g. drug, dialysis, transfusion etc) was the patient receiving when and before the specimen was collected?

Antibiotics: Contrast dye: Valproic Acid: Others:

Specimen

Urine: Serum:

Plasma: CSF:

Others:

Contact Person

Name:

Cell No:

Address

Screening Test	Volume, ml	Container
plasma glucose	0.3	yellow cap(fluoride tube)
plasma ammonium**	0.75	orange cap(heparin tube, sent on ice
blood gases	0.3	heparinised capillary
anion gap		calculated based on routine electrolyte & RFT and blood acid/ base results. (Chloride ought to be requested)
plasma lactate**	0.3	yellow cap(fluoride tube, sent on ice)
plasma amino acids	2 x 0.75	orange cap (heparin tube)
urine for metabolic screening	spot urine*	pink top sterile container

Peri-mortem Protocol

In the event that a patient with suspected inherited metabolic disease dies before a specific diagnosis can be made, the following procedure is suggested.

Perimortem collection: Please state the period after death when the specimen was collection.

(a) Blood collection

- 2 x 5 ml in heparin tube
- 5 ml in EDTA tube
- 2 x 1 ml in fluoride tube
- 10 ml in plain tube (for storage)

(b) 903 S& S filter paper for collection of capillary blood.

(c) Urine collection: as much as possible and put in pink top sterile container.

7. Post-mortem Protocol

Bile: Can be used for post-mortem diagnosis of organic acidaemias.

Transport Of Samples(for indications, material, transport conditions etc.:)

Requested Investigations
Basic investigations: organic acids, aminoacids, acylcarnitines, simple metabolic tests *(urine, plasma, serum, dried blood spot)*
Basic investigations + additional investigations according to clinical picture *(urine, plasma, serum, dried blood spot)*
Tests
Aminoacids in plasma
Aminoacids in urine
Aminoacids in CSF + plasma
Acylcarnitine profile
Carnitine
Guanidinoacetate, creatine, creatinine
Urinary Metabolites
Miscellaneous: ____________________

Special Investigations

Biochemical Diagnostics	Indication	Material Transport/ preservation	Technique
Organic Acids Basic investigation	Metabolic screening, Unexplained decompansation, coma, unclear hepatopathy unclear neurological disorder	Spot urine Add 2 drops of(5-10 ml) chloroform or layer 5 ml on Whatman Filter paper 3	**GCMS**
Aminoacids in plasma Basic Investigation	Susp. of aminoacidopathy hyperammonaemia, mitochondriopathy	EDTA-plasma Centrifuge, (1 ml, 4-6 h send supernatant frozen after last meal) (dry ice)	**HPLC**
Aminoacids in urine	Renal disorders, hyperammonaemia	Spot urine Add 2 drops of (5-10 ml) chloroform or sodium azide	**HPLC**
Aminoacids in CSF	Susp. of neurometabolic disorders, esp., epileptic encephalopathy	CSF + EDTA-plasma Plasma centrifuged(1 ml each) supernatant and CSF (freeze directly at -70°C)	**HPLC**
Acylcarnitine profile Basis investigation	Susp. of organic aciduria or fatty acid oxidation defect	Dried blood spot Room temperature	**TMS**
Aminoacids in dried blood spot	Suspicion of Aminoacidopathy	Dried blood spot Room temperature	**TMS**

ICMR TASK FORCE ON INBORN METABOLIC DISORDERS

Task Force on Inborn Metabolic Disorders

HIGH RISK CONSENT FORM

Mrs. ________________________ W/o ____________________. I hereby give my consent to give my babies blood for newborn screening. I have been explained about the procedure and likely benefits of screening. I also understand that I may need to visit my doctor again and retesting of the baby may be required. I have also been explained that I can withdraw from the study at any time without compromising the care of my child. My participation in the study is totally voluntary.

Yes /No — I agree to have the remaining blood spots be stored and used for research purposes and the confidentiality will be maintained.

Mother's signature / Thumb impression

Name ____

W/O ____

Address ____

(Present & Permanent)

____

____

Phone # ____

Email ____

Fax # ____

Father'/Mother's signature/Thumb impression :

Signature of the study Doctor/Study staff ______________________________
Name in block letters

Date ______________________

Annexure – Hindi Consent Form

मैं, श्रीमान/श्रीमती ________________ **माँ/पिता** ______________, अपने बच्चे ______________ के रक्त/मूत्र परीक्षण के लिए सहमति देता हूँ। मुझे इस परीक्षण के लाभ के विषय में समझाया गया है। मुझे यह भी बताया गया है कि मुझे बच्चें को पुनः परीक्षण के लिए लाना पड़ सकता है। मुझे यह भी ज्ञात है कि मै किसी भी समय इस सोध में भाग लेने से मना कर सकता/सकती हूँ और इससे मेरे बच्चे की चिकित्सा में कोई बाधा नहीं आयेगी। मैं इस शोध में स्वैच्छिकता से भाग ले रहा/रही हूँ।

पिता/माता के नाम व हस्ताक्षर / अंगूठा

दिनांक :

शोधकर्ता के हस्ताक्षर :

नाम :

दिनांक :