

Received By:

*** Clinical Details (Mandatory):** Please indicate whether each feature is PRESENT by putting a tick mark on the indication mentioned below:

S. No	Indications	
1	CENTRAL NERVOUS SYSTEM	Developmental Delay / Mental retardation/ Hypotonia / Spasticity/Movement disorder/ Autistic Features/ Encephalopathy/ Lethargy/ Migraines/ Stroke, Ischemic episodes/ Ataxia/ Episodic Coma/ Seizures/ Pyramidal Signs/ Hemiparesis/ Self-Injury/ Speech Delay/ Feeding Problems/ Sleep Disturbance.
2	NEUROMUSCULAR	Peripheral Neuropathy/ Exercise Intolerance/ Muscle Weakness/ DMD-like/ Muscle Cramps after Exercise/ Easy Fatigability/ Cardiomyopathy/ Arrhythmia/ Ptois/ Cardiomegaly.
3	VISCERAL	Vomiting/ Gastrointestinal Reflux/ Diarrhea/ Constipation/ Cyclic Vomiting/ Hepatic Failure/ Elevated Transaminases/ Renal Tubular Disease/Apnea/Hypoventilation/ Respiratory Deficiency/ Renal Dysfunction/ Liver Carcinoma/ Jaundice/ Splenomegaly/ Hepatomegaly.
4	METABOLITES/ METABOLIC	Abnormal Newborn Screen/ Keosis/ Lactic Acidosis/ High CSF Lactate/ Acyl Carnitine profile/ Elevated Pyruvate/ Elevated Alanine / Hyperammonemia/Hypoglycemia/ Hyperglycemia/ Unusual Color/Odor
5	SENSORY	Cataract/ Sensorineural Hearing Loss/ Cherry Red Spot/Eye/ Corneal Opacity/ Ectopia Lentis
6	ENDOCRINE	Diabetes/ Exocrine/ Pancreatic Deficiency/ Gonadal Failure/ Hypothyroidism/ Hypoparathyroidism/ Hypo/Hyper-adrenal Function/ Short Stature/ Adrenal Calcification/ Hydrops Fetalis
7	OTHER CLINICAL	Failure to Thrive/ Microcephaly/ SIDS/ Unexplained Death/ Congenital Anomalies/ Dysmorphic Features/ Immunodeficiency/ Neutropenia/ Hypertrichosis/ Alopecia/ Acrocyanosis/ Macrocephaly/ Coarse Features/ Skeletal Anomalies/ Angiokeratoma/ Ichthyosis/ Arthritis/ Pregnant
8	IMAGING/ OTHER STUDIES	Posterior Stroke/ Leukodystrophy/ MRI/ Lactate Peak
9	TREATMENT (if any)	

INSTRUCTIONS FOR SAMPLE COLLECTION & TRANSPORTATION

Patient Registration: Fill up the requisition form with the required details and get the consent form signed by the patients/parent. (It is required to get detailed clinical history and consent along with the sample).

Labeling instructions: Label the tube/ container/ filter paper with the patient details including name, age, gender, collection date. Label with correct details to avoid sample switch over. Consent and patient clinical information need to be provided.

Transport instruction: Transport the sample as early as possible, according to biological sample transport guidelines. Sample should reach the lab in 48-72 hrs once the sample is collected.

Sample packing instructions: Place frozen gel packs on top & bottom of the sample in a thermocol box. Place samples in the sample holders. To avoid direct contact of gel pack with the sample, place thin sponge or filter paper/ paper between sample and gel pack. Duly filled test requisition and patient consent forms have to be placed in zip lock bag and send along with the sample.

Sample pickup: For sample pick up in case of outstation patients, send email to genomefoundation@gmail.com; Mobile No: 9704899766

Sample type	Volume/ Quantity	Collection instructions	Transport instructions
Whole blood - EDTA (Purple top)	4 -7 ml One tube	As soon as the sample is collected, gently invert the tube for 6-8 times to prevent clotting.	Refrigerate (4° C) the sample until transport. Transport with 2 gel packs (<i>Avoid direct contact</i>). <i>Do not freeze the sample.</i>
Plasma collected from Sodium heparin (Green top)	1 - 2 ml	Immediately centrifuge the sample at 2000 RPM for 10 min. Collect plasma in another tube.	Freeze the sample until transport. Transport with 2 gel packs (<i>Avoid direct contact</i>)
Serum - Plain tube (Red top)	1 - 2 ml	Allow sample to settle. Centrifuge the sample at 2000 rpm for 10 min. Collect serum in another tube.	Freeze the sample until transport. Transport with 2 gel packs (<i>Avoid direct contact</i>)
Skin biopsy with transport media (Orange top tube)	2-4 mm wide 2 mm depth	Using sterile instrument excise disinfected skin and immediately place in transport media provided (HAM'S F10 media). Do this procedure in sterile conditions.	Place the sample at room temperature. Transport as soon as possible at room temperature. <i>Do not freeze or refrigerate the sample.</i>
Cultured cells	25 or 75 cm ² flask	Fill the flask completely up to the neck with the appropriate medium.	Transport the sample at room temperature <i>Do not freeze or refrigerate the sample.</i>