THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

DEPARTMENT OF BIOCHEMICAL GENETICS
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CYSAB ISO 15189:2012 No. of Cert. L061

DEPARTMENT OF BIOCHEMICAL GENETICS – REQUEST FORM

Department Code: 30

F07.02.03.BG – Request Form_v1_16012024

Patient Details

Surname:	**Name:**
D.O.B.: ____/____/____	I.D. No.:
Nationality:	Gender: ☐ Male ☐ Female
CING No.:	Hospital File No.:
Address:	
City: / Code: / Country:	
Phone:	Email:

Patient Status

GESY: ☐	Government-Non GESY: ☐
Private-Non GESY: ☐	Hospital Card No.:
Outpatient: ☐	Inpatient: ☐

Case Information

Clinical Summary (also fill form at the back >>>PTO)

Referring clinician's/scientist's details

Surname:	**Name:**
Hospital/Clinic:	
CING: ☐ / Government (ΟΚΥπΥ): ☐ / Private-GESY: ☐	
Private-Non GESY: ☐	GESY No.:
Address:	
City:	Code:
Country:	Email:
Phone:	Fax:
Signature:	Date: ____/____/____

The referring physician undertakes and confirms understanding and compliance in respect of the mutual obligations as these are determined under the GDPR.

Sample details

Date and time of sample collection:

Blood: ☐ CSF: ☐ Urine: ☐ Muscle: ☐

Other (please specify):

First Investigation: ☐ Repetition: ☐

Test Requested (Code No.) *(Please tick ☑ accordingly)*

■ cannot be requested directly - subject to the results of other tests

Biochemical Assays

- ☐ *Lactate blood (1.1)**
- ☐ Lactate CSF (1.2)**
- ■ Pyruvate (2)
- ☐ *Ammonia (5)**
- ☐ Creatine Kinase (7)**
- ☐ *Amino Acids plasma (9.1)
- ☐ *Amino Acids CSF (9.2)
- ☐ *Amino Acids urine (9.3)
- ☐ *Acylcarnitines plasma (10)
- ☐ Biotinidase (15)
- ☐ Hexosaminidase A & B (16)
- ☐ Lysosomal Enzymes single (17)

  Please specify: _____
- ☐ Lysosomal Enzymes Screen (18)
  - ☐ Neurodegenerative
  - ☐ Dysmorphic features
  - ☐ Disorders involving liver & spleen
- ☐ Urine Screen-Stick (13)
- ☐ Reducing Substances (14.1)**
- ☐ Nitroprusside test for Cystine/Homocystine (14.2)
- ☐ *Mucopolysaccharides-Quantitative (19)
- ■ *Mucopolysaccharides-Electrophoresis (21)
- ☐ *Urine Organic Acids (31)
- ■ Sugar Chromatography-TLC (66)
- ☐ Oligosaccharides-TLC (67)
- ☐ Plasma methylmalonic acid (31.2)
- ☐ C26:0-LysoPC in plasma (private only) (24)
- ☐ Other _____ _____

Vitamins**

- ☐ *Vitamins A & E (80)
- ☐ *Vitamin B12 (69)
- ☐ *Folate (70)
- ☐ *Total Homocysteine (65)
- ☐ *Homocysteine-B12-Folate screen (71)

Muscle Biochemistry

- ☐ Mitochondrial Enzymes (41)
- ☐ Muscle Enzymes single (42)

  Please specify: _____

Westernblot:
- ☐ Dystrophin (46)
- ☐ Dysferlin (private only) (47)
- ☐ Calpain (private only) (49)
- ☐ Dysferlin - Calpain (priv. only) (50)

Exome Sequencing

- ☐ Metabolic Disorders WES
  - ☐ Single (32.6) ☐ Trio (32.7)
- ☐ Sanger sequencing for confirmation of NGS result (32.8)

Please check the box if diagnostic NGS tests will be requested and fill in the following fields:

☐ Current NGS request is for diagnostic purposes only.

Please state if any other diagnostic tests have been performed for this patient **Yes/ No**

If **Yes** state which tests: _____

If **No** state why patient is referred for NGS as a first-tier test: _____

For laboratory use only

Sample received on: _____ at: _____ Received by: _____

Comments: _____

Lab No.: _____

***Accredited Test** ****Can only be requested as part of an investigation for inherited metabolic disorders**

>>>PTO

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IMPORTANT!

To assist in the interpretation of laboratory results and facilitate diagnosis please give as much information as possible.

I. GENERAL PHYSICAL ABNORMALITIES

P 3 10 50 90 length _____ cm
P 3 10 50 90 weight _____ kg
P 3 10 50 90 headcir _____ cm

☐ abnormal face
☐ hepatomegaly
☐ splenomegaly
☐ ascites
☐ oedema
☐ icterus
☐ tachypnea
☐ hyperventilation
☐ hair + nail abnormalities
☐ skin abnormalities
☐ deafness
☐ strange smell
Other:_____
_____

II. NEUROLOGICAL ABNORMALITIES

☐ mental retardation
☐ motor retardation
☐ ataxia
☐ spasticity
☐ hypotonia
☐ hypertonia
☐ muscle dystrophia / weakness
☐ nystagmus
☐ choreo-athetosis
☐ convulsions
☐ lethargy / coma
☐ behavioural abnormalities
☐ exercise intolerance
☐ abnormal CT/MRI
Other:_____
_____

III. GASTROINTESTINAL ABNORMALITIES

☐ vomiting
☐ diarrhea
☐ refusal of nutrition
☐ constipation
Other:_____
_____

IV. NEPHROLOGICAL ABNORMALITIES

☐ renal stones
☐ polyuria
☐ strange colour / smell urine
Other:_____
_____

V. X-RAY ABNORMALITIES

☐ bone-age retardation
☐ skeletal abnormalities
☐ osteoporosis
☐ rachitis
Other:_____
_____

VI. IMMUNOLOGICAL ABNORMALITIES

☐ recurrent infections
Other:_____
_____

VII. HAEMATOLOGICAL ABNORMALITIES

☐ anaemia
☐ neutropenia
☐ thrombopenia
☐ thrombo-embolic abnormalities
☐ bleeding tendency
☐ lymphocyte vacuoles
☐ leucocyte granula
☐ abnormal bone-marrow
Other:_____
_____

VIII. CARDIAC ABNORMALITIES

☐ cardiomegaly
☐ cardiomyopathy
Other:_____
_____

IX. LABORATORY RESULTS

☐ acidosis / aniongap _____
☐ hypoglycaemia_____
☐ hyperglycaemia _____
☐ bilirubin _____
☐ liver enzymes_____
Other:_____
_____

X. NUTRITION

☐ oral
☐ parenteral
1 2 3 4 5 g/kg protein int.
☐ breast milk only
☐ special formula _____
☐ normal diet for age
☐ MCT oil
☐ vitamins
☐ carnitine
time of last meal_____
Other:_____
_____

XI. MEDICATION

☐ anti-epileptic _____
☐ antibiotics _____
Other:_____
_____

XII. FAMILY DETAILS

☐ nationality _____
☐ consanguinity
☐ metabolic disease in family
Other:_____
_____