THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

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GUIDELINES FOR SAMPLE COLLECTION - EXTENDED VERSION

Important Note:

A. All samples sent to the Biochemical Genetics Department must be accompanied by a request form properly filled with all personal and clinical details, including other available biochemical or genetic results. This is important for the interpretation of results and the selection of further tests.
B. For patients older than 5 years, blood sampling can be performed at the Cyprus Institute of Neurology & Genetics. For an appointment please call Mrs Eftychia Gaglia at 22-392737 after 10.00 am.
C. Tests marked with an asterisk (*) are offered on a private basis. The cost of these tests is covered either by the patient or the Ministry of Health following a relevant request.

1. ACID MALTASE (MUSCLE) *(Test code: 42)* TAT: 3-4 weeks

Muscle: 30-50mg rapidly frozen, stored and sent frozen.

2. ACYLCARNITINES *(Test code: 10)* TAT: 1-2 weeks

3ml of fasting blood in lithium heparin gel tube (**light green**). For newborns and babies, blood should be collected immediately prior to their next meal.
Send to the lab as soon as possible (keep cool) but not later than 2 hours after collection. If this is not possible, centrifuge immediately and freeze the plasma. Send to the lab frozen or at least cold.

Please provide information about carnitine supplementation or any other medication together with the clinical details to assist interpretation.

3. AMINO ACIDS *(Test codes: 9.1, 9.2, 9.3)* TAT: 1-2 weeks

For non-urgent investigations take a fasting sample. For urgent investigations take one sample immediately and repeat later.

For newborns and milk-fed babies take at least 4 hours after a feed.

Blood: 3ml of blood in lithium heparin gel tube (**light green**). Send to the lab as soon as possible (keep cool) but not later than 2 hours after collection. If this is not possible, centrifuge immediately and freeze the plasma. Send to the lab frozen or at least cold.

CSF: 2ml in a clean container. Keep cool and send to the lab immediately. If this is not possible, centrifuge immediately and freeze supernatant. Send to the lab frozen.

Urine: Fresh random urine sample (5-10ml) in a clean container **without preservative** (universal urine container).

4. AMMONIA *(Test code: 5)* TAT: 1-2 days

Fasting sample for routine analysis. Draw blood from stasis-free vein; the vein must not be compressed for more than 30 secs.

3ml of blood in EDTA tube (lavender). **Send to the lab immediately, preferably on ice**. For accurate results sample must reach the lab within **30 minutes** from collection. If this is not possible, centrifuge immediately and send plasma to the lab frozen.

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5. BIOTINIDASE *(Test code: 15)* TAT: 2-4 weeks

3ml of blood in a lithium heparin tube **(dark green)**. **Send to the lab immediately**. If this is not possible, centrifuge immediately and freeze the plasma. Send to the lab frozen or at least cold.

6. CREATINE KINASE (CK) *(Test code: 7)* TAT: 3-5 days

3-5 ml of blood in a plain tube **(red)**. Send to the lab as soon as possible (keep cool) but not later than 2 hours after collection. If this is not possible, centrifuge immediately and freeze the serum. Send to the lab frozen or at least cold.

7. DIP STICK / SPOT TESTS *(Test codes: 13, 14.1, 14.2)* TAT: 1-2 days

Fresh random sample (5 ml) taken in a clean container without preservative (standard urine container).

The **dip stick** includes pH, glucose, protein, blood, ketones, nitrite and sulphite.

The urine spot tests are:

- Reducing substances (detection of sugars: galactose, fructose, glucose)
- Nitroprusside test for cystine/homocystine

8. DNA ANALYSIS (NGS & Sanger sequencing) *(Test codes: 32.3-32.8)* TAT: 6-8 weeks

2ml of blood sample in EDTA tube **(lavender)**. Send to the lab as soon as possible and no later than 48 hours at room temperature.

9. DYSTROPHIN / DYSFERLIN / CALPAIN / DYSFERLIN-CALPAIN *(Test codes: 46, 47, 49, 50)* TAT: 4-8 weeks

Important! *(Test codes: 47, 49, 50)* private only*.

Muscle: 30-50mg rapidly frozen, stored and sent frozen.

10. LACTATE/PYRUVATE *(Test codes: 1.1, 1.2)* TAT: 3-5 days

The patient should be in a fasting and resting state.

Blood: Draw blood with minimum of stasis; the veins must not be compressed for more than 30 secs. Add 1ml of blood in the tube provided by the laboratory **(not to exceed the mark)**. Mix (it turns brown) and send to the lab. If the tube contains less than 2ml of reagent do not use.

CSF: 2 ml in a clear container. Send to the lab immediately.

11. LYSOSOMAL ENZYMES *(Test code: 18)* TAT: 4-6 weeks

Lysosomal enzymes can be measured as a group, according to the clinical features, or individually if there is a strong clinical suspicion for a specific disorder.

9ml of blood in sodium heparin tube **(dark green)**. If it is not possible to collect 9ml of blood from a newborn/baby/child, please prioritize enzymes according to the most probable diagnoses, since the sample will likely not be sufficient for all enzymes.

Send to the lab as soon as possible and not later than 48 hours, at **room temperature**.

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A. Neurodegenerative Screen	
ENZYME	**DISORDER**
Hexosaminidase-total & HexA (WBC)	Tay-Sach's / Sandhoff
β-galactosidase (WBC)	GM1 Gangliosidosis / Galactosialidosis / Morquio B
Arylsulphatase A (WBC)	Metachromatic Leukodystrophy
α-fucosidase (WBC)	Fucosidosis
β-glucuronidase (plasma)	Sly disease
I-cell screen (plasma)	I-cell disease
β-mannosidase (plasma)	β-Mannosidosis

B. Dysmorphic features (not Mucopolysaccharidosis)	
ENZYME	**DISORDER**
β-galactosidase (WBC)	GM1 Gangliosidosis /Galactosialidosis/Morquio B
Arylsulphatase A (WBC)	Metachromatic Leukodystrophy / Multiple sulphatidosis
α-fucosidase (WBC)	Fucosidosis
α-mannosidase (WBC)	α-Mannosidosis
β-glucuronidase (plasma)	Sly disease
I-cell screen (plasma)	I-cell disease
β-mannosidase (plasma)	β-Mannosidosis

C. Disorders involving liver and spleen	
ENZYME	**DISORDER**
β-galactosidase (WBC)	GM1 gangliosidosis / Galactosialidosis / Morquio B
α-mannosidase (WBC)	α-Mannosidosis
β-fucosidase (WBC)	Fucosidosis
β-glucosidase (WBC)	Gaucher disease
β-glucuronidase (plasma)	Sly disease
I-cell screen (plasma)	I-cell disease
β-mannosidase (plasma)	β-Mannosidosis

D. Other enzymes	
ENZYME	**DISORDER**
α-galactosidase (WBC)	Fabry disease
α-glucosidase (WBC)	Pompe disease
chitotriosidase (plasma)	Significantly increased in Gaucher disease. May also be increased in other lysosomal storage disorders e.g. Niemann Pick, GM1 gangliosidosis, Wolman, CESD, Krabbe.

If you are interested in a lysosomal disorder <u>not</u> included in the above list please contact the lab.

12. METHYLMALONIC ACID (MMA) *(Test code: 31.2)* TAT: 2-4 weeks

3ml of fasting blood in lithium heparin tube (**light green**). For newborns and babies, blood should be collected immediately prior to their next meal.
Send to the lab as soon as possible (keep cool). If this is not possible, centrifuge immediately and freeze the plasma. Send to the lab frozen or at least cold.

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13. MITOCHONDRIAL ENZYMES *(Test code: 41)* TAT: 4-8 weeks

Respiratory Chain Enzymes:

NADH Dehydrogenase (Complex I)
Succinate Dehydrogenase (Complex II)
Succinate Cytochrome C Reductase (Complex II & III)
NADH Cytochrome C Reductase (Complex III)
Cytochrome Oxidase (Complex IV)
Citrate Synthase (used for normalization)

Muscle: At least 50 mg rapidly frozen, stored and sent frozen.

14. MUCOPOLYSACCHARIDES - QUANTITATIVE *(Test code: 19)* TAT: 2-4 weeks
- ELECTROPHORESIS *(Test code: 21)*

Fresh random urine sample (5-10ml) in a clean container without preservative (standard urine container).

15. ORGANIC ACIDS *(Test code: 31)* TAT: 2-4 weeks

Fresh random urine sample (5-10ml) in a clean container **without preservative** (universal urine container).

- Pre-breakfast or fasting urine for suspected fatty acid oxidation defects.
- Non-fasting sample for organic acidaemias.
- Collect during the acute illness if possible.
- Provide information on clinical details, drugs and diet.

16. C26:0-lysophosphatidylcholine (C26:0-lysoPC) (*Test code: 24*) TAT: 2-4 weeks

Important! Private only*

3ml of fasting blood in EDTA tube **(lavender)**. For newborns and babies, blood should be collected immediately prior to their next meal.
Send to the lab as soon as possible (keep cool). If this is not possible, centrifuge immediately and freeze the plasma. Send to the lab frozen or at least cold.

17. SUGARS/OLIGOSACCHARIDES (*Test codes: 66, 67*) TAT: 2-4 weeks

Fresh random urine sample (5-10ml) in a clean container **without preservative** (standard urine container).

18. VITAMINS A & E *(Test code: 80)* TAT: 2-4 weeks
VITAMIN B12-FOLATE-TOTAL HOMOCYSTEINE *(Test codes: 65, 69, 70, 71)* TAT: 3-5 days

The patient should be fasting. If the patient has been on vitamin supplementation the sample should be taken at least one week after the last dose.

3-5 ml of blood in a plain tube **(red)**. The sample must be sent to the lab within two hours. If this is not possible, the sample should be centrifuged and the serum or plasma should be frozen and sent to the lab frozen or cold. Samples for vitamin E should be protected from light (aluminium foil).

FOR URGENT ANALYSES
PLEASE CONTACT

22392642 / 22392643 / 22392645

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