

Instruction sheet for tests to be performed in metabolic emergencies

This sheet should be customised by each reference centre or specialist centre

1 Acylcarnitine analysis

Two types of samples can be used: The reference or specialist centre should select the appropriate sample type according to the laboratory it normally works with.

PLASMA

Sampling conditions

- When: as close as possible to the acute episode, preferably before specific treatment.
- Type: heparin tube with or without gel separator
- Minimum volume: 300 µL of whole blood

Storage/transport

Store the primary tube at +4°C until centrifugation and decantation (< 3 hours). Then send the plasma to the performing laboratory at -20°C

WHOLE BLOOD on blotting paper

Sampling conditions

- When: as close as possible to the acute episode, preferably before specific treatment
- Type: whole blood on blotting paper
- Minimum volume: 2 spots

Storage/transport

Store at room temperature, protected from moisture. Send the blotting paper to the performing laboratory at room temperature, protected from moisture

Performing laboratory:

Billing code B [laboratory test]: or BHN [unlisted laboratory test]:

Laboratory contact details:

Catalogue link:

2 Plasma amino acid analysis

Sampling conditions

- When: as close as possible to the acute episode, preferably before specific treatment and on an empty stomach
- Type: heparin tube with or without gel separator
- Minimum volume: 300 µL of whole blood

Storage/transport

Store the primary tube at +4°C until centrifugation and decantation (< 3 hours) (or at room temperature < 1 hour). Then store and send the plasma to the performing laboratory at -20°C

Performing laboratory:

Billing code B [laboratory test]: or BHN [unlisted laboratory test]:

Laboratory contact details:

Catalogue link:

View the Emergency section of the G2M website



3 Urinary organic acids chromatography (including orotic acid)

Sampling conditions

- As close as possible to the acute episode, preferably before specific treatment
- Type: preservative-free urine
- Minimum volume: 1 to 2 mL

Storage/transport

Store in a dry preservative-free container, vial or tube at room temperature for < 6 hours. Then freeze and send to the performing laboratory at -20°C

Performing laboratory:

Billing code B [laboratory test]: or BHN [unlisted laboratory test]:

Laboratory contact details:

Catalogue link:

3 Determination of δ -aminolevulinic acid (ALA) and porphobilinogen (PBG)

Sampling conditions

- As close as possible to the acute episode, preferably before specific treatment
- Type: preservative-free urine, **to be protected from light immediately (opaque container, opaque bag or wrapped in aluminium foil)**
- Minimum volume: 1 to 2 mL

Storage/transport

Room temperature for < 6 hours, then frozen and sent to the performing laboratory at -20°C, protected from light

Performing laboratory:

Billing code B [laboratory test]: or BHN [unlisted laboratory test]:

Laboratory contact details:

Catalogue link:



EMERGENCY SAMPLE DISPATCH

For the sample dispatch, send them **as an emergency** during working hours via your usual courier and **notify the sorting centre**. If a sample is taken at night, ensure it is sent first thing the next morning.

Always inform the performing laboratory when a sample needs testing as an emergency.

Special case of hospital facilities under contract with the private laboratories CERBA or BIOMNIS:

These emergency samples may be sent to the performing laboratories (biochemistry services in the network, specialising in metabolic disorders) via CERBA or BIOMNIS. CERBA and BIOMNIS will only provide emergency transport to the performing laboratory, using Bioterfly for CERBA and Fastlab for BIOMNIS. Inform these laboratories that the samples must be sent as an emergency to the performing laboratories and **notify the performing laboratory** that the sample must be tested as an emergency.

LABORATORY CONTACT DETAILS

Secretarial inquiries should be made by phone through the laboratory office during the week or by email.

For on-call services, the laboratory's on-call telephone numbers for metabolic emergencies are: