Amsterdam UMC
University Medical Centers

Laboratory Genetic Metabolic Diseases

Protocol for collecting CSF samples for metabolic screening

Includes the following analyses:

Neurotransmitter metabolites, 5-methyltetrahydrofolate (5-MTHF), amino acids, GABA, homocarnosine, B6 vitamers, pipecolic acid and pterins.

General considerations

- Preferably collect CSF samples between 8 and 10 h in the morning, before any medication is taken.
- Always mention the actual medication on the request form.
- Collect the CSF samples in small clean tubes without additives.
- Note the fraction numbers on the CSF samples. Reference values for neurotransmitter metabolites are determined for this specific fraction.

Sample collection and storage

Collect fractions in numbered tubes according to the following scheme:

Fraction	Volume	Instruction	Analysis
1	1 ml	On ice	Amino acids + special requests
2	1 ml	On ice	Neurotransmitter metabolites, 5-MTHF, pterins
3	0.5-1 ml	On ice	GABA, homocarnosine, pipecolic acid, B6 vitamers
4	0.5-1 ml	On ice and deproteinized	Lactate, pyruvate

- For newborns collect at least 2 fractions of 1 ml.
- Transport the CSF samples on ice to the lab, as fast as possible.
- In case of blood contamination immediately centrifuge samples prior to freezing and transfer the clear supernatants to labeled clean tubes.
- For lactate and pyruvate measurements deproteinize the sample according to protocol, see www.amc.nl/lgmd/forms
- **Freeze samples immediately at -80°C and store until shipment** (if not possible, freeze at minimally -20°C).

Shipment

- Send the frozen samples on sufficient dry ice to our laboratory by (international) courier together with a completed test request form Metabolite Diagnostics (available at www.labgmd.nl).

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