

## Laboratory Genetic Metabolic Diseases

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### Protocol for collecting CSF samples for metabolic screening

#### Concerns the following analyses:

- Biogenic amine neurotransmitter metabolites
- Pterines
- 5-methyl-tetrahydrofolate (5-MTHF)
- Amino acids
- GABA
- Pilocarpic acid

#### 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.
- Due to the ventriculo-lumbar concentration gradient for homovanillic acid (HVA) and 5-hydroxyindoleacetic acid (5-HIAA), lumbar CSF samples should be taken in fractions in order to analyze the right fraction for these neurotransmitter metabolites

#### Sample collection and storage

- For newborns up to six months of age: collect a sample of 1-2 ml.
- For ages over six months collect fractions in separate numbered tubes according to the following scheme:

| Age      | Fraction 1<br>(# ml) | Fraction 2<br>(# ml) | Fraction 3<br>(# ml) |
|----------|----------------------|----------------------|----------------------|
| < 2 j    | 2                    | 1                    | 1                    |
| 2 - 16 j | 4                    | 2                    | 2                    |
| > 16 j   | 8                    | 4                    | 4                    |

- In case of blood contamination immediately centrifuge samples prior to freezing and transfer the clear supernatants to labeled clean tubes
- Because of the light sensitivity of the pterins, wrap all tubes in aluminum foil.
- **Freeze samples immediately at -80 °C** and store until shipment.

#### 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](http://www.labgmd.nl)).