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Laboratory Genetic Metabolic Diseases

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Protocol for collection and pretreatment of blood/CSF samples for the determination lactate, pyruvate, acetoacetate and 3-hydroxybutyric acid

- *Blood*: collect a minimum of 0.5 ml of blood in a fluoride or EDTA tube, mix well
- *CSF*: collect a minimum of 0.5 ml of CSF in a clean tube without additives
- Pipette 0.5 ml of blood / CSF in a conical tube of at least 10 ml
- Directly add an equal volume of 1.0 M perchloric acid to the sample
- Mix by gently tapping the tube (Do not vortex!)
- Place the mixture on ice for 10 minutes
- Centrifuge for 10 minutes at 4°C minimally 1700 x *g*
- Transfer the supernatant to a new tube
- Centrifuge again for 10 minutes at 4°C at 1700 x *g*
- If the supernatant is clear transfer it to a new tube suitable for transport; otherwise centrifuge again
- Store the sample at -20°C (-80°C if available) until shipment
- Send the frozen samples on sufficient dry ice, together with a completed laboratory form (available at www.labgmd.nl) to our laboratory by (international) courier