



**LABORATORIUM VIR AANGEBORE  
METABOLIESE DEFEKTE  
LABORATORY FOR INHERITED METABOLIC  
DEFECTS**

**SKOOL VIR BIOCHEMIE / SCHOOL FOR BIOCHEMISTRY**

**NOORDWES-UNIVERSITEIT (POTCHEFSTROOM KAMPUS) /  
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# HANDLING OF SAMPLES

Contact person for all enquiries:  
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## URINE SAMPLES:

- All urine samples must please be **FROZEN** and send on an ice pack or dry ice in a cooler box.
- NO PRESERVATIVES must be added to the urine sample
- For **CYSTINE & HOMOCYSTINE** analysis we need a 24h sample [volume must please be provided to calculate the excretion per day]

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## SERUM SAMPLES:

- Please avoid using red top tubes for blood collection, especially if the sample is not spun down and separated immediately (as required). ✘
- The blood sample must please be spun down, the serum/plasma transferred to another tube, frozen and send on an ice pack or dry ice in a cooler box. Even if a SST / EDTA tube is taken, the sample must still be spun down, the serum/plasma transferred and frozen – BEFORE transport to our laboratory.
- If the sample is haemolized, no analysis can be done, because haemolized samples could give false positive results. **ⓘ IMPORTANT**



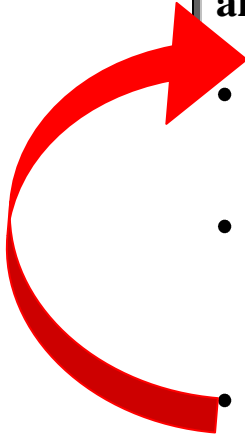

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## BLOOD CARD SAMPLES :

- All circles must please be completed.
- The sample must be taken by heel or finger prick and done in one circular movement. Be careful not to 'overlay' the sample!
- Please see instructions for taking of the sample at the back of the blood card.
- Allow samples to dry in a horizontal position (contact [brenda.klopper@nwu.ac.za](mailto:brenda.klopper@nwu.ac.za) for more specific information on New Born Screening requirements and Information)
- CARBOHYDRATE DEFICIENT GLYCOPROTEIN SCREENING & BIOTINIDASE ENZYME ACTIVITY DETERMINATION – allow samples to dry in a horizontal position (see instructions at the back of the blood card – Guthrie card)


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**CFS (cerebrospinal fluid) and SERUM SAMPLES for GLYCINE SPECIFIC ANALYSES (both required for this analyses):**

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- **CSF SAMPLE must be frozen and transported on DRY ICE**, since protocol requires that the sample does not defrost / thaw out during transport because it is only **viable** for analysis when **received frozen**.
  - **SERUM SAMPLE must be collected according to specifications on page 5 of this document** (as for other serum / plasma samples), **separated, frozen and send together with the CSF sample on DRY ICE**.
  - **IMPORTANT:** *For GLYCINE SPECIFIC analyses – the CSF + SERUM SAMPLES must be taken on the same date and time.*


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**OTHER SAMPLES FOR SPECIFIC ENZYME ANALYSIS:**

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- Please contact our laboratory, since the blood samples should be collected according to certain specifications and protocol.

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**FIBROBLAST SKIN CULTURE:**

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- Please contact our laboratory for skin biopsy protocol and relevant information regarding any specific analysis requested on fibroblast skin culture samples.
  - **PRIOR ARRANGEMENTS** must be made, in order to avoid possible contamination of collected samples during transport.

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**MUSCLE BIOPSY – MITOCHONDRIAL ACTIVITY DETERMINATION:**

- Please contact our laboratory for muscle biopsy protocol that should be followed and for **PRIOR ARRANGEMENTS**.
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