



Zentrum für Labormedizin
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SCHWEIZERISCHES REFERENZ-
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Stoffwechselfeldiagnostik Labor
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Arbeitsanweisung für Leberbiopsien für Harnstoffzyklus Enzyme (Englisch)

AW

Procedure for urea cycle enzymes analysis

We perform determinations of urea cycle enzymes in liver and erythrocytes, in order to help you delineate the cause of hyperammonemia in your patient and provide adequate treatment. In the past, we have experienced a variety of problems with unsuitable samples and packings. We, therefore, would like to specify the best way, in our experience, to send samples (Liver or Erythrocytes, see below).

Contact for technical questions (lab): +41 31 632 95 47 or orphan-disease.zlm@insel.ch

Contact for clinical questions: Prof. Dr. med. Jean-Marc Nuoffer, +41 31 632 95 69 or jean-marc.nuoffer@insel.ch

Specimen Information

Liver biopsy (NAGS, CPS, OTC, ASL)

20 - 50 mg of liver tissue are needed, which may be obtained by needle biopsy (Menghini needle, 1.2 or 1.4 mm diameter). Open laparoscopic biopsy should only be used if not enough material can be ascertained by needle biopsy. Blot the tissue on filterpaper, put it (without paper) into a small plastic vial or tube (e.g. Eppendorf tube) and close it tightly. Do not use glass tubes and do not wrap specimen in parafilm, Saran, aluminum, or similar foil. Immediately place the tube in dry ice and store the liver at -80 °C until shipment, if possible. Send in dry ice.

Erythrocytes (ASL, arginase)

Red blood cells from 5 ml blood should be washed at room temperature three times with a tenfold volume of physiological saline (0.9 %). After the last washing centrifuge sharply, remove the supernatant carefully and freeze the red cells.

Shipping information

Check if all tubes are properly closed and label them using a waterproof pen. If you use a label, make sure that it firmly sticks to the tube. Put the tube(s) in a styrofoam box together with dry ice (ca. 5 kg), wrap the package and attach an accompanying letter listing all the samples (preferably in a waterproof envelope). Indicate the value of the parcel as 20 Swiss Francs.

For mailing we recommend to use the services of a special parcel delivery agency. Please check that costs of transportation include delivery to the laboratory. Our laboratory is open at all times but it may be helpful to mail parcels at the beginning of the week to avoid delays at customs. Please inform us of the mailing and the approximate time of arrival. If you have any questions, please do not hesitate to contact us.

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