Laboratory of Clinical Biochemistry and Metabolism
Dr. phil. U. Berger/Dr. med. Dr. sc. nat. A. Schumann
University Children`s Hospital
Breisacher Str. 62
79106 Freiburg
Germany

Phone: **+49- 761- 270- 43680**
Switchboard: **+49- 761- 270- 43000**
Fax: **+49- 761- 270- 9645270**
E-Mail: zkj.stoffwechsellabor@list.uniklinik-freiburg.de

University Children`s Hospital
Department of Pediatrics and
Adolescent Medicine
Prof. Dr. med. U. Spiekerkötter
Chair and Medical Director of Pediatrics

Request form

Patient data (block letters)

Name:

First name:

Date of birth:

Gender: ☐ **f** ☐ **m** ☐ **n**

Date of sample collection:

Clinical information/diagnostic indications (essential for interpretation of test results)

Medication/infusions	☐ **no**	☐ **yes** (please specify)
Special diet	☐ **no**	☐ **yes** (please specify)

Clinical findings:

Ataxia	☐
Autism	☐
Coma	☐
Dystonia	☐
Epilepsia	☐
Encephalitis	☐
Failure to thrive	☐
Hepato-/Splenomegaly	☐
Cardiomyopathy	☐
Macro-/Microzephaly	☐
Mental retardation	☐
(Muscular) hypotonia	☐
Recurrent vomitting	☐

Pathological findings:

Acidosis [pH, BE]	☐_____
Anion gap [mmol/l]	☐_____
CK [U/l]	☐_____
Hyperammonaemia [µmol/l]	☐_____
Hypoglycaemia [mg/dl]	☐_____
Ketonuria	☐_____
Lactic acidaemia [mmol/l]	☐_____
Liver enzymes [U/l]	☐_____
Miscellaneous:	☐_____

Sender (name, full postal address)

Name of referring physician (block letters)

Phone

E-Mail address

Date/Signature

Metabolic requests:

☐ **Basic screening:** Acylcarnitines (DB), amino acids (S)[1], organic acids (U)[4], simple metabolic tests (U)[4]

Special requests:

☐ Acylcarnitines (DB)
☐ Acylcarnitines (S)[1]
☐ Acylcarnitines (U)[4]
☐ Adenosindesaminase 2 (ADA 2)-activity (WB)[3,5,9] (Requires declaration of consent on page 3)
☐ S-Adenosylmethionine/S-Adenosylhomocysteine (SAM/SAH) (P)[2,7]
☐ Alpha-aminoadipate semialdehyde (U)[4,8]
☐ Amino acids quantitative (CSF)[1,7]
☐ Amino acids quantitative (S)[1]
☐ Amino acids quantitative (U)[4]
☐ CDG-diagnostics (S)[1]
☐ 7-Dehydrocholesterol (S)[1]
☐ Enantiomeric separation (☐ Glyceric acid, ☐ 2-Hydroxy-glutaric acid, ☐ Lactate) (U)[4]
☐ Fatty-acid oxidation:

☐ Enzymatics (WB)[3,5,9] (Requires declaration of consent on page 3)
☐ MCAD ☐ VLCAD

☐ Genetics (WB)[3,5,10] (Requires declaration of consent from pediatric genetics)
☐ CACT (*SLC25A20*) ☐ Carnitine transporter (*OCTN2*) ☐ CPT 1 (*CPT1A*)
☐ CPT 2 (*CPT2*) ☐ MAD (*ETFA; ETFB; ETFDH*) ☐ LCHAD/MTP (*HADHA; HADH*)
☐ MCAD (*ACADM*) ☐ SCAD (*ACADS*) ☐ VLCAD (*ACADVL*)

☐ Glutathione (P/S/U)[2,7], (VB)[3,9], (F)
☐ Homocysteine (S/P)[1]
☐ Creatine deficiency syndromes (U)[4]
☐ Lysosomal diagnostics:
  ☐ Fabry disease (DB[6], only after consultation!)
  ☐ Gaucher disease (DB[6], only after consultation!)
  ☐ Mucopolysaccharidosis:
    ☐ Screening assay (U)[4]
    ☐ Electrophoresis (U)[4]
    ☐ Enzymatics (Type I, II, IIIB, IVA, VI, VII), (DB[6], only after consultation!)
  ☐ Niemann-Pick disease (DB[6], only after consultation!)
  ☐ Pompe disease (DB[6], only after consultation!)
  ☐ Wolman disease (DB[6], only after consultation!)
☐ Methylmalonic acid (S/U)[2,4]
☐ Mono-/Disaccharides (U)[4]
☐ Organic acids (U)[4]
☐ Orotic acid (U)[4]
☐ Peroxisomal disorders (VLCFA, Phytanic acid) (S)[2]
☐ Phenylalanine-/Tyrosine concentration (DB)
☐ Purines-/Pyrimidines (U)[4]
☐ Remethylation defects:
  ☐ Enzymatics (CblC, CBS, MTHFR), only after consultation!
  ☐ Remethylation profile (homocysteine, cysteine, methionine, cystathionine) (P/U)[1,4]
☐ Sulfocysteine (U)[4,7]

Legend for requested tests:

(CSF) Cerebrospinal fluid
(DB) Dried blood spots (allow to dry for 2 hrs at rt)
(F) Fibroblasts
(P) EDTA-plasma
(S) Serum
(U) Urine, conservation with 4-6 drps. of dichloromethane, ship at rt
(WB) EDTA whole blood

1. 0,5 ml
2. 1 ml
3. 2 ml
4. 5-10 ml
5. Informed consent mandatory (German law § 8, Abs. 1)
6. At no charge through different diagnostic initiatives
7. Immediately transfer to and ship on dry ice
8. Store at -20 C° until shipment, shipment of the frozen specimen together with cold pack
9. Arrival within 48 h after blood draw at the latest
10. Prenatal diagnostics only after consultation

Declaration of consent for enzymatic testing
according to the *German Genetic Diagnostics Act (GenDG)*

UNIVERSITÄTS KLINIKUM FREIBURG
ZKJ ZENTRUM FÜR KINDER- UND JUGENDMEDIZIN

Facility/Physician/Stamp	**Patient data** Name: Date of birth: First name: Street/number: Postcode/place:

The *German Genetic Diagnostics Act* requires detailed information and written consent for all molecular genetic analyses. Genetic counseling is additionally required prior to prenatal and predictive (predictive) analyses. Depending on the question, high-throughput methods such as exome sequencing are also used. According to the *German Genetic Diagnostics Act*, consent is also required for gene product analyses, such as **biochemical/enzymatic** tests.

Desired analysis/question (diagnosis, gene or biochemical-enzymatic investigation):

.....

Please read this consent carefully and mark the answers that apply to you:

O I consent to the findings of the analysis(s) being forwarded to Dr.:

.....

O If necessary, my findings/ may be used for the consultation/ examination of the following relatives:

.....

I hereby give my consent for blood and/or tissue taken from me or my child to be examined for genetic changes using molecular genetic/biochemical/enzymatic methods and consent to the processing of the genetic sample and genetic data. I have been informed about the purpose, nature, extent and significance of the requested molecular genetic/biochemical/enzymatic analysis and have had sufficient opportunity to discuss any open questions.
The possible consequences of the results of the molecular genetic/biochemical/enzymatic analysis for me or other family members were also discussed in detail (genetic counseling). I have been informed that I may revoke my consent in whole or in part at any time without giving reasons, either orally or in writing, without incurring any disadvantages. I have been informed that I have the right not to take note of the results of the examination in whole or in part, but to have them destroyed (“right not to know”).

Please mark with a cross where applicable:

O In genetic examinations, results may occur that are not limited to the above diagnosis (incidental findings). I also request that medically significant incidental findings be reported.
O The results of the examinations do not have to be destroyed after 10 years in accordance with the legal requirements, so that they may also be available to my family at a later date if necessary.
O I agree to the storage of the examination material for possible additional examinations to find a diagnosis or for laboratory-analytical quality control measures.
O I agree that data collected on the disease in question may be used in encrypted form (pseudonymized) for scientific purposes and published anonymously in scientific journals.

.....
Place, date Name (printed) Signature of patient or legal representative

.....
Place, date Name (printed) Signature of informing physician + stamp