

Phone: +49-761-270--43740 (secretary),
-45270 (Fax)
-43700 (PD Dr. Sara Tucci)
E-mail: fettsaeureoxidation@uniklinik-freiburg.de

requisitions can be downloaded:
<http://www.uniklinik-freiburg.de/kinderklinik/live/labor.html>

Confirmation diagnostics in case of suspected fatty acid oxidation disorders

Patient data
Name:

Sender (stamp)

First name: female male

Date of birth:

Invoice address:

Physician in charge (stamp):

Phone/Fax:

SUSPECTED DIAGNOSIS:

CLINICAL DATA:

Positive newborn screening result: yes no
Concentration of key metabolites:

Symptoms: yes no, asymptomatic

CLINICAL FINDINGS

- | | | |
|--|--|--|
| <input type="checkbox"/> Acute metabolic decompensation (11) | <input type="checkbox"/> Hepatopathy (12) | <input type="checkbox"/> Lethargy, Coma (13) |
| <input type="checkbox"/> Reduced clinical condition (15) | <input type="checkbox"/> Tachycardia/Arrhythmia (20) | <input type="checkbox"/> Seizures (34) |
| <input type="checkbox"/> Feeding difficulties (19) | <input type="checkbox"/> Cardiomyopathy (21) | <input type="checkbox"/> Muscular hypotonia (22) |
| <input type="checkbox"/> Other (60): _____ | | |

LABORATORY RESULTS

- | | | |
|---|--|--|
| <input type="checkbox"/> Metabolic acidosis (61) | <input type="checkbox"/> Elevated transaminases (65) | <input type="checkbox"/> Lactate _____ mmol/l (68) |
| <input type="checkbox"/> NH ₃ ↑: _____ μmol/l (64) | <input type="checkbox"/> Elevated creatine kinase (66) | <input type="checkbox"/> Alkalosis (74) |
| <input type="checkbox"/> Hypoglycemia, _____ mg/dl (63) | <input type="checkbox"/> Ketonuria (77) | <input type="checkbox"/> Other (90): _____ |

REQUESTED ANALYSIS:

Enzyme assay MCAD VLCAD CPT II

Please send at least 2 ml EDTA blood at room temperature, ideally at the beginning of the week. The sample should reach the lab within 48 h.

Genetic analysis MCAD LCHAD / MTP Carnitin transporter
 VLCAD CPT I CACT (Carnitin-Acylcarnitin-Carrier/ Translocase)
 SCAD CPT II ETFA/ ETFB/ ETFDH (Multiple acyl-CoA dehydrogenase deficiency)
 FAOD diagnostic panel (26 genes; complete gene list at request)

Please send 2 ml EDTA blood or dried blood spots as well as a consent form for genetic analyses signed by the parents.

SAMPLE: Date of sample collection: ____ . ____ . 20 ____

- EDTA blood
 dried blood spots (filter paper)

PLEASE INFORM US BEFORE SENDING SAMPLES! THANK YOU!