

**MVZ Labor PD Dr. Volkmann und Kollegen GbR
– Consultation Porphyria –**

Family name, first name Date of birth

Mailing Address

Sample taking – date / time

Urine volume ml	Sample period h	Body height cm	Body weight kg
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Prof. Dr. med. M. O. Doss
Dr. rer. nat. T. Stauch
Gerwigstr. 67
D-76131 Karlsruhe
Tel. +49-721 85000165
Fax +49-721 85000115
www.porphyrie.de
www.porphyrie.com

Address of sender / seal or stamp

Porphyria Diagnostics

Porphyrin precursors (5-aminolevulinic acid and porphobilinogen) in urine, porphyrines in urine and faeces, fluorescence scan in plasma or serum, enzymes of haeme biosynthesis in heparin blood and molecular genetic analyses

10a

Please fill in your address / contact details and consider also the second page of the form

Requesting physician or contact person: _____ phone _____

Sample material:

(please try to send all mentioned materials;)

- Urine: 10 ml spot sample (**without** additives / stabilizers), protected from light with aluminum foil
 or: 10 ml urine sample from a 24hour-collection with specification of the collected volume (protected from light).
 Faeces: 4 – 5 g (ml), protected from light with aluminum foil
 Heparin blood: 2 sample tubes ammonium- or lithium heparinate. Please use collection tubes without separator gel and send whole blood samples (tubes must not be centrifuged). Light protection also recommended

5701 **Screening / Primary diagnostics**

- because of clinical suspect (see below)
 because of positive family history

Symptoms during the time of sample taking?

- 5712 Yes (please specify below)
 5713 No

Do you or did you observe / record the following symptoms or complaints? (* ,see back page)

- | | |
|--|---|
| 5714 <input type="checkbox"/> Abdominal pain / colics | 5722 <input type="checkbox"/> Changes in personality, depression |
| 5715 <input type="checkbox"/> Nausea / vomiting | 5723 <input type="checkbox"/> Hepato-/Splenomegaly |
| 5716 <input type="checkbox"/> Constipation | 5724 <input type="checkbox"/> Vesication / Blistering on light-exposed skin areas |
| 5717 <input type="checkbox"/> Tachycardia, irregular heartbeat | 5725 <input type="checkbox"/> Hypertrichosis (temple, cheeks, eyebrows) |
| 5718 <input type="checkbox"/> Hypertonia | 5726 <input type="checkbox"/> skin fragility, bad healing capacity |
| 5719 <input type="checkbox"/> Otherwise unexplained ache of back or limbs | 5727 <input type="checkbox"/> Skin erosions, lesions, pigmentary abnormalities |
| 5720 <input type="checkbox"/> Paresthesia or numbness (fingers, palms, hips, thighs) | 5728 <input type="checkbox"/> Burning pain following light exposure |
| 5721 <input type="checkbox"/> Paralysis, movement disorders | 5729 <input type="checkbox"/> Oedema and/or erythema after light exposure |
| | 5730 <input type="checkbox"/> Chronical anaemia, haemolysis |

5702 **control examination or confirmation analysis following conspicuous screening results**

in: Urine Faeces EDTA-/Heparin blood Plasma

Which parameters were elevated? 5-Aminolevulinic acid Porphobilinogen Porphyrins (urine) Porphyrins (faeces)

Others: _____
 (Please send a copy of the reports, if possible) Please also specify information on symptoms (see above)

5703 **Follow-up examination**

- in case of known/diagnosed porphyria (the disorder was at least once in the past clinically active/symptomatic)
 in case of genetic predisposition to porphyria (so far no symptoms or complaints reported)
 0355 only state of metabolic activity in case of acute hepatic porphyria requested (determination of porphobilinogen)

Which kind of porphyria or porphyria predisposition is on hand or was diagnosed, respectively?

- | | |
|--|--|
| 5731 <input type="checkbox"/> Acute intermittent porphyria (AIP) | 5737 <input type="checkbox"/> Congenital erythropoietic porphyria (Morbus Guenther, CEP) |
| 5732 <input type="checkbox"/> Variegated Porphyria (VP) | 5738 <input type="checkbox"/> Hepatoerythropoietic porphyria (HEP) |
| 5733 <input type="checkbox"/> Hereditary Coproporphyria (HCP) | 5739 <input type="checkbox"/> Erythropoietic protoporphyria (EPP) |
| 5734 <input type="checkbox"/> Doss porphyria (ALAD-DP) | 5740 <input type="checkbox"/> X-linked protoporphyria (XLPP) |
| 5735 <input type="checkbox"/> Porphyria cutanea tarda (PCT) | 5741 <input type="checkbox"/> Atypical porphyrinogenic Response (APR) |
| 5736 <input type="checkbox"/> Lead intoxication/-exposure | |

5704 **Molecular genetic analysis** (exclusively after preceding consultation; genetic counselling and informed consent required)

Further clinical and anamnestic data or information (please report laboratory results with measuring unit only):

Medications / hormones / alcohol / drugs of abuse:

Liver- and/or bile diseases / hyperbilirubinaemia (GOT/GPT/ γ -GT/Bili):

Haemoglobin Concentration (blood):

Serum iron / Transferrin saturation / Ferritin:

Serum sodium / -magnesium:

Discolouration of urine:

Infectious diseases:

Neurological findings:

Psychiatric findings or -peculiarities:

Dermatologic assessment:

* depending on the overleaf marked symptoms or complaints the following tests will be performed:

- Nos. 5714 – 5722: 5-aminolevulinic acid (urine), porphobilinogen (urine), Total porphyrins (urine), fluorescence scan (serum/plasma), Total porphyrins (faeces), porphobilinogendesaminase activity (blood)
- No. 5723: 5-aminolevulinic acid (urine), porphobilinogen (urine), Total porphyrins (urine), fluorescence scan (serum/plasma), Total porphyrins (faeces), porphobilinogendesaminase activity (blood), protoporphyrins (blood)
- Nos. 5724 – 5726: 5-aminolevulinic acid (urine), porphobilinogen (urine), Total porphyrins (urine), fluorescence scan (serum/plasma), Total porphyrins (faeces)
- No. 5727: 5-aminolevulinic acid (urine), porphobilinogen (urine), Total porphyrins (urine), fluorescence scan (serum/plasma), Total porphyrins (faeces), protoporphyrins (blood)
- Nos. 5728 – 5729: Total porphyrins (urine), Coproporphyrin isomers (urine), Total porphyrins (faeces), protoporphyrins (blood)
- No. 5730: Total porphyrins (urine), fluorescence scan (serum/plasma), protoporphyrins (blood)

In case of conspicuous results (increased excretion values), we will perform further differentiations if considered necessary

Literatur:

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