MVZ Labor PD Dr. Volkmann und Kollegen SE & Co. eGbR
– Consultation Porphyria –

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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

Address of sender / seal or stamp

Porphyria Diagnostics

Porphyrin precursors (5-aminolevulinic acid and porphobilinogen) in urine, porphyrines in urine and faeces, fluorescscence 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/mail** _____

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 suspicion (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 ☐ Abdominal pain / colics
5715 ☐ Nausea / vomiting
5716 ☐ Constipation
5717 ☐ Tachycardia, irregular heartbeat
5718 ☐ Hypertonia
5719 ☐ Otherwise unexplained ache of back or limbs
5720 ☐ Paresthesia or numbness (fingers, palms, hips, thighs)
5721 ☐ Paralysis, movement disorders
5722 ☐ Changes in personality, depression
5723 ☐ Hepato-/Splenomegaly
5724 ☐ Vesication / Blistering on light-exposed skin areas
5725 ☐ Hypertrichosis (temple, cheeks, eyebrows)
5726 ☐ skin fragility, bad healing capacity
5727 ☐ Skin erosions, lesions, pigmentary abnormalities
5728 ☐ Burning pain following light exposure
5729 ☐ Oedema and/or erythema after light exposure
5730 ☐ 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 ☐ Acute intermittent porphyria (AIP)
5732 ☐ Variegate Porphyria (VP)
5733 ☐ Hereditary Coproporphyria (HCP)
5734 ☐ Doss porphyria (ALAD-DP)
5735 ☐ Porphyria cutanea tarda (PCT)
5736 ☐ Lead intoxication/-exposure
5737 ☐ Congenital erythropoietic porphyria (Morbus Guenther, CEP)
5738 ☐ Hepatoerythropoietic porphyria (HEP)
5739 ☐ Erythropoietic protoporphyria (EPP)
5740 ☐ X-linked protoporphyria (XLPP)
5741 ☐ Atypical porphyrinogenic Response (APR)

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 – 5276: 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

Literature:

– Anderson KE, Bloomer JR, Bonkovsky HL, Kushner JP, Pierach CA,Pimstone NR, Desnick RJ (2005): Recommendations for the diagnosis and treatment of the acute porphyrias. Ann Intern Med 142:439–450
– Bonkovsky HL, Guo JT, Hou W, Li T, Narang T, Thapar M (2013): Porphyrin and heme metabolism and the porphyrias. Compr Physiol 3:365–401
– Stölzel U, Stauch T, Doss MO in: Blau N, Duran M, Gibson KM, Dionisi-Vivi C (Hrsg): Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases, Springer: 1115-1128 (2022); https://doi.org/10.1007/978-3-030-67727-5_57
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– Stölzel U, Doss M (2009) Porphyrias. In: Dancygier H (ed) Clinical hepatology, principles and practice of hepatobiliary diseases, vol 2. Springer, Berlin, pp 1077–1092