



## Lab-form porphyria analyses (1/2)

<p><b><u>Patient (or vignette):</u></b></p> <p>First name(s): _____</p> <p>Last names(s): _____</p> <p>Date of birth: _____ <input type="checkbox"/> male <input type="checkbox"/> female</p> <p>Street, Nr.: _____</p> <p>ZIP code, place: _____</p> <p>Invoice to (health insurance membership number): _____</p> <p>Sample collection (Date/time): _____</p>	<p><b><u>Sender:</u></b></p> <p>_____</p> <p>_____</p> <p>_____</p> <p>_____</p> <p><b><u>! Contact details urgent results:</u></b></p> <p><b>Name:</b> _____</p> <p><b>Tel:</b> _____</p>
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<b>Acute attack (acute hepatic porphyrias):</b>				
Max. 5 days after last symptoms (otherwise see p.2). Urine <u>might</u> have a reddish/brown colour.				
Symptoms can include abdominal pain, paralysis, nausea, vomiting, obstipation.				
Possible forms of porphyria: <i>AIP, VP, HCP, ADP</i> (list of abbreviations see p.2)				
	Analysis	Material	Shipment	Comment
<input type="checkbox"/>	Aminolevulinic acid (ALA) und porphobilinogen (PBG), quantitative (qn.)	Spot urine, native, protected from light, 5-10 ml	A-Post, ambient temperature <b>In case of an emergency: Express/dispatch</b>	<b>Please inform the lab in case of an emergency: +41 44 416 56 40 (24/7)</b>  TP: 73.8

<b>Cutaneous symptoms:</b>				
- Photodermatosis with bullae, skin fragility ( <i>PCT, VP, HCP</i> )				
- Acutely painful photodermatosis, with or without visible symptoms ( <i>EPP, XLEPP</i> )				
- phototoxic mutilations ( <i>CEP, HEP</i> )				
	Analysis	Material	Shipment	Comment
<input type="checkbox"/>	Plasma-fluorescence scan	Heparin blood (without gel), protected from light, 4ml (alt.: Plasma)	A-Post, ambient temperature	Screening test for all forms of cutaneous porphyrias  TP: 54

<b>Reddish/brown urine in the newborn:</b>				
CEP or HEP <u>must be excluded</u> in a specialized laboratory before icterus therapy with blue light!				
	Analysis	Material	Shipment	Comment
<input type="checkbox"/>	Differentiation of porphyrins in the urine	Spot urine, native, protected from light, at least ml	<b>Emergency! Express/dispatch</b> Ambient temperature	<b>Emergency! Inform lab: +41 44 416 56 40 (24/7)</b> Cave: ALA und PBG are not elevated!  TP: 185.5



## Lab-form porphyria analyses (2/2)

### Abbreviation:

#### Acute hepatic porphyrias:

AIP : Acute intermittent porphyria <sup>Δ</sup>  
VP : Porphyria variegata <sup>Δ,\*</sup>  
HCP : Hereditary coproporphyria <sup>Δ,\*</sup>  
ADP : ALAD-Deficiency /Doss porphyria <sup>Δ</sup>

#### Non-acute porphyrias:

PCT : Porphyria cutanea tarda \*  
CEP : Congenital erythropoietic porphyria \*  
HEP : Hepato-erythropoietic porphyria \*  
EPP : Erythropoietic protoporphyria \*  
XLEPP : X-linked EPP \*

<sup>Δ</sup> **Medicines can trigger acute attacks!**

\* *Cutaneous symptoms*

ALA : Aminolevulinic acid

TP :Tarif points (Taxpunkte)

PBG : Porphobilinogen

### Synonyme:

ALA = delta-aminolevulinic acid, delta-aminolevulinat, 5-aminolevulinic acid

HMBS = Hydroxymethylbilane-synthase, PBG-deaminase, Urosynthase

<b>Parameters for monitoring of the porphyrias etc.:</b>				
Please provide us with information on the clinic. We recommend contacting the laboratory and/or the porphyria outpatient clinic for the most cost-effective diagnostic approach.				
	Analysis	Material	Shipment	Comment
<input type="checkbox"/>	Aminolevulinic acid (ALA) und porphobilinogen (PBG), qn.	Spot urine, native, protected from light, 5-10 ml	A-Post, ambient temperature <b>In case of an emergency, p.1</b>	Monitoring of the diseases activity (AIP, VP, HCP) TP: 73.8
<input type="checkbox"/>	Differentiation of porphyrins in the urine	Spot urine, native, protected from light, 5-10 ml	A-Post, ambient temperature	Monitoring of the disease activity (AIP, VP, HCP, PCT) TP: 185.5
<input type="checkbox"/>	Differentiation of the fecal porphyrins	5g sample of feces (size of a plum), without additives, protected from light	A-Post, ambient temperature	Differentiation of the porphyrias/ latent phase TP: 185.5
<input type="checkbox"/>	Enzymatic activity of HMBS	Heparin blood (without gel), 5ml, <b>no centrifugation</b>	A-Post, ambient temperature, <b>max. 24h</b>	Differentiation of the porphyrias/ latent phase TP: 131.5
<input type="checkbox"/>	Free, zinc- and total erythrocyte protoporphyrin	Heparin blood (without gel), 5ml protected from light, <b>no centrifugation</b>	A-Post, ambient temperature	Confirmation / monitoring of EPP und XLEPP TP: 184.5
<input type="checkbox"/>	Differentiation of porphyrins in the plasma (please see comment!)	Heparin blood (without gel), protected from light, 4ml (alt.: Plasma)	A-Post, ambient temperature	Patients on hemodialysis who developed bullae TP: 184.5

- ◆ Results are provided with an interpretation and recommendations.
- ◆ For medical questions and advice regarding therapeutic interventions please contact the porphyria outpatient clinic .

► More information: Swiss reference centre for porphyrias:

[https://www.stadt-zuerich.ch/triemli/de/index/kliniken\\_institute/swissporphyriacentre.html](https://www.stadt-zuerich.ch/triemli/de/index/kliniken_institute/swissporphyriacentre.html)