Protoporphyrin

Classification of Porphyria:

Primary Porphyria	
Erythropoietic forms	Inherited erythropoietic porphyria, Morbus Günther: Uroporphyrinogen-III co-synthase deficiency, extremely rare, red coloring of teeth, in early childhood light dermatosis, hemolytic anemia, hepatosplenomegaly and deep red urine;
Hepatic forms	Acute intermittent porphyria (AIP): uroporphyrinogen synthase deficiency, the most frequent (approx. 60% of all porphyrias) and most serious porphyria, more women than men affected (4:1), predominant manifestation age: 20-40th year of life, colic-like abdominal pains, vomiting, crises after drug intake;
Inherited coproporphyria	coproporphyrin oxidase deficiency, very rare, porphyrin in stool increased in latency phases as well;
Porphyria variegata (PV)	protoporphyrinogen oxidase deficiency, rare, manifestation between 10 and 30 years, frequently latent, fecal porphyrin excretion increased;
Porphyria cutanea tarda (PCT)	uroporphyrinogen decarboxylase deficiency, second most frequent porphyria, approx. 35% of all porphyrias, more men than women concerned (3:1), onset usually after 40 years, often after alcohol abuse. Liver function tests are pathological, fecal porphyrins only slightly increased, heptacarboxyporphyrin detectable in stool;
Erythrohepatic forms	Erythrohepatic protoporphyria (EPP): ferrochelatase deficiency, onset at any age, high protoporphyrin excretion in stool, coproprophyrins and uroporphyrins in erythrocytes increased or normal;

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Medication induced porphyria	aluminum salts, antiepileptics (carbamazepine, phenytoin, primidone), meprobamate, antirheumatics (gold derivatives, phenacetin, phenylbutazone), danazol, diclofenac, furosemide, griseofulvin, hypnotics and sedatives (esp. barbiturates), local anesthetics, alpha-methyldopa, metopirone, muscle relaxants, anesthetics, spironolactone, sulfonyl urea substances, sulfonamide antibiotics, theophylline, valproic acid, cytostatics;

Secondary Porphyria	
Intoxications	alcohol, chemicals, heavy metals, (esp. lead: ALS and copropor-phyrins in urine, protoporphyrin in urine, stool and in erythrocytes increased);
Liver disorders	cirrhosis, hepatitis, fatty liver, cholestasis, liver changes due to alcohol, porphyrinuria syn-drome, hemochromatosis, medication induced damages;
Blood disorders	hemolytic, medicament induced, sideroachrestic, sideroplastic and aplastic anemias, malignant anemia, leukemia;
Medication side effects	analgesics, antibiotics, hypnotics, anesthetics, estrogens, sedatives, sulfonyl ureas;
Other causes	diabetes mellitus, neoplasia, iron metabolism disturbances, hepatic cellular bilirubin transport disturbances;

Follow up tests: blood differential, hematocrit, iron, lead, glucose, (postprandial), bilirubin, GOT, GPT, γ-GT.

Laboratory values in Porphyrias:

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	inherited erythro-poietic porphyria	acute inter-mittent porphyria (AIP)	Hereditary copro-porphyria	Porphyria variegata (PV)	porphyria cutanea tarda (PCT)	erythrohep. Proto-porphy-ria (EPP)
Inheritance	autosomal recessive	dominant	dominant	dominant	dominant	dominant
Forms	erythropoietic	hepatic	hepatic	hepatic	hepatic	erythro-he-patic
Photosensitive skin	yes	no	rare	yes	yes	yes
Abdominal pain attacks	no	yes	yes	yes	no	no

Urine						
δ-ALA	n	(+++)	(+++)	(+++)		n
PBG	n	(+++)	(+++)	(+++)	n	n
URO	+++	++	+	+	+++	n
COPRO	++	n	+++	++	+	(+/++)
Stool						
COPRO	+	n	+++	+	(+)	(+)
PROTO	+	n	+	+++	n	+++
Erythrocytes						
COPRO	+++	n	n	n	n	+
URO	++	n	n	n	n	n

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PROTO	(+)	n	n	n	n	+++

N = normal

() = often increased only during a crisis

+ = slightly increased

++ = moderately increased

+++ = significantly increased

ALA = alpha-ALA, delta aminolevulinic acid

PBG = Porphobilinogen

PROTO = Protoporphyrin

URO = Uroporphyrin

COPRO = Coproporphyrin

General:

Examinations if suspecting porphyria:

Urine: δ-aminolevulinic acid, porphobilinogen (PBG) and complete porphyrins. PV (also in latency phase), hereditary coproporphyria, AIP are detected.

Stool: Porphyrins. EPP, latency phases of PV and hereditary coproporphyria are included.

Examinations in diagnosed porphyria: Porphyrins complete in urine, porphyrins differentiated in urine, δ-aminolevulinic acid in urine, porphobilinogen in urine, porphyrins erythrocytic, zinc protoporphyrin erythrocytic, coporphyrin in stool and protoporphyrin in stool.

The following tests are available:

- **Porphyrins, total, in urine**

  Indication: Acute intermittent porphyria (AIP), hereditary erythropoietic porphyria (Morbus Guenther), erythro-hepatic protoporphyria (EPP), porphyria variegata (PV). Porphyria cutanea tarda (PCT), hereditary coproporphyria, para-neoplastic porphyria (in prostate glan and liver tumors), acute and chronic lead poisoning induced porphyria (e.g. hexachlorbenzene, polychlorinated biphenyls, vinylchlorid, TCDD)

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Material: 10 (20) ml urine
Preanalytics: 24 hour urine, collect light protected, no spontaneous urine, please indicate urine quantity
TAT: 7-10 days*
Method: RECH
Units: µg/24 h
Ref.- range: up to 150

- **Porphyrins, total, in stool**

Material: 1 g stool
TAT: 7-10 days*
Units: µg/g stool
Ref.- range: up to 34.0

- **Porphyrins, total, in RBC**

Material: 2.7 ml EDTA blood
TAT: 7-10 days*
Method: HPLC
Units: µg/dl Ery.
Ref.- range: up to 60.0

- **Porphyrins, differentiated in urine**

Indication: Acute intermittent porphyria (AIP), hereditary erythropoietic porphyria (Morbus Guenther), erythro-hepatic protoporphyria (EPP), porphyria variegata (PV). Porphyria cutanea tarda (PCT), differentiation of positive total porphyrins, e.g. heptacarboxy-, hexacarboxy-, pentacarboxy porphyrins.
Material: 20 ml urine
Preanalytics: 24 hour urine cool, light protected and collected without additives, no spontaneous urine!
TAT: 7-10 days*
Method: HPLC
Units: µg/24 h

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- **Delta-aminolevulinic acid, δ-ALA, in urine**

General: Delta-aminolevulinic acid (5-aminolevulinic acid, δ-ALA) is an intermediate product in porphyrin biosynthesis. δ-ALA is generated from succinyl-coenzyme A, glycine and pyridoxal phosphate. Porphobilinogen is formed by condensation of two molecules ALA and catalyzed by the enzyme PBG-synthetase.

Indication: Suspicion of porphyria, lead intoxication

Material: 24 h collection period urine

Preanalytics: 24-hour urine with 10 ml of 10% acetic acid, of which 10 ml should be sent indicating the collected quantity.

TAT: 7-10 days*

Method: RECH

Units: mg/24 h

Ref.- range: <5.0

Note: The combined determination of porphobilinogen, porphyrine and δ-ALA is recommended

- **Porphobilinogen, PBG, in urine**

Material: 20 ml urine

Preanalytics: 24 hour urine, cool and light protected and collected without additives, no spontaneous urine!

TAT: 7-10 days*

Method: RECH

Units: mg/24h

Ref.- range: up to 3.0

Note: Porphobilinogen should always be requested together with δ-ALA

- **Uroporphyrin, coproporphyrin, pentaporphyrin, hexaporphyrin, heptaporphyrin in urine**

Material: 10 ml urine

TAT: 7-10 days*

- **Uroporphyrin, protoporphyrin, coproporphyrin, pentaporphyrin, hexaporphyrin, heptaporphyrin in stool**

Material: 5 g stool

TAT: 7-10 days*

- **Zinc protoporphyrin, in RBC**

Material: 3 ml EDTA blood

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TAT:	7-10 days*
Method:	HPLC
Units:	µg/g Hb
Ref.- range:	0.70 - 4.00

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

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