

#PORPHYRIA TOGETHER



WHAT IS PORPHYRIA?

por·phyr·ia | \ pòr-'fir-ē-ə

Porphyria is a group of mostly inherited genetic disorders caused by alterations in the eight enzymes in the pathway that makes

H E M E

H E M E

is part of essential hemoproteins, such as **hemoglobin**, **myoglobin**, and **cytochromes**

Which intermediates accumulate in the different porphyrias determines the type of symptoms – cutaneous or neurological (usually with acute attacks)

Then intermediates from the pathway accumulate, including porphyrins, porphobilinogen (PBG) and delta-aminolevulinic acid (ALA), depending on the type of porphyria

Each type of porphyria is caused by an inherited or acquired alteration in one of these enzymes

THEREFORE, PORPHYRIAS ARE COMMONLY CLASSIFIED AS “ACUTE” OR “CUTANEOUS”. THEY ARE ALSO CLASSIFIED AS “HEPATIC” OR “ERYTHROPOIETIC” DEPENDING ON WHETHER INTERMEDIATES FROM THE PATHWAY ACCUMULATE FIRST IN THE LIVER OR BONE MARROW.

ACUTE HEPATIC PORPHYRIAS

Characterized by
neurovisceral
attacks

COMMON SYMPTOMS

- Severe abdominal pain
- Vomiting, nausea
- Muscle weakness
- Blistering after sun exposure can occur in VP and HCP

- Tachycardia
- Confusion, hallucinations
- Seizures
- Insomnia

The Acute Porphyrias are classified as “Hepatic” because the ALA, PBG and porphyrins are overproduced and accumulate initially in the liver.

Certain triggers such as first hormones, diet, stress, and infection can trigger neurovisceral attacks.

TYPES

- Acute Intermittent Porphyria (AIP)
- Variegate Porphyria (VP)
- Hereditary Coproporphyria (HCP)
- ALAD-Deficiency Porphyria (ADP)

CUTANEOUS PORPHYRIAS

Characterized by skin
blistering or phototoxic
reactions

COMMON SYMPTOMS

- PCT, CEP, HEP: Chronic blistering and scarring
- EPP, XLP: Acute, painful, nonblistering phototoxicity avoided by altering life activities

PCT and HEP are hepatic porphyrias

EPP, XLP and CEP are erythropoietic porphyrias

TYPES

- Erythropoietic Protoporphyria (EPP) and X-Linked Porphyria (XLP)
- Congenital Erythropoietic Porphyria (CEP)
- Porphyria Cutanea Tarda (PCT)
- Hepatoerythropoietic Porphyria (HEP)

S

E

U

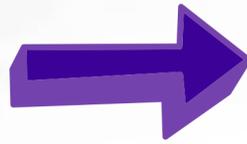
S

S

I

IMPORTANT

**DELAYED
DIAGNOSIS**



Patients can go undiagnosed for years even though diagnostic testing is available

PAIN



Pain is especially severe in the acute porphyrias. It is often inadequately treated during attacks and can become chronic.

**ACCESS TO
TREATMENT**



Barriers restrict access to approved treatments

**FUNDING FOR
RESEARCH**



New knowledge and treatments are needed!

**AWARENESS
CAN HELP!**

**TOGETHER,
we can raise awareness
about porphyria!**