#PORPHYRIA TOGETHER

WHAT IS PORPHYRIA?

por·phyr·ia | \ pȯr-ˈfir-ē-ə

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

HEME

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

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

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

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

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)

DISSIMILAR 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!