MAYO CLINIC
Mayo Medical Laboratories

Porphyria (Cutaneous) Testing Algorithm*

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Symptom: Skin manifestations restricted to sun-exposed areas

Possible cutaneous porphyria:
- Porphyria cutanea tarda (PCT)
- Erythropoietic protoporphyria (EPP)
- Variegate porphyria (VP)[a]
- Hereditary coproporphyria (HCP)[b]
- Congenital erythropoietic porphyria (CEP)
- X-linked dominant protoporphyria (XLDPP)

Symptoms:
- Edema
- Sun-induced erythema
- Acute painful photodermatitis
- Urticaria

PPFE / Protoporphyrins, Fractionation, Whole Blood

Symptoms:
- Blistering lesions or bullae
- Skin fragility
- Scarring
- Hyper/hypopigmentation
- Possible hypertrichosis

Normal → Excludes EPP → If clinically indicated

Increased zinc protoporphyrin → Consider:
- Iron deficiency anemia
- Heavy metal intoxication
- Anemia of chronic disease

Increased free protoporphyrin →
- EPP
- Family studies may be warranted

Increased free and zinc protoporphyrins (>40% zinc protoporphyrin) → Suspicious of XLDPP

FECHZ / Ferrochelatase (*FECH*) Gene, Full Gene Analysis

Mutation indentified → Confirms EPP

No mutation found

PQNRU / Porphyrins, Quantitative, Random Urine (includes PBG)

Normal → Excludes PCT and CEP → If clinically indicated

Increased coproporphyrin and/or PBG and uroporphyrin

Increased uroporphyrin and coproporphyrin

Increased uroporphyrin and heptacarboxylporphyrin

FQPPS / Porphyrins, Feces

UPGC / Uroporphyrinogen III Synthase (Co-Synthase) (UPG III S), Erythrocytes

UPGD / Uroporphyrinogen Decarboxylase (UPG D), Whole Blood

Increased coproporphyrin III/I ratio (<10) and protoporphyrin
- VP
- Family studies may be warranted

Increased coproporphyrin III/I ratio (>10) and coproporphyrin III
- HCP
- Family studies may be warranted

Normal → Excludes VP and HCP

Decreased → Confirms CEP

Normal → Excludes CEP

Decreased →
- PCT type II
- Family studies may be warranted

Normal →
- Excludes PCT type II
- Does not exclude PCT type I (sporadic) or PCT type III[c]

PPOXZ / *PPOX* Gene, Full Gene Analysis

CPOXZ / *CPOX* Gene, Full Gene Analysis

Legend
a. 80% of patients with VP have cutaneous symptoms
b. 20% of patients with HCP have cutaneous symptoms
c. Erythrocyte uroporphyrinogen decarboxylase is normal in PCT types I and III

*Interpretive report provided for all tests in this algorithm

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