

Symptom: Skin manifestations restricted to sun-exposed areas

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

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

PPFE / Protoporphyrins, Fractionation, Whole Blood

- Normal
- Increased zinc protoporphyrin
- Increased free protoporphyrin
- Increased free and zinc protoporphyrins (>40% zinc protoporphyrin)

Excludes EPP  
If clinically indicated

- Consider:
- Iron deficiency anemia
  - Heavy metal intoxication
  - Anemia of chronic disease

- EPP
- Family studies may be warranted

Suspicious of XLDPP

Consider CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies (ALAS2, FECH Gene List ID: IEMCP-XM9G9M)

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

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

Normal

Excludes PCT and CEP

Increased coproporphyrin and/or PBG and uroporphyrin

Increased uroporphyrin and coproporphyrin

Increased uroporphyrin and heptacarboxylporphyrin

If clinically indicated

FQPPS / Porphyrins, Feces

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

UPGD / Uroporphyrinogen Decarboxylase, Whole Blood

Increased coproporphyrin III/I ratio (<10) and protoporphyrin

Increased coproporphyrin III/I ratio (>10) and coproporphyrin III

Normal

Decreased

Normal

Decreased

Normal

- VP
- Family studies may be warranted

- HCP
- Family studies may be warranted

Excludes VP and HCP

Confirms CEP

Excludes CEP

- PCT type II
- Family studies may be warranted

- Excludes PCT type II
- Does not exclude PCT type I (sporadic) or PCT type III<sup>c</sup>

Consider CGPH (PPOX Gene List ID: IEMCP-XD2GBN) or PCGP / Porphyria Comprehensive Gene Panel, Varies

Consider CGPH (CPOX Gene List ID: IEMCP-BQFCG2) or PCGP

Consider CGPH (UROS Gene List ID: IEMCP-8W4945) or PCGP

Consider CGPH (UROD Gene List ID: IEMCP-376B8T) or PCGP

Consider PCGP