INDICATIONS FOR TESTING
Clinical suspicion of porphyrias

Neurologic and abdominal symptoms
Consider AIP, HCP, VP

Cutaneous photosensitivity
AND neurologic and abdominal symptoms
Consider HCP or VP

Cutaneous photosensitivity
Blistering
Burnling, itching, redness
Consider PCT, HCP, VP, CEP

ORDER
Urine PBG test
ORDER
Urine porphyrins and PBG tests
ORDER
Fractionated urine porphyrins test
ORDER
Erythrocyte porphyrin test

INITIAL TESTING

Strong suspicion
Consider ADP (rare)

Negative
Porphyria unlikely if symptomatic

Positive
FOCAL porphyrins test

Positive
Porphyria unlikely

Negative
Repeat testing when active symptoms are present

ADP probable

Negative
Repeat testing when active symptoms are present

Positive

Confirmatory testing
• PBG deaminase
• Variant analysis (PBGD gene)

Confirmatory testing
• Total plasma porphyrins

Positive

Other testing
• Total plasma porphyrins (use to monitor disease)
• Fecal porphyrins
• Variant analysis (UROD gene)

Other testing
• Fecal porphyrins
May consider
• Erythrocyte porphyrin (EP)
• Variant analysis (UROS gene)

Other testing
• Variant analysis (FECH gene, ALAS2 gene)

Negative
Other testing
• Variant analysis (CPOX gene)

Other testing
• Variant analysis (PPOX gene)

Pseudoporphyria, immunobullous disease, or connective tissue disease possible

CONSIDER
Skin biopsy for direct immunofluorescence and serum for indirect immunofluorescence
Connective tissue disease workup

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Abbreviations
ADP Aminolevulinic acid dehydratase-deficiency porphyria
AIP Acute intermittent porphyria
ALAD Aminolevulinic acid dehydratase
CEP Congenital erythropoietic porphyria
EPP Erythropoietic protoporphyria
HCP Hereditary coproporphyria
PBG Porphobilinogen
PCT Porphyria cutanea tarda
VP Variegate porphyria
XLP X-linked protoporphyria

"Result pattern, as interpreted by a medical expert, distinguishes porphyria type.
"If asymptomatic, consider repeating testing when symptoms are present.