Porphyrias

William W. Huang, MD, MPH

Dr. Huang is Assistant Professor of Dermatology, Wake Forest University School of Medicine, Winston-Salem, North Carolina. The author reports no conflict of interest.

Diagnosis (Inheritance Pattern)	Enzyme Defect[a]	Testing	Other
Acute intermittent porphyria (AD)	Porphobilinogen deaminase (C)	Elevated urinary porphobilinogen; elevated δ-aminolevulinic acid in plasma and urine; normal fecal and erythrocyte porphyrins	Second most common porphyria; no skin lesions; acute gastrointestinal, neurological, and psychiatric attacks; precipitated by certain medications (eg, griseofulvin, hormones, sulfonamides, barbiturates), physical stress, infection, and alcohol
Congenital erythropoietic porphyria (also known as Gunther disease)(AR)	Uroporphyrinogen III synthase (C)	Elevated urinary, fecal, and erythrocyte uroporphyrins and coproporphyrins; stable fluorescence of red blood cells	Dark red urine at birth; early photosensitivity, bullae, scarring, milia, and hypertrichosis; erythrodontia; porphyrin deposition in gallstones; growth retardation, osteopenia, bone fractures, deformed hands, and loss of eyelashes and eyebrows; anemia and thrombocytopenia
Porphyria cutanea tarda (AD, sporadic)	Uroporphyrinogen decarboxylase (C)	Urine with pink fluorescence under Wood lamp; elevated urinary porphyrins; uroporphyrins to coproporphyrins ratio, ≥3:1	Most common porphyria; associated with hepatitis C virus, lupus, hemochromatosis, and HIV; photosensitivity, bullae, scarring, milia, and hypertrichosis; increased risk for hepatocellular carcinoma
Hepatoerythropoietic porphyria (AR)	Uroporphyrinogen decarboxylase (C)	Elevated erythrocyte protoporphyrins; elevated urinary uroporphyrins; elevated fecal coproporphyrins	Recessive form of porphyria cutanea tarda; dark red urine at birth; early photosensitivity, bullae, scarring, milia, and hypertrichosis; erythrodontia; resembles congenital erythropoietic porphyria without hematologic abnormalities
Hereditary coproporphyria (AD)	Coproporphyrinogen oxidase (M)	Elevated fecal coproporphyrins > protoporphyrins; elevated urinary porphobilinogen, δ-aminolevulinic acid, and coproporphyrins only during acute attacks	Photosensitivity (1/3 of patients); acute gastrointestinal, neurological, and psychiatric attacks; precipitated by certain medications (eg, griseofulvin, hormones, sulfonamides, barbiturates) and physical stress, infection, and alcohol; clinically similar to variegate porphyria
Variegate porphyria (AD)	Protoporphyrinogen oxidase (M)	Plasma fluorescence at 626 nm; elevated fecal protoporphyrins > coproporphyrins; elevated urinary coproporphyrins > uroporphyrins (can help distinguish variegate porphyria from porphyria cutanea tarda)	Photosensitivity, bullae, scarring, milia, and hypertrichosis (like porphyria cutanea tarda); acute gastrointestinal, neurological, and psychiatric attacks (like acute intermittent porphyria); precipitated by certain medications (eg, griseofulvin, hormones, sulfonamides, barbiturates) and physical stress; common in South Africa
Erythropoietic protoporphyria (AD)	Ferrochelatase (M)	Normal urinary porphyrins; elevated erythrocyte protoporphyrins; transient fluorescence of red blood cells	Early photosensitivity, bullae, and scarring; waxy thickening of skin; porphyrin deposition in liver and gallstones
Pseudoporphyria	N/A	Normal porphyrin profile	Furosemide, NSAIDs (eg, naproxen), tetracyclines, dapsone, amiodarone, and retinoids; associated with hemodialysis and UVA exposure
Transient erythroporphyria of infancy	N/A	Transient elevated erythrocyte coproporphyrins and protoporphyrins	Blood transfusion associated; exposure to blue light for indirect hyperbilirubinemia; skin purpura

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; HIV, human immunodeficiency virus; N/A, not available; NSAID, nonsteroidal anti-inflammatory drug.

[a]Classified as cytoplasmic (C) or mitochondrial (M).

Practice Questions

1. Which of the following enzymes of the heme biosynthesis pathway is found in the mitochondria?

a. δ-aminolevulinic oxidase
b. porphobilinogen deaminase
c. protoporphyrinogen oxidase
d. uroporphyrinogen decarboxylase
e. uroporphyrinogen III synthase

2. A patient with variegate porphyria uniquely has plasma fluorescence at the following wavelength:

a. 311 nm
b. 366 nm
c. 410 nm
d. 626 nm
e. 630 nm

3. What laboratory result would be expected to be abnormal in a patient with a defect in ferrochelatase?

a. elevated δ-aminolevulinic acid
b. elevated erythrocyte protoporphyrins
c. elevated fecal coproporphyrins
d. elevated urinary porphobilinogen
e. elevated urinary uroporphyrins

4. What clinical feature would be present in a newborn diagnosed with transient erythroporphyria of infancy?

a. dark red urine
b. immediate burning photosensitivity
c. red teeth that fluoresce under Wood lamp
d. skin purpura
e. urine that fluoresces pink under Wood lamp

5. What medication should be avoided in a patient with a defect in porphobilinogen deaminase?

a. captopril
b. furosemide
c. griseofulvin
d. naproxen
e. tetracyclines

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