From a single urine sample*, order:
Urine Porphobilinogen (PBG), Quantitative, random (test code PBQTR) AND
Delta-Aminolevulinic Acid, Quantitative (ALA), random (test code ALAQR)

Order:
Porphyrins, Fractionated, random urine (test code PORFUR) AND
Porphyrsins, Fractionated, Fecal (test code MOLT) AND
Porphyrsins, Fractionated, Plasma (test code MOLT) AND
PBG Deaminase, RBC (PBGD) (test code UPGS)

*Random urine preferred for quantitative measurements during an acute attack to prevent delay in diagnosis

Normal PBG and ALA

Acute Porphyrias Excluded
(May consider repeat testing during acute episode if clinical suspicion remains high)

Increased PBG

Order:
Porphyrsins, Fractionated, random urine (test code PORFUR) AND
Porphyrsins, Fractionated, Fecal (test code MOLT) AND
Porphyrsins, Fractionated, Plasma (test code MOLT) AND
PBG Deaminase, RBC (PBGD) (test code UPGS)

Normal PBG, Increased ALA

Possible ALA Dehydratase Deficiency

Normal Fecal Porphyrin Profile
Decreased PBG Deaminase, RBC

Acute Intermittent Porphyria (AIP)

Increased Fecal Porphyrins
Normal PGB Deaminase, RBC
Increased Plasma Porphyrins

Variegate Porphyria (VP) or Hereditary coproporphyria (HCP)

Decreased ALAD

ALA Dehydratase Deficiency

Consider other disorders:
Lead poisoning or Hereditary tyrosinemia type I

Reviewer: Theodore Kurtz, MD. Last updated: 02/28/2014