**INDICATIONS FOR TESTING**

Clinical suspicion of porphyrias

**Symptoms:**
- Neurological
- Abdominal
- AND
- Cutaneous photosensitivity

**Consider**
- PCT
- EPP/XLPP

**ORDER**
- Erythrocyte Porphyrin (EP), Whole Blood
- Porphyrins, Fractionation and Quantitation, Urine

**Consider acute porphyria**
- AIP
- HCP
- VP

**ORDER**
- Porphobilinogen (PBG), Urine
- Porphyrins, Fecal

**Confirmatory testing**
- Aminolevulinic Acid Dehydratase (ALAD), Blood
- Mutation analysis (ALAD gene)

**Interpretation by medical expert necessary to distinguish the overproduction in the heme pathway**

**ORDER**
- Aminolevulinic Acid (ALA), Urine

**Consider**
- ADP (rare)

**ORDER**
- Porphobilinogen (PBG), Urine

**Consider**
- EPP/XLPP

**ORDER**
- Erythrocyte Porphyrin (EP), Whole Blood

**Consider**
- HCP
- VP

**ORDER**
- Porphyrins, Total, Plasma or Serum

**Consider**
- PCT
- CEP

**ORDER**
- Porphyrins, Fractionation and Quantitation, Urine

**Consider**
- ADP (probable)

**ORDER**
- Porphobilinogen (PBG) Deaminase, Erythrocyte
- Mutation analysis (PBGD gene)

**Consider**
- PCT or VP

**Consider**
- HCP or VP

**Consider**
- AIP

**Consider**
- EPP

**Consider**
- VP

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (UROS gene)

**Other testing**
- Erythrocyte Porphyrin (EP), Whole Blood
- Mutation analysis (UROS gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (UROD gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (FECH gene, ALAS2 gene)

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

**Other testing**
- Porphyrins, Total, Plasma or Serum

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (PPOX gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (CPOX gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (UROD gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (UROS gene)

**Other testing**
- Porphyrins, Fecal
- Mutation analysis (FECH gene, ALAS2 gene)

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