

Longitudinal Study of Porphyrias

Status: Recruiting

Eligibility Criteria

Sex: All

Age: Not specified

This study is NOT accepting healthy volunteers

Inclusion Criteria:

- Individuals with a documented diagnosis of a porphyria.
- For each type of porphyria, the inclusion criteria are based on
- Biochemical findings, as documented by laboratory reports (or copies) of porphyria-specific testing performed after 1980 (Absolute values are preferred for diagnostic biochemical thresholds. Fold increases in comparison to an upper (or lower) limit of normal (ULN or LLN) are also acceptable, but are complicated by considerable variation between laboratories in normal limits. Equivocal biochemical measurements may require confirmation by a consortium reference laboratory;)
- molecular findings documenting the identification of a mutation in a porphyria-related gene.
- In addition, an individual or a parent or guardian must be willing to give written informed consent or assent, as appropriate.
- Provision is made for enrolling relatives who may not have symptoms but have biochemical or molecular documentation of a porphyria, or in the case of recessive disorders carry a disease-related mutation.

Exclusion Criteria:

- Cases with elevations of porphyrins in urine, plasma or erythrocytes due to other diseases (i.e. secondary porphyrinuria or porphyrinemia), such as liver and bone marrow diseases;
- Patients with a prior diagnosis of porphyria that cannot be documented by review of existing medical records or repeat biochemical or DNA testing.

Conditions & Interventions

Conditions:

Acute Porphyrias, Cutaneous Porphyrias

Keywords:

porphyria, acute intermittent, coproporphyrin, variegate, erythropoietic, protoporphyria, hepatoerythropoietic, cutanea tarda, AIP, HCP, VP, ADP, ALAD, PCT, HEP, CEP, EPP, XLP, homozygous dominant, acute hepatic, AHP

More Information

Description: The objective is to conduct a longitudinal investigation of the natural history, complications, and therapeutic outcomes in people with acute and cutaneous porphyria.

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Phase: NA

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