Variegate Porphyria (VP) & Hereditary Coproporphyria (HCP)

OVERVIEW & SYMPTOMS

VARIEGATE PORPHYRIA (VP)

VP is caused by mutations in the gene of the protoporphyinogen oxidase (PPOX) enzyme. Over most of the world, it is less common than AIP. In South Africa, however, a prevalence of 3 in 1,000 individuals has been estimated, most of the cases arising in Whites of Dutch ancestry. Acute attacks in VP are identical to those in AIP. Blistering skin lesions are much more common than in HCP, are indistinguishable from those of PCT and may be chronic. There is no remedy for VP photosensitivity other than use of protective clothing. Unlike PCT, iron-depletion and chloroquine are not helpful.

HEREDITARY COPROPORPHYRIA (HCP)

The clinical presentation of HCP is similar to that of VP. About 10% develop blistering photosensitivity. The incidence of HCP appears to be at most 2 per 1,000,000. The deficient enzyme is coproporphyrinogen oxidase (CPOX).

TREATMENT & MANAGEMENT

Acute attack symptoms, long-term complications, treatment and prognosis, and management are the same as AIP for VP and HCP patients. Avoidance of sunlight is recommended for all individuals diagnosed with HCP or VP who have blistering photosensitivity. Both VP and HCP patients can present with acute attacks alone, blistering skin symptoms alone, or both.

VP & HCP are both autosomal dominant and recommendations for familial testing are the same for AIP.