

Title: Hereditary Coproporphyrria *GeneReview* – Differential Diagnosis

Authors: Bissell DM, Wang B, Cimino T, Lai J

Initial posting: December 2012

Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Nonspecific coproporphyrinuria is common and often misconstrued as HCP by the non-specialist, as in the following case:

*A 35-year-old woman seeks help for recurrent abdominal pain. She has visited the ER multiple times and has been hospitalized. An extensive evaluation has failed to yield a diagnosis. The patient does some web research, finding that her symptoms are a match for porphyria. Her conclusion about the diagnosis is reinforced by a “urine porphyrin screen”, which reveals a coproporphyrin level that is twice the upper limit of normal. Her physicians agree that the clinical picture and lab findings are consistent with acute porphyria, specifically HCP. They proceed to manage her pain episodes with opiates, glucose infusion, and intravenous hematin. The results are variable and often unimpressive. Symptom relief, when achieved, is of short duration – hours to a few days only. Overall the attacks become more frequent, to the point that pain is present on most days. Her use of opiates escalates, raising questions of narcotic dependence. She pursues a high-carbohydrate diet, having read that this may preempt symptoms in porphyria, and gains 90 lb. She is unable to work. She is referred to a porphyria specialist, who obtains genetic testing. The results are negative for any form of acute porphyria.*

The history illustrates that misdiagnosis of HCP can have life-altering consequences.