Condition: Acute intermittent porphyria

Inheritance:
Autosomal dominant.

Genetic etiology:
Mutation in *HMBS* gene, encoding porphobilinogen deaminase.

Frequency:
1-2/100,000 in Europe, 1/10,000 in Sweden.

Clinical features:
Intermittent episodes of abdominal pain, nausea, vomiting, abdominal distention, constipation or diarrhea; progressive peripheral neuropathy leading to weakness; progressive psychiatric disorder. Attacks precipitated by exposure to specific environmental agents, including alcohol and many medications.

Management:
Avoidance of exposure to inciting agents; acute attacks treated with intravenous dextrose and hemin.

Genetic counseling:
Based on autosomal dominant inheritance; molecular genetic testing available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.