

## ACUTE INTERMITTENT PORPHYRIA AND CYCLOID PSYCHOSIS

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**Introduction:** Acute intermittent porphyria (AIP) is a rare autosomal dominant inherited metabolic disease characterized by mutations in the porphobilinogen deaminase gene. This mutation may provoke neurotoxic levels of delta-aminolevulinic-acid and porphobilinogen, potentially resulting in an acute life-threatening clinical syndrome, characterized by psychiatric, in particular atypical psychotic, symptoms as well as severe neurological and gastrointestinal symptoms. Since the clinical presentation varies and symptoms are nonspecific, diagnosis is often made late.

**Objectives:** Naming of alarm symptoms based on a recent case study.

**Methods:** Description of a recent case supplemented with data from the literature.

**Results:** The patient is a 46 year old woman who was admitted in 2007 with abdominal pain, an epileptic seizure and weakness, interpreted as a Guillain-Barre syndrome. In 2011 she was readmitted with severe abdominal pain, diarrhea, volatile psychotic symptoms and seizures, following a short period of excessive alcohol consumption. During admission she developed progressive weakness in the upper arms, shooting pains in the limbs and a feeling of tightness. Impaired abdominal breathing was suspected. Again, Guillain-Barré syndrome was considered, but additional studies did not support this diagnosis. Because of the recurrent character of symptomatology following alcohol abuse, acute (intermittent) porphyria was considered diagnostically. The dark-colored urine indeed contained significantly increased delta-aminolevulinic-acid and porphobilinogen concentrations. Additional (genetic) diagnosis follows.

**Conclusion:** A recurrent disease course with severe gastrointestinal, neurological and psychiatric symptoms, following alcohol abuse, is suspect for AIP.