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FROM METABOLISM TO PSYCHIATRIC SYMPTOMS: PSYCHOSIS AS A
MANIFESTATION OF ACUTE INTERMITTENT PORPHYRIA

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Psychiatric symptoms may represent the main feature of some metabolic diseases. Acute intermittent porphyria (AIP) is the most common of the four forms of neuroporphyria. This is an heterogeneous group of hereditary illnesses with common alterations along the heme biosynthesis pathway. AIP mimics a variety of disorders, and it difficults a correct diagnosis. We report a case of confirmed diagnosis of AIP in a 19-years-old woman who was admitted to hospital due to a sudden acute psychosis. We have done also a complete bibliographic research about this illness and its relation to psychiatric symptoms.

In this case the differential diagnosis included schizophrenia and substance-induced psychosis. However, the presence of abnormal abdominal pain, port-wine color urine, sensory loss and muscular weakness gave us the clue for investigate AIP. Moreover, some anamnestic details about the use of anticonceptive pills, drug abuse and starvation increased our suspicion. Given the fact that the patient and her sister were both adopted, no family history was available.

The main laboratory findings supporting the diagnosis of AIP were a decrease in porphobilinogen deaminase (PBGD) activity in erythrocytes concomitantly with the detection of IVS7 (+33) G>T mutation of this enzyme's gene.

Despite the prevalence of AIP is not high around Mediterranean countries, this possibility should be kept in mind in order to elucidate the origins of a not so low number of acute psychotic episodes. They can be effectively and specifically treated, as well as easily prevented by controlling porphirinogenic factors.