Malady Of Kings In A Wine Drinker From Lombardy
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Citation

Abstract
Acute intermittent porphyria is a rare autosomal dominant disorder that has an incidence of 1-5/100,000. Acute attacks usually present with a variety of symptoms in particular, abdominal pain, neurological and psychiatric disturbances. It is quiescent until puberty and can be triggered by drugs such as phenytoin or oral contraceptives and fasting, alcohol and infection. The clinical diagnosis of acute intermittent porphyria is difficult and can be made more complicated by associated biochemical abnormalities. We present a case where a diagnostic dilemma is posed.

CASE REPORT
A previously fit 21 year old Italian male with no history of abdominal pain presented to our A&E department in November 2002 with a five day history of central abdominal pain, colicky in nature associated with vomiting and constipation. His abdominal examination revealed a soft and centrally tender abdomen, with no guarding or rebound and normal bowel sounds. The rest of the physical examination was normal, except for hypertension with blood pressures of 140/110mmHg and 183/114mmHg. There was no evidence of renal artery bruits or hypertensive retinopathy.

He was admitted because of the severity of the abdominal pain but after both surgical and medical reviews, no definite diagnosis was made. Most of his initial investigations were normal, except for polycythaemia with Hb of 21g, RBC of 6.46 x 1012 and hyponatraemia with Na 127mmol/L.

Radiological investigation revealed a normal chest x-ray, and a plain abdominal film showed dilatation of the transverse and descending colon with faecal loading of the ascending colon. There was no evidence of small bowel obstruction. A gastrograffin enema excluded an obstructive lesion.

As a surgical cause was now excluded, a diagnosis was proposed of acute porphyria. His hypertension was associated at the time of presentation with abdominal pain, nevertheless even in the absence of classical symptoms of phaeochromocytoma, his urine was screened for both catecholamines and porphyrins.

He was then started on a dextrose IV infusion, high carbohydrate and low protein diet. He made a good recovery over 5 days in hospital and on discharge his blood pressure was 135/70mmHg with a Na 134mmol/L.

DISCUSSION
The urinary porphyrins were diagnostic for an attack of acute intermittent porphyria (AIP). AIP is an autosomally dominant disorder resulting in a deficiency in the enzyme uroporphyrinogen I synthetase: an enzyme of the haem biosynthetic pathway, converting PBG to hydroxymethylbilane. This results in overproduction of PBG and its precursor delta-aminolaevulinic acid (ALA), and accounts for the increased urinary -ALA and PBG, which is diagnostic of an acute attack of AIP (Meyer et al. 1972). Our case had some of the features found in AIP, in particular the abdominal pain and constipation. Hypertension is also described but its mechanism remains controversial (Massey et al. 1980). Psychological manifestations and peripheral neuropathy, mimicking Guillain-Barre syndrome are also
described (Massey et al. 1980), but were not a feature here.

Interestingly, urinary analysis showed catecholamines elevated to levels suggestive of a phaeochromocytoma. This therefore posed a dilemma as to the cause of the hypertension. Essential hypertension was thought less likely, as there was no past history of hypertension and no evidence of end organ damage, with a normal chest x-ray and ECG. His BP also returned to normal over his stay in hospital. A phaeochromocytoma was also considered because he had hypertension associated with abdominal pain (25.8% : Ross et al. 1989). But the more characteristic symptoms of headache (59.9%), sweating (52.2%), palpitations (49.2%), palor (42.9%) and tremor (33.5%) were absent, so it was not considered to be a likely diagnosis. Nevertheless, repeat 24 hour urinary catecholamines on two further occasions followed by a clonidine suppression test would have helped to differentiate between AIP and phaeochromocytoma. Clonidine reduces plasma catecholamines in patients with neurogenically induced hypercatecholaminaemia, but not in patients with phaeochromocytoma. (Bravo et al. 1981). Consequently, we did not -block the patient, and instead treated for AIP. Repeat 24hour urinary catecholamine and porphyrin levels were normal.

We then reviewed the literature which suggests that hypercatecholaminaemia is the cause of hypertension in AIP (Suarez et al. 1997). Various mechanisms have been proposed. Flint et al. (1977) propose that patients with uroporphyrinogen I synthetase deficiency have an occult defect in catecholamine uptake into adrenergic neurons that is un-masked only in an attack of AIP by the high levels of ALA and PBG. Post mortem studies (see Suarez et al. 1997) found abnormalities in many components of the autonomic nervous system including the hypothalamus, and peripheral sympathetic ganglia. The real mechanism behind the increased catecholamine release may be multi-factorial.

**CONCLUSION**

Medical causes of abdominal pain are few and should always be considered when an atypical presentation of an acute abdomen occurs. They include AIP, lead poisoning, diabetic ketoacidosis and phaeochromocytoma. Our case was unusual in that there was a suggestion that he had both AIP and a phaeochromocytoma. But the hypercatecholaminaemia in this case were secondary to AIP. Should difficulties arise with the diagnosis; a clonidine suppression test can be used to differentiate.

**References**
