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ACUTE PORPHYRIA PRESENTING AS MAJOR TRAUMA: CASE REPORT AND LITERATURE REVIEW

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□ **Abstract—Background: Acute porphyria is historically known as "the little imitator" in reference to its reputation as a notoriously difficult diagnosis. Variegate porphyria is one of the four acute porphyrias, and can present with both blistering cutaneous lesions and acute neurovisceral attacks involving abdominal pain, neuropsychiatric features, neuropathy, hyponatremia, and a vast array of other nonspecific clinical features. Case Report: A 40-year-old man presented to the Emergency Department (ED) as a major trauma call, having been found in an "acutely confused state" surrounded by broken glass. Primary survey revealed: hypertension, tachycardia, abdominal pain, severe agitation, and confusion with an encephalopathy consistent with acute delirium, a Glasgow Coma Scale score of 13, and head-to-toe "burn-like" abrasions. Computed tomography was unremarkable, and blood tests demonstrated hyponatremia, acute kidney injury, and a neutrophilic leukocytosis. The next of kin eventually revealed a past medical history of variegate porphyria. The patient was experiencing an acute attack and received supportive management prior to transfer to intensive care, subsequently making a full recovery. Why Should an Emergency Physician Be Aware of This?: This case highlights the importance of recognizing acute medical conditions in patients thought to be suffering from major trauma. Acute porphyria should be considered in any patient with abdominal pain in combination with neuropsychiatric features, motor neuropathy, or hyponatremia. Patients often present to the ED without any medical history, and accurate diagnosis can be essential in the acute setting to minimize morbidity and mortality. The label of the major trauma call must be taken with great caution, and a broad differential diagnosis must be maintained throughout a diligent and thorough primary survey.**

□ **Keywords—porphyria; variegate; variegata; acute; trauma; medical crises; major trauma call**

INTRODUCTION

A PubMed search for variegate porphyria (VP), or the alternative, porphyria variegata, reveals only 15 case reports in the last 10 years. Variegate porphyria is one of four acute porphyrias, a group of rare inherited disorders of heme biosynthesis that all present with acute and potentially life-threatening neurovisceral attacks (1,2). The symptoms of an acute attack of porphyria are almost always dominated by severe abdominal pain, but the clinical presentation is relatively nonspecific. Acute porphyria has historically been described as "the little imitator," a reference to its reputation as a notoriously

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difficult diagnosis (3,4). This case highlights how acute porphyria can easily be misdiagnosed, as well as the importance of recognizing acute medical crises in patients presumed to be suffering from major trauma.

CASE REPORT

A 40-year-old man presented to the Emergency Department (ED) of a local trauma unit, brought in by ambulance and accompanied by police. He had been found in an acutely confused state outside his ground floor apartment surrounded by broken glass, having apparently fallen through a window. No past medical history was known for this patient. Primary survey revealed no inadequacy of airway or breathing, and no apparent injury to the cervical spine. The patient was hypertensive (with readings as high as 220/110 mm Hg) and tachycardic (105 min-1). He complained of abdominal pain, although his abdomen was soft and without peritoneal signs on examination. His Glasgow Coma Scale score was 13 or 14 (depending upon when he was scored) with E 3 or 4, V 4, and M 6. The patient was encephalopathic with acute delirium and severe agitation. The computed tomography (CT) scanner in the local trauma unit was temporarily unavailable, so the patient was immediately transferred to a Major Trauma Centre for urgent reassessment and imaging. The patient was acutely confused and unable to give any explanation for his injuries. What appeared to be head-to-toe "burn-like" abrasions were present (Figure 1), as well as a 5-cm laceration to the occiput. A prolapsed stoma was present in the right iliac fossa, and there was a visible midline laparotomy scar. There were no obvious signs of a focal neurologic deficit or systemic infection. A general anesthetic was given and the patient intubated to facilitate safe transfer for CT. The patient was managed as a trauma case, but results of biochemical tests began to raise suspicion that an underlying medical condition was the primary acute issue. The next of kin was contacted and eventually revealed a past medical history of alcohol and morphine abuse, ulcerative colitis, chronic pancreatitis, and VP. After his CT scan and supportive treatment in the ED, he was admitted to Intensive Care.

Over the following days it emerged that the patient had visited a general practitioner with blistering skin lesions less than a month previously and had subsequently seen a dermatologist, but miscommunication led to an assumption that this was porphyria cutanea tarda, a cutaneous porphyria, in which acute attacks do not take place. A week later the patient was admitted to the hospital after presenting with vomiting and severe abdominal pain; a diagnosis of acute-on-chronic pancreatitis was made after the urinary amylase was measured at 1051 IU/L (24-400 IU/L), and he was discharged after 9 days. His next of kin reported that he had been "acting strangely" in the few hours between his discharge from the hospital and being discovered by the police outside his apartment. Within a matter of hours he was being transferred as a major trauma call in the midst of an unknown acute attack of VP.

Venous blood gas demonstrated a sodium of 117 mmol/L (135–145 mmol/L) and a potassium of 2.9 mmol/L (3.5–5.0 mmol/L). Subsequent blood tests (Table 1) revealed marked hyponatremia, acute kidney injury (AKI), hyperphosphatasemia, and hypomagnesemia. The latter two results were likely a result of high stoma output. The stoma had been created 10 years previously after a colonic resection was necessary to manage the patient's ulcerative colitis. Liver function tests and serum amylase were raised, consistent with the patient's past medical history of alcohol abuse; he had been drinking heavily in the weeks building up to admission. There was a significant neutrophilic leukocytosis and a raised C-reactive protein; results typical of an underlying infection. Blood cultures were later positive for *Klebsiella*

Table 1. Blood Results and Porphyria Screen on Admission

Test	Result (Normal Range)
Full blood count	
WBC (↑)	26.19 (4.00–11.00 $\times 10^9$/L)
Hemoglobin	150 (130–165 g/L)
Mean cell volume (↑)	95.4 (77.0–95.0 fL)
Platelets	190 (150–450 $\times 10^9$/L)
Neutrophils (↑)	24.17 (2.2–6.3 $\times 10^9$/L)
Lymphocytes (↓)	0.92 (1.3–4.0 $\times 10^9$/L)
Renal profile	
Potassium	3.6 (3.5–5.0 mmol/L)
Urea (↑)	18.6 (3.3–6.7 mmol/L)
Creatinine (↑)	159 (45–120 umol/L)
Estimated GFR	34 (mL/min)
Sodium (↓)	121 (135–145 mmol/L)
Bone profile	
Corrected calcium	2.16 (2.15–2.6 mmol/L)
Phosphate (↑)	1.55 (0.80–1.40 mmol/L)
Magnesium (↓)	0.62 (0.7–1.0 mmol/L)
Liver profile	
Albumin	43 (35–50 g/L)
Bilirubin (↑)	99 (3–20 μmol/L)
Alkaline phosphatase	73 (30–130 IU/L)
Aspartate transaminase (↑)	225 (10–50 IU/L)
Gamma-glutamyl transferase (↑)	75 (1–55 IU/L)
Other blood results	
Amylase (↑)	127 (<100 IU/L)
Creatine kinase (↑)	2496 (<150 IU/L)
C-reactive protein (↑)	25.3 (<5 mg/L)
Random total urine porphyrin screen	
Total porphyrin excretion (↑)	429 (<35 nmol/mmol creatinine)
PBG excretion (↑)	24.9 (0–1.5 umol/mmol creatinine)
ALA excretion (↑)	11.2 (0–3.8 umol/mmol creatinine)

WBC = white blood cell; GFR = glomerular filtration rate; PBG = porphobilinogen; ALA = δ-aminolevulinic acid.