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Intelligent Malingering in the Setting of Porphyria Variegata: A Rare Occurrence on Both Fronts

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ABSTRACT

Malingering can be a difficult diagnosis to discern, especially in patients with well-crafted stories presenting with signs and symptoms that align directly with the literature. This can further become a challenge when a patient is malingering in the setting of a rare disease, where many complaints can be subjective in nature and not entirely testable by physical exam alone. Malingering is responsible for billions of dollars of healthcare waste every single year, and this report can serve as a guide of history elements, signs and symptoms to look out for with patients malingering in the setting of the porphyrias. It is important to recognize when patients are malingering, and when they are not, so that they may receive the appropriate care to help with their condition. This report can also serve as a guideline for what laboratory tests and studies to order in the setting of a suspected porphyria case, in order to confirm the diagnosis and get the patient the appropriate treatment regimen. Intelligent malingering is a growing problem, especially with the amount of access the general public has to medical information, and it is important for us to be able to identify when a patient is truly suffering from a rare disease and when they are malingering.

INTRODUCTION

Porphyria Variegata is a very rare disorder, affecting about 1 in 200,000 of the general population. It is a genetically inherited metabolic disorder in which Protoporphyrinogen Oxidase (PPOX)1, the enzyme that catalyzes the penultimate step of heme synthesis, is defective leading to the buildup of porphyrin precursors that manifest themselves as neurological, dermatological and abdominal symptoms. Medically, it is managed with pain control as well as Hemin infusions which inhibit ALA Synthase[1], the rate-limiting step of heme synthesis that converts succinyl-CoA and glycine to aminolevulinic acid, so that toxic heme precursors do not build up and cause pain or neuro-visceral symptoms. Acute attacks can be brought about in times of stress[2], or by use of contraceptives containing progesterone and antibiotics. Here, we discuss a patient presenting with an acute porphyria attack determined to be malingering. We will discuss tactics for management of patients with extremely intelligent malingering strategies.

CASE PRESENTATION

34 year old female with a reported PMH of Porphyria Variegata diagnosed at age 8, PTSD, anxiety, and right thigh port who presented with diffuse 9/10 intractable abdominal pain, nausea, vomiting, fatigue, right sided weakness and claimed photosensitive dermatitis. The patient reported that 5 days prior she was sexually assaulted

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by her father and was evaluated at an outpatient clinic where she was given antibiotics and contraceptives. She endorses being unable to return home during that period to receive her medications including "Dilaudid 2mg for panic attacks". She endorses developing nausea, vomiting, abdominal pain, weakness and dermatitis "exactly" similar to previous episodes of acute porphyria attacks. During the admission process and encounter, she was given her reported doses of pain medications. We were unable to independently verify any medical information for this patient initially, as she had no forms of identification upon presentation and could not remember her social security number. We were unable to obtain hospital records from locations she had been previously treated, and there was no record of the physician that she claimed had treated her previously. We were unable to verify her family members that she had reported, as well as the address that she gave. We were also unable to find any records of the patient in the PDMP database. As a diagnostic effort, urine testing was ordered to confirm porphobilinogen levels, but the patient refused to give a sample. The patient also refused to give a urine spot test to confirm her diagnosis. Hematology questioned whether her skin lesions accurately reflected photosensitive dermatitis as well. Nursing reported that the patient, when alone, was resting comfortably, but once surrounded by providers immediately shifted to a tearful and anxious affect. She would not let physicians examine her, and was demanding increases in her pain medication, not allowing pain management to see her. The patient was finally convinced to give a urine sample, but again demanded increases in pain medication for intolerable pain. Seeing as though she was not in acute distress, the decision was made to switch her medications to PO rather than IV. Once this decision was made the patient left AMA. Several days after leaving AMA, the patient's urine porphyrins came back as abnormal, with uroporphyrin 1 being elevated at 21.4, but the other urine porphyrins measured were all within normal limits. Urine porphobilinogen was 11.5, within normal limits.

DISCUSSION AND CONCLUSIONS

The porphyria's are rare inherited disorders, with subjective complaints that can be taken advantage of in the setting of malingering. The prevalence of the disease is estimated at 1 in 200,000 but in groups with South African or Dutch heritage the prevalence is estimated as 1 in 300 likely due to a founder mutation in the PPOX gene[1]. Recent studies have shown that in 20% of cases, both cutaneous lesions and neuro-visceral symptoms are present, similar to the presentation in our case, whereas up to 60% of cases present with cutaneous symptoms only[2]. The subjective nature of porphyria symptoms[2] make it difficult to distinguish between a drug seeking patient and a patient that truly needs medical intervention. The major presenting complaints in our case: GI upset, neurological complaints including numbness[3] and photosensitive dermatitis, are difficult to confirm in a patient reporting such pain that they do not want to be examined[3] or have lights on them. Our differential as this patient's course progressed included malingering, factitious disorder, somatoform disorder, and acute porphyria attack. We were able to differentiate between malingering and factitious disorder[4] due to the identifiable source of personal gain (Dilaudid), whereas in factitious disorders there is the patient has an uncontrollable desire[4] to take on the sick role[4], which this patient did not portray to us. Somatoform disorders of involuntary nature were also ruled out for this patient due to the patient's behaviors and symptoms reported[4]. Our case was complicated by lack of medical records and a carefully woven story that included all the details we as physicians look for in a porphyria attack. The workup to confirm an acute attack includes a urine porphobilinogen spot test and urine porphyrins[5]. Patients that refuse to give urine samples over the course of their time in the ED should raise some minor suspicion about their situation. Patients requesting exact dosages of narcotic medications should also raise minor suspicion as well. The story elements that directed us to a porphyria workup[6] are the reported history of porphyria, and the claim that she was given a rape kit at a clinic. During this patient's course, some history elements[6] could not be confirmed, at which point it can be recommended to switch IV medications to PO. Malingering patients may try to extend their hospital stay by reporting increasing pain, putting physicians in a tough spot for cases of actual porphyria attacks. In the case of suspected malingering for narcotics, our goals should be to completely evaluate all patients so that we may verify a diagnosis and ensure we deliver the best medical care possible.

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