Diagnosing deception: How doctors solved a woman’s dramatically faked condition

Enlarge / A health care worker in a medical intensive care unit.

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Diagnosing medical conditions is not easy. Patients can have nondescript symptoms that could point to common problems as easily as rare or poorly understood ones. They can sprinkle in irrelevant details while forgetting crucial ones. And they can have complex medical histories and multiple conditions that can muddy the diagnostic waters.

But then, there are the rare cases of pure deception. Such was the case of a woman seen at Massachusetts General Hospital for intense pain and jerking movements. The woman’s case record, published this week in the New England Journal of Medicine, documents the thorough investigation of her dramatic condition. Doctors’ initial alarm at her symptoms led to puzzlement as inconsistencies and oddities piled up.

It began when the woman presented to another hospital complaining of abdominal pain, jerking motions in her right arm and leg that she worried were seizures, as well as confusion, agitation, a rash on her chest, and a dislocated jaw bone. She told doctors at that hospital that she had a history of acute intermittent porphyria and that her symptoms matched previous flares of the condition.

Porphyrias are rare disorders caused by genetic mutations that are usually inherited. The mutation affects an enzyme involved in turning compounds called porphyrins and porphyrin precursors into heme, which makes up hemoglobin, the iron-containing red protein in blood responsible for transporting oxygen. In people with porphyrias, the heme precursors build up, causing disease that can present as abdominal pain, arm and leg pain, paresthesia, weakness, and tachycardia.

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The woman was admitted to the first hospital and began receiving treatment. But, the hospital was short on hemin—the standard treatment for porphyria—so she was transferred to Massachusetts General.

There, she told doctors a similar story, and they began treating her with hemin and other drugs, including morphine for the pain. She told doctors she was 25, though they noted in her records that she appeared older. She told them she had been diagnosed with porphyria 13 years ago and that the condition ran in her family. Her maternal grandmother had the condition, and one of her seven siblings was a silent carrier. She also noted that though she had been born in New England, she moved to the United Kingdom 15 years ago and was only in the area at the time to visit family.

Oddities

During the next two days, oddities started piling up. Despite doctors giving her the standard treatment for porphyria, her symptoms didn’t improve. And her urinary PBG and porphyrin levels—which are typically elevated in cases of porphyria—were normal.

The doctors began to doubt that porphyria was behind the woman’s symptoms. Instead, they considered bowel obstruction, biliary colic, appendicitis, or pancreatitis that could explain the abdominal pain. They thought about a medication or

toxin, such as lead poisoning, causing some symptoms. There was also a consideration of withdrawal syndrome from being off morphine before her admission. But, the woman’s symptoms also weren’t improved by the morphine, ruling that possibility out. Nothing quite fit.

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