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Case Report

Attack of Acute Intermittent Porphyria in a 34-Year-Old Female Patient after a Lithotrypsy

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INTRODUCTION

Hereditary porphyrias are a group of rare, metabolic disorders caused by haem biosynthesis defects. The clinical presentation, severity and prognosis depend on alteration of specific enzymatic step. One of the most common porphyrias is acute intermittent porphyria, inherited in autosomal dominant pattern. [1] It is characterized by overproduction of δ-aminolevulinic acid (ALA) and porphobilinogen deaminase (PBG) which are neurotoxic. These upstream metabolits are released from the liver to the circulatory system. [2,3] Most of acute attacks present as a combination of abdominal pain, mild psychiatric symptoms and autonomic dysfunction. However, severe attacks can be life threatening and present with seizures and psychosis. Acute attacks might be triggered by specific factors, usually ingestion of drugs metabolized by cytochrome P-450 but also undergoing surgery. The simplest and most specific way to diagnose an acute attack of porphyria is measurement of ALA and PBG levels in patient's urine. Hydration, carbohydrate administration and haem in therapy lead to reduction of ALA and PBG production. [4]

CASE REPORT

A 34-year-old female was admitted to the emergency department (ED) on 24th of June presenting paresthesia and muscle weakness in the lower extremities. Generalized seizure was observed. The blood pressure was 155/90 mmHg and the heart rate- 90 bpm. Physical examination showed a soft, tender abdomen with no peritoneal symptoms.

A complete neurological examination was performed. According to the medical history, the patient underwent a recent (on 19th of June) urological surgery- URSL + DJ catheter of the right kidney and since then was presenting a persistent abdominal pain and constipation. Nonsteroidal anti-inflammatory drugs (NSAIDs) turned out to be insufficient. Laboratory investigations revealed severe hyponatremia (107 mmol/l). cerebral oedema most likely caused by low blood sodium level. Due to persistent neurological symptoms, a computer tomography (CT) scan of head was ordered. The CT examination revealed a She was admitted to the nephrology department with recommendations for the further hyponatremia treatment. The patient's condition deteriorated rapidly. Despite the implemented fluid therapy, the level of sodium in the blood did not increase. The urine sample turned dark after being exposed to the light. Moreover, hypertrichosis on the patient's face was noted. The above raised the strong suspicion of porphyria. Urine quantitative determination of porphobilinogen (PBG) and δ-aminolevulinic acid (ALA) was commissioned. The urine test showed an increased excretion of PBG and ALA. This confirmed the initial diagnosis of acute intermittent porphyria (AIP). The patient was transferred to the Department of Disorders of Haemostasis and Internal Diseases for further treatment. After the symptoms ceased, she was discharged home and remains under the care of the clinic mentioned above.

CONCLUSION

The diagnosis of AIP is often delayed due to nonspecific symptoms and similarities to other diseases. Clinical presentation, characteristic to porphyria was initially attributed only to recent surgical intervention. This is indeed a trigger for the acute attack. Absence of symptoms in family history does not exclude a diagnosis of porphyria. This disease in 90% of cases has no clinical presentation. The patient's age, characteristic symptoms and recent surgery should raise strong suspicions of porphyria.

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