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Case Report

Porphyria and anorexia: cause and effect

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Porphyrias are hereditary disorders related to impaired biosynthesis of heme and characterized by multisystemic manifestations. Acute intermittent porphyria (AIP) is the most common acute subtype of the disease, and often associated with psychiatric symptoms. We here report a patient who developed acute flaccid paralysis after remarkable weight loss, which was related to an eating disorder (anorexia nervosa). After an extensive neurologic workup, he was diagnosed with AIP. This case emphasizes a deleterious vicious cycle between AIP and anorexia: porphyria may lead to anorexia and the carbohydrate restriction may lead to recurrent porphyric attacks. Therefore, an interruption of this cycle with psychiatric approaches to the eating disorders is crucial for long-term therapeutic efficacy.

INTRODUCTION

Acute intermittent porphyria (AIP) is an inborn error of metabolism caused by the accumulation of neurotoxic intermediates of heme, particularly aminolevulinic acid (ALA) and porphobilinogen (PBG). The main features are peripheral neuropathy, abdominal pain and psychiatric disorders, which are remarkably variable [1]. Here, we report a case of AIP and anorexia nervosa, highlighting this psychiatric manifestation either as a symptom related to porphyria or as a trigger to acute porphyric attacks.

CASE REPORT

A 19-year-old man previously diagnosed with anorexia nervosa (he had history of food restriction, rigorous physical exercise programs, induced vomiting and use of laxatives) lost 80 pounds in just 6 months. After that, he suddenly developed acute symmetrical flaccid lower limb weakness that progressed to the upper limbs in a few days, and was associated with paresthesias in all four limbs. At this time, the patient also suffered from abdominal pain, constipation and recurring episodes of 'red' urine. He had positive family history of psychiatric conditions (both his mother and maternal grandmother suffered from major depressive disorder), but no family history of neurologic illness. He was then diagnosed with Guillain–Barré syndrome in a different institution, but he refused treatment with intravenous immunoglobulin or plasmapheresis. He was left with severe disability and was wheelchair bound since then.

After 5 months, he was admitted to the psychiatric unit of UNICAMP with a clinical picture of worsening aggressiveness and antisocial personality disorder, which were present (but not treated) since childhood. At the time, the patient was severely disabled with quadriplegia, reduced muscle stretch reflexes, tactile and vibratory hypoesthesia with a distal gradient, combined with altered conscious proprioception. Nerve conduction studies and needle electromyography performed in the acute phase revealed a sensorimotor axonal polyneuropathy. He had normal vitamin B12 levels and there was no albumin–cytologic dissociation in cerebrospinal fluid analyses. Urinary PBG levels were found significantly increased, so that AIP was finally biochemically diagnosed. We opted not to treat him with hematine, because the clinical picture was stable with no signs of recent neurologic decline.

He was discharged with optimized psychiatric treatment that included the introduction of selective serotonin reuptake inhibitor (SSRI), as well as individual cognitive–behavioral

therapy and dietary guidelines under the supervision of a nutritionist.

The patient continues to be regularly monitored at the psychiatry and neurology clinics at UNICAMP. Although there has been no motor improvement to date, the eating disorder is now stable and he had no further attacks of AIP.

DISCUSSION

AIP is an autosomal dominant disorder caused by a deficiency of the enzyme PBG deaminase that leads to the accumulation of toxic substrates of heme [2]. Prevalence of porphyria has been estimated in 0.01% of the population and in 0.21–0.45% of all psychiatric patients [3]. Traditionally, AIP appears as a triad of abdominal pain, peripheral neuropathy and mental disorders. Abdominal pain is the most prevalent symptom, occurring in up to 90% of cases [4]; axonal neuropathy presents as weakness and hyporeflexia, and occurs in ∼20% of patients [5]. Neuropsychiatric disorders are found in 19–58% of patients heralding as depression, anxiety, acute confusional state, insomnia and psychosis [6].

Acute attacks are precipitated by events that increase the enzyme ALA synthetase or the demand for heme synthesis in the liver, including carbohydrate restriction and weight loss [5], which could explain the present case, that was triggered by carbohydrate restriction and weight loss in the context of eating disorders, which can be classified as a psychiatric manifestation related to porphyria.

Interestingly, it is possible that porphyria itself may be the cause for anorexia, which can lead to further porphyric crises due to weight loss and carbohydrate deficiency. Although uncommon, this correlation has been identified in other patients [7], confirming that both the association of porphyria and AIP is bidirectional: AIP influences the outcome of anorexia, whereas anorexia can cause attacks of porphyria.

To summarize, we conclude that the correct diagnosis and proper treatment of eating disorders are instrumental to reduce the chances of AIP exacerbation, preventing disability and improving the prognosis of these patients.

CONFLICT OF INTEREST STATEMENT

None declared.

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