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ACUTE INTERMITTENT PORPHYRIA CASE IN CLINICAL PRACTICE INTERNIST

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Abstract

Porphyria is group of eight metabolic disorders characterized by defects in hemebiosynthesis. The presentation of porphyria is highly variable, and the symptoms are non specific, which accounts in part for delay sinestablishing a diagnosis. Inthisstudy, were port the characteristics of one case acute porphyria patients in Ukraine. Case patient is femal, and the age was 20 years. The most frequent presenting symptoms was abdominal pain, dyspnoe, paresis and convulsion. Hyponatremia was the most common electrolyte abnormality. Testing provided a precise diagnosis of the patients:determination of coproporphyrins in the urine (the survey was conducted three times) three times the figures were slightly increasing, the highest 172 (provision do 120)nmol / day,determination of porphyrins in the blood of 248.5 (rule 60) mcmol / day and intheurine of 422 mcg / L (normal 150mcg / L), and excretion in the urine δ-

aminolevolinovoyacid 700 (normal 8-53) mcmol / day. Treatment. Drugs for the treatment of porphyria are not registered in Ukraine, because patients in stable condition was transferred to the Department of porphyria treatment Institute of Hematology and Transfusion (Warsaw, Poland).

Key words: porphyria, symptoms, testing, treatment.

Introduction. Porphyria is group of eight metabolic disorders characterized by defects in hemebiosynthesis. The presentation of porphyria is highly variable, and the symptoms are non specific, which accounts in part for delay sinestablishing a diagnosis. In this study, were port the characteristics of one case acute porphyria patients in Ukraine. **Case reports.** Patient M., female, born in 1993, was 01/14/2013 was admitted to one of the district hospitals of Lviv region, with complaints of fever up to 39ºS, back pain and abdominal pain, vomiting, constipation, muscle weakness and seizures. Disease began acutely 2 days before menstruation, with episodes of seizures before menstruation in the past. The genetic history. Brother of mother died at 3 years of age.

History of disease. In the CRH previously has been diagnosed with acute pyelonephritis, but the effect of treatment was absent, seizures, muscle weakness and respiratory insufficiency were progressing. 29.01.2013 due to the deterioration the patient was transferred to the intensive care unit №1 of Lviv regional hospital, where the council of physicians established the preliminary diagnosis of Guillain–Barré syndrome. The patient was transferred to the neurological department, but treatment was not effective, symptoms progressed. Chief of neurological department suspected porphyria because of cherry color of urine, and again the patient was transferred to the intensive care unit №1. Due to the absence of spontaneous breathing and limb paresis mechanical ventilation and parenteral nutrition were prescribed.

Objective examination: general condition is severe. Skin pale-yellow. Respiratory system: MV withRR 18/minute. Heart sounds muffled, rhythmic. Pulse - 90 per minute, blood pressure 120/80 mm Hg. Abdominal palpation: soft, peristalsisis weakened. Neurological status: reduction of tendon reflexes,tetraparesis.

Labs: a) complete blood count (29.01.2013) erythrocytes - 3.1 T / l, hemoglobin - 94 g / l, leukocytes - 5.4 G / l. Eosinophils - 1%, bounds - 5% segmented - 84%, lymphocytes - 10% ESR - 37 mm / h;b) blood chemistry: ALT - 34 mmol / L, urea - 7.4 mmol / L, K - 4.5 mmol / l, Na - 138 mmol / L (normal 145-155 mg / d L);

c) urinalysis: protein - 0.99 mmol / l , 25-35 leukocytes in sight, erythrocytes 6-8 in sight;d) coproporphyns in the urine (three times) - slightly increasing, the highest - 172 (normal up to 120nmol / day);d)porphyrins in the blood - 248.5 (normal up to 60 mcg / day). in urine - 422 (normal 150mcg / L);g) urinary excretion of δ-aminolinolenic acid - 700 (normal 8-53mmol / day);

Consulted online by porphyria expert from Moscow (Russian Federation).

He recommended:

A diet rich in carbohydrates (bread diet in obese patients rarely provokes attacks);

40% glucose; potassium (Panangin, Kalipoz); riboksin; plakvenil (skin changes);

normosan (arhinatheme) – is not registered in Ukraine.

Diagnosis. Acute intermitent porphyria.

Treatment. Council of physicians (chief of LRH, chief doctor of the medical work, hematologists, endocrinologists, neurologists, gynecologists, emergency physicians, GP, immunologist identified: there is no center of treatment patients with porphyria and no registered drugsin Ukraine, so patient in stable condition 27/03/2013 was transferred to a Institute of Hematology and Transfusion, Warsaw (Poland).

Monitoring. During 2013-2017 years patient experienced 16 acute attacks of AIP, 2013 – 6 episodes, 2014 – 4, 2015 - 5, 2016 – 0, 2017 (January-February) - 1. Triggers were menstruation, poisoning, stress, weight loss. The patient lives and works in Warsaw (Poland). Thus Poland provides free drugs. Without government assistance parents of the patient could not afford treatment.

Summary. Acute intermittent porphyria (AIP) - a hereditary autonomously dominant disease caused by deficiency of porphobilinogendeaminase and is characterized by accumulation of precursor of heme synthesis: δ-aminolinolenic acid and porphobilinogen [1, 2]. In Ukraine the disease is rare, so the records of these patients is not conducted. AIP is not endemic and widespread throughout the world. The incidence rate of acute manifestations of the disease in developed countries, according to various data, ranging from 1: 10,000 to 1: 50,000, while asymptomatic AIP carriers occur much more frequently (1: 1000). In today's world nearly 80 patients. Mainly women suffer. Peak incidence occurs at age 20-30. Often the only way to diagnose the disease is genetic study that can detect mutations in the gene PBDA. Biochemical disturbances associated with the disease is the result of the primary defect in the PBHD gene and are accompanied by increased levels of the liver enzyme aminolinolenic acid synthase (ALAS). The result is excessive production of heme precursors excreted in the urine. Menstruation ceasing is recommended using synthetic analogue of gonadotropin-releasing

hormone - hozerelin (Zoladex) 3.6 mg p / w 28 days or methyltestosterone 50 mg sublingually 1 per day. A characteristic feature of porphyria is that some drugs arecontraindicated [3, 4].

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