recommended to improve result reliability and clinical applicability.

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PP 20_ Case report

A CASE OF FAMILIAL PORFIRIA

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Introduction: Porphyria is a group of metabolic diseases caused by hereditary defects in the enzymatic system of heme biosynthesis. We present three cases of familial porphyria in two sisters (50 and 45-years-old) and a brother (39-years-old). The clinical symptoms of these patients were analyzed, and it was found that all patients have the same symptoms of the disease since childhood: constantly dark urine, burn-like changes on the skin of the face and ears, long-lasting wounds, moderate splenomegaly, and deformity of the joints on the fingers. **Clinical case:** Here, we present the some clinical and laboratory data one of the three patients. This patient was initially diagnosed with scleroderma, but due to the splenomegaly, she was referred to a hematologist to clarify the diagnosis. It was found that since childhood she had black urine, poorly healing wounds, and burn-like changes on the face and on the hands, which most often appeared after exposure to the sun. A physical examination revealed splenomegaly (+2–3 cm). The urine was initially intensely yellow-colored – when exposed to bright sunlight, the color changed to a dark yellow color. Total porphyrins in daily urine were 1.93 mg/L (norm 0–0.15 mg/L), uroporphyrinogen in single probe urine is 13 mg/L (norm 0–2 mg/L), and δ-aminolevulinic acid is 16.7 mg/L (norm 0.1–4.5 mg/L). The patient's sister and brother also were examined for quantitative tests for porphyrins in the urine. Both had similar porphyrin samples in their urine: total porphyrins in single probe urine – 1.23–1.3 mg/L, uroporphyrinogen in single probe urine – 12–13 mg/L, δ-aminolevulinic acid – 17.1 mg/L. **Conclusions:** Increased excretion of porphyrin precursors is one of the permanent signs of the disease and is observed not only during acute manifestations of the disease but also during the period of remissions, among the people with the latent form of the disease. Analyzing the results of our discussions, we can draw the following conclusions. Porphyria is a rare disease with very variable clinical symptoms, which causes certain difficulties in its timely diagnosis. To verify the diagnosis, even in the presence of a characteristic clinical picture, it is necessary to study the excretory profile of porphyrin metabolism with quantification of porphyrin precursors and fractions, as well as a comprehensive genetic examination.

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Adult Hematology Abstract Categories

Myeloproliferative Neoplasms

PP 21_ Case report

CHIC2 DELETION-ASSOCIATED HYPEREOSINOPHILIA AND SUBSEQUENT JAK2 V617F POSITIVE THROMBOCYTOSIS

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Background and aim: Myeloid/lymphoid neoplasms with eosinophilia and tyrosine kinase gene fusions (MLN-TK) are myeloid or lymphoid neoplasms driven by rearrangements involving genes encoding specific tyrosine kinases. These BCR::ABL1-negative diseases have long been recognized for their sensitivity to tyrosine kinase inhibitors. Herein, we present an interesting case diagnosed as myeloid neoplasm with abnormality of PDGFRA. **Case presentation:** A 63-year-old female patient with known glaucoma, hypothyroidism, and vitiligo was admitted to our clinic 8-years ago with fatigue. The patient had splenomegaly and eosinophil-predominant (10×10^9/L) leukocytosis (70.8×10^9/L). Secondary causes of eosinophilia (rheumatologic, infectious and immunologic) were excluded. Although not directly attributed to the patient's hypereosinophilia, echocardiographic left ventricular contraction abnormality was observed. Bone marrow aspiration and biopsy were hypercellular and showed 35%–40% eosinophils. With a preliminary diagnosis of hypereosinophilic syndrome, the patient was treated with steroids and then with hydroxyurea. Karyotype analysis was normal and FISH for t(9;22) was negative. The FISH panel revealed a 76% CHIC2 (4q12) deletion, but was negative for abnormalities of PDGFRB, FGFR1, and FIP1L1::PDGFRA fusion. The patient was switched to imatinib treatment. While the patient was followed in hematological remission for a long time with imatinib, thrombocytosis (807×10^9/L) was detected in the patient six months ago. The patient had suppressed erythropoietin level (1.94 mu/mL) and JAK2 V617F mutation. Low-dose hydroxyurea was combined with imatinib. Hematological remission was regained. **Discussion:** The majority of MLN-TK cases associated with PDGFRA rearrangements have cytogenetically cryptic deletion of 4q12 resulting in FIP1L1::PDGFRA. Although FIP1L1::PDGFRA fusion could not be demonstrated in this patient, it was thought that the patient had a myeloid neoplasm with abnormality of PDGFRA class due to CHIC2 deletion and typical clinical findings. The fact that the patient responded to treatment for many years seems to be evidence of this. The detection of JAK2 mutation during follow-up raised the question of whether this clone was present in the patient from the beginning or was acquired later.

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