Abstract: PB2569

Title: ALAS! PORPHYRIA OR SIDEROBLASTIC ANEMIA? A CASE REPORT

Abstract Type: Publication Only

Session Title: Iron metabolism, deficiency and overload

Background:

A 73 year old man presented in 01.2023 in the Hematology department with asthenia, adynamia, vertigo syndrome, mucocutaneous pallor, intermittent abdominal pain and lower limb polyneuropathy disorder, intolerance to sun exposure

Family history: Brother with MDS-RARS 2012-2022, COPD, chronic pulmonary heart disease, NYHA II heart failure, stage III kidney failure; passed away in 09.2022 due to cardiorespiratory and renal decompensation (3 dialysis sessions)

Medical history: Since 2005, hospitalizations for lead poisoning/professional saturnism (worked with lead-based paints and lacquers). Received chelation treatment with EDTA for lead and later remained without treatment. Since 2009, the patient is retired and has no contact with lead-containing substances, but symptoms have been progressing and worsening during these 14 years.

Other diagnoses: lead-induced polyneuropathy, gastropathy due to lead poisoning, orthostatic hypotension, single syncope episode, single paroxysmal tachycardia episode, carotid atheromatosis, cervical spondylosis, hepatic steatofibrosis, left inguinal hernia, first-degree atrioventricular block, high blood pressure with level III risk, secondary hemochromatosis.

Aims:

Accurate diagnosis and specific treatment.

Methods:

> Laboratory tests: CBC, Folic acid, Vitamin B12, Peripheral blood smear, Bone marrow biopsy and aspiration
>
> Fibroscan

Porphyria genetic testing

Results:

Biological Data: WBC=5940/mm3; Platelet count=283000/mm3;

Differential count: Ne 1%, Ly 61%, Mo 3%,Eo 1%, Ba 27%, atypical Ly 8%. Hb=8.2 g/dl; Ht=27%; MCV=59.9 fl; MCH=17.6 pg.

PBS: moderate anisocytosis with microcytosis and marked hypochromia up to anisocytosis; moderate poikilocytosis with target cells, elliptocytes, ovalocytes, irregular contracted erythrocytes (3%); hypogranular granulocytes.

HbA2 normal - excludes beta-thalassemia.

Serum Iron=249 ug/dL (VN 65-175); Serum ferritin=3660 ng/mL (22-322); Hemochromatosis gene negative - exclude hereditary hemochromatosis.

Folic acid 2.49 ng/mL (3.8-26.8), serum vitamin B12 normal.

Urea=72 mg/dL (17-49), creatinine=1.57 mg/dL (0-1.3); eGFR 43 mL/min, uric acid=7.4 mg/dL (3.5-7.2).

Glucose=89 mg/dL (83-110); TGO=60 U/L (0-45); TGP=38 U/L (11-34); GGT=109 U/L(11-50); FAS=132 U/L (Normal value).

Viral and tumor markers negative; FIBROSCAN: S1, F3-4!

PCR COVID positive CT 24 - on the day of admission (treated at home - mild form molnupiravir).

Bone marrow aspiration: Myelodysplastic syndrome, refractory anemia with ringed sideroblasts; 86% sideroblasts; 65% ringed.

Arguments for genetic testing for porphyria: Classic MDS is typically associated with normo-macrocytosis!

Microcytosis is only seen in iron deficiency anemia and hemoglobinopathies, both of which have been ruled out by the elevated serum iron and ferritin levels and hemoglobin electrophoresis. The persistence of symptoms, after 14 years of no exposure to lead, is noteworthy.

Genetic testing for porphyria: The following mutation was detected in hemizygous state: ALAS2 (NM_000032.5):c.1701G>C(p.(Met567Ile)).

Summary/Conclusion:

The detection of this mutation confirms X-sideroblastic anemia or erythropoietic protoporphyria, depending on the loss of function or gain of function, respectively. In order to put the right diagnosis, we had the patient tested for Zn protoporphyrin, free erythrocyte protoporphyrin with the results still pending.

Treatment:

Rbc transfusion: 2 U (anemia not well tolerated clinically)

Genetic testing of relatives:

- daughter most likely a carrier
- direct grandchildren: boy and girl
- nephews

Will undergo surgery for inguinal hernia.

Keywords: Mutation, Anemia, Gene mutation