Group (A) 11 patients (30–49 ys) and (B) 5 patients (51–70 ys). This analysis confirmed, better than BMI, an excess of fat mass (FM) in 64% of patients in A (7 on 1) and 40% in B (2 on 5). Excessive consumption of simple sugars, SFA results in an increase of 36% (4 on 7) in A and 80% (4 on 1) in B of blood LDL levels above the reference range (<100 mg/dL).

The HDL level is above the normality (x=83±20.3, >45 mg/dL) even though it is consumed by 7 on 16 AIP in low amounts with diet. Moreover, HDL correlates with age (r=0.56, p=0.02). Recent literature demonstrates that low and high HDL values have the same predisposition to mortality changing the paradigm that high HDL levels provide greater protection against cardiovascular diseases. This could suggest that high HDL may not play a completely protective role in AIP. In addition, by detecting BIA water distribution we found the ratio of extracellular water (ECW) to total body water (TBW) was above the normal range in 9 patients in A and 4 in B (v.n.0.36–0.4), indicating over-hydration in ECW. This data suggests inflammatory and stress status, as supported by the ECW/TBW ratio correlation with cortisol (r=0.67, p=0.008). In contrast, we did not find a correlation with ALA, PBG, or total porphyrins vs data on nutrition and BIA.

In conclusion, we confirm our hypothesis that AIP patients excessively consume simple sugars and SFA. This behaviour is reflected in their body composition and biochemical markers. This finding highlights the importance of providing nutritional support to prevent metabolic syndrome. Particular attention should be paid to a larger sample size cohort to assess the significance of high HDL and the TBW/ECW ratio as markers of disease progression based on age or stress.

Session 10 – Rational approach(es) to diagnosis and care in cutaneous porphyrias (September 25th, 2024)

Thematic area: diagnosis and care in porphyrias

04092 LIVER FUNCTION, IRON STATUS, AND HEMOPOIESIS IN ERYTHROPOIETIC PROTOPORPHYRIA (EPP): INSIGHTS INTO A COMPLEX INTERPLAY

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10.1136/bmjgast-2024-ICPP.24

Background Despite its dramatic cutaneous manifestations, EPP is a systemic disease with multi-organ involvement. In fact, protoporphyrin IX (PPIX) accumulates in multiple sites beside the skin, chiefly the bone marrow and the liver. Therefore, EPP patients display mild hematological alterations and are at risk of developing cholestatic liver disease, which may evolve to liver cirrhosis. Several patients show thrombocytopenia with increased spleen dimensions, which in the presence of hepatic alterations may raise the suspicion of portal hypertension.
Aim To characterize the phenotype of clinically stable EPP from a multi-system perspective.
Methods To test phenotypic correlations, 15 patients were enrolled: liver status, hematology and spleen parameters, and PPIX levels were collected at the same timepoint.
Results Increased spleen dimensions (SplD: bipolar diameter, BP; area-at-hilus, Ah) are common in EPP (7/15). Free PPIX levels predicted liver stiffness (LS, β=0.71, p=0.004), erythropoietin levels (β=0.63, p=0.020), SplD (BP: β=0.76, p=0.001; Ah: β=0.81, p<0.001), and platelet count (β=−0.60, p=0.017); SplD predicted platelet count (BP: β=−0.64, p=0.014; Ah: β=−0.82, p<0.001); when both SplD and PPIX levels were tested, platelet count was significantly correlated to SplD only (Ah: β=−0.95, p=0.008). Spleen stiffness was within normal ranges in all patients and not significantly associated with SplD. Instead, it showed significant correlations with hemoglobin (Hb, β=0.62, p=0.03), hypochromic red blood cell percentage (HypoRBC, β=−0.77, p=0.003), and ferritin (β=0.60, p=0.038). Hb was inversely related to HypoRBC, (β=−0.64, p=0.010). HypoRBC was inversely related to reticulocyte Hb content (β=−0.65, CHr, p=0.008) and mean corpuscular Hb (MCH, β=−0.51, p=0.047). Serum ferritin predicted the iron status of red blood cell lineage (HypoRBC: β=−0.64, p=0.010, CHr: β=0.72, p=0.002), but did not significantly reflect on Hb. Alterations in liver biochemistry were significantly associated with increased LS.
Discussion Taken together, these findings support the following phenotype model: in EPP, hypersplenism is likely an expression of compensatory extramedullary erythropoiesis, independent of protoporphyric hepatopathy; thrombocytopenia may be directly linked to increased spleen dimensions, rather than direct PPIX-driven impairment on platelet production; the erythropoietic drive is directly dependent on the disease activity; HypoRBC, MHC, and CHR could support a diagnosis of iron-restricted erythropoiesis better than serum ferritin or transferrin saturation.
Conclusion Although further studies are needed to confirm these findings, this is the first description of a complex multi-system interplay in protoporphyria. These findings could help better define the phenotype of EPP patients, particularly in the context of protoporphyric hepatopathy with spleen or hematological alterations.

Thematic area: recent advances in the pathophysiology of porphyrias

04112 QUANTITATIVE ANALYSIS OF POLY-UNSATURED FATTY ACIDS PROFILES IN ERYTHROPOIETIC PROTOPORPHYRIA

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10.1136/bmjgast-2024-ICPP.25

Background Erythropoietic Protoporphyria (EPP) is an inherited metabolic disorder due to ferrochelatase activity deficiency and it is characterized by the accumulation of protoporphyrin