demonstrated elevated total red blood cell porphyrin of 36.1 umol/L RBC (reference range <1.8) with normal levels of zinc protoporphyrin 1.3 umol/L RBC (reference range <1.6). Liver function tests were unremarkable. She was mildly anaemic, in keeping with MDS. A diagnosis of erythropoietic protoporphyria was made. She was able to cease prednisolone and antihistamines. We counselled her on the need to avoid sun exposure and implement strict sun protection strategies. Presently, she continues venetoclax monotherapy. A bone marrow transplant would potentially be curative for both MDS and EPP however, she has previously been deemed inappropriate due to her age.

While acquired EPP is a rare entity, our case exemplifies the need to screen for EPP in patients of any age where there is clinical suspicion, especially in patients with an underlying haematological disorder. Treatment of the abnormal bone marrow clone with chemotherapy temporarily reduces EPP symptoms.

Poster 59

04093 PORTAL HYPERTENSION IN ADVANCED PROTOPORPHYRIC HEPATOPATHY: A CASE REPORT

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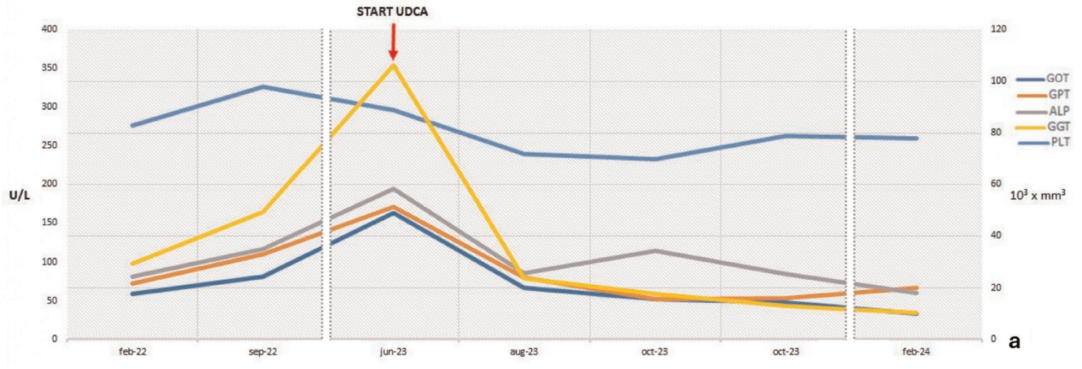


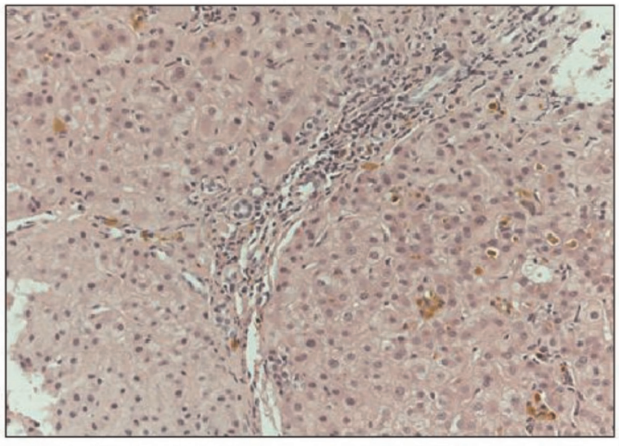
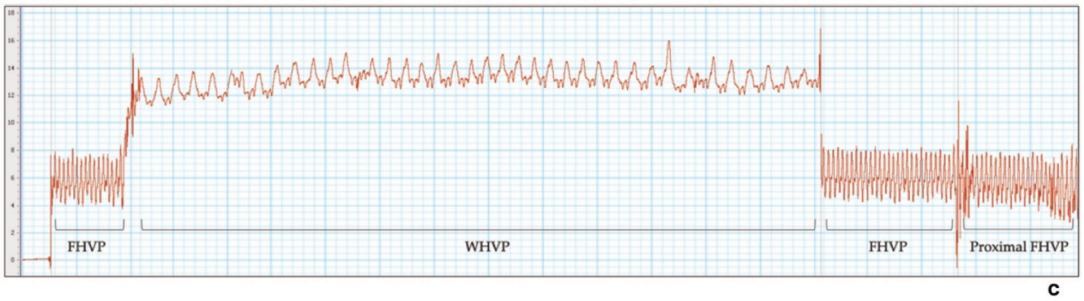


Figure 1. a) Follow-up in liver biochemistry in a patient with advanced protoporphyric hepatopathy. A dramatic improvement in liver biochemistry was noted after starting UDCA. *Please note that, for graphic purposes, GGT values have been halved.* b) Liver biopsy with fibrous septum and cholestasis (courtesy of Dr Pamela Sighinolfi, Pathology Department, Policlinico of Modena). c) Pressure waveform recorded during HVPG measurement (FHVP: 5 mmHg, WHVP: 12 mmHg). HVPG measurement of 7 mmHg indicates the presence of non-clinically significant sinusoidal portal hypertension. Permanent tracings were obtained with PowerLab (ADInstruments, Inc, Colorado Springs, CO).

Abbreviations: ALP, alkaline phosphatase; FHVP, free hepatic vein pressure; GOT, serum glutamic oxaloacetic transaminase; GPT serum glutamic pyruvic transaminase; GGT, gamma-glutamyl-transpeptidase; PLT, platelets. HVPG, Hepatic venous-portal gradient; WHVP, wedge hepatic vein pressure.



Abstract 04093 Figure 1

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Protoporphyric hepatopathy (PPHep) can be a dreaded, long-term complication of erythropoietic protoporphyria (EPP). In fact, PPHep may go unnoticed for years until advanced chronic liver disease (ACLD) develops, leading to cirrhosis and portal hypertension (PH). We present a case of a 31-year-old, lean (BMI 18 Kg/m2) Caucasian male EPP patient, who presented at our Centre with altered liver biochemistry parameters (figure1a) and normal liver function (MELD score 7, Child-Pugh score 5). He had microcytic anaemia, low platelet count (<80000/mm3), and marked splenomegaly (spleen bipolar diameter 15.9 cm, area 93.8 cm2). Although an abdominal ultrasound did not provide clear evidence of ACLD, a liver biopsy was performed, which revealed a picture of cholestatic cirrhosis with fibrous septa, slight chronic inflammation, and diffuse cholestasis, both intracanalicular and intraductal (figure 1b). A gastroscopy detected mild congestive gastropathy without varices. The patient underwent hepatic venous-portal gradient (HVPG) measurement, which revealed a gradient of 7 mmHg, indicating the presence of non-clinically significant sinusoidal portal hypertension (figure 1c). No evidence of vein-to-vein communications was found during venography, which could have led to an underestimation of the gradient. Liver and spleen stiffness were 17.6 kPa and 27.7 KPa, respectively, consistent with the HVPG and other findings. After 3 months of follow-up, during which therapy with ursodeoxycholic acid was started, a dramatic improvement in liver biochemistry abnormalities was observed (figure 1a) with a reduction in liver stiffness (11.6 KPa). Although a definitive confirmation of the type of PH would have required a direct puncture of the portal vein, this case provide indirect evidence that PH in PPHep appears to be strictly sinusoidal, unlike other cholestatic liver diseases such as primary biliary cholangitis.

Poster 61

04109 THE IMPACT OF ERYTHROPOIETIC PROTOPORPHYRIA (EPP) ON FAMILY QUALITY OF LIFE

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Erythropoietic protoporphyria (EPP) adversely affects the quality of life (QoL) of patients, due to the requirement for strict sun protection measures and debilitating pain during acute episodes, often limiting educational and career choices. Few data exist regarding the effect of EPP on the QoL of close relatives. There are no previous published reports of the FROM-16 or FDLQI in EPP families.

Close family members of n=252 adults with EPP in England and Wales were surveyed during summer 2023. Questionnaires included: the 16 item Family Reported Outcome Measure (FROM-16), utilised across medical and surgical specialties, comprising emotional and personal/social life domains (score 0–32). The Family Dermatology Life Quality Index (FDLQI) includes 10 items assessing the secondary impact of skin disease on the families of dermatology patients. FDLQI scores range from 0–30, higher scores indicating a greater impact on QoL.

The response rate was 29.4% (n=74), the mean age was 51.5 years (SD 15.8, range 20 to 91) and 52.7% were female. Spouses/civil partners represented 67.6% (n=50), parents 23% (n=17) and adult son or daughter 6.8% (n=5). The majority of respondents were White British (90.5%, n=67), with 5.4% (n=4) White other background, and 2.8% (n=2) Asian or Asian British Indian and Other ethnicity.

The mean total FDLQI score was 6.8 (SD 5.9, range 0–21). The highest impact was on recreation and leisure activities, social life and emotional distress. There was no significant gender difference in overall scores, however males reported having to do significantly more extra housework than females due to their relative's EPP (p=0.03).

A very or extremely large impact on QoL was seen in 13.6% (N=10) for the FROM-16 and a moderate impact was seen in 37.8% (N=28). There was no significant gender difference. Highest mean scores were for problems going on holiday, followed by adverse impact on family activities. The lowest impact reported in both tools was the effect of EPP on the family member's work and studying.

We have shown a previously undocumented, substantial impact of EPP on patients' family members. A greater understanding of the effect of EPP on patients' relatives may lead to improved holistic care of this rare, lifelong and severe photodermatosis.

Poster 62

04194 IN-DEPTH BIOCHEMICAL INVESTIGATIONS IN A NEW CASE OF ALAD DEFICIENCY: BETTER UNDERSTANDING FOR BETTER TREATMENT

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Introduction Delta-aminolevulinic acid deshydratase (ALAD) deficiency porphyria (ADP) or Doss porphyria is an autosomal recessive disease caused by a profound deficiency of the second enzyme in the heme biosynthesis pathway. ADP is the rarest porphyria with only eight documented cases. Different therapeutic strategies aimed at slowing down heme biosynthesis have been described with varying level of efficacy, suggesting a complex pathophysiology.

Clinical Description We report the case of a 3-year-old boy whose two unrelated parents came from the same village in Cape Verde. He was hospitalized in neuropediatric department for recurrent generalised tonic-clonic seizures in a viral

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