Sun, alcohol, and skin lesions

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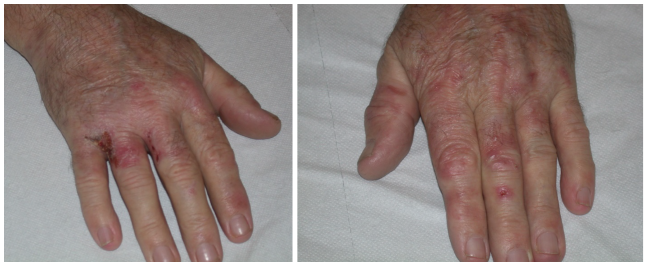

A 65-year-old man presented to the emergency room with blistered not itching skin lesions on the dorsal surface of both hands, which developed recurrently after exposure to sun and solved spontaneously with scarring. The patient had a history of hypertension, diabetes mellitus type 2 and hypercholesterolemia. His medications included ramipril, metformin, simvastatin, and acetylsalicylic acid. He denied smoke, but he used to consume a large amount of alcohol (1 Lt of red wine daily). His body mass index was 29 (overweight). Blood exams revealed: altered glucose metabolism (fasting and post-prandial blood glucose, respectively 172 mg/dL and 267 mg/dL), macrocytosis (mean cell volume 100.6 fL), increased transaminases (AST 62 U/L and ALT 79 U/L, normal value 10-37) and gamma-glutamyl transferase (GGT 255 U/L, normal value 7-40), and an iron assessment as follows: serum ferritin 2234 ng/mL, transferrin saturation 40%, serum iron 138 mcg/dL. Hepatitis C, hepatitis B, and HIV were excluded. Autoimmune screening was negative. Point of care ultrasound documented a grade 3 liver steatosis.

Question

Given the patient's history, what is the most likely diagnosis?

1. epidermolysis bullosa acquisita (EBA)
2. porphyria cutanea tarda (PCT)
3. cutaneous adverse drug reaction (CADR)
4. bullous systemic lupus erythematosus (BSLE)

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Availability of data and materials: all data underlying the findings are fully available upon reasonable request to Erika Poggiali, E.Poggiali@ausl.pc.it

Ethics approval and consent to participate: as this was a descriptive case report and data was collected without patient identifiers, ethics approval was not required under our hospital's Institutional Review Board guidelines.

Informed consent: the patient provided consent for the access to medical records at the time of admission.

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Answer

Our patient was diagnosed as having PCT and hereditary haemochromatosis according to his medical history, typical skin lesions, and supportive laboratory findings. Total urine porphyrins were elevated (6481 mcg/24 hours, normal value <150), particularly uroporphyrin. Molecular diagnostics revealed compound heterozygosity for the C282Y and H63D mutation within the hemochromatosis HFE gene. Phlebotomy was started as treatment regimen (450 cc/every 2 weeks) until iron depletion. The patient progressively reduced and stopped his alcohol intake. Sun exposure was restricted or allowed with total sunscreen. Full remission of skin lesions and liver damage was achieved after 6 months.

PCT is the most common type of porphyria worldwide It is a metabolic disorder of the heme biosynthesis pathway caused by decreased activity of hepatic uroporphyrinogen decarboxylase (UROD). This results in an accumulation of photosensitive by-products, such as uroporphyrinogen, which leads to the fragility and blistering of sun-exposed skin and liver damage. Most of the cases (80%) are acquired and strictly related to iron overload with HFE gene mutations,[1] alcohol abuse, viral infections (HCV, HIV), and use of cytochrome P-450 inhibitors or oestrogens. Familial PCT is rare (20%) and due to autosomal dominant UROD mutations.[2] Acquired PCT is more common in males after the age of 30, and it involves both the skin and the liver. Skin manifestations included increased fragility, erosions, bullae, milia, scars and keratosis on sun-exposed areas, particularly face and hands.[3] Liver damage might present in a wide range of ways from liver function test abnormalities to advanced fibrosis and hepatocellular carcinoma.[4] The toxic effect of iron plays a role in liver damage pathogenesis. Screening for hereditary haemochromatosis and HCC using ultrasound examination are always recommended in PCT patients, especially with cirrhosis and advanced fibrosis.[5] Phlebotomy is the first line treatment to be continued until ferritin concentration is less than 20 to 25 ng/mL. When phlebotomy is contraindicated, 100 mg hydroxychloroquine orally twice a week can be prescribed.[2] Broad-spectrum sun protection, alcohol cessation, and hepatitis C treatment must be always recommended to prevent disease progression and relapses.

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