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RESEARCH ARTICLE

"Chronic alcoholic with cutaneous blisters: Have a look at the urine"

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Abstract

Porphyria cutanea tarda (PCT) encompasses a heterogeneous group of disorders that can be acquired or inherited. It is usually characterised by photosensitivity and skin fragility over sun exposed parts and red coloured urine. We hereby report a 46-year-old chronic alcoholic who presented with blisters and itchy skin lesions over the sun exposed parts since 3 months. He had history of alcohol consumption for past 20 years. Cutaneous examination revealed few vesicles, crusting, hyperpigmentation and thickening of skin over sun exposed parts. Urine appeared red on gross examination and showed coral pink fluorescence under Wood's lamp. Biopsy taken from a vesicle revealed subepidermal blister with festooning of dermal papillae. DIF studies showed deposits of IgG at the dermo-epidermal junction and around blood vessels which were PAS positive. Based on the above findings a presumptive diagnosis of porphyria cutanea tarda was made. Any case presenting with cutaneous blisters along with history of chronic alcoholism should arouse the suspicion of Porphyria cutanea tarda. Gross examination of urine usually reveals a diagnostic clue in such cases which should be followed by a workup of porphyrin profile to rule out other acute porphyrias and other causes of photo-induced bullous dermatosis. Precipitating factors are invariably found in cases of PCT which should be thoroughly investigated as the avoidance of these factors play a major role in the management.

Introduction

Introduction:

Porphyria cutanea tarda (PCT) encompasses a heterogeneous group of disorders that can be acquired or inherited. It is caused by low levels of an enzyme (uroporphyrinogen decarboxylase or UROD) involved in heme synthesis. It is characterised by photosensitivity, skin fragility over sun exposed parts and red coloured urine. It is usually precipitated by certain factors like alcoholism, intake of aromatic polyhalogenated hydrocarbons, iron, and infection by hepatitis B, hepatitis C and Human Immunodeficiency (HIV) virus.

Case report:

A 46-year-old man presented with blisters and itchy skin lesions over the sun exposed parts since three months. He had similar skin lesions since the last 3 years. He also had history of photosensitivity and blisters which were aggravated during summer season. The patient had history of alcohol consumption (approximately 180ml daily) for the past 20 years. There was no history of similar complaints in the family members. Cutaneous examination revealed few vesicles, crusted erosions, hyperpigmentation and skin thickening over the face, extensors of forearms and dorsa of hands (Figure 1 and 2).

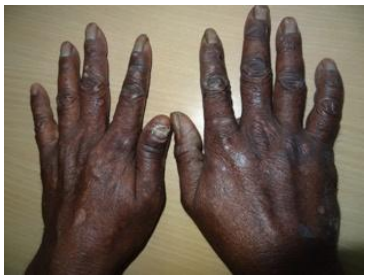

Fig 1 showing crusted vesicles, crusted erosions and scarring over the dorsa of hands.

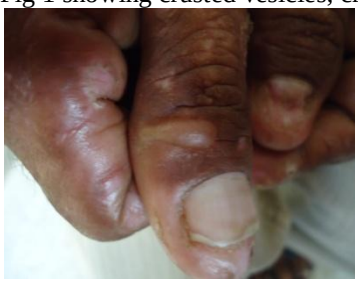

Fig 2 showing vesicles on the fingers.

Nails showed dystrophic changes. There was no hypertrichosis and milia. Routine investigations including ultrasound whole abdomen were normal except for liver function tests which showed elevated levels of alanine transaminase and aspartate transaminase. Urine appeared red on gross examination and showed coral pink fluorescence under Wood's lamp (Figure 3).

Fig 3 showing coral red fluorescence of patient's urine sample compared with control sample of urine on Wood's lamp examination

Histopathological examination of biopsy taken from a vesicle on the dorsum of right hand revealed subepidermal blister with festooning of dermal papillae (Figure 4).

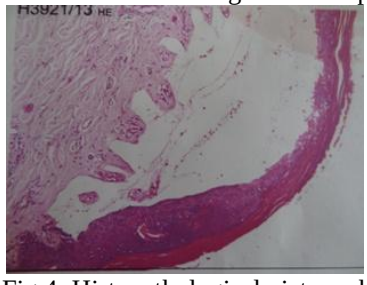

Fig 4: Histopathological picture showing subepidermal blister with festooning of dermal papillae.

Periodic Acid Schiff (PAS) stain showed the subepidermal bulla and deposition of PAS positive material around the blood vessels in the dermis. DIF studies showed deposits of IgG at the dermo-epidermal junction (Figure 5).

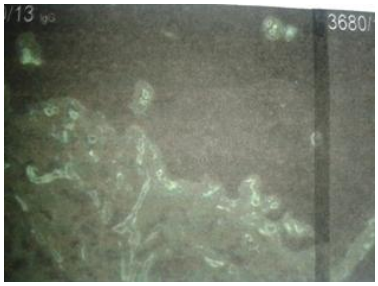

Fig 5: DIF studies showed deposits of IgG at the dermo-epidermal junction.
Based on the above findings, a presumptive diagnosis of porphyria cutanea tarda was made.

Discussion:

The porphyrias are a group of infrequent metabolic diseases caused by partial deficiencies of the activity of seven sequentially acting enzymes in the biosynthetic pathway of heme. They are inherited diseases with the exception of sporadic PCT (Type I). PCT is the result of inactivation of hepatic UROD enzyme.[1-2]

Symptoms are manifested by skin fragility with erosions appearing with minimal trauma and bullous photo-induced lesions. These lesions evolve to scarring and slowly progressive skin thickening with mutilating lesions especially on acral areas such as fingers, nose and ears. Some patients also present with hypertrichosis and milia on the face.[2]

Gross examination of urine may show reddish discoloration and Wood's lamp examination will show coral pink fluorescence. Porphyrins in their oxidized forms are reddish in colour and are also fluorescent. When exposed to light at certain wavelengths, they emit light with a different wavelength. This makes them visible with a Wood's lamp, and enables them to be measured accurately with a spectrofluorometer.[2-4] Accumulation of uroporphyrin and protoporphyrin in tissues allows production of phototoxic reaction upon light exposure. This phototoxicity is the basis of cutaneous lesions through the generation of reactive oxygen species and lipid peroxidation which leads to degranulation of mastocytes, liberation of inflammatory mediators, complement activation and increased collagen synthesis.[5-6]

Precipitating factors include alcoholism, intake of aromatic polyhalogenated hydrocarbons, iron, and infection by hepatitis B, hepatitis C and HIV which were excluded in our case except alcohol consumption.[1]

Role of alcohol in PCT:

Acquired PCT (type 1) can arise due to alcohol consumption which could be the possible inducing factor in our case. It is thought to do so by causing oxidative damage to liver cells, resulting in oxidized species of uroporphyrinogen that inhibit the activity of hepatic UROD. Alcohol induces the first and rate-limiting enzyme in the pathway, delta-aminolevulinic acid synthase which causes the accumulation of the porphyrin products. So abstinence from alcohol is a therapeutically and prophylactically important measure in PCT.[2]

Differential diagnosis includes other porphyrias, pseudoporphyria, epidermolysis bullosa acquisita, phototoxic and bullous drug eruptions. Detailed history and investigative profile helps in ruling out these diseases and confirming the diagnosis of porphyria cutanea tarda.[2,7]

Diagnostic workup includes complete blood count, liver and kidney function tests, blood sugar, iron profile, screening for hepatitis and HIV viruses, ultrasound abdomen, porphyrin profile, histopathology and direct immunofluoresence study of skin lesions, analysis of enzyme levels involved in the heme synthesis and DNA analysis for mutations. Treatment measures include phlebotomy, low dose chloroquine, desferrioxamine, erythropoietin, plasmapheresis and plasma exchange. Intake of iron supplementation and aromated polycyclic hydrocarbons should be avoided.[2,8,9]

Conclusion:

Any case presenting with cutaneous blisters along with history of chronic alcoholism should arouse suspicion of porphyria cutanea tarda. Gross examination of urine usually reveals a diagnostic clue in such cases which should be followed by a workup of porphyrin profile to rule out other acute porphyrias and other photo-induced bullous dermatosis. Precipitating factors are invariably found in cases of PCT which should be thoroughly investigated and managed accordingly which plays a major role in the management.

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