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ERYTHROPOIETIC PROTOPORPHYRIA (REPORT OF THREE CASES)

M Shafi, M L Khatri

Three Libyan patients developing clinical manifestations of erythropoietic protoporphyria (EPP) at 5 years, 7 years and 3 years of age are reported. All of them had variable degree of photosensitivity leading to pitted scarring. Red blood cells in peripheral blood smear revealed positive pink fluorescence in all. First patient responded well with chloroquine therapy and the 3rd patient showed improvement with oral beta-carotene therapy, but the second patient did not show significant response to either.

Key Words : Erythropoietic protoporphyria, Porphyria, Beta-carotene

Introduction

Erythropoietic protoporphyria (EPP) is a genetic disorder of porphyrin metabolism due to decreased activity of ferrochelatase, resulting in increased levels of protoporphyrin in red blood cells, plasma and stools. It is clinically manifested as variable degree of photosensitivity developing in childhood.[1]

We have seen 3 Libyan cases of EPP in last 10 years. The clinical features of whom are summarized in this report.

Case Report

Case I : A male petient, 25 years, presented with recurrent erythematous maculopapular and vesicular lesions with burning sensation and moderate itching on the cheeks, since the age of 5 years. One of his brothers also had similar complaints but in milder form (not seen by us). At the time of assessment he had diffuse erythema of both cheeks with small pitted scars in bewteen.

Case II : This boy aged 11 years presented with 4 years history of intense burning sensation over face and dorsum of hands on exposure to sun. He had significant erythema over cheeks, forehead and dorsum of hands. Small areas of atrophy were also present on the cheeks and bridge of nose. Later he developed marked atrophy on these sites and also developed multiple papular lesions with waxy appearance on the dorsum of hands. Moderate cheilitis was also observed during summer months.

Case III : A male patient aged 28 years came with complaints of having recurrent erythematous, urticarial, vesicular and crusted lesions on the face and dorsum of hands, since the age of 3 years, more severe during summer. At the time of evaluation, he was having multiple pitted rounded and irregular scars and diffuse hyperpigmentation on nose, cheeks and ears (Fig. 1). There were multiple small hyperkeratotic smooth and rough closely set papular lesions with atrophy in between on the dorsum of the hands (Fig. 2).

All the patients were investigated thoroughly. The results of complete blood counts, ESR, liver function tests, blood urea, creatinine, electrolytes, sugar, serum protein electrophoresis, rheumatoid factor

From the Department of Dermatology, Central Hosptial and Faculty of Medicine, Al-Fateh University of Medical Science, Tripoli, Libya.

Address correspondence to : Dr M L Khatri, PO Box 13457, Tripoli.

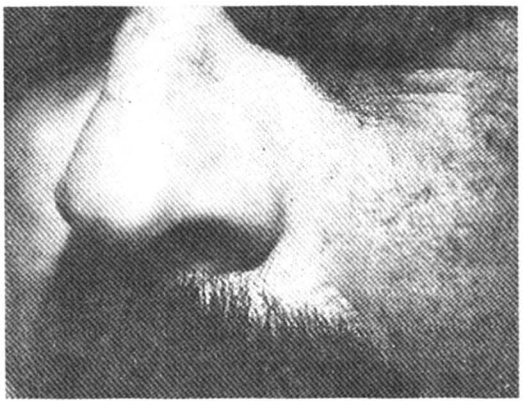

Fig .1. Case III : Multiple pitted rounded and irregular scars and diffuse hyperpigmentation.

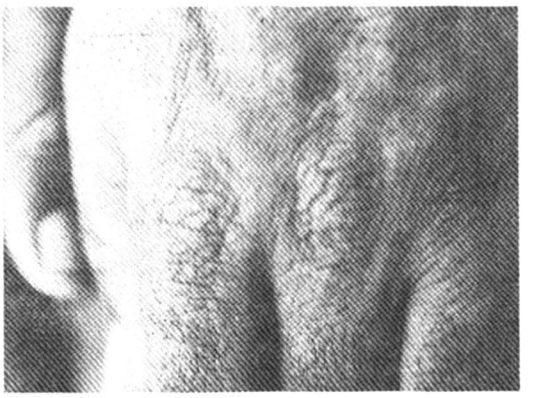

Fig .2. Case III : Multiple small hyperkeratotic smooth and rough clsoely set papular lesions with atrophy in between.

and LE cell phenomenon did not reveal any abnormality. The routine urine and stool tests also did not show any abnormality. Urine test for porphyrin were also negative in all the patients.

Examination of peripheral blood smear under fluorescent microscope showed intense pink fluorescence in majority of red blood cells in all the patients. Elder brother of patient No. II who did not have any symptoms also showed such a fluorescence.

Biopsy in all the cases showed deposition of hyalin material around the blood vessels in the dermis. In cases II and III thickened collagen bundles were also seen.

Although the parents were not affected by EPP, a history of consanguinity was present in all. One brother of patient I had similar clinical features and one brother of patient III showed positive fluorescence of red blood cells without clinical manifestations.

Treatment and follow-up

All the patients were prescribed sunscreens, mainly 5% zinc oxide cream and were advised to avoid exposure to sun light. All the patients were initially given chloroquine therapy, 150 mg twice daily (first week), once daily (second week) and later on alternate days. The patient I showed good response and symptoms almost sudsided within 6 weeks. On discontinuation of chloroquine therapy symptoms started reappearing after 3 weeks. This patient was given interrupted courses of chloroquine during summer and showed reasonably good control for 2 years. Later this patient was lost to follow-up. The patients II and III did not show any significant improvement with chloroquine therapy, hence these patients were given beta-carotene 120 mg/day. The patient III showed significant improvement after a month's therapy. Later he was given interrupted courses of beta-carotene during summer months only. This patient is under control. Patient II did not show improvement even after 3 months therapy with beta-carotene, so it was discontinued and he was kept on sunscreening creams only. In this patient the lesions are progressing fast resulting in severe scarring.

Discussion

In initial genetic study it was concluded

that EPP is transmitted by an autosomal dominant gene.[1,2] Later workers[3] hypothesized more complex mechanisms of inheritance. Reed et al[4] suggested that the mode of inheritance is autosomal dominant with variable penetrance. One homozygous case of EPP has been reported.[5] None of the parents of our patients had clinical manifestations of EPP but their marriages were consanguineous.

The clinical features of our patients were almost similar to the previously reported cases.[1] All of our patients developed pitted scars, while DeLeo et al1 reported scarring only in 19% of their patients. Even after repeated investigation on follow-up, we could not reveal any systemic involvement. De Leo et al[1] reported anaemia in 7, cholelithiasis in 4 and abnormal liver function studies in one patient out of their 32 patients.

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