Scleritis in congenital erythropoietic porphyria – infective or inflammatory?

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Key words: Congenital erythropoietic porphyria, necrotizing scleritis, scleritis

Congenital erythropoietic porphyria (CEP) is an autosomal recessive disorder with deficiency of uroporphyrinogen III cosynthase. Accumulation of uroporphyrin I is responsible for the oxygen-dependent phototoxic dermatological, skeletal, visceral, and ocular damage.[1,2] The dermatological changes include skin ulceration and extensive mutilation in the sun exposed area.

Ocular complications of CEP include nodular scleritis, necrotizing scleritis with inflammation or scleromalacia perforans. Necrotizing scleritis and scleromalacia perforans are among the most commonly reported ocular complications.[3,4]

In our case, the patient had visual acuity of 6/12 in both eyes with scleritis, scleral thinning, and scleral calcific plaques evident on slit lamp examination [Figs. 1a and b; Fig. 2a and b]. On systemic examination, patient had hypertrichosis, hyperpigmentation, depigmentation, fibrosis, and mutilation of ear, nose, and fingers [Fig. 3]. Dental examination revealed brownish discoloration of teeth. Examination of hand and foot revealed acromiria, contracture of fingers, and atrophy of phalanges [Figs. 4 and 5]. Although the history and clinical findings suggested a possible diagnosis of scleritis with inflammation, the diagnosis was still missed by the primary treating physician. CEP can be associated with devastating ocular complications. Necrotizing scleritis is one such complication, which if managed in time can avoid blinding outcomes as highlighted by dramatic healing in our case with topical prednisolone acetate, moxifloxacin, and lubricants at 1-week follow-up [Fig. 1c and d; Fig. 2c and d].

This case highlights the importance of awareness about the ocular complications in CEP that every general practitioner must be acquainted with and the fact that such a presentation should not be confused with an infective etiology.

Conclusion

CEP is a rare disorder but can be associated with serious ocular complications, such as scleral melting and blindness. The treating physician must be aware of the ocular complications of this entity to ensure timely referral to an ophthalmologist for accurate diagnosis and prompt treatment.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients

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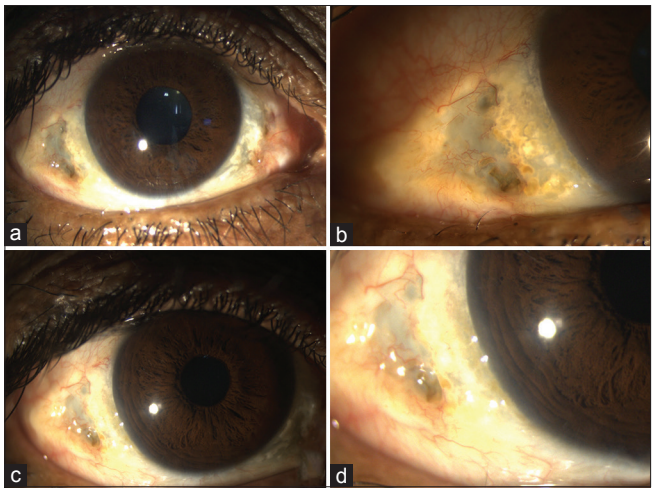

Figure 1: (a) Slit lamp photograph of the right eye showing scleral necrosis in the inter-palpebral area; (b) Slit lamp photograph of temporal area of scleral necrosis with thinning; (c and d) Slit lamp photograph of the right eye at 6 weeks follow-up showing decrease in sclera necrosis and thinning

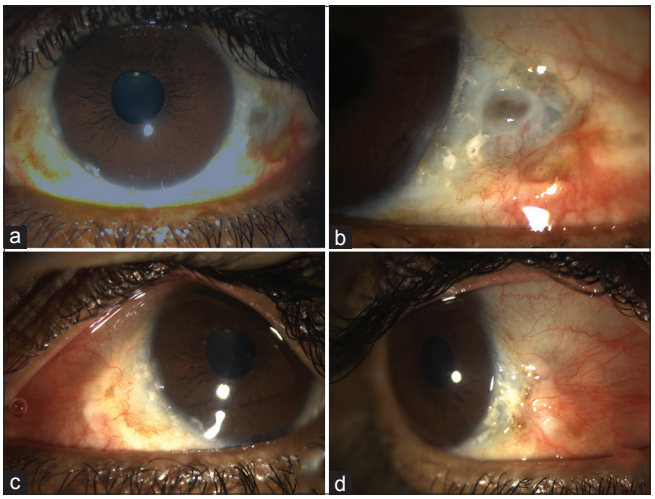

Figure 2: (a) Slit lamp photograph of the left eye showing sclera necrosis in the inter-palpebral area; (b) Slit lamp photograph of temporal area of sclera necrosis with thinning; (c and d) Slit lamp photograph of the left eye at 6 weeks follow-up showing decrease in sclera necrosis and thinning

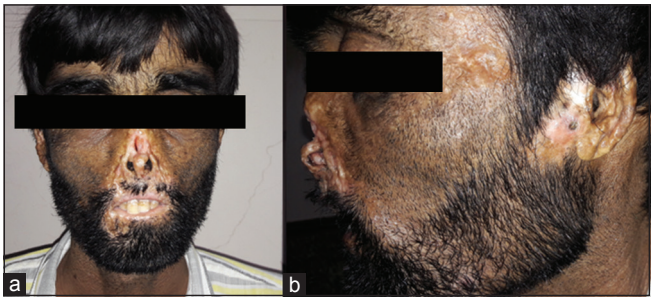

Figure 3: (a) Face photograph of the subject with evident hypertrichosis and mutilation of the nose; (b) Face photograph of the subject with evident mutilation of the ear lobule, skin hyperpigmentation, and depigmentation

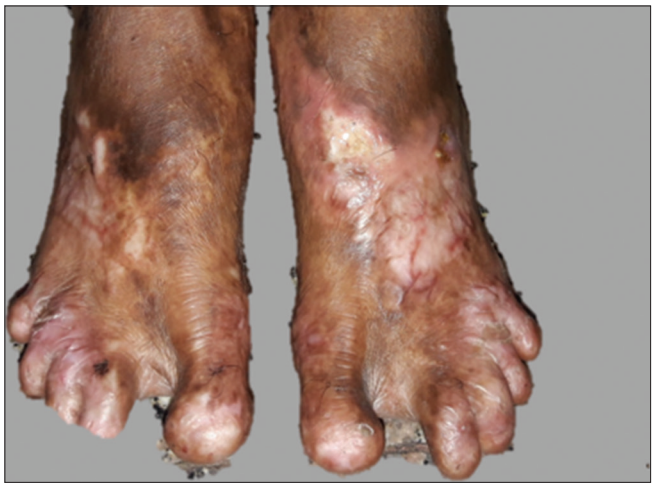

Figure 5: Clinical photograph of both foot showing hyperpigmentation and depigmentation of skin with resorption of phalanges

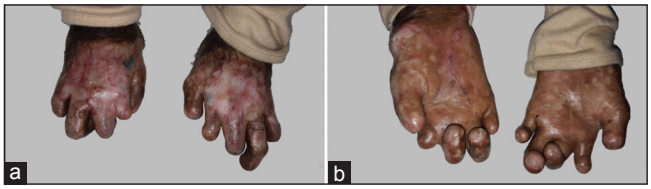

Figure 4: Clinical photograph of both hands, (a) back surface, (b) front surface showing acromicria, hyperpigmentation, and depigmentation of skin, contracture of fingers, and resorption of phalanges

understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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