CASE REPORT

Ocular manifestations in porphyria cutanea tarda

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SUMMARY

A 24-year-old man presented with pain, sticky discharge and loss of vision in the right eye. He has had typical skin manifestations of porphyria cutanea tarda (PCT) since 6 years and ophthalmological symptom for 6 weeks. On ophthalmological examination, visual acuity was light perception in the right eye and 6/12 in the left. There were bilateral, symmetrical temporal scleromalacia along with temporal corneal melting in both eyes and perforation in the right eye. Ultrasonography B-scan (USG B-scan) revealed a retinal detachment in the right eye. Artificial tear instillation was started every hour along with topical antibiotic coverage in both eyes. Additionally, ultraviolet protective sunglasses and hat for photo-protection was advised. The vision in the right eye improved to 5/60 along with subsidence of retinal detachment on repeat USG B-scan after 3 weeks.

BACKGROUND

Sight-threatening ocular manifestations are rare in porphyria cutanea tarda (PCT). There are very few case reports highlighting ocular problems and their management in patients with PCT. We describe a case of PCT presenting with corneal perforation, scleromalacia perforans and retinal detachment. A

PubMed search revealed very few case reports about ocular complications of PCT from around the world and just one has been reported previously from the Indian subcontinent.

CASE PRESENTATION

A 24-year-old man presented with multiple blisters and pigmented, slightly depressed scars on the skin, especially in the sun-exposed parts such as face, hands and legs. The face showed melasma-like hyperpigmented patches and hypopigmented scars ([figure 1](#)). Indurated, waxy, yellowish plaques that resemble lesions of scleroderma were seen in pre-auricular and temporal area along with patches of growth of lanugo type of hair in these regions suggestive of hypertrichosis ([figure 2](#)). Digital shortening, atrophy and contractures were seen in hands, legs and the pinna ([figure 3](#)).

Ocular symptoms were those of pain, sticky discharge and loss of vision in the right eye since 6 weeks. On examination, visual acuity was perception of light in the right eye and 6/12 in the left. Lids of both eyes showed hypopigmented patches, milia-like lesions, erythema, scarring with tylosis and matting of eyelashes ([figure 4](#)). Severe conjunctival chemosis and congestion was seen in the right

Figure 1 Hands and front of the face showing skin changes.

Figure 2 Hands and side face showing skin changes.

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Figure 3 Hands and feet showing digital shortening and scarring.

eye. There were bilateral, symmetrical areas of punched out scleral thinning with uveal show temporally in the interpalpebral fissures in both eyes ([figure 5](#)). In the right eye, sclera was inflamed with a corneal melt, perforation and irregular anterior chamber depth. Fundus examination was not possible in the right eye due to corneal haze and was normal in the left eye.

According to the history, skin manifestations started at the age of 6 years in the form of hyperpigmented patches which gradually increased in size and severity. Gradually, other skin manifestations started showing too along with digital atrophy and loss. Owing to the disfigurement and social awkwardness, patient left school at 14 years of age and started working in his farm; subsequent to which his skin problems aggravated. He is not an alcoholic and there is no history of any drug abuse. Moreover there was no positive family history.

Urine examination showed markedly raised porphyrin levels and since there was no hepatosplenomegaly, a probable diagnosis of PCT was made.

INVESTIGATIONS

Ultrasonography B-scan (USG B-Scan) of the right eye showed retinal detachment most probably exudative secondary to the overlying scleral necrosis ([figure 6](#)). Routine urine examination

Figure 4 Eyelids showing melasma-like lesions, matting of lashes and hypopigmented patches.

Figure 5 Bilateral temporal scleral craters along with severe congestion and corneal involvement in the right eye before treatment.

revealed maroon-coloured urine which showed bright pink fluorescence under Wood's light after acidification with hydrochloric acid. It also showed a markedly raised porphyrin level (18 050 nmol/L). Determination of urine porphyrins by high-performance liquid chromatography showed marked increase in uroporphyrins and 7-carboxyl porphyrin levels. Plasma showed only trace amounts of porphyrins. Serum iron was 70 µg/dL and ferritin was 1980 ng/L. Serum transaminase and γ-glutamyl transpeptidase levels were also raised. The patient was HIV, hepatitis-B virus (HBV) and HCV negative.

DIFFERENTIAL DIAGNOSIS

Markedly raised porphyrin levels in urine with just trace amounts in plasma and absence of hepatosplenomegaly helped us to differentiate it from other forms of porphyria namely variegate porphyria and hereditary coproporphyria.

Moreover, onset of skin lesions after 5 years of age, absence of telangiectasia in sun-exposed areas, no waning of skin lesions during winter and chronic nature of photosensitivity helped us differentiate from xeroderma pigmentosa.

The presence of severe skin photosensitivity manifesting in childhood along with blistering, scarring, hypertrichosis and pigment changes, closely resembling congenital erythropoietic porphyria pointed to the clinical diagnosis of hepatoerythropoietic porphyria (HEP) that is the homozygous form of familial PCT. Confirmation of clinical diagnosis of HEP by molecular studies that demonstrate mutations affecting both uroporphyrinogen decarboxylase (UROD) alleles could not be conducted due to financial and facilities constraints.

TREATMENT

The patient was advised instillation of tear substitutes in both eyes every hour during the day and gel at night before sleeping along with antibiotic coverage. Steroid eye drops were avoided

Figure 6 Ultrasonography B-scan showing retinal detachment.

Figure 7 Healing scleral melt patches of both eyes after treatment.

in view of the scleral melt. Moreover, effective photoprotection in the form of good ultraviolet (UV) protective sunglasses and hat along with avoidance of outdoor activities in bright sunlight was advised. In consultation with a dermatologist, low-dose chloroquine therapy (125 mg chloroquine phosphate twice a week) was started for skin lesions.

OUTCOME AND FOLLOW-UP

There was drastic improvement in the condition of the scleral melt of both eyes within 3 weeks of starting the aforementioned simple but rational treatment (figure 7). There was marked reduction in pain and congestion of the right eye. Vision in the right eye improved to 5/60. Repeat USG B-Scan of the right eye showed subsidence of retinal detachment (figure 8).

DISCUSSION

Porphyrias are a group of disorders of heme-metabolism with deficiency in the enzymes of the hemebiosynthetic pathway resulting in excess porphyrin production. Of the porphyrias, PCT is a hepatic porphyria characterised by deficient UROD activity and can be either sporadic (type 1) or familial (type 2).¹ HEP is the homozygous or compound heterozygous form of familial PCT characterised by markedly reduced activity of hepatic UROD to usually less than 20% of normal as measured in erythrocytes. Patients are either homozygous for one UROD mutation or compound heterozygous for a different UROD mutation inherited from each parent.

Blisters on the skin that are followed by erosions, crust formation and scarring are the major clinical feature.² They mainly occur in the sun-exposed and trauma-prone areas such as forearms, face, legs and feet. Skin friability and small white papules

Figure 8 Post-treatment ultrasonography B-scan showing subsidence of retinal detachment.

termed milia are common, especially on the back of the hands and fingers. Other skin changes include diffuse hyperpigmentation, sclerodermiform plaques, with scarring and calcification resembling systemic sclerosis. Digital shortening and atrophy are very rare.

Ocular manifestations are caused by accumulation of photoactive porphyrins in the ocular tissues.^{3–5} External findings include ectropion, symblepharon, eyelid ulcerations and depigmentation. Anterior segment ocular stigmata include scleromalacia, conjunctival and scleral necrosis with vesicle formation and corneal scarring, ulceration and vascularisation.⁶ Scleral involvement is seen in the form of painless, bilateral, symmetric areas of scleral thinning without surrounding inflammation, with a bluish uveal hue in the base and calcareous degeneration in the adjacent area.^{6–8} These areas of scleromalacia perforans are usually found in the sun-exposed interpalpebral fissure. The adjacent cornea may become secondarily involved with corneal opacification, thinning, melt or perforation. The corneal changes seen can be attributed first, to the conjunctival scarring causing a disturbance of the mucin layer of the tear film which in turn leads to a dry eye syndrome. Second, the dry eye syndrome is further aggravated by severely scarred eyelids with incomplete lid closure leading to lagophthalmos and exposure.⁹ Posterior segment involvements include retinal haemorrhages, cotton wool exudates, papilloedema and choroiditis with pigment migrations.¹⁰

Besides wearing UV-protective glasses and hat, frequent application of artificial teardrops produces a mechanical effect of washing out the accumulating porphyrins on the ocular surface and may improve healing of scleral necrosis. Scleritis is treated with oral steroids, frequent instillation of topical lubricants and prevention of exposure to sunlight. Advanced cases with scleral perforation require surgical intervention in the form of scleral patch graft.

Learning points

- ▶ Ophthalmic disease in the porphyrias is probably under-recognised.
- ▶ Dermatologists should be aware that patients with porphyria cutanea tarda (PCT) can have severe ophthalmic problems which need prompt detection and urgent intervention to save eyesight.
- ▶ Ophthalmologists see patients with porphyria rarely but should consider the diagnosis in cases of photosensitive ocular disease.

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