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Sir,

Sight threatening complications in porphyria cutanea tarda

The porphyrias are a group of disorders of haem metabolism with deficiency in the enzymes of the haem biosynthetic pathway resulting in excess porphyrin production.[1] Porphyria cutanea tarda (PCT) is the most common of eight subtypes with a predilection among black Southern Africans.[1] Clinical manifestations are predominantly dermatological with photoactive porphyrins depositing in the skin causing bullae, hyper- and hypo-pigmentation, pseudoscleroderma, and hypertrichosis in s un-exposed areas.[2] Sight-threatening ocular manifestations are rare and we describe a case of PCT presenting with corneal perforation and scleromalacia perforans.

A 54-year-old black female presented with a 6-week history of pain and loss of vision in the right eye, and progressive darkening and coarseness of her skin.

Examination revealed dark skin and sclerodermatous-like facial features (Figure 1). Visual acuity was hand movements right and 6/5 left. There were bilateral, symmetrical areas of punched out scleral thinning with choroidal show temporally in the interpalpebral fissures. The sclera was moderately inflamed in the right with a thin cornea, central perforation, and flat anterior chamber (Figure 2).

She had a tender hepatomegally and no systemic features suggestive of collagen-vascular disease.

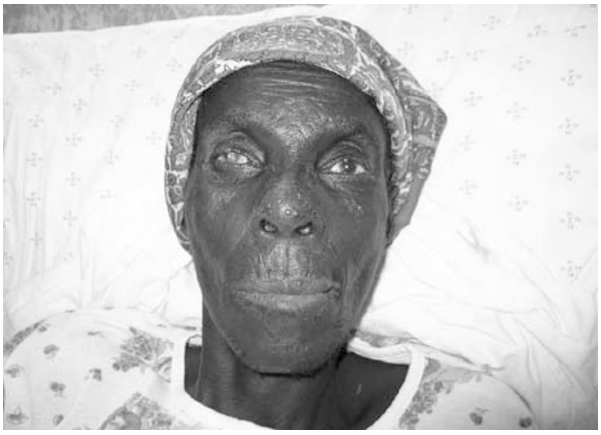

Figure 1 Sclerodermatous features in porphyria cutanea tardia.

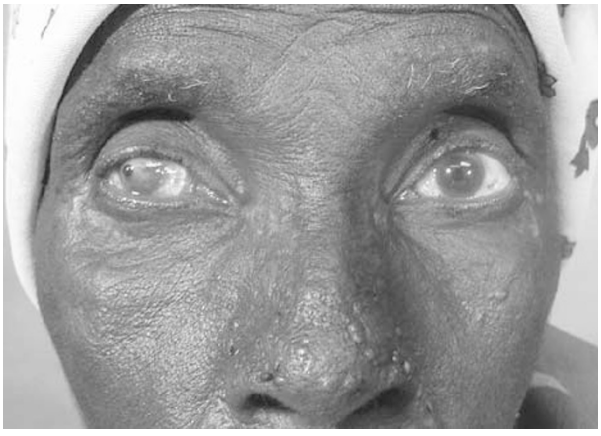

Figure 2 Perforated right cornea and bilateral scleromalacia.

Urine porphyrins were 14 035 nmol/l, serum iron 58.3 μmol and ferritin 1790 ng/l confirming a diagnosis of PCT and iron overload.

Treatment included oral prednisolone 60 mg daily and cyclosporine 2% drops QDS, avoiding topical steroids because of the risk of scleral perforation. Oral steroids were tapered and topical cyclosporine and lubricants continued. There has been steady improvement in scleral thickness and no sign of disease progression. The right eye is comfortable with a sclerosed cornea. She has undergone serial phlebotomy and is avoiding alcohol and sun exposure.

PCT is a hepatic porphyria characterised by deficient uroporphyrinogen decarboxylase activity and may be autosomal dominantly inherited or may occur sporadically.[1] The predilection in black Southern Africans may be due to the increased incidence of haemochromatosis caused by ingestion of traditional tribal beer brewed in iron pots.[1] Hereditary PCT is characterised by enzyme deficiency in all tissues while in acquired PCT, the deficiency is isolated to the liver and

may be precipitated by external factors like iron overload, alcohol, and oestrogens.

Ocular manifestations are caused by deposition of photoactive porphyrins in ocular tissues with lid scarring, ectropion, lacrimal scarring, scleromalacia, and corneal thinning described.[3–7]

Treatment involves avoidance of precipitating factors and UV exposure, while success has been achieved with phlebotomy and iron chelating agents.

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Sir,

Segmental neurofibromatosis

Neurofibromatosis (NF) is classified under phakomatoses, which are a group of disorders where neurological abnormalities are combined with congenital defects of skin, retina, and other organs. Localised orbital neurofibromas are discrete, space-occupying lesions within the orbit and may be difficult to differentiate from other orbital tumours; multiple tumours can occur. We report here a case of a patient with orbital neurofibromas in a segmental pattern unassociated with systemic von Recklinghausen's disease.

Case report

A 52-year-old Caucasian woman was referred to the orbital eye clinic for management of a 10-year history of exophthalmos of her left eye (Figure 1). This was not associated with any diplopia in primary position or painful eye movements.

Examination revealed a systemically healthy woman, with no signs of thyroid status abnormality. The best-corrected visual acuities were right 6/6 and left 6/5. Palpation revealed a soft tender mass in the superior nasal region of the left orbit producing a 5 mm proptosis and 10 mm downward displacement. Extraocular movement examination showed a restriction of up-gaze of the left eye especially in the adducted position. Slit-lamp biomicroscopy showed normal anterior segments; no signs of optic nerve compromise were seen and fundus examination was normal.

Orbital magnetic resonance imaging demonstrated three discrete tumours in the left orbit (Figure 2). This prompted the diagnosis of neurofibromas and the patient was examined and found to have no stigmata or family history of NF 1 or NF 2.

The two more anterior masses were successfully removed, while removal of the more deeply seated tumour was abandoned owing to the close proximity to the neurovascular bundle in the inferior orbital

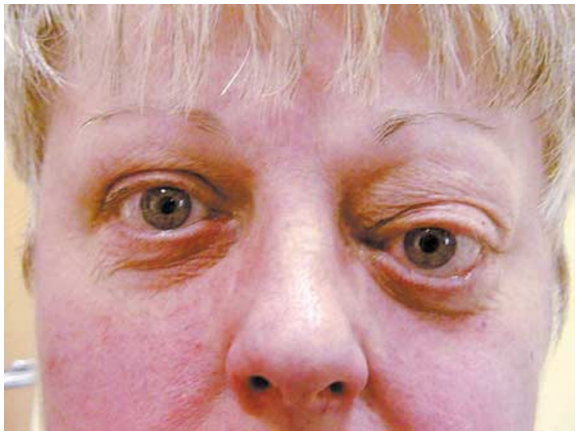

Figure 1 Clinical photograph showing exophthalmos and downward displacement of the left eye.