

Retinitis pigmentosa and Coats's disease

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The association of two rare ocular conditions of unknown aetiology presents the clinician with an opportunity to study interrelating factors. The association of retinitis pigmentosa with Coats's disease has been previously reported in three patients (Zamorani, 1956; Morgan and Crawford, 1968), in all of whom bilateral involvement occurred with severe visual loss.

The present case which has been fully documented demonstrates the association of bilateral retinitis pigmentosa with retinal telangiectasia (Coats's disease) and drusen of the optic disc.

Case report

A 15-year-old girl was seen at the Medical Ophthalmology Unit in July 1973 with a 10-year history of impaired vision and recent further deterioration.

HISTORY

The patient's birth had been normal although two weeks premature and her early development had also been normal. At the age of five years she was found to be deaf in her right ear and her parents also noticed she had poor vision in dim light. In July 1970 at the age of 12 years she was admitted to the Medical Ophthalmology Unit. Visual acuity was 5/60 and N8 in the right eye and 6/18 and N8 in the left; colour vision was reduced in both eyes. Peripheral visual fields were markedly constricted.

Fundus examination showed exposed drusen of the right disc and buried drusen of the left, and a widespread pigment epithelial disturbance with a few clumps of intraretinal pigment deposition. The choroidal vessels were visible and a serous detachment was present inferiorly in the right eye.

There was no family history of retinitis pigmentosa or consanguinity although electrodiagnostic tests performed on her two younger sisters showed a slightly subnormal light rise in the electro-oculogram (light rise right eye 228 per cent, left eye 210 per cent in the 12-year-old sister; right eye 243 per cent, left eye 200 per cent in the 10-year-old sister).

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OCULAR FINDINGS

In July 1973 the visual acuity in the right eye was 5/60 and N8 and in the left 6/60 and N8 with correction. The peripheral visual fields showed marked constriction to 5° from fixation on each side, and the central fields showed severe loss of visual function including severely diminished macular thresholds (Friedmann analyser). Fundus examination showed exposed drusen of the right optic disc and widespread degeneration of the retina and pigment epithelium including the macular region. There were bilateral inferior serous detachments with dilated and tortuous retinal vessels. Fluorescein angiography (Fig. 1) demonstrated good filling of the retinal arterioles which were of normal calibre, and the inferior vessels appeared relatively larger than the superior vessels. Choroidal fluorescence was visible through the deficient pigment epithelium. The retinal vessels in the inferior fundus on both sides were dilated and tortuous particularly in the periphery, with capillary dilatation and microaneurysm formation. Extensive retinal and subretinal leakage of dye was visible (Fig. 2).

Electrodiagnostic studies

Both the scotopic and photopic electroretinograms were almost totally extinguished with the highest intensity evoking only a very small response. The electro-oculogram was flat in both eyes. Laboratory data including plasma electrolytes, plasma proteins, Wassermann reaction and Kahn's test were all normal.

Discussion

The association of retinitis pigmentosa and Coats's disease was first described by Zamorani (1956) in a 16-year-old girl. Zamorani's patient had a fully developed cataract in the eye so that fundus changes were visible on only one side, and drusen of the disc was not observed. Electrodiagnostic tests were not performed.

Two further cases were described by Morgan and Crawford (1968) with typical pigment epithelial degeneration and retinal arteriolar narrowing. The inferior vessels were dilated and irregular and there were inferior serous detachments. Electroretinograms were extinguished and fluorescein angiography was not performed.

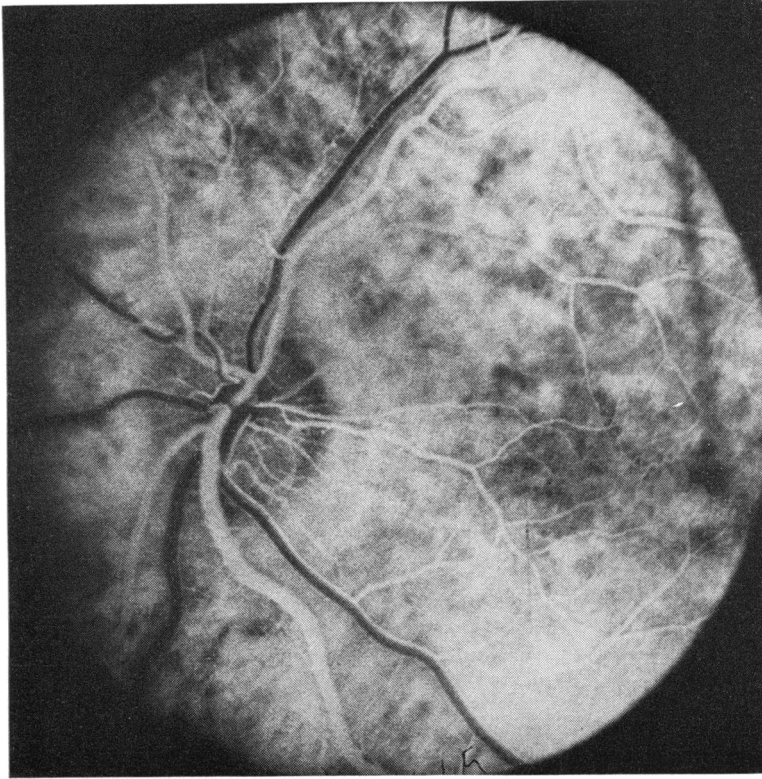


FIG. 1 *Arterial and early venous phase of the fluorescein angiogram, inferior retinal vessels appear larger than superior vessels. Note widespread pigment epithelial disturbance and the patchy choroidal fluorescence*

The case now reported shows the association of pigmentary degeneration with inferior retinal telangiectasia and massive exudation (Coats's disease). Fluorescein angiography shows the dilated and tortuous arteries and veins with numerous microaneurysms. Electro-oculography in this family showed mild impairment of light rise in two sisters, although a brother similarly tested at the same time had normal results. These findings raise the possibility that the two sisters are carriers suggesting a recessive mode of inheritance.

Retinitis pigmentosa is a bilateral hereditary disorder of the retinal receptors and pigment epithelium which may be present from birth. Numerous associations with systemic conditions are reported (Alstrom, 1959), and in some a specific metabolic disorder is described, as in Refsum's syndrome (Refsum, 1946). Drusen of the disc may be associated in a few (0.3 to 2 per cent) cases (Lorentzen, 1966). Pathological evidence in the tapeto-retinal disorders suggests a primary degeneration of the receptors and pigment epithelium. Several aetiological mechanisms have also been implicated for drusen of the disc including degeneration of axons (Seitz and Kersting, 1962), degeneration of neuroglia (Fuchs, 1926), and

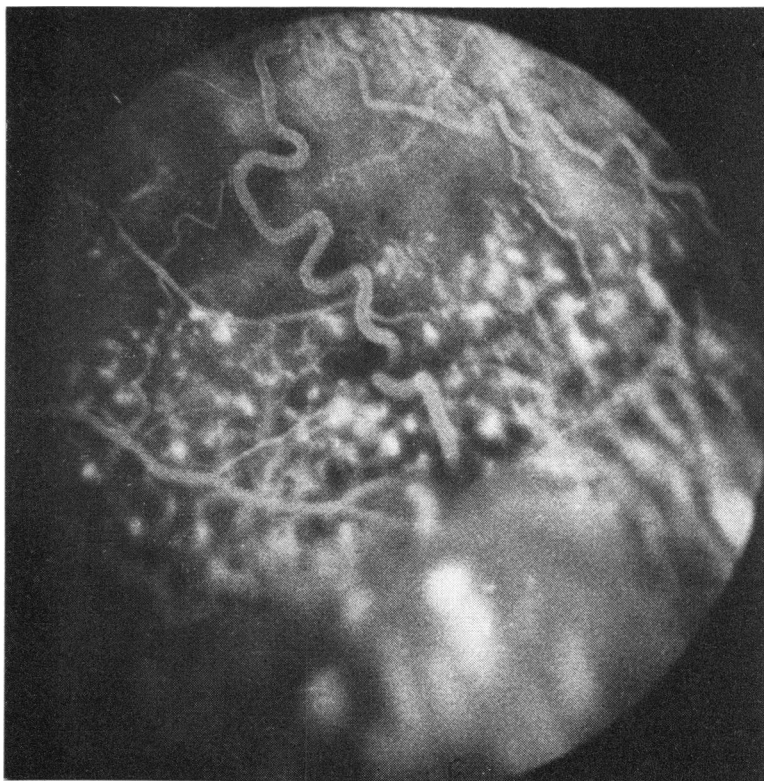
migration of the pigment epithelium (Lauber, 1921).

In contrast Coats's disease is a rare unilateral disease without any hereditary pattern or systemic associations which develops in the second and third decades. Recent electron microscopical findings, however, suggest a primary defect in the vascular endothelium producing increased permeability (Tripathi and Ashton, 1971) and resultant massive exudation. Dilatation of the retinal feeder vessels in our patient might be explained by the increased perfusion. The presence of bilateral retinal telangiectasia with tapeto-retinal degeneration and drusen in a number of reports suggests a possible linked association. Although speculative, this could be explained as a primary degeneration involving the retinal vessels, the retinal axons, the retinal receptors, and pigment epithelium.

Summary

The association of bilateral retinitis pigmentosa, Coats's disease, and drusen of the optic disc has been described. Previous reports are discussed and the association could be explained as a degenerative condition involving retinal vessels, retinal receptors, and axons.

FIG. 2 Venous phase and the dilated tortuous inferior temporal retinal vein is clearly seen with a shunt vessel



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