The fundal appearances have remained unchanged since the patient was first seen in 1995. Both parents are deceased; fundal appearances of the patient's brother, sister, son and daughter were normal.

Comment

The chorioretinal atrophy observed could represent a congenital abnormality related to choroidal inflammation or an unusual chorioretinal dystrophy. The most common cause of well-defined chorioretinal scarring in the United Kingdom is inflammation caused by *Toxoplasma gondii*.[2] The absence of detectable (undiluted) serum antibodies suggests that ocular toxoplasmosis is highly improbable.[3] Well-defined atrophy such as this occurs in bifocal chorioretinal atrophy[4] and North Carolina macular dystrophy,[4] both of which are dominantly inherited, although the site is quite different from that described in this report. Whilst it is tempting to assume that these abnormalities may represent an unusual chorioretinal dystrophy no other living family members have been similarly affected. We are not aware of any reports documenting a similar phenotype and therefore are unable to establish the diagnosis.

We are grateful to Professor Alan Bird for his help in preparing the manuscript.

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Raymond Radford, MRCP, FRCOphth ✉
Department of Ophthalmology
Royal Eye Hospital
Oxford Road
Manchester M13 9WH, UK
Tel: +44 (0)161 2761234
Fax: +44 (0)161 2736354

Mark Wright, FRCSEd
Department of Ophthalmology
Princess Alexandra Eye Pavilion
Chalmers Street
Edinburgh EH3 9HA, UK

Sir,

Mitochondrial DNA disease masquerading as age-related macular degeneration

Pigmentary changes in the retinal periphery are well-recognised features of mitochondrial disease.[1] We report here a patient with the MELAS (mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes) syndrome[2] who presented with a pigmentary retinopathy characterised by predominant posterior pole and macular involvement.

Case report

The patient was first noted to have retinal pigmentary abnormalities aged 44 years during a routine visit to her optician. She was visually asymptomatic and had no parental history of any ophthalmic disease. Four years previously she had developed mild gradual-onset bilateral sensorineural deafness in the absence of other neurological disease. Past medical history was otherwise unremarkable and she was systemically well.

Visual acuity was 6/9 in both eyes. Lid position and function were normal and extraocular movements full. No pathology was detected on anterior segment examination. Indirect ophthalmoscopy revealed symmetrical irregular patches of retinal pigment epithelial atrophy and pigment aggregation predominantly involving the posterior pole and macular region (Fig. 1). This was confirmed on fluorescein angiography (Fig. 2). Both optic discs were pink and healthy. Electrophysiological studies, including flash electroretinograms and visual evoked potentials, were all within normal limits. The clinical picture was thought to be consistent with geographic age-related macular degeneration. In view of her relatively young age, periodic review was arranged. During the next 7 years there was no deterioration in her visual acuity or progression of her macular changes.

Seven years after her initial presentation the patient had a generalised seizure. Preliminary investigations revealed elevated fasting lactate levels and diabetes mellitus. A CT showed multiple occipito-parietal infarctions and calcification of her basal ganglia and posterior thalami. This clustering of clinical features and biochemical abnormalities suggested a diagnosis of mitochondrial cytopathy. A muscle biopsy was performed showing the presence of 2% ragged-red fibres. Subsequent molecular genetic studies confirmed the

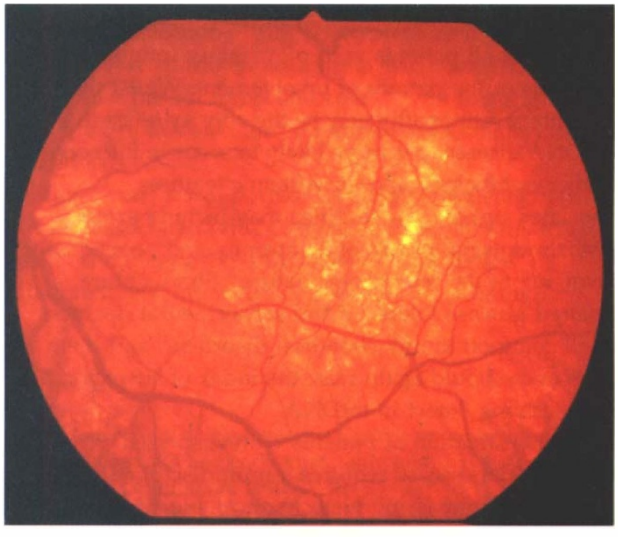

Fig. 1. *Fundal appearance at initial presentation.*

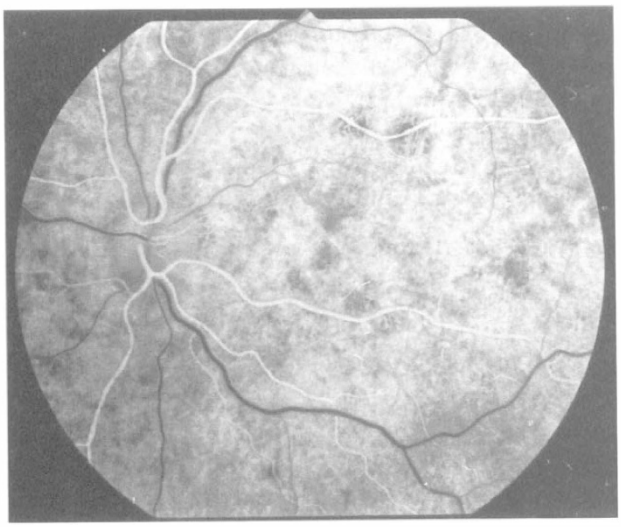

Fig. 2. *Fluorescein angiogram confirming pigmentary macular disruption. A similar picture was also observed in the fellow eye.*

presence of a heteroplasmic *A3243G* mutation in mitochondrial DNA (mtDNA) consistent with the MELAS syndrome.[2]

Comment

A peripheral 'salt and pepper' retinopathy is well-recognised in patients with mitochondrial disease.[1] Predominant macular disruption has been thought to be an uncommon or variable finding, but two recent series both report this pattern of involvement in up to 50–60% of individuals with a variety of defects in mtDNA.[3,4] In many cases these findings were subclinical and detected only on fluorescein angiography performed after the initial diagnosis of a mitochondrial cytopathy had been made. Because angiography is not routinely performed on patients with mitochondrial disease in the absence of overt fundal changes, it is likely that the prevalence of macular involvement in mitochondrial disorders has been significantly underestimated in the past.

In our patient the pigmentary maculopathy was an early manifestation of the disease and moreover readily identifiable on direct ophthalmoscopy. Together with other studies[3,4] this suggests that a mitochondrial aetiology should be considered in the differential diagnosis of all patients with pigmentary retinopathy regardless of the pattern of involvement. Whilst the pigmentary retinopathy may be the only manifestation of a mtDNA disease, it is more likely to occur in the context of other ophthalmic and/or systemic features. Associated ophthalmic findings may include external ophthalmoplegia, ptosis, optic atrophy and occasionally narrowing of the retinal vasculature.[1] Short stature, impaired glucose tolerance and sensorineural deafness are all common in mitochondrial disorders[5] and their coexistence should again raise the index of suspicion for an underlying defect in mtDNA.

The investigation of patients with suspected mitochondrial disease requires a multidisciplinary approach encompassing histochemical, biochemical and molecular genetic studies.[5] Every effort should be made to establish an accurate diagnosis, as a mtDNA disorder not only has implications in relation to genetic counselling[6] but suggests the possibility of underlying and potentially life-threatening complications including cardiac conduction defects.[5]

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R.M. Andrews[1]
B.J. McNeela[3]
P. Reading[2]
P.G. Griffiths[1]
P.F. Chinnery[2]
D.M. Turnbull[2]
Departments of [1]Ophthalomology and [2]Neurology
University of Newcastle upon Tyne
Newcastle upon Tyne, UK
[3]Department of Ophthalmology
North Riding Infirmary, Middlesborough, UK

Mr Richard Andrews ✉
Department of Neurology
The Medical School
Framlington Place
Newcastle upon Tyne NE2 4HH, UK

Tel: +44 (0)191 2228334
Fax: +44 (0)191 2228553
e-mail: r.m.andrews@newcastle.ac.uk

Sir,

Candida endophthalmitis: a diagnostic dilemma

Candida endophthalmitis has varied presentations and the differential diagnosis is often large so a high index of suspicion is necessary. We outline the case of a man who presented with severe uveitis and a negative vitreous biopsy who subsequently developed an intralenticular abscess. This is the second reported case of an intralenticular abscess associated with *Candida* endophthalmitis. The mechanisms of this are discussed and current treatment options for *Candida* endophthalmitis are outlined.