subsequent observations have suggested the outer retina to be the location of the pathology.[2,5,6] One or two retinal flame-shaped haemorrhages are also often seen.[2] A recent report showed disturbance of the inner/outer segment junction on ultra-high-resolution OCT in a proposed case of AMNR,[7] but the features of the case were not typical (age over 50 years, unilateral non-classical lesion, and no predisposing event). The high-resolution OCT images in our patient clearly show focal abnormality in the photoreceptor layer, corresponding to the lesions on infra-red imaging, with preservation of inner retinal architecture. Whether a direct viral effect or immunologic phenomenon is responsible remains unclear, although abnormalities of choroidal perfusion may be responsible for cases associated with the use of adrenaline or noradrenaline (with or without antecedent acute systemic hypotension). This report is further evidenced that the pathology in this unusual condition is located in the outer retina/photoreceptors and not, as was originally thought, in the inner retina. The condition may perhaps be more accurately described as 'acute macular outer retinopathy'.

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EH Hughes[1], Y-C Siow[1] and AP Hunyor[1,2]

[1]Vitreoretinal Unit, Sydney Eye Hospital, New South Wales, Australia
[2]Retinal Therapeutics Research Group, Save Sight Institute, University of Sydney, New South Wales, Australia
E-mail: edward.hughes1@gmail.com

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Sir,

Flecked retina associated with ring 17 chromosome

Ring chromosome 17 was first described in 1970. There have been 4 previous reports of flecked retina associated with ring chromosome. We report another case of ring chromosome 17 with flecked retina.

Case report

We present a 25-year-old boy, who presented 23 years ago with myoclonic seizure, learning disability, and developmental delay. There were no signs of dysmorphism or any skin lesions. CT scan of the head showed no definite abnormalities. No family history of seizures or visual problems were reported.

Ocular examination at the age of 16 years revealed VA of 6/6 in both eyes. Low-frequency jerk nystagmus was apparent on dextroversion. Fundoscopy revealed well-defined white foveal flecks at the level of retinal pigment epithelium in both eyes (Figure 1).

Karyotype analysis (Figure 2) showed 46 XY, r(17)(p13.3q25) with no obvious loss of genetic material. Fluorescence *in situ* hybridisation studies (Figure 2) using the Oncor D17s379 probe from the Miller–Dieker chromosome region on 17p13.3, revealed no deletion. Probes within 300 kb of each telomere showed signals on normal chromosome 17 only, suggesting that at least 300 kb of material has been lost from each arm.

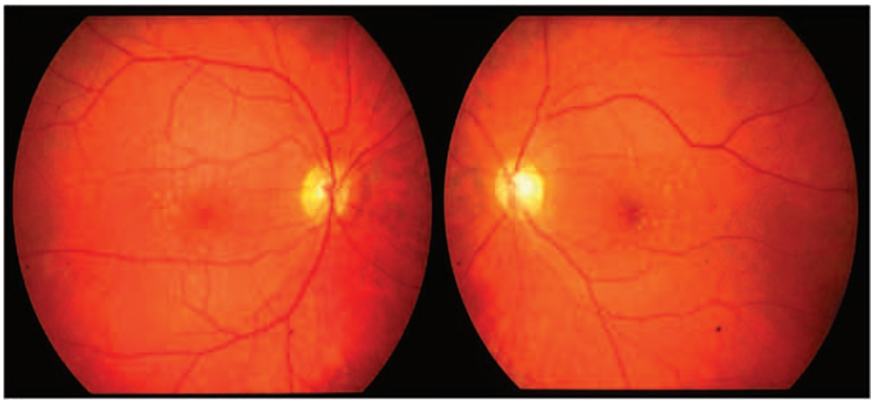

Figure 1 Fundus photographs of reported patient showing well-defined white foveal flecks at the level of the retinal pigment epithelium.

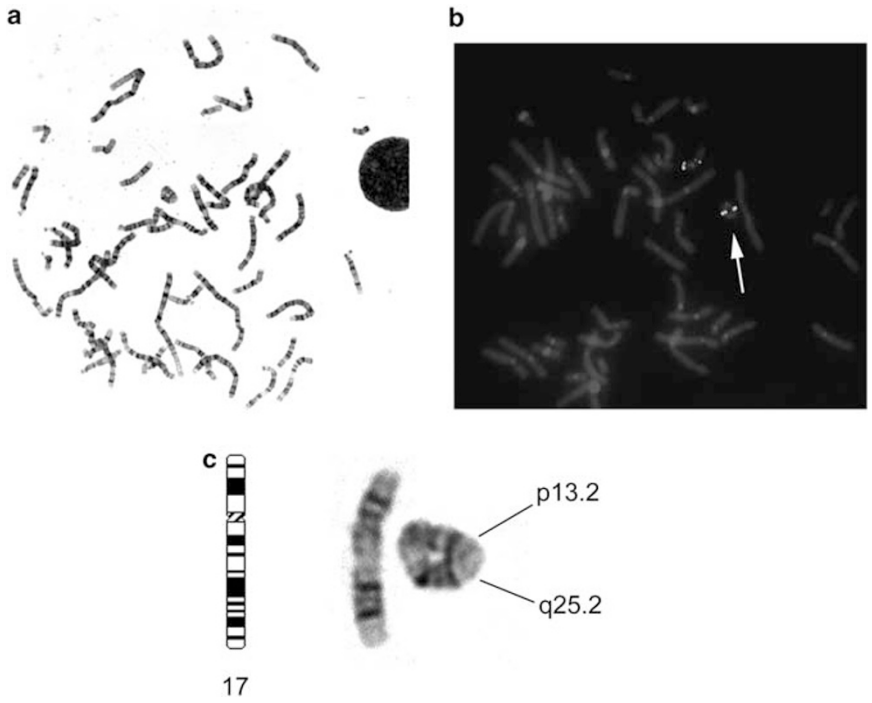

Figure 2 Karyotypic analysis showing ring chromosome 17. (a) G-banded karyotype showing ring chromosome 17 (arrowed). (b) Fuorescence *in situ* hybridisation with probes from 17q21 (RARA) and 17p13.3 (Oncor D17s379, Miller Dieker region) showed that both probes are present on ring chromosome. This confirms that there is loss of little chromosome material from the P arm of chromosome 17. (c) Ideogram of the ring 17 chromosome.

No abnormality in parental chromosome confirmed this as a *de novo* change.

Comment

Four other cases of flecked retina associated with ring 17 chromosome have been reported.[1–4] The reported cases were associated with short stature, mental retardation, epilepsy, and café-au-lait spots. Neurofibromotosis can be misdiagnosed in these cases due to the presence of café-au-lait spots. Fundus fluorescein angiogram showed normal retinal and choroidal vascular filling.[1–4]

Retinal spots or flecks are seen in a range of heritable ocular disorders, including age-related macular degeneration, pattern dystrophy, Stargardt disease, fundus flavimaculatus, Bietti's crystalline dystrophy, as well as being part of multisystemic phenotypes, such as Alport disease, primary hyperoxaluria, and Gaucher disease. Such flecks may be caused by an isolated abnormality of receptor or RPE metabolism leading to the accumulation of abnormal material within the RPE or Bruch's membrane.[2,3] Retinal flecks have also been reported in cases of ring chromosome 14.[5] This may be a non-specific consequence of ring chromosome formation. However, it remains possible that loci on chromosome 17 may be involved in the regulation of RPE or photoreceptor function. Future detailed mapping of the chromosomal imbalance in such patients will be critical in defining whether this is the case.

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R Kumari[1], G Black[1,2], J Dore[2] and IC Lloyd[1]

[1]Department of Ophthalmology, Manchester Royal Eye Hospital, Nelson Street, Manchester, UK
[2]Medical Genetics Research Group, University of Manchester and Central Manchester and Manchester Children's Hospital Trust, St Mary's Hospital, Manchester, UK
E-mail: docreena2003@yahoo.co.in

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