PostScript

LETTERS

Choroidal infiltration in juvenile myelomonocytic leukaemia

Juvenile myelomonocytic leukaemia (JMML), previously called juvenile chronic myelogenous/myeloid leukaemia, is a disorder of infancy and early childhood associated with clonal overproduction of immature myeloid cells, leading to organ infiltration, infection, and failure. Hallmarks include leucocytosis, often with monocytosis, hepatosplenomegaly, increased HbF levels, and no Ph chromosome.^{1,2} Allogeneic bone marrow transplant is considered the only curative therapy.³ Ocular involvement has only been reported on one occasion.⁴

Case report

A 3 year old boy with a 2 year history of JMML, with remission and subsequent relapse after allogeneic haematopoietic cell transplant, was incidentally found to have a fixed, dilated left pupil at 6 mm, unreactive to light, and no perception of light. Family members reported 2 months of left eye erythema. The boy was otherwise asymptomatic. Examination showed no evidence of a third cranial nerve palsy. Fundus findings in the left eye included subtle inferotemporal spicular changes, consistent with previous exudative retinal detachment. Magnetic resonance imaging (MRI) revealed a meniscus at the left posterior retina, adjacent to the optic nerve and macula, consistent with haemorrhage versus leukaemia infiltration (fig 1). Complete blood count revealed 29% blasts, and lumbar puncture revealed leukaemic cells.

The patient was treated urgently with radiotherapy, in an attempt to return left eye function and to treat occult central nervous system disease. He additionally developed fevers, and was treated with antibiotics despite negative blood cultures. Over 5 days, the patient received 200 cGy per day (6 MV photons) to the left eye, for a total of 1000 cGy. At the end of the treatment course, the patient continued to have a fixed, dilated left pupil, unreactive to light, and no

Figure 2 B-scan ultrasound, transverse 3 o'clock, shows highly reflective anterior surface of shallow exudative retinal detachment in the left eye of a patient with juvenile myelomonocytic leukaemia.

perception of light. Ultrasound of the eye revealed a shallow, exudative retinal detachment, with moderate to high reflectivity, consistent with leukaemia infiltration (fig 2). The clinical examination supported this, with the observation of loculated fluid superiorly with spicules. Optic atrophy of the left eye was also noted. On follow up 1 month later, the patient had continuing fevers, but the visual acuity of the left eye had improved. It was also weakly reactive to light, and the patient was averse to light shone into the eye. The child died 4 months after presentation.

Comment

JMML is well associated with organ infiltration, but ocular involvement had not been implicated until Nambu *et al* presented a 1 month old boy with JMML, bilateral ocular infiltration, and eventual complete blindness.⁴ His MRI scan revealed a high density area only in the right ocular fundus.⁴ We are unaware of any other report of JMML with ocular involvement. In our patient, using MRI and ultrasound, we observed retinal exudative detachment, suggesting retinal leukaemic infiltrate, further supported by the regain of some ocular function after radiation therapy. The fixed dilated pupil may be the result of either unsuspected

leukaemia infiltration or subsequent ischaemia. Family members refused autopsy.

In summary, ocular infiltration is a rare but devastating complication of JMML. Any visual complaints in these patients should be investigated urgently and thoroughly by an ophthalmologist.

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Occult retinal and choroidal lesions in neurofibromatosis type 1

Neurofibromatosis 1, a common inherited disorder,¹ is considered a neurocristopathy characterised by pathological hamartomatous proliferations of neural crest derived tissues.^{1,2} Neurofibromatosis can be observed in any tissue with neural crest cells throughout the body, including the eye. However, retinal and choroidal lesions had been considered unusual in eyes with this disease,³ although Lisch nodules are frequently observed.⁴ Because choroidal abnormalities especially have been reported in up to 100% of cases during examinations using infrared monochromatic light and the confocal scanning laser ophthalmoscope (SLO),⁵ choroidal involvement is thought to be common in neurofibromatous 1.

Conventional optical coherence tomography (OCT) can non-invasively obtain optical cross sectional retinal images with histological detail.⁶ Newly developed three dimensional (3-D) OCT operates in a so called transverse fashion and yields OCT images originating at the same axis and plane as the retina.^{7,8} Because this instrument simultaneously produces SLO and interferometric OCT images, it readily allows detection of an area of pathology in the fundus.⁹ The system also scans laterally along a line to the retina to produce cross sectional longitudinal OCT images with resolution of less than 10 µm. We report on the lesions associated with neurofibromatous 1 detected using this technology.

Case report

A 19 year old Japanese man with neurofibromatosis type 1 was referred to us because of ocular complications. He had no visual

Figure 1 Magnetic resonance imaging in the left eye. A meniscus level is seen. Hyperintense signal on T1 (left) and hypointense signal on T2 weighted (right) images were consistent with either haemorrhage or leukaemia infiltration. Fundus findings precluded the presence of haemorrhage.

symptoms. The bilateral visual acuity was 20/20 and no remarkable scotoma was observed by microperimetry. There were several Lisch nodules bilaterally. No remarkable fundus abnormalities were observed by ophthalmoscopic and biomicroscopic fundus examinations or fluorescein angiography. The transverse retinal images obtained using 3-D OCT showed small, round, isolated, high intensity lesions near the optic disc (fig 1A); the longitudinal OCT images showed that the lesions were in the retinal nerve fibre layer (fig 1B). Areas of multiple, bright patches were observed non-invasively with SLO infrared imaging (fig 2A) using the direct confocal mode. Two regions of bright and dark patches using the indirect mode (fig 2B) were observed. Only the dark patches corresponded to the hypofluorescent area observed by indocyanine green angiography (fig 2C).

Comment

We detected small, round, isolated high intensity retinal lesions on the transverse images using 3-D OCT. We also evaluated the retinal layer in which the lesions were located by longitudinal imaging and found that the lesions were in the retinal nerve fibre layer. The retinal lesions were undetected by conventional means—that is, funduscopy or fluorescein angiography, because they were too small and had no neovascularisation. Therefore, these occult lesions could hardly be detected by longitudinal imaging without the availability of transverse images. Astrocytic hamartomas had been reported as a rear retinal manifestation of neurofibromatosis.³ Because astrocytes are distributed mainly within 5–6 mm of the optic disc, astrocytic hamartomas have been reported close to the disc and in the nerve fibre layer or ganglion cell layer. Therefore, the retinal lesions that could be detected using 3D OCT might be astrocytic hamartomas. Our findings suggested that retinal lesions might not be as rare as previously thought but rather are undetectable by conventional means.

Figure 1 (A) Transverse OCT image and (B) longitudinal OCT image. The transverse OCT image shows the area of the lesion (arrow), and the longitudinal OCT image shows that the lesion is in the retinal nerve fibre layer (arrowhead).

In the present case, multiple, bright patches were observed with infrared imaging using the SLO direct confocal mode, the same as have been reported. Because no remarkable abnormalities were observed by fluorescein angiography, the lesions would not be caused by retinal or choriocapillaris circulation failure. Furthermore, a bright reflex using the SLO with a confocal aperture indicated the presence of highly reflective tissue. Yasunari *et al*⁵ reported this to be choroidal neurofibromas or choroidal nodules because of histological observations. These bright patches showed two types of lesions using the indirect mode. When using the indirect mode, the ring aperture blocks the reflex from the focal plane and allows imaging of deep structures. The finding that only the dark patches corresponded to the hypofluorescent area might be because the choroidal region close to the retina blocks fluorescence from the underlying choroidal vessels and the deeper region does not. Therefore, these differences might indicate the depth of the lesions in the choroid. SLO infrared imaging using the indirect mode demonstrates choroidal lesions and may provide information about their depth.

3-D OCT and SLO facilitate non-invasive analysis of retinal and choroidal pathologies in eyes with neurofibromatosis type 1. The advanced technology enables us to observe structures that were previously undetectable using conventional methods. Retinal and choroidal lesions in eyes with neurofibromatous 1 might not be as rare as previously thought.

Figure 2 (A) Confocal SLO infrared image, (B) indirect SLO infrared image, and (C) indocyanine green angiography. (A) Several bright patches, (B) bright patch (arrowhead), and dark patch (arrow). (C) The area of hypofluorescence (arrow) is only observed corresponding to the dark patch in the indirect SLO infrared image.

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Disseminated keratoconjunctival epitheliopathy in a patient with systemic lupus erythematosus

Conjunctivocorneal involvement in systemic lupus erythematosus (SLE) is quite infrequent.^{1–3} We report the impression cytological (IC), immunohistochemical (IHC), and transmission electron microscopic (TEM) features of a case with unusual keratoconjunctival epitheliopathy refractory to medical treatment.

Case report

A 76 year old woman was referred with a long history of bilateral foreign body sensation. She had been on topical antibiotics, artificial tears, corticosteroids, hyaluronate, and aciclovir without improvement. At initial examination, biomicroscopy revealed diffuse, translucent, disseminated peripheral corneal intraepithelial coin lesions with a cystic appearance (fig 1A, B), also in the bulbar conjunctiva over the limbus (fig 1C). Fluorescein staining revealed increased permeability with negative staining. Schirmer's test was normal in both eyes. The patient had systemic and dermatological features diagnosed as SLE and had been taking prednisolone 7.5 mg/day for 1 year. Conjunctival cultures were negative for infectious processes. A thorough systemic evaluation for other systemic diseases and serum antibodies for herpes simplex virus (HSV-1, HSV-2), varicella zoster virus (VZV), and Epstein-Barr virus (EBV) were negative.