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.