Diagnostic dilemmas

Neurosarcoiosis masquerading as glioma of the optic chiasm in a child

KL Ng, N McDermott, CAJ Romanowski, A Jackson

Summary
We present a case of sarcoiosis in a 14-year-old girl who presented with a short history of visual disturbance. Computed tomography and magnetic resonance imaging (MRI) demonstrated enlargement of the optic chiasm and pre-chiasma optic nerves. Post-contrast MRI showed marginal enhancement of the affected areas of the optic pathways. A diagnosis of optic nerve glioma and arachnoid gliomatosis was made; surgical confirmation was not sought due to the risk to vision associated with biopsy. A rapid clinical deterioration led to repeat MRI which demonstrated extensive enhancing soft tissue throughout the basal cisterns with extension into the brain. Biopsy confirmed a diagnosis of sarcoiosis.

Keywords: neurosarcoiosis, optic glioma, magnetic resonance imaging

Neurosarcoiosis is a rare disease which is most common in adults with evidence of systemic disease. It may present with a diverse range of clinical features including meningitis, cranial nerve palsies, epilepsy, hydrocephalus, and transverse myelitis. Magnetic resonance imaging (MRI) and to a lesser extent computed tomography (CT) will demonstrate sarcoiosis of the meninges and of the brain itself but are non-specific. A wide range of radiological appearances have been described and neurosarcoiosis may be mistaken for other diseases of the meninges (including his-tiocytosis, tuberculosis, and meningo-kiotosis), for diffuse white matter diseases (particularly multiple sclerosis), or for mass lesions (such as lymphoma, glioma, or metastasis). The correct diagnosis may be further delayed when the disease occurs in childhood or without systemic signs. We describe a case of sarcoic leptomeningitis in a child which clinically and radiologically masqueraded as a primary glioma of the optic chiasm.

Case report
A 14-year-old girl who presented with a two-year history of intermittent headaches and a two-month history of right visual disturbance. Examination revealed diminished visual acuity (4/6) and an afferent pupillary defect of the right eye. The right optic disc was swollen and there were fundal haemorrhages. The remainder of the neurological examination including that of the left eye was normal. Chest radiograph, biochemical, haematological and endocrine investigations were normal.

A CT brain scan demonstrated an enhancing midline suprasellar mass in the position of the optic chiasm. T1-weighted MRI (SE 500/25 and GE 500/25) confirmed enlargement of the optic chiasm and pre-chiasmal optic nerves (figure 1). Following intravenous contrast administration (Gadolinium DTPA) there was marked enhancement around the periphery of the chiasm and pre-chiasmal optic nerves. Small areas of enhancement were also noted along the infundibulum, on the base of the hypothalamus and in the region of the right lateral geniculate body (figures 2 and 3). Differential diagnoses of optic chiasm glioma or sarcoiosis were considered, however, in view of the patient’s age and the absence of other stigmata of sarcoiosis, a diagnosis of optic chiasm glioma was considered most likely.

Neurosarcoiosis

Demographic features:
- Age range: 3 months–35 years
- Commonest between 20 and 55 years
- Equal sex incidence
- Present in 5–16% of systemic sarcoiosis
- Presenting feature in 0.3–2.5% of cases

Pathological features:
- Basal leptomeningitis
- More diffuse leptomeningeal and epidendral enhancement
- Intraxial granulomatous masses
- Granulomatous angiitis

Clinical features:
- Raised intracranial pressure
- Hypothalamic and pituitary dysfunction
- Multiple cranial nerve palsies
- Uni- or multi-focal neurological deficits
- Epilepsy
- Stroke

Radiological features:
- Hydrocephalus
- Leptomeningeal thickening and enhancement
- Intraxial mass lesions
- Extraaxial mass lesions
- Multiple high signal white matter lesions on T2-weighted MRI

Box 1
likely. A biopsy was not performed due to the risk of visual deterioration and a decision was made to monitor the lesion using MRI unless the clinical condition deteriorated.

Three months later, the patient developed generalised headaches and examination demonstrated left-sided papilloedema. CT scan showed marked enlargement of the suprasellar mass with obstructive hydrocephalus and bilateral ventriculo-peritoneal shunts were inserted. T1-weighted MRI showed a large ill-defined soft tissue mass arising in the suprasellar cistern and extending to involve the hypothalamus, basal ganglia, and medial temporal lobes (figure 4). T2-weighted images (SE 2000/80) demonstrated extensive oedema around the mass and extending along major fibre tracts which were involved in it (figure 5). Following contrast administration, diffuse enhancement of the optic chiasm, basal leptomeninges, ependyma of the third ventricle and of the basal ganglia (figure 6) could be seen. Anteriorly, the mass was encasing the anterior cerebral arteries, whilst posteriorly, it extended to the lateral geniculate bodies without involvement of the optic radiations. A revised diagnosis of an aggressive neoplastic or inflammatory process was made and the most likely diagnosis was felt to be a high grade primary neoplasm.

The patient deteriorated rapidly with fluctuating confusion, extensive somnolence and worsening visual acuity in both eyes. The endocrine profile revealed inappropriate antidiuretic hormone secretion and hypogonadotrophic hypogonadism. A biopsy was therefore taken from the right intra-cranial optic nerve and anterior optic chiasm. Histology revealed multiple granulomata and inflammatory cell infiltrate with perivascular cuffing but no acid-fast bacilli and a diagnosis of sarcoidosis was made. The patient was commenced on oral steroids supplemented by weekly pulses of intravenous methylprednisolone, in addition to demeclocycline and

Figure 1 Sagittal T1-weighted MRI showing enlargement of the optic chiasm (arrow)

Figure 2 Coronal T1-weighted MRI following contrast showing marginal enhancement of the optic chiasm and a small area of leptomeningeal enhancement on the base of the hypothalamus (arrow)

Figure 3 Oblique axial T1-weighted MRI following contrast showing enlargement of the optic chiasm and pre-chiasmal optic nerves (small arrows). There is also a small area of enhancement in the region of the lateral geniculate body (long arrow)

Figure 4 Sagittal T1-weighted MRI three months after presentation showing marked enlargement of the optic chiasm mass with invasion of the adjacent hypothalamus (arrows)
Neurosarcoidosis masquerading as glioma of the optic chiasm in a child

Enlargement of the optic chiasm in children is most commonly a result of optic nerve glioma. Fifty per cent of cases present before the age of five years and 90% before the age of 20. They are common in females and up to 25% of cases are associated with neurofibromatosis (box 2). They are extremely slow-growing glial tumours with a 40–50%, 20-year survival rate. Approximately 45% originate in the chiasm and spread by direct extension along the optic pathways. They are commonly associated with precocious puberty or other hypothalamic disturbance and may also cause secondary hydrocephalus due to invasion of the ventricular system. Histologically there is infiltration of the optic pathways with glioblasts of various sizes and occasional astrocytes. Hyperplasia of the overlying arachnoid mater, known as arachnoid gliomatosis, is a well recognised feature. Radiologically MRI is the optimal modality for investigation and demonstrates smooth enlargement of the chiasm and affected portions of the optic nerve. Intravenous contrast administration results in variable degrees of enhancement which is frequently patchy and which may occur on the margins of the lesion due to arachnoid gliomatosis. Surgical excision is inappropriate since the overall prognosis is good and radiotherapy appears to have little effect on survival. Chiasmal biopsy is contra-indicated since it has been identified as the main cause of visual deterioration and has also been associated with a significant but unexplained mortality.

Although the incidence of neurosarcoidosis is not documented it has been described as

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<tr>
<th>Optic glioma</th>
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<td>Demographic features:</td>
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<tr>
<td>• 50% below the age of 5 years</td>
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<td>• slightly commoner in females</td>
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<td>• 15–25% associated with neurofibromatosis</td>
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<td>Pathological features:</td>
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<td>• infiltrative tumour enlarging the optic nerve (50%), optic chiasm (45%), or optic tract (5%)</td>
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<td>• direct spread along optic nerves, optic chiasm, and optic tracts</td>
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<td>• tumour limited by dura</td>
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<td>• slow growth</td>
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<td>Clinical features:</td>
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<tr>
<td>• slowly progressive visual deterioration</td>
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<td>• hypothalamic and pituitary dysfunction</td>
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<td>Radiological features:</td>
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<td>• enlargement of the affected areas of the optic pathway</td>
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<tr>
<td>• mild, usually homogeneous enhancement</td>
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<tr>
<td>• rarely: peripheral enhancement due to arachnoid gliomatosis</td>
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<td>• other features of neurofibromatosis in 5–15%</td>
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Figure 5 Coronal T2-weighted images at the levels of the chiasm (A) and optic radiations showing extensive cerebral oedema (B, high signal) and mass effect. Extensive oedema is also seen within the optic tracts and visual radiations (arrows)

Figure 6 Coronal T1-weighted MRI following contrast showing extensive enhancement of the basal meninges and ependyma of the third ventricle (arrow)
Neurosarcoidosis is most commonly seen in patients with systemic disease but can, rarely, be the presenting feature of the disease and may remain confined to the nervous system. The age of onset has ranged from a three month old child to the 8th decade but the vast majority of cases are between the ages of 20 and 55. In the nervous system, sarcoid granulomas develop in the leptomeninges giving rise to thickening of the arachnoid mater particularly around the optic chiasm and basal cisterns but sarcoid meningitis may occur anywhere over the surface of the brain or spinal cord. Granulomata are also commonly seen in the ependymal linings of the ventricles and in the choroid plexus. Parenchymal lesions may occur anywhere within the nervous system and are particularly common in the periventricular regions and in the Virchow–Robin spaces where they may form granulomatous masses up to several centimetres in diameter. Sarcoid vasculitis may also occur and give rise to cerebral infarction.1,2,9

Clinically and radiologically neural sarcoid is one of the great mimics of other pathological processes. Involvement of the leptomeninges may present with meningitic symptoms or more classically, with multiple cranial nerve palsies.2 Intracranial lesions may cause raised intracranial pressure, epilepsy or focal neurological deficits and sarcoid vasculitis may lead to presentation as stroke.

MRI is the most useful form of radiological investigation and intravenous contrast media appear to be invariably associated with enhancement in active lesions.3,4,10 Despite this the MRI appearances of neurosarcoidosis are seldom distinctive. Diffuse neurosarcoidosis is most common in the corona radiata and periventricular regions and may be indistinguishable from the plaques of multiple sclerosis. Solitary enhancing mass lesions within the neural axis are very difficult to differentiate from lymphoma, metastatic or primary malignant disease, while similar lesions in the spinal cord may also mimic glioma, ependymoma or acute inflammatory myelitis.1,4,5

In the present case, a leptomeningitis affecting principally the optic chiasm and pre-chiasmal optic nerves led to an incorrect diagnosis of optic nerve glioma. The presence of chiasmal enlargement gave rise to an appearance identical to that of optic chiasm glioma on unenhanced MRI. Marked enhancement around the margins of the lesion was felt to represent arachnoid gliomatosis and enhancement in the region of the lateral geniculate body was also in keeping with a diagnosis of optic nerve glioma. The presence of small areas of leptomeningeal enhancement on the base of the hypothalamus and of enhancement of the infundibulum were noted on the original MRI examination and should, in retrospect, have suggested more extensive leptomeningeal disease.

In conclusion, leptomeningeal sarcoidosis affecting the optic chiasm may closely mimic optic nerve glioma. The presence of atypical radiological features such as leptomeningeal enhancement or peripheral enhancement of an enlarged chiasm should suggest an inflammatory process. If the risk of chiasmal biopsy is considered too great then disease progression should be monitored frequently using contrast enhanced T1-weighted MRI.

Learning points
- neurosarcoidosis may occur in the absence of systemic involvement
- neurosarcoidosis may mimic a wide range of neurological disorders
- neurosarcoidosis has a wide range and may occur in childhood
- leptomeningeal enhancement on MRI should suggest a diagnosis of sarcoidosis even in the presence of mass lesions, diffuse white matter abnormality, or focal ischaemia

Box 3

Neurosarcoidosis masquerading as glioma of the optic chiasm in a child.

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