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Sir,

Familial Arnold-Chiari Type I malformation

Arnold-Chiari Type I malformation (CMI) is a congenital malformation of unknown incidence. It occurs in a small posterior fossa and constitutes 3–5 mm herniation of the cerebellar tonsils into the spinal canal.[1] In all, 25% of patients have cerebrospinal fluid displacement into the spinal cord canal, causing syringomyelia.[1,2]

Familial clustering of CMI with and without syringomyelia has been documented in American scientific literature but this case of siblings appears to be the first instance reported in the UK.

Case report

Case 1: A 49-year-old gentleman presented to ophthalmic outpatients in 1996 with an 8-month history of horizontal diplopia, vertigo, left arm and thigh pain, a change in gait, and occipital headaches. The patient smoked 20 cigarettes/day, but there was no past medical and, at that stage, no known family history.

Snellen acuity was 6/9 right (amblyopia), 6/6 left. Examination demonstrated a small right esotropia with limited abduction of the right eye. Horizontal nystagmus with a vertical component on right gaze, and reduced convergence were present. There was no sign of optic nerve dysfunction. Humphrey visual fields were full. Intraocular pressures were 15 bilaterally and fundoscopy was normal.

Neurological examination demonstrated broad-based gait and a positive Rhomberg's test. Tone and reflexes were normal.

Routine blood tests and vitamin B12 and folate levels were normal, and VDRL/TPHA screen was negative. Initial MRI scan was normal and differential diagnoses were scan-negative demyelinating disease, Arnold–Chiari malformation, basilar invagination, and paraneoplastic syndrome.

In late 1997, the patient developed oscillopsia and had increasing difficulty walking. Ophthalmic examination further revealed downbeat and rotatory nystagmus in all gaze positions.

Repeat MRI in 1997 demonstrated Chiari Type I malformation, the cerebellar tonsils having herniated to the posterior arch of C2 (Figure 1a and b).

The patient underwent foramen magnum decompression in early 1998 with marked symptom resolution. He presented to ophthalmology again in mid-1998 with recurrence of diplopia and had a small right VI nerve palsy on examination. Further MRI showed no recurrence of hindbrain disease. Following extraocular muscle surgery 1 year later, the patient has remained asymptomatic.

Case 2: Aged 52 years, the elder sister of the aforementioned patient presented to neurologists in 1995 with intermittent shaking and weakness of the left hand. She was an asthmatic on bronchodilators; nothing else was significant medically. Neurological examination and CT brain scan were normal, the history was felt to be suggestive of focal motor seizures and the patient was commenced on Phenytoin 100 mg tds.

In 1996, the patient developed left thenar eminence fasciculation and in 1998 described upper limb paraethesiae. Examination revealed hypertonic lower limbs, hyper-reflexia, right extensor plantar response, and decreased pin prick sensation in the C2–C8 distribution. Power remained full throughout.

MRI scan showed extensive syringomyelia, from C1 to the mid-dorsal region with associated Chiari Type I malformation, the cerebellar tonsils having herniated to the arch of C1 (Figure 2a and b).

In early 1999, the patient underwent foramen magnum decompression with good symptom resolution. She later became ataxic and developed an upper limb tremor.

Repeat MRI scan in 2003 demonstrated that despite satisfactory decompression, the caudal end of the syrinx remained. The patient awaits further neurosurgical intervention.

Comment

The first reported case of familial CMI was in the United States in 1990, involving siblings aged 18 months and 2

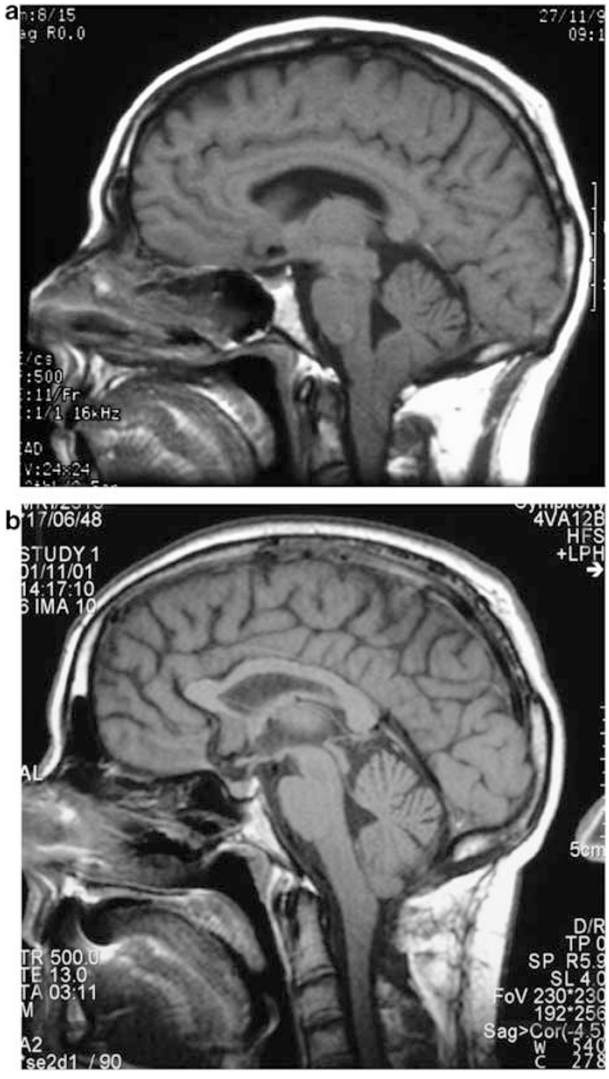

Figure 1 (a) Case 1: Chiari Type I malformation: herniation of the cerebellar tonsils into the spinal canal. (b) Case 1: postforamen magnum decompression — improved position of cerebellar tonsils.

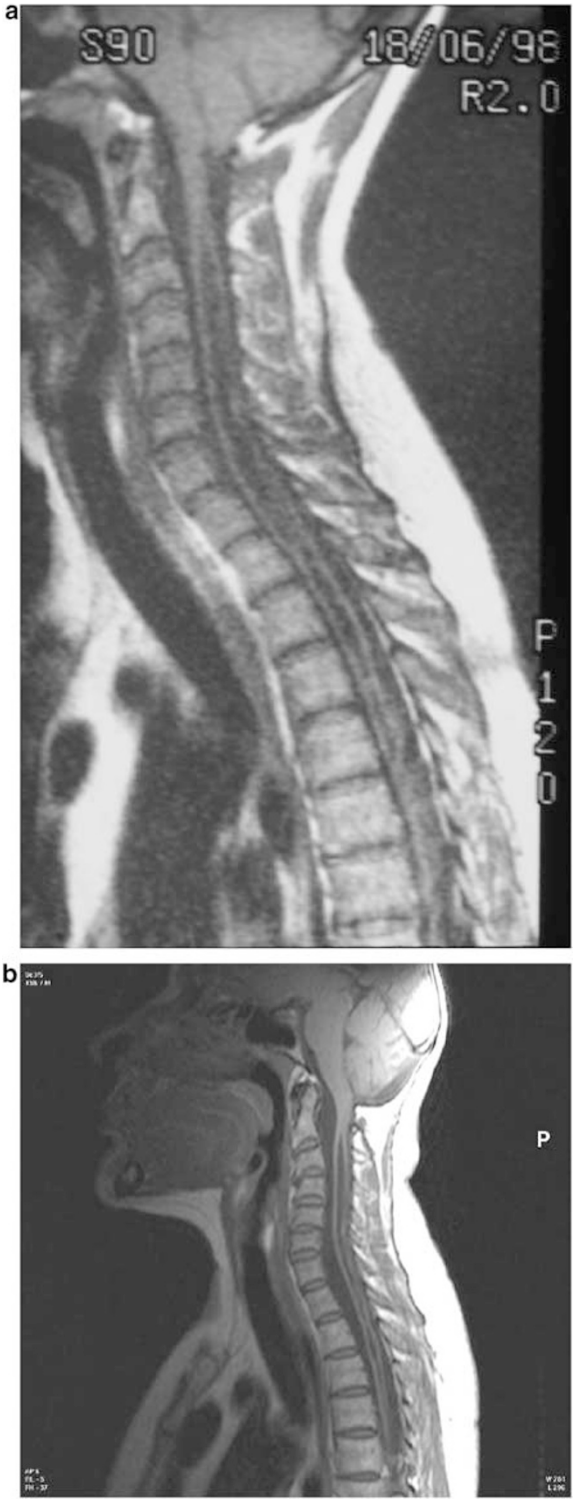

Figure 2 (a) Case 2: Chiari Type I malformation with syringomyelia: herniation of cerebellar tonsils associated with fluid cavity into the spinal cord. (b) Case 2: postforamen magnum decompression–persistence of syrinx.

years.[3] Speer *et al*[2] demonstrated familial aggregation in a large patient cohort including 28 sibling pairs. Additionally one-fifth of asymptomatic relatives were diagnosed with CMI±syringomyelia following neuroimaging. No cases of isolated syringomyelia were noted, adding weight to the growing belief that the two conditions are part of a continuum. Milhorat *et al* demonstrated 12% of 364 patients having one close relative with either CMI or syringomyelia.[1] Inheritance patterns suggest autosomal dominant transmission,[2,4,5] although autosomal recessive patterns with reduced penetrance are also reported.[1] Concordance has been reported in monozygotic twins and triplets.[1,2] The potential genetic basis is reinforced by the recognised association of several

genetic syndromes with CMI, including many of skeletal abnormality, such as achondroplasia.[1,2]

CMI is thought to be a skeletal mesodermal disorder resulting from faulty division embryonic somites forming the skull base and cranoicervical junction.[2] Governing this process is the gene *Pax-1*, whose malfunction causes vertebral fusion and a small posterior cranial fossa.[2] Altered cerebrospinal fluid flow in this constricted environment produces the varied symptoms, the commonest being suboccipital headache. Ophthalmic symptoms occur in the majority of patients and include retro orbital pain, floaters, photopsia, photophobia, diplopia, and visual field loss. Hearing loss and vestibular impairment causing vertigo, oscillopsia, and nystagmus, as well as symptoms of spinal cord dysfunction, are prevalent.[4]

Extensive literature search has failed to identify another reported case of familial CMI in the United Kingdom. Learning points illustrated herein include the diagnostic challenges posed by CMI, and the importance of assessing family history to increase surveillance of potentially affected relatives.

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Sir,

A brief history of punctoplasty: the 3-snip revisited

We read with interest the article by Caesar *et al*[1] on 'A brief history of punctoplasty: the 3-snip revisited'. In the article, the authors reviewed the development of various surgical methods for treating punctal stenosis and reported their results of 3-snip punctoplasty. We feel that some issues that may affect the outcomes warrant further discussion.

All the patients were assessed for subjective improvement of epiphora at 1 week after operation. This relatively short-term symptomatic evaluation may not be reliable because some patients may, in contrary, experience increased epiphora in the early postoperative period due to surgical wound and associated inflammation.[2] In the four cases (8%) without improvement of epiphora, the reasons could be restenosis of punctum, coexistence of other obstructions in the lacrimal drainage system, lacrimal pump failure, tear hypersecretion, or even dry eye.[3] Information on patient selection, and outcome measures including anatomical success would be relevant in interpreting the results. To avoid operating on patients with symptoms of 'epiphora' caused by dry eye, we would recommend preoperative Shirmer test and fluorescein staining test.

Punctoplasty is usually performed under topical with or without adjacent subcutaneous anaesthetic agents.[4] We observed that some patients may still experience variable degree of intraoperative pain. It is probably due to inadequate penetration of anaesthetic agent into the surgical field that involves excision of a tissue block from the posterior lamella of eyelids. We found that a small amount of local anaesthetic agent such as 2% lignocaine hydrochloride (IMS, CA, USA) into the subcaruncle area instead of subcutaneous injection provides excellent anaethetic result.

We congratulate Caesar and co-workers for their good work. We hope that the discussion would enhance our understanding and treatment of punctal stenosis.

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