CASE REPORT

Chiari type 1 malformation in a pseudotumour cerebri patient: is it an acquired or congenital Chiari malformation?

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SUMMARY

Chiari malformation type 1 (CM1) is a developmental abnormality of the cerebellar tonsils. Patients with CM1 commonly present with headache. Papilloedema is rarely seen in CM1. However, a 52-year-old woman presented to the hospital with a headache and her ophthalmological examination revealed bilateral papilloedema. Her cranial MRI was compatible with borderline CM1. Bilateral papilloedema and headache suggested idiopathic intracranial hypertension (IIH) as the preliminary diagnosis. IIH is a rare case in CM1. This article argues about this association and discusses as to whether it is an acquired or congenital Chiari malformation.

BACKGROUND

Chiari malformation type 1 (CM1) is cerebellar tonsillar herniation at least 3 mm below the base of the foramen magnum.¹ CM1 can cause headaches, pseudotumour-like episodes, visual disorders, endolymphatic hydrops, Meniere's disease-like syndrome, lower cranial nerve palsies, syringomyelia and hydrocephalus.² The most common symptom in CM1 is headache, as in IIH. Headaches are cough type and are seen in the occipital-suboccipital region in CM1.³

Intracranial pressure rises unknowably in IIH.⁴ Intracranial mass, cerebral venous thrombosis, meningeal processes or any other reasons must be ruled out to diagnose IIH. It is frequent in obese women at childbearing age.⁴ High-body mass index (BMI), untreated obstructive sleep apnoea, hyperthyroidism, hypothyroidism, iron deficiency anaemia and various medications can cause IIH.⁴ Visual disturbances, diplopia, headaches, tinnitus, retrobulbar pain and diplopia may occur in IIH.⁵ Retro-orbital eye pain during eye movements is helpful in differentiating it from other headache disorders.⁶ IIH is managed medically, surgically or both. Medical treatment consists of weight reduction, diuretic therapy or oral acetazolamide treatment. Surgical options include neurosurgical shunting or optic nerve sheath fenestration.

Symptoms of headache and papilloedema in a patient with CM1 indicate IIH as the diagnosis. Sinclair *et al*⁷ described a case of IIH without ventricular dilation in a 12-year-old patient with CM1. IIH and CM1 have been detected with an overall incidence of 1.3%, rising to 2.7% in patients with MRI scanning.⁷ MRI discovers CM1 in one-third of patients who are asymptomatic.³ This article presents a patient with IIH which is thought to be an acquired CM1.

CASE PRESENTATION

A 52-year-old female patient presenting with recurrent headaches for 1.5 months referred to the eye clinic last year. Her ophthalmic examination revealed visual acuities of 20/20, bilateral normal intraocular pressures, isocoric pupils and positive light reflexes in both eyes with no relative afferent pupillary defect in her eyes. Colour vision was normal on Ishihara charts. Extraocular eye movements were free in each direction with no restriction and no diplopia. No focal neurological deficit was noticed. Bilateral early nuclear sclerosis and bilateral early papilloedema were detected as positive proof. Humphrey visual field analysis showed bilateral superior and inferior arcuate-type defects with mean deviations of -11.76 dB in the right and -13.11 dB in the left eye. Optical coherence tomography (OCT) indicated bilateral optic nerve fibre layer elevation with global averages of 144 µm on the right eye and 190 µm on the left eye. Visual fields and OCT images are shown in figures 1 and 2.

INVESTIGATIONS

MRI indicated cerebellar tonsillar descent of 5 mm, distension of the optic nerve sheaths, flattening of posterior globe contour and vertical tortuosity of the optic nerves that were consistent with IIH, except tonsillar herniation. MRI films are shown in figure 3. Cerebellar tonsillar descent was thought to be a component of borderline CM1. There was no evidence of any intracranial mass, cerebral sinus thrombosis or meningeal disease. Empty sella appearance, flattening of the pituitary gland, tight subarachnoid spaces and ventricular dilation were absent. The patient said she had gained excessive weight after she had given up smoking. Medical investigation revealed subclinical hypothyroidism, mild iron deficiency anaemia and obesity with a BMI of 33.24 kg/m². The patient was applied lumbar puncture drainage. Opening pressure was over 30 cm H₂O with normal cytological and biochemical component.

DIFFERENTIAL DIAGNOSIS

Bilateral papilloedema and headache with no ventricular dilation in a patient with CM1 suggested IIH as a preliminary diagnosis. Obesity, subclinical hypothyroidism and mild anaemia were compatible with IIH. Intracranial mass, tumours, subarachnoid haemorrhages, subdural haematomas, meningeal inflammations, central retinal artery and vein

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Figure 1 Computerised visual field analysis shows bilateral superior and inferior visual field constriction in the early stages of papilloedema.

occlusions, optic neuritis and anterior ischaemic optic neuropathy must be ruled out in bilateral optic disc oedema.⁸

TREATMENT

Lumbar puncture, oral acetazolamide 250 mg two times a day and paracetamol 500 mg a day were administered. The first opening pressure was over 33 cm H₂O and the second was 30 cm H₂O, 30 cc fluid was discharged in each one.

Papilloedemas and visual fields healed bilaterally and significantly. Control MRI was not performed because the patient refused progressive investigation.

OUTCOME AND FOLLOW-UP

The patient recovered from her headaches in a few days. Control visual field analysis showed improvement. She continued oral acetazolamide treatment for 1 year. Control visual

Figure 2 Bilateral increased retinal nerve fibre thicknesses are seen in optic coherence tomography of early stage papilloedema with average thickness of 144 µm in the right and 190 µm in the left.

Figure 3 Empty sella appearance is seen in transverse sections of MRI. Distension and tortuosity of optic nerve sheath are seen in orbital sections of cranial MRI. Cerebellar tonsillar descent is seen in coronal T1-weighted MRI. Cerebellar tonsillar descent of 5 mm below the foramen magnum is seen in sagittal T2-weighted sections of cranial MRI.

fields showed mean deviations of -8.16 dB in the right eye and -9.85 dB in the left eye. Repeated tests showed mean deviations of -2.32 dB in the right eye and -4.01 dB in the left eye in 3 months, -1.32 dB in the right eye and -1.73 dB in the left eye in 6 months and -0.76 dB in the right eye and -1.02 dB in the left eye in 12 months. Control visual fields are shown in figure 4. Optic disc oedemas resolved without optic atrophy. Optic disc photos are seen in figure 5.

DISCUSSION

The modern concept accepts IIH as an increase in the volume of brain mass rather than an increase in the volume of cerebrospinal fluid.⁹ A decreased posterior fossa volume leads to overcrowding and consequently cerebellar herniation.¹⁰ Cerebellar descent can be congenital or acquired. The acquired type has been reported by Payner *et al*¹¹ and they mention that craniospinal pressure gradient creates cerebellar tonsil descent. It is

difficult to differentiate between CM1 with papilloedema and IIH due to similarities in patient presentations.

Tonsillar herniation may be a result of diffuse cerebral oedema and IIH, or it may be a cause that triggers IIH and diffuse cerebral oedema. Vaphiades and Braswell emphasised that they, as well, could not differentiate between these.¹² They noted that cerebellar tonsillar herniation was seen in increased intracranial pressure and acetazolamide treatment resolved CM1 and herniation.¹² According to Lancione and Kosmorsky, tonsillar herniation is a well-documented complication of lumboperitoneal shunt revision. Symptoms of IIH in a normal lumbar opening pressure arouse suspicion of CM1 with a history of multiple lumboperitoneal shunt revisions.¹³

Headaches in CM1 were correlated with a degree of tonsillar herniation independent of cerebrospinal fluid disturbances according to Pascual *et al*.¹⁴ Stowner noted that headaches resulted from the tension of pain-sensitive nerve dendrites in

Figure 4 Visual field controls at 1, 3, 6 months and at 1 year show complete resolution of superior and inferior visual field constrictions.

Figure 5 Early stage papilloedema and resolution of oedema after the treatment are seen in optic disc photographs.

the veins or meninges due to mechanical downward displacement of the tonsils when the patient coughed or made a Valsalva action.¹⁵

Sometimes tonsillar herniation is the result of intracranial hypotension like in pseudo-Chiari. Pseudo-Chiari intracranial hypotension is a result of previous lumbar puncture or past epidural anaesthesia. Orthostatic headaches are seen in pseudo-Chiari and contrast cranial MRI confirms diagnosis by absent posterior fossa displasia and meningeal colouration.¹⁶

Visual symptoms of IIH occur because of the transient ischaemia secondary to increased tissue pressure in the optic nerve head.¹⁷ Amount of visual loss cannot be predicted from the degree of papilloedema because papilloedema may decrease due to apoptosis of the axonal nerve fibres. So axonal nerve death may occur while papilloedema is healing. The OCT may mislead the physician in the diagnosis of IIH because oedema of retinal nerve fibre may seem normal in OCT. Computerised visual field analysis is essential and more valuable in evaluating IIH.⁴ OCT is a helpful instrument in evaluating axonal nerve fibres but it could be preferred when the papilloedema is resolved. Serial automated perimetry remains as the primary method of patient monitoring.⁴

The cerebellar tonsillar position in patients with IIH is significantly lower than in age-matched controls according to Aiken *et al*.¹⁸ The incidence and morphology of cerebellar tonsillar ectopia in IIH has not been described in the radiology literature previously. When present, tonsillar ectopia in IIH may be confused with CM1 and spontaneous intracranial hypotension.¹⁸ The surgical literature describes a subset of pretreatment patients with IIH and cerebellar tonsillar ectopia, meeting the criteria for CM1. Banik *et al*¹⁹ observed tonsillar ectopia in 24% of patients with IIH and tonsillar ectopia of >5 mm in only 10% of patients with IIH. Aiken *et al*¹⁸ suggest that a low cerebellar tonsil position is present in patients with IIH, possibly due to chronically elevated intracranial pressure. A significant proportion (21%) of patients with IIH had cerebellar tonsillar ectopia of >5 mm, which was mimicking CM1.¹⁸

We postulated that our patient was an acquired CM1 secondary to cerebral oedema and IIH. Increased cerebral tissue mass secondary to cerebral oedema of IIH and intracranial pressure gradient caused posterior fossa overcrowding and cerebellar tonsillar

descent. Later, lumbar puncture headaches and visual fields healed rapidly with no complication of acute herniation. Acetazolamide treatment and lumbar puncture are effective methods in treating IIH. We did not comment on tonsillar herniation after the treatment because we could not obtain the control MRI. Finally, we are of the opinion that medical treatment of acquired CM1 and IIH is a safe and effective method. More studies are required to confirm this data. When tonsillar ectopia is identified, careful radiographic and clinical consideration of IIH is warranted to avoid misdiagnosis as congenital CM1.

Learning points

- Acquired-cerebellar tonsillar descent may be seen in idiopathic intracranial hypertension (IIH).
- Papilloedema and IIH are rarely seen in Chiari malformation type 1 (CM1).
- It is often difficult to differentiate between CM1 with papilloedema and IIH clinically due to the similarities in patient presentations.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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