SHORT REPORT

Idiopathic intracranial hypertension in female homozygous twins

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Figure 1 Case 1 (left)
and case 2 (right). A
direct coronal CT shows
thin bony structures at the
frontal skull base in both
cases. There is an orifice
located in the right anterior
base of the skull in case 1
(arrow).

Abstract

The authors report on female homozygous twins with idiopathic intracranial hypertension. At the age of 12 years, both twins simultaneously developed visual disturbances with photophobia. At the age of 19 years, an ophthalmological examination disclosed papilloedema in both their eyes. At the age of 22 years, a lumbar puncture showed raised CSF pressure over (200 mm H₂O) in both twins. Their neurological and radiological examinations were extremely similar; both of them had severely impaired visual acuity and impaired visual field, bilateral optic nerve atrophy, intracranial hypertension, an enlarged and partial empty sella turcica, digital markings of the calvarium, and an enlarged frontal subarachnoid space. This is the first case report describing idiopathic intracranial hypertension occurring in homozygous twins.

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Idiopathic intracranial hypertension was first described by Quinke in 1897.¹ This syndrome

is defined as increased intracranial pressure without neurological, laboratory, or radiological evidence of an intracranial space occupying lesion or hydrocephalus. An annual age adjusted incidence rate of idiopathic intracranial hypertension was estimated to be about 0.9 per 100 000 population.² Therefore, it is not an uncommon disorder and is most often seen in women of childbearing age either with obesity or recent weight gain.¹ When obesity was considered the incidence increased to 19.3 per 100 000 for women aged 20 to 44.³

However, the occurrence of familial intracranial hypertension is rare. To our knowledge, only 10 case reports concerning familial intracranial hypertension have been reported in the literature.¹⁻¹² The disorder was found in two pairs of heterozygous twins previously,^{5,10} but not in homozygous twins. This paper describes idiopathic intracranial hypertension occurring in female homozygous twins.

Case reports**CASE 1**

A 22 year old woman was referred to our hospital with a 10 month history of watery nasal discharge. The patient was born in 1973 as a homozygous twin. The delivery was six weeks premature. At the age of 12 years, she experienced photophobia and developed progressive loss of visual acuity. The corrected visual acuity

Case 1

Case 2

was 0.07 in the right eye and 0.2 in the left eye. At the age of 19 years, an ophthalmological examination disclosed papilloedema and visual field defects in both eyes. At the age of 21 years, a watery nasal discharge from the right side of the nose occurred without any history of head injury, surgical procedure, or sinus disease.

On admission, the patient was well nourished, but not obese. She had had no menstrual irregularity, oral contraceptive use, pregnancy, or thyroid disease. And she had no headache. An ophthalmological examination disclosed optic atrophy in both eyes with decreased visual acuity; corrected acuities were 0.06 in the right eye and 0.09 in the left eye. A skull radiograph showed digital markings on the calvarium. Direct coronal CT (fig 1) showed a tiny orifice in the right anterior fossa and clouded ethmoidal sinus. Sagittal MRI showed an enlarged and partially empty sella (fig 2). Axial T2 weighted MRI showed wide subarachnoid spaces in the frontal region and minimally enlarged lateral ventricles (fig 2).

The patient underwent a bifrontal craniotomy through the right frontal skull base to repair the CSF leakage. After surgery the rhinorrhoea ceased, but headache, nausea, and vomiting gradually developed. A lumbar puncture showed a high opening pressure of 340 mm H₂O with normal cell count and normal concentrations of glucose, protein, albumin, prealbumin, and γ -immunoglobulin. Because of the high intracranial pressure, a ventriculo-peritoneal shunt was placed. The patient's clinical course after the surgery was uneventful and the visual function remained stable.

CASE 2

The twin sister of this patient was also seen at the age of 22 years. A blood type analysis confirmed that they were homozygous twins. She presented a nearly identical clinical history to her sister. At the age of 12 years, she experienced photophobia and developed a progressive loss of visual acuity; the corrected visual acuities were 0.04 in both eyes. At the age of 19 years, an ophthalmological examination disclosed papilloedema and visual field defects

Figure 2 Case 1 (left) and case 2 (right). Brain MRI shows a partially empty sella and a slight enlargement of the bilateral frontal subarachnoid space in both cases.

Case 1

Case 2

Case 1

Case 2

in both eyes. Like her sister, she was not obese and had had no menstrual irregularity, oral contraceptive use, pregnancy, thyroid disease, or headache. An ophthalmological examination showed optic atrophy in both eyes with decreased visual acuity; corrected acuities were 0.04 in both eyes. A lumbar puncture showed a pressure of 210 mm H₂O. The CSF study was normal. A skull radiograph showed digital markings on the calvarium. A direct coronal CT (fig 1) showed thin bony structures in the anterior skull base and deep olfactory grooves. Sagittal MRI showed an enlarged and partially empty sella (fig 2). An axial T2 weighted MRI showed wide subarachnoid spaces in the frontal region and minimally enlarged lateral ventricles (fig 2). The patient underwent a ventricle-peritoneal shunt operation. Lately she has been free from deterioration of visual functions.

Discussion

Idiopathic intracranial hypertension is a condition that occurs predominantly in women of childbearing age with obesity and recent weight gain; there is a peak incidence in the fourth decade.⁹ Neither of our patients were obese.

The confirmed diagnosis of idiopathic intracranial hypertension requires (1) a documented increase in intracranial pressure, more than 200 mm H₂O in the non-obese patient; (2) a normal neurological examination, except for papilloedema with deficits of visual acuity and an occasional abducens nerve palsy; (3) the absence of a space occupying lesion or ventricular enlargement, except for an empty sella on CT or MRI; and (4) normal CSF composition.^{13 14 15}

Our cases are consistent with these criteria. Although the sizes of the lateral ventricles were slightly large, a diagnosis of hydrocephalus was equivocal. In addition, the MRI showed minimal enlargement of the bifrontal subarachnoid space. Both patients might have developed an empty sella because of high intracranial CSF pressure for several years. Slight enlargements of the lateral ventricles, the sella, and the frontal subarachnoid space are not uncommon radiological findings for idiopathic intracranial hypertension.^{5 9 13 16}

Johnston and Morgan⁶ state that familial intracranial hypertension supports the idea of dysfunctional CSF absorption in this disease. Because of the relatively late onset of intracranial hypertension, Cardoso *et al* postulate that age might account for disturbed CSF absorption. In infants, impaired CSF circulation is more likely to dilate the ventricles but in adults, complete myelination and mature brains resist ventricle dilatation. So intracranial hypertension occurs after a long asymptomatic history and without ventriculomegaly.

Ten previous case reports have described familial idiopathic intracranial hypertension. The cases include one mother and son,⁸ a mother and two of four daughters and a son,⁶ three mother-daughter pairs,^{7 9 12} three pairs of siblings,^{14 11} and two pairs of heterozygous twins.^{5 10}

To our knowledge, ours is the first report of idiopathic intracranial hypertension occurring in homozygous twins, and the findings support the hypothesis of genetic transmission of this disease.

The simultaneous occurrence of visual disturbance in the twins is interesting. Torlai *et al* also reported male twins who became symptomatic simultaneously and followed similar clinical courses for more than two years.¹⁰ We think that the intracranial hypertension was not present in early childhood in our patients, but that it gradually developed at the beginning of their adolescence. Many studies have emphasised the association of idiopathic intracranial hypertension with menstrual irregularities, pregnancy, oral contraceptive use, and a heterogeneous group of endocrine diseases.^{2 18 19} Although we have not investigated hormonal concentrations in our patients, the aetiology of idiopathic intracranial hypertension may relate to a balance of secretion of gonadotrophin(s).

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