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Basal Meningoencephalocele, Anomaly of Optic Disc and Panhypopituitarism in Association with Moyamoya Disease

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Key Words
Basal meningoencephalocele · Moyamoya disease · Optic disc anomaly · Panhypopituitarism

Abstract
Basal meningoencephalocele is frequently associated with midfacial anomaly, optic disc anomaly, brain anomaly, cerebrospinal fluid rhinorrhea, chiasma syndrome, and endocrinologic disturbance. The combination of basal meningoencephalocele and moyamoya disease is extremely rare. A 29-year-old man had basal meningoencephalocele (transsphenoidal type), anomaly of the optic disc (morning glory syndrome), panhypopituitarism and moyamoya disease. The patient was treated by hormone replacement, but surgical intervention was not required. Basal meningoencephalocele and moyamoya disease are a possible combination of the diseases.

Introduction

Basal meningoencephalocele is a rare congenital disease comprising 1.2% of all cranial meningoencephaloceles [1]. Basal meningoencephalocele is frequently associated with midfacial anomalies, ocular anomalies, brain anomalies, cerebrospinal fluid rhinorrhea, chiasma syndrome and hypothalamic-pituitary dysfunction [2–11].

Moyamoya disease is characterized by steno-occlusive changes at the terminal portions of the bilateral internal carotid arteries as well as development of so-called moyamoya vessels at the base of the brain. An association of basal meningoencephalocele with moyamoya disease was emphasized recently by ophthalmologists [12], but there have been no previous reports in the Western neurosurgical literature. We report a patient with this combination of diseases and review such cases, including our case, to emphasize this combination.

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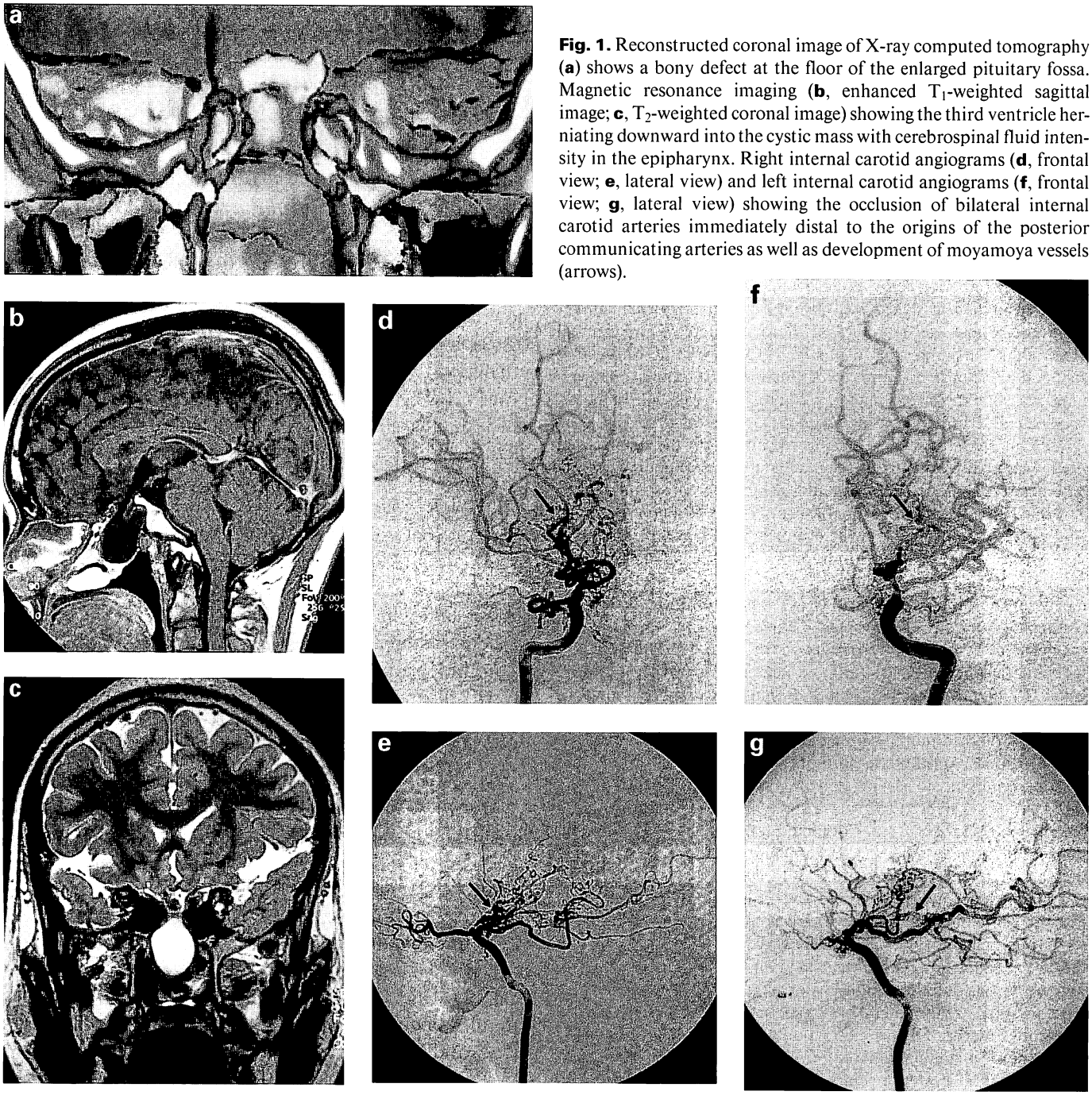

Fig. 1. Reconstructed coronal image of X-ray computed tomography (**a**) shows a bony defect at the floor of the enlarged pituitary fossa. Magnetic resonance imaging (**b**, enhanced T_1-weighted sagittal image; **c**, T_2-weighted coronal image) showing the third ventricle herniating downward into the cystic mass with cerebrospinal fluid intensity in the epipharynx. Right internal carotid angiograms (**d**, frontal view; **e**, lateral view) and left internal carotid angiograms (**f**, frontal view; **g**, lateral view) showing the occlusion of bilateral internal carotid arteries immediately distal to the origins of the posterior communicating arteries as well as development of moyamoya vessels (arrows).

Case Presentation

A 29-year-old man was admitted for re-evaluation of a known basal meningoencephalocele, ocular anomaly, panhypopituitarism and moyamoya disease. The patient, a second child, was the product of a normal spontaneous delivery at full term. Gestation was normal. Birth weight was 3,000 g. Development was normal except for short stature. There was no history of leakage of the cerebrospinal fluid or meningitis. The patient had no respiratory disturbance or history of snoring. Family history was not contributory. He had been treated with human growth hormone injections between the ages of 9 and 20 years, and with cortisone acetate, dried thyroid or levothyroxine

sodium and desmopressin acetate since the age of 9. Human chorionic gonadotropin had been given since the age of 19, and human menopausal gonadotropin since the age of 22. Height was 93.2 cm (–6 SD) at the age of 8, 144.5 cm (–5 SD) at the age of 20 and 161.6 cm (–2 SD) at the age of 24. Surgical treatment was not attempted until the present.

The patient had slightly short stature, 161.8 cm, and weighed 52.4 kg on admission. Smell sensation was intact bilaterally. Visual acuity of the right eye was light perception and that of the left eye was 0.15. Left homonymous hemianopsia was observed. Intraocular pressure was normal bilaterally. The right eye had congenital cataract, and the left eye had an enlarged, excavated optic disc and a mottled pigmentation of the retina. There was no hypertelorism since the interpupillary distance was 60 mm. Except for visual impairment, the patient was neurologically normal. There was no external abnormality, including that of the face. There were no cafe-au-lait spots on the skin. Pubic and axillary hair were normal. Testes volume, measured by orchidometer, was 4 ml, bilaterally. Otorhinolaryngological examination using an endoscope revealed the pulsating midline mass covered by a mucous membrane in the epipharynx. There was no abnormality in the oral cavity, including the soft and hard palates. There was no apparent leakage of cerebrospinal fluid.

Laboratory data were normal except for the results of hormone studies. Baseline value of growth hormone was below 0.05 ng/ml, and there was no response to growth-hormone-releasing factor. Luteinizing-hormone-releasing hormone test results were negative, since luteinizing hormone was always below 0.4 mIU/ml and follicle-stimulating hormone was always below 0.2 mIU/ml. Thyrotropin-releasing hormone test results were negative for thyroid-stimulating hormone (always below 0.5 μU/ml) and negative for prolactin (always below 2.1 ng/ml). Results of corticotropin-releasing hormone tests were hyporesponsive (cortisol was always below 2.4 μg/dl and adrenocorticotropic hormone was 5 pg/ml before provocation and 36 pg/ml at 30 min). Antidiuretic hormone test was not performed, but without desmopressin acetate, polyuria was evident.

Skull X-rays showed platybasia and a bony defect at the floor of the enlarged pituitary fossa. Computed tomography (CT) scan also showed the same bony defect and an enlarged pituitary fossa filled with a nonenhancing mass with a fluid density (fig. 1a). Magnetic resonance (MR) imaging showed the invaginated third ventricle extending into the epipharynx, which was consistent with the diagnosis of transsphenoidal meningoencephalocele (fig. 1b, c). Agenesis of the corpus callosum or septum pellucidum was not observed. There were no lesions of infarction or hemorrhage. Small signal void spots were observed in the bilateral basal ganglia, which were suggestive of moyamoya vessels. Digital subtraction angiography showed the occlusion of the bilateral internal carotid arteries immediately distal to the origins of the posterior communicating arteries and the development of moyamoya vessels (fig. 1d–g). Spontaneous anastomosis between the external carotid system (branches of the middle meningeal arteries) and intracranial vasculature was also observed bilaterally. These findings were consistent with a diagnosis of moyamoya disease. Cerebral blood flow study using a technetium-99m-labeled hexamethyl-propyleneamine oxime showed normal regional blood flow without any perfusion deficits.

Due to stable ocular signs, lack of cerebrospinal fluid leakage or respiratory difficulty, lack of cerebrovascular events attributable to moyamoya disease for at least 10 years after the first diagnosis at the age of 19 years and normal cerebral blood flow, conservative treatment of hormone replacement with desmopressin acetate, cortisone acetate, levothyroxine sodium and gonadotropic hormones (human chorionic gonadotropin and human menopausal gonadotropin) was continued.

Discussion

Clinical Manifestation of Basal Meningoencephalocele

Suwanwela and Suwanwela [13] classified basal meningoencephalocele into four types on the basis of anatomical position of the herniation: transethmoidal, sphenoethmoidal, transsphenoidal and frontosphenoidal. The transsphenoidal type is more frequently complicated by a greater variation of anomalies than the transethmoidal type [11]. The clinical characteristics of the transsphenoidal type are as follows: (1) midfacial anomaly (median cleft lip, cleft palate, broad nasal root, bifid nose, hypertelorism), (2) ocular abnormalities (anophthalmos, microphthalmos, optic nerve dysplasia including hypoplasia, optic pit, optic coloboma, megalopapilla, morning glory syndrome, missing optic chiasm), (3) cerebral anomalies (agenesis of the corpus callosum, agenesis of the septum pellucidum, hydrocephalus especially colpocephaly), (4) cerebrospinal fluid rhinorrhea and recurrent meningitis and (5) endocrinologic disturbance (hypothyroidism, growth hormone deficiency, hypogonadotropic hypogonadism, panhypopituitarism, diabetes insipidus) [2–11].

Optic Disc Anomalies and Moyamoya Disease

Morning glory syndrome is characterized by an enlarged, funnel-shaped optic disc containing a central white dot of glial tissue, surrounded by an elevated annulus of chorioretinal pigmentary disturbance [14]. Two patients with a combination of morning glory syndrome and unilateral moyamoya disease have been reported. None had basal meningoencephalocele. A 5-year-old Asian boy had left morning glory disc anomaly with ipsilateral steno-occlusive changes of the internal carotid artery and the development of moyamoya vessels [15]. A 9.5-year-old girl with right morning glory anomaly had stenosis of the right middle cerebral artery with the development of moyamoya vessels on the right side [16]. This patient had a dolichoectatic carotid artery on the left side. These two cases may suggest an underlying pathognomonic association between congenital optic disc anomalies and moyamoya disease.

Basal Meningoencephalocele and Moyamoya Disease

There have been three cases, including ours, in the literature of a combination of basal meningoencephalocele

and moyamoya disease [12, 17]. Bakri et al. [12] reported a 10-year-old girl with 'sphenopharyngeal' meningoencephalocele, developmental anomalies of optic discs, a chorioretinal coloboma and moyamoya disease. Kobayashi et al. [17] reported a 6-year-old boy with 'sphenoidal' meningoencephalocele, morning glory syndrome, hypopituitary dwarfism, hypertelorism as well as intracranial vascular abnormalities, i.e. stenosis at the distal portion of the internal carotid artery, a development of moyamoya vessels on the right side and irregular filling of the middle cerebral artery on the left side. The vascular abnormalities in this boy were not typical for moyamoya disease because the steno-occlusive change in the internal carotid artery and the development of moyamoya vessels were observed unilaterally. These 3 patients with basal meningoencephalocele and moyamoya disease have optic disc anomalies as well [12, 17]. Less than 40 cases of transsphenoidal meningoencephaloceles have been reported in the literature. Among these, 3 had moyamoya disease or a related condition, which may indicate that the combination of basal meningoencephalocele and moyamoya disease may not be a chance occurrence.

Pathogeneses of Basal Meningoencephalocele, Optic Disc Anomalies and Moyamoya Disease

Various theories on the pathogeneses of basal meningoencephalocele have been proposed: (1) failure of the ethmoid plate to close around the olfactory nerves, (2) localized increased intraventricular pressure, (3) failure of the neuroectodermal separation at the anterior neuropore from the surface ectoderm, preventing the subsequent development of the mesodermal elements, (4) persistence of the craniopharyngeal canal and (5) developmental failure of ossification centers in the sphenoid bone [3, 7, 10, 11, 18–20]. Congenital optic disc anomalies, including morning glory syndrome, are thought to be caused by defective closure of the fetal optic fissure [7]. Thus, it can be said that both basal meningoencephalocele and optic disc anomalies are the result of developmental failure in the early embryological stage.

The pathogenesis of moyamoya disease is still unknown. This disease was once thought to be an acquired disease, but there is increasing support for complex etiologies both on genetic and environmental bases [21, 22]. Although the progression of the pathological processes and timing of clinical manifestations of basal meningoencephalocele and optic disc anomalies and moyamoya disease are different, there might be underlying common, but unknown, genetic and environmental (acquired) factors.

Neuroradiological Diagnosis

In the diagnosis of basal meningoencephalocele, CT and MR imaging are useful in addition to conventional skull X-ray films and X-ray tomograms [23]. Bony defects are well demonstrated by CT, and herniated midline structures are shown by the sagittal and coronal MR images. Conventional cerebral angiography demonstrates a downward deflection of the proximal portions of the anterior cerebral arteries into the sella turcica [3, 8]. Since the vascular anomalies including moyamoya disease can be demonstrated by the less invasive MR angiography, it should be carried out first to rule out this possible vascular disease in the cases of basal meningoencephaloceles.

In conclusion, basal meningoencephalocele is frequently associated with midfacial anomaly, optic disc anomaly, brain anomaly, cerebrospinal fluid rhinorrhea, chiasma syndrome and endocrinologic disturbance. In addition to these, basal meningoencephalocele and moyamoya disease are a possible combination of the diseases.

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