Delayed presentation of morning glory disc anomaly and transsphenoidal encephalocele: A management dilemma

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ABSTRACT

Background Morning glory disc anomaly (MGDA) is a rare developmental abnormality of the optic disc that is associated with many other neurological and vascular conditions. Most cases are diagnosed in childhood.

Case Report We report a 57-year-old woman who presented to the ophthalmology department for assessment of long-standing poor vision in the left eye and exotropia. Examination showed a left MGDA and bitemporal hemianopsia. These findings prompted magnetic resonance imaging, revealing a transsphenoidal basal meningoencephalocele with herniation of the optic chiasm and inferior hypothalamus into the dural sac. Due to the eloquence of the neurovascular structures it contained, a decision was made not to reduce the meningoencephalocele. Instead, a ventriculoperitoneal shunt was placed. The patient's ophthalmologic examination remained stable over the following year.

Conclusion While rare, MGDA can be first diagnosed in late adulthood and a thorough evaluation should be completed to assess for midline cranial defects, vascular abnormalities, and other associated abnormalities. Patients presenting late in life with basal encephalocele, herniation of the optic chiasm, and bitemporal hemianopsia present a management dilemma. In this case, a ventriculoperitoneal shunt was placed with the intention of lowering intracranial pressure to prevent further herniation and reduce the risk of cerebrospinal fluid leak.

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Background

The morning glory disc anomaly (MGDA) describes a funnel-shaped excavation of the posterior fundus that incorporates the optic disc and is surrounded by an elevated annulus of chorioretinal pigment. This is said to resemble the morning glory flower, giving this condition its characteristic name. The retinal blood vessels extend from the disc radially and there is a central core of white glial tissue located in the normal position of the cup.¹ The clinical features of MGDA were first delineated in 1970.^{1,2}

The correct diagnosis of MGDA is important, as it can be associated with many other neurological and vascular conditions.³ Associated conditions include moyamoya disease, basal encephalocele, Chiari malformation type I, agenesis of the corpus callosum, and

neurocutaneous disorders such as PHACES syndrome (posterior fossa malformations–hemangiomas–arterial anomalies–cardiac defects–eye abnormalities) and neurofibromatosis. Ocular associations include retinal detachment, strabismus, amblyopia, leukocoria, decreased visual acuity, relative afferent pupillary defect, and visual field defects such as hemianopia and central or paracentral scotomas. Visual acuity is variable with most patients in the 20/100–20/200 range. There is characteristically no family history associated with MGDA. It typically presents unilaterally, is more common in females, and is usually diagnosed during childhood.^{2,3}

The pathogenesis of MGDA is not well understood, but it is thought to result from a stochastic process during embryogenesis that results in poor development of the posterior

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sclera and lamina cribrosa during gestation.^{2,3} We report a unique case of late MDGA occurrence associated with basal encephalocele and compressive optic neuropathy.

Case report

A 57-year-old woman presented with long-standing poor vision in the left eye and exotropia. Her past medical history included diabetes and hypertension. Her past surgical history included only cholecystectomy and appendectomy. She denied a family history of ophthalmologic difficulties and did not recall having previous eye examinations or eye surgery. She denied diplopia, as well as constitutional and neurological symptoms. On examination, vision in the right eye was 20/25 and she was able to count fingers accurately in the left eye at four feet. The intraocular pressure was normal in both eyes. There was a left relative afferent pupillary defect, left hypotropia, and left exotropia with full motility. The dilated exam revealed MGDA in the left eye and was unremarkable in the right (Figure 1). She had a flat nose and hypertelorism. Automated visual field testing revealed bitemporal hemianopsia (Figure 2). Magnetic resonance imaging (MRI), including angiography, showed a basal encephalocele (Figure 3) with herniation of the optic chiasm. Optical coherence tomography of the macula and peripapillary area did not reveal any subretinal fluid or breaks in the retina.

The patient was referred for neurosurgical evaluation. Lumbar puncture was obtained to measure the cerebrospinal fluid (CSF) opening pressure, which was 17 cm H₂O. Typically, cranial base defects with associated meningoencephaloceles are repaired surgically with reduction or resection of the dural and neural tissue and then repair of the osseous defect. In this patient, the meningocele contained the optic nerves, chiasm, and a portion of the hypothalamus. Therefore, no attempt was made at its manipulation or direct repair. After extensive conversations with the patient regarding management options, including observation with serial ophthalmologic evaluations, repair of the cranial base defect, and decreasing the intracranial pressure (ICP), which was believed to be a potential contributing factor

Figure 1. Colour fundus photo of the left eye demonstrating the morning glory disc anomaly.

to the meningoencephalocele formation and associated visual symptoms, the patient opted for the latter. Therefore, a right frontal ventriculoperitoneal shunt with a programmable shunt valve was placed in hopes of lowering the ICP and indirectly reducing the defect. At 1 year follow-up, the patient has demonstrated visual stability with serial Humphrey visual field (HVF) exams and no radiographic increase in the size of the meningoencephalocele (Figure 4, 5).

Discussion

Basal encephaloceles are very rare, occurring in approximately 1 in every 35,000 live births.^{4,5} They can be classified into distinct categories based on their anatomical site of herniation, including transsphenoidal, spheno-orbital, sphenoethmoidal, transethmoidal, and sphenomaxillary. Transsphenoidal encephaloceles are recognized as the least common of the encephalocele subtypes, occurring in approximately 1 in 700,000 live births. The osseous defect is located within the sphenoid bone body, allowing neural tissue to herniate into the sphenoid sinus and possibly into the nasal cavity. The classification can be further broken down into intrasphenoidal encephaloceles and true transsphenoidal

encephaloceles based on the extent of neural tissue herniation.⁴ By definition, intrasphenoidal encephaloceles are confined within the sphenoid sinus cavity and do not extend into the nasopharynx. True transsphenoidal encephaloceles extend through the floor of the sphenoid sinus into the nasopharynx.⁶ Intrasphenoidal and

transsphenoidal can even be further subdivided based on if the neural tissue herniates medially (perisellar) or laterally (through a defect proximal to the sphenoid recess).⁷

Basal encephalocele occurrence in association with MGDA is well documented^{5,8}, though the prevalence is not known. While regarded as the

Figure 2. Humphrey visual field of the right (OD) and left (OS) eyes showing bitemporal hemianopsia.

Figure 3. Preoperative A) coronal T2WI magnetic resonance imaging (MRI) showing meningoencephalocele (asterisk) containing optic nerves, chiasm, and tract. Preoperative B) sagittal T1WI MRI with meningoencephalocele (asterisk) extending through the sphenoid sinus and into the nasopharynx. Additionally, the anterior cranial base is overall low-lying.

Figure 4. A) Preoperative and B) postoperative sagittal T1WI MRIs, demonstrating radiographic stability after ventriculoperitoneal shunt placement.

Figure 5. A) Preoperative and B) postoperative sagittal T2WI MRIs, demonstrating radiographic stability after ventriculoperitoneal shunt placement.

least common encephalocele classification in general, transsphenoidal basal encephaloceles are most frequently reported in association with MGDA.³ They are most often diagnosed and treated in infancy or early life after symptom presentation or as an incidental finding.^{4–6}

Cases with delayed presentation of basal encephalocele are rare but have been well documented.^{4,6,9–12} Presentation in adulthood is often delayed until CSF rhinorrhea, visual field deficit, an epipharyngeal mass, or hormonal abnormality is identified. These findings can be subtle and remain subclinical for long periods of time.^{4,6} In our described case, the abnormality was not detected until the patient had reached 57 years of age. This created a therapeutic dilemma, as

this functionally monocular patient presented with subjective visual deterioration and automated visual fields demonstrating a bitemporal hemianopsia. Unfortunately, despite her long-standing visual difficulties, no prior examination or ophthalmologic data was available to determine if this represented a true progression of her visual defects. Progressive herniation and compression of the optic chiasm was considered as basal encephaloceles have been reported to displace the third ventricle, elements of the hypothalamic pituitary axis, anterior cerebral arteries, and optic chiasm.⁶ Long-term ophthalmological follow-up with repeated visual field testing was recommended to the patient in addition to neurosurgical follow-up. She declined

initial nonsurgical management, however, due to her worsening symptoms and the bitemporal defect on her automated visual fields, referring her ophthalmologic disorder to the sellar/parasellar region, as demonstrated by the MRI.

The appropriate neurosurgical treatment for this case was debated, as it was difficult to assess if there had truly been progression of her symptoms and if surgical intervention was warranted. Surgical treatment of basal encephaloceles is complex and both endoscopic and transcranial approaches for repair have been described, depending on the location and severity of herniation and the affected surrounding anatomy.^{4,7,11} We describe and review relevant literature regarding both the typical surgical management of basal encephaloceles, and the relationship between ICP and cranial based defects, which is more relevant to our treatment choice.

Surgical management of basal encephaloceles

Surgical intervention of basal encephaloceles is indicated with CSF rhinorrhea, epipharyngeal respiratory obstruction, or progressively worsening neurological deficits.^{9–11,13} All cases of spontaneous CSF rhinorrhea should be treated surgically because of the increased risk for developing meningitis.¹⁰ Relatively asymptomatic patients with no dural defect and thus no CSF rhinorrhea are evaluated on an individual basis. The risk of observation must be weighed against the risk of surgical intervention in these patients. The natural history of basal encephaloceles and therefore the outcomes of nonsurgical management have not been well described.⁷

The subclassification of transsphenoidal encephalocele has been shown to have significant implications on the surgical management of the defect.^{6,7} Jabre et al advocated for the use of an endoscopic transsphenoidal approach for the treatment of intrasphenoidal encephalocele reporting successful treatment in 11 of 12 patients.⁶ Landreneau et al reported that in 3 of 4 patients with true transsphenoidal meningoceles, a craniotomy was finally required for repair because of failed endoscopic attempts. The authors stated that the minimally invasive nature of endoscopy may be outweighed by its decreased efficacy in

true transsphenoidal encephaloceles.¹⁴ In contrast, Lai et al advocated for an endoscopic approach for the repair of all transsphenoidal encephaloceles reporting successful repair in 11 of 12 cases. With endonasal endoscopic repair, the potential morbidity associated with craniotomies, including large external incisions, required ICU monitoring, temporal lobe retraction, and possible damage to surrounding delicate skull base anatomy, may be avoided. Lai et al further delineated which endoscopic approach may be more successful for medial and lateral transsphenoidal encephaloceles. A purely transsphenoidal approach may be utilized to repair medial/parasellar transsphenoidal encephaloceles. Lateral transsphenoidal encephaloceles are better accessed through transethmoidal transsphenoidal transpterygoid approaches to gain better visualization of the lateral sphenoid sinus.⁷

With these approaches, basal encephaloceles or meningoencephaloceles are resected and reduced so that an adequate cranial base repair can be performed. This can typically be done as the contents of the dural sac is typically nonfunctional meninges or frontal or temporal lobe. In our described case, however, a direct reduction could not be performed without devastating neurologic morbidity due to the contents of the meningoencephalocele.

Association of ICP and cranial base defects

Prior CSF studies have established a normal ICP range of 5–15 cm H₂O and suggested that neurologic symptoms can manifest at pressures exceeding 15–20 cm H₂O.¹⁵ Intracranial hypertension has been well established as a potential aetiology of cranial base defects, including CSF fistulae and meningo(encephalo)celes. While CSF pressures greater than 25 cm H₂O are required for the diagnosis of disorders such as idiopathic intracranial hypertension, it has been suggested that an active CSF leak or osseous cranial base defect may act as a release valve, lowering the ‘true’ ICP; after surgical repair of these disorders, subsequently increased CSF pressures may cause new neurologic symptoms.^{16–18} Additionally, elevated ICP in basal encephaloceles has been reported to induce spontaneous CSF rhinorrhea.^{4,10}

Although a CSF opening pressure of 17 cm H₂O does not fulfill standard accepted criteria for intracranial hypertension, we believed our patient meningoencephalocele to be acting as a potential 'release valve' for ICPs. Even if the patient did not suffer from elevated CSF pressures, however, we did believe there was a benefit to further decreasing the ICP through permanent CSF diversion, in hopes of relieving any force on the meningoencephalocele sac that may provoke its enlargement. With no prior ophthalmologic or radiographic evaluations for this patient, there was a concern as to what the natural history of her disorder would be without intervention. Rupture of the dural sac and resultant CSF fistula would likely prompt direct surgical treatment, which could prove neurologically devastating in such a patient for the reasons previously cited. Therefore, we elected to indirectly reduce the defect through a ventriculoperitoneal shunt, allowing for CSF diversion and decreased pressure upon the dural defect. This approach allowed for clinical and radiographic stability at 1 year postoperatively.

Conclusion

While rare, MGDA may not be diagnosed until late adulthood and a thorough evaluation should be performed to assess for midline cranial defects, vascular abnormalities, and other associated abnormalities in these patients. Adult patients presenting with basal encephaloceles should be evaluated and treated on an individual basis based on the associated symptoms and any associated progression. The risk of observation must be weighed against the risk of surgical intervention for patients with subclinical symptoms or those without current surgical indications. Surgical intervention, if deemed necessary, must then be tailored to that individual's respective anatomical defect.

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