Case Report

A Silent Congenital Ethmoidal Encephalocele Progressing into Frontoethmoidal Meningoencephalocele with Episodic Seizures in Adulthood: A Case Report and Literature Review

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ABSTRACT

Background: Frontoethmoidal meningoencephalocele (FEM) is rare congenital malformations that has specific geographic distribution. This condition is depicted with herniation of brain tissue as well as meninges via a defect in the cranium at the frontal, orbital, nasal as well as ethmoidal regions. Although several cases have been reported, none of them have a silent and progressive nature with episodic seizures. Case Presentation: We present a 20-year-old man with a swelling on the nasal bridge which was notice a few years after birth. The swelling initially progressed slowly without any symptoms from childhood through adolescents. Both CT scan and MRI revealed a defect in cranium at the frontal and ethmoidal regions with herniation of meninges and brain matter into the nasal cavity but no fistula. We successfully reduced the encephalocele as well as watertight closed the dura mater. The skull defect repaired, and left orbital defect also reconstructed via surgery. Conclusions: Aflatoxins, generated by one genus of a defective fungi, aspergillus could be responsible for the development of this malformation. We suggest that pregnant women in areas with high incidence of the malformation be screen thorough with ultrasound scan to detect the malformation earlier. We also suggest that further studies on the malformation be geared toward using 3D ultrasound to study the pathophysiology of this malformation.

INTRODUCTION

Frontoethmoidal meningoencephalocele (FEM) is a congenital malformation that encompass the herniation of meninges and cerebral tissue via a defect in the anterior cranium, mostly between the frontal as well as ethmoidal bones(1-3). The herniation could also occur at the points of meeting of the nasal, maxillary, as well as lacrimal bones(1). Although rare, the condition has been reported worldwide with high incidence or occurrences in particular geographical location (1, 4, 5). FEM usually presents with swellings on the upper face which are either sessile or pedunculated. On palpation they may vary from solid and firm to soft and cystic. The skin over such masses may be normal in appearance, thin and shiny or thick and wrinkled (6, 7). Congenital abnormalities such as hypertelorism, middle nasal fissure, wide nasal root, as well as cleft lip or palate is associated in about a third of patients. Also, optic malformations such as anophthalmia/microphthalmia, colobomas, retinal idiosyncrasies, morning glory syndrome, as well as optic nerve or chiasm hypoplasia have been associated with most FEM. Furthermore, cerebral malformations such as corpus callosum agenesis have been reported in about 50% of cases. Also, hydrocephalus and pituitary dystopia/hypoplasia has been reported(8-10). Although most authors have speculated that without typical facies, the diagnosis can be hidden up to adolescence or adulthood, none of them have reported a case or case series demonstrating the silent and progressive nature of this congenital malformation from childhood to adulthood. No data has also associated seizure with this adult presentation although a case series by Mahapatra et al reported a few pediatric cases with convulsions(11). We therefore present a case of silent congenital ethmoidal encephalocele progressing into FEM with episodic seizures in adulthood and concise literature review on the pathogenesis.

CASE REPORT

We present a 20-year-old man with a swelling on the nasal bridge which was notice a few years after birth. The swelling increased in size gradually without any symptoms until
adulthood when he started having episodic seizures. "He first presented at a local hospital in Yunnan Province and was referred to West China Hospital for further assessment and management." The obstetric history of this man was uneventful and childhood was equally uneventful. His parents did not seek any health intervention initially because he was very well until two years ago when he started having episodic seizures. Family history of this disease was unremarkable. Other children of his age group do not have such abnormalities in the community in which he lives. On examination we found a soft mass on the nasal bridge measuring 5x6cm with ocular hypertelorism. He could not stand steadily. Other physical examination of nervous system was unremarkable. All other systems where normal. Routine laboratory investigations done were all normal.

Both computed tomography scan (CT-scan) (Figure 1, A & B) and magnetic resonance imaging (MRI) done revealed depression of the anterior cranial fossa. The upper and lower frontal sinus walls, the left fronto-anterior wall and the lateral wall were defective resulting in the formation of a frontal hernia. The frontal lobes and meninges were seen intruding into the nasal vestibule as well as the adjacent left anterior orbit. There were no bilateral eye abnormalities. The optic nerve as well as the rectus muscles were visible with no obvious abnormalities. The brain tissue and the ventricular system were normal. However, at posterior fossa, there was congenital atlanto-occipital fusion deformity, posterior displacement of the odontoid process, cervical spine narrowing but no cerebellar tonsillar herniation (Figure 2, A-F).

The aim of surgical intervention was to reduce the encephalocele, achieve watertight closure of the dura mater, closure of the skull defect, as well as reconstruct left orbital defect. After general anesthesia, the patient was put on supine position and the head fixed in the Mayfield three keys. A V-sharp scalp marking was done followed by draping with antiseptic solution and povidone iodine. A bifrontal craniotomy was done and through left fronto-orbital angle, the content of hernia in the nasal vestibular (dura and brain tissue) were carefully retracted back. The defective dura mater was repaired with autogenous fascia, and ethmoid bone defect and frontal sinus repaired with titanium mesh. The orbital defect was also repaired with a mesh. Intra-operatively we noted that the herniated part of frontal lobe was short of blood supply. Post-operative CT-scan as well as MRI (Figure 3, A-D) revealed total closure of the defects. He recovered markedly after operation and was discharged home on the seventh day. There were no major complications after surgery. Scheduled out-patient reviews were arranged on every sixth month after operation up to two years.

**DISCUSSION**

FEM is usually made up of anterior skull defects that originate between the fronto-ethmoidal bones and end outwardly on the facial bone(1, 2). The incidence and prevalence of FEM is meticulously linked to particular geographical location. A birth assessment in Bangkok revealed a prevalence of 1:6000 of FEM in every life birth(1, 12). Similar prevalence were reported in a community study in Thailand(1, 12) and by postal surveys in Burma(1, 4). Epidemiological we think our case is coming from Yunnan Province which share borders with the above geographical location hence may have similar predisposing factor with cases reported from these regions.

Thu and Kya proposed that aflatoxins, generated by one genus of a defective fungi, aspergillus could be responsible for the development of this malformation(1, 4). In Thailand, these toxins have been isolated in food samples obtained from markets, secretions as well as tissues of normal and sick kids. Ochratoxin A which is one of the aflatoxins, has proven to stimulate a range of gross idiosyncrasies like exencephaly, anophthalmia as well as cleft lip-in mouse fetuses. (1, 14).
It has been proven that these toxins may find their way into foodstuffs during humid months when foodstuffs are stored for long periods in poorly ventilated areas. The toxins are not destroyed by heat hence cooking doesn’t eliminate them (1).

De Klerk proposed that adhesions between the neuroectoderm and the surface ectoderm may prevent the normal ingrowth of mesoderm to form a normal skull (4). Embryonically, encephalocele occurs as a result of none fusing of neural tubes at the level of the anterior neuropore during the 25th day of gestation (15, 16). The non-fusing of neural tube is as a result of viral infection, radiation, salicylates, hypervitaminosis, trypan blue, hyperthermia, as well as hypoxia. It has been shown that neuroschisis occurs on the neuroepithelium, followed by a 'neurocystic bleb' and subsequently expansion and rupture at the surface of the neuroectoderm (15, 17).

Many theories have been hypothesized for the development of an anterior encephalocele (6, 18): i) Primary osseous defect resulting in non-closure the ethmoidal plate near the olfactory nerve and subsequently herniation of the brain at a later stage. ii) Augmented ventricular pressure in the embryo could force the budding brain through poorly formed osseous structures. iii) Jeffrey-Saint-Hillaire theory is by far the most satisfactory: “The skull originates from two portions,” the endochondral cranial floor as well as the intrembranous cranial vault. In infant, the frontal and ethmoidal bones are fused, but during embryogenesis they separated. Therefore, a potential defect may occur between the frontal and ethmoidal bones resulting in herniation. iv) A tenacious craniopharyngeal canal may facilitate the formation of an encephaloceles through the sphenoid bone, but an early herniating of the brain through this canal may result in obstinacy v) Lack of ossification centers in the sphenoid bone is another possible cause of encephaloceles in this area. FEM also highly linked to holotelencephaly, backing the notion of an imperfect stage during embryogenesis (1, 19).

Surgical exploration and Postmortem studies have shown that the opening in the cranium in FEM is usually found between frontal and ethmoidal bones. Also, cases of anterior to the crista galli and cribriform plate at the locus of the foramen caecum has been reported (1, 20, 21). The opening can be single at the midline, bilateral on each side of the midline or unilateral (1, 21). The herniating content often extrudes through the skull in various directions. The most frequent locations usually at the meeting of the nasal, maxillary, as well as the lacrimal bones. This was the typical presentation in our case. In some cases, the hernia exerts direct pressure on orbital structures resulting in anophthalmia or microphthalmia. On the other hand, in these eye idiosyncrasies occurs at contralaterally side of the herniation. The pathogenesis of hydrocephalus is still a matter of debate. Obstruction of the foramen of Monroe has been observed as the cause of herniation in some cases (1)

MRI of the brain is effective in determining contents of the sac while a non-contrast CT-scan with 3D reconstruction is valuable in reconstruction of the skull base defect. MRI or CT angiography is also very helpful in visualizing of the vascular system (8). In our institution, MRI or CT angiogram practice is routine radiology investigation for every patient that has to undergo craniotomy to rule of vascular malformations. We advocate endocrine assessment should be done in every patient. This is because hypothalamic pituitary dysfunction is often found in transsphenoidal encephaloceles. Also, deficiency of anti diuretic hormone as well as growth hormone has been seen in some cases (8, 22, 23).

Mahapatra et al with a large case series observed three cases with FEM who were above 10 years of age (11). However, they did not indicate if there were adults. Netsiri et al in the very recent study with large series fail to explain further on the progressive nature of this congenital defect. They also did not give details of the congenital disease in adulthood although they indicated that the age range and mean age of their patients were 5 days to 43 years and 7 years 9 months, respectively (2). Their recent classification also did not include associated convulsion although Mahapatra et al and our very recent case have associated convulsions. Mahapatra et al further indicated that three of their cases who were all children had recurrent convulsions for which they required long term antiepileptic drugs. They also observed hydrocephalus in two of their cases in whom they passed ventriculoperitoneal shunts (11). They however did not indicate the incidence of convulsion in any of their case that was over 10 years old. Furthermore, our case did not have hydrocephalus so we propose that his seizures could be as a result of the brain matter extending into the frontal defect or due to the associated posterior fossa malformation. FEM can cause recurrent meningioencephalitis due to direct connection of the central nervous system (CNS) with the external environment which accelerates the entry of pathological microorganisms (6).

Also, cases of FEM with craniofacial deformity with related interorbital hypertelorism has been reported. This presentation is very rare but true orbital hypertelorism/telorbitism occur because, the medial orbital walls are usually widened (6). Secondary trigonocephaly could lead to
CONCLUSION

Aflatoxins, generated by a defective fungus, Aspergillus, could be responsible for the development of this malformation. CT scan and MRI allow particular education of FEM and associated malformations. The aim of surgical intervention should be focus on reduction of encephalocele, watertight closure of the dura mater, closure of the skull defect, and reconstruction of associated defects. We suggest that pregnant women in the geographical location where the congenital malformation is prevalent screen thoroughly during antenatal care to detect this malformation early as well as study the pathogenesis of the malformation with ultrasound scan.

DECLARATION

Ethics approval and consent to participate: The ethical committee of the West China Hospital full approved our case study. The patient and his relatives were informed about our intention to involve him in a case study and he/she agreed to partake in the study. He/she signed the concern form before the operation was carried out according to all surgical protocols.

Consent for Publication: The patient and his relatives were dually informed about our intention to publish his case and he/she fully concerted to the use of his documents. The hospital also concerted to the use of his information for publication.

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