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Clinical Record

Frontoethmoidal encephalomeningocoele with colpocephaly: case report and clinical review

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Abstract

Frontoethmoidal encephalomeningocoele is a rare congenital disease in which an intracranial mass protrudes through a midline defect from the anterior cranial fossa into the facial skeleton. The condition affects patients in South East Asian countries, such as Thailand, Burma, Malaysia and Indonesia, with frequency of 1 in 5000. The pathogenesis of encephalocoeles may be regarded as a 'late' neurulation defect during the fourth gestational week.

We present a case of frontoethmoidal encephalomeningocoele with corpus callosal agenesis and colpocephaly; this may well be the first report of this combination. The patient had a bulging mass in the middle frontonasal area, with broadening of the nasal bridge and hypertelorism. Computed tomography scans delineated the skull defect and associated brain anomalies. A one-stage, combined transfacial–transcranial approach, correctional procedure was performed. We present here a discussion of the findings, with special reference to the condition's pathogenesis, morphological classification and evolving surgical treatments.

Early diagnosis and referral, involving multidisciplinary teamwork, are of paramount importance because of the distorting influence of the extruding mass on facial growth.

Key words: Encephalocele, Frontal; Congenital Malformations, Nervous System; Skull Base; Tomography, Computerized

Introduction

Frontoethmoidal encephalomeningocoele is a herniation of the intracranial contents through a defect in the skull at the junction of the frontal and ethmoidal bones. Encephalocoeles usually occur in the mid-sagittal plane anywhere from the frontonasal region to the occiput; they may be anterior (when found anterior to the coronal suture), parietal (when found between the coronal and lambdoid sutures) or occipital (when found at or posterior to the lambdoid suture).[1]

Frontoethmoidal encephalomeningocoeles are anterior encephalocoeles and are generally named according to their extracranial pathway and facial presentation. Depending on the facial component of the defect, a frontoethmoidal encephalomeningocoele is classified as nasofrontal, nasoethmoidal or naso-orbital, although there may be some overlap or multiplicity. They constitute a well defined clinical entity with remarkable epidemiologic peculiarities, which affects patients in South East Asian countries, such as Thailand, Burma, Malaysia and Indonesia, with a frequency of 1 in 5000. A disturbance in the separation of the neural and surface ectoderm may be the pathogenetic basis of the frontoethmoidal encephalocoele.[2]

Patients with frontoethmoidal encephalomeningocoele present at birth with soft tissue swelling at the nasal bridge, an elongated face and telecanthus. The size of the swelling increases as the child grows. Patients may display a wide range of abnormalities, including ophthalmological problems, intracerebral malformations, shunt-dependent hydrocephalus and meningitis. Anosmia is infrequent and patients generally show normal psychomotor development.

We present a patient with frontoethmoidal encephalomeningocoele who displayed corpus callosal agenesis and colpocephaly; this may well be the first report of this combination. The investigation and treatment of this case involved a multidisciplinary approach.[3]

Case report

A one and half year old boy was born with a small bulging mass, completely covered by skin, in the middle frontonasal area, which gradually increased in size, leading to broadening of the nasal bridge and hypertelorism. There was no hereditary or personal antecedent history. The swelling measured about 1 cm in diameter and was soft to firm in consistency, with the centre of the swelling appearing unhealthy. The patient's parent gave a history of seizures and delayed developmental milestones. Thorough clinical and radiographic investigations were performed, including three-dimensional computed tomography (CT) scans.

Thorough clinical and radiographic investigations, including three-dimensional CT scanning, were performed. Non-enhanced CT scans showed a frontoethmoidal cephalocoele, containing tissue iso-dense with normal brain, and also a bony defect. Intracranial pathological findings of interest included a hypoplastic frontal horn of the left lateral ventricle, a pointed configuration of the frontal horn of the right lateral ventricle, and colpocephaly involving the occipital horn of the right lateral ventricle and the

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temporal horn of the left lateral ventricle, with wide separation of the occipital horns. Corpus callosal agenesis was evident. Left frontal lobe focal grey matter heterotropia was also seen. The third ventricle was continuous with the interhemispheric fissure. The cerebellum and brainstem appeared normal.

A diagnosis was made of frontoethmoidal encephalomeningocoele with corpus callosal agenesis with colpocephaly and focal grey matter heterotropia in the left frontal lobe. The case was surgically managed by a combined ENT and neurosurgical team, using a one-stage, combined transfacial–transcranial approach correction.

Discussion

We report a rare variety of frontoethmoidal encephalomeningocoele with intracranial malformation. Encephalomeningocoele, especially in the frontoethmoidal region, is a form of neural tube defect. It is a rare condition, and only a few large series have been published in the literature. The reported incidence in the West is between 1:35 000 and 1:40 000 live births; however, the incidence is much higher in South East Asian countries. In Thailand, the incidence is as high as 1:5000 live births, which accounts for 15 per cent of all neural tube defects.[4] Sadewa *et al.* reported frontoethmoidal encephalomeningocoele to be the most common form of neural tube defect in East Java, Indonesia.[5] Its underlying cause is not known, but teratogenic environmental agents and nutritional deficiency (an established cause of spina bifida) cannot be excluded. The condition does not show an increased risk of recurrence in siblings and offspring, unlike other cephalocoeles and neural tube defects.

The pathogenesis of frontoethmoidal encephalomeningocoele is primarily based on a disturbance in the separation of the neural and surface ectoderm at the site of final closure of the rostral neuropore, during the final phase of neurulation in the fourth week of gestation. This disturbance in separation may be caused by insufficient apoptosis. Such non-separation results in a midline mesodermal defect, causing a median skull defect. The outgrowth of the nasal septum, with concomitant forward displacement of epidermis (surface ectoderm) and attached brain tissue (neural ectoderm) may act as a herniating force.[6] The craniofacial deformity may consist of hypertelorism, orbital dystropia, elongation of the face or dental malocclusion,[7] associated with cerebrospinal fluid rhinorrhoea, poor visual development, recurrent meningitis and brain abscess.

Boonvisut *et al.* have categorized such skull base defects as follows.[8] Type I defects comprise a single external opening between the frontal, nasal, ethmoidal and orbital bones; in type IA, the opening is limited to between two bones within this area, while in type IB the opening extends transversely or cephalad to involve adjacent structures. Type II comprises multiple external openings in the region; in type IIA, all of the openings are limited, while in type IIB one or more of the openings is extended and involves adjacent structures. This classification was helpful in understanding the herniation pathway in our patient.

Frontoethmoidal encephalomeningocoele can present as a unilateral hydrocephalus, albeit rarely, and careful attention is required to visualise the obscured hemisphere.[9] Frontoethmoidal encephalomeningocoele may be associated with intracerebral malformations. In our case, CT scanning revealed corpus callosal agenesis with colpocephaly. Abnormal enlargement of the occipital horns of the lateral ventricle occurs when there is underdevelopment or lack of thickening of the white matter in the posterior cerebrum. The cause of colpocephaly is unknown, and it is often misdiagnosed as hydrocephalus. Absence of the frontal sinus is also a predominant finding in frontoethmoidal encephalomeningocoele.[10] Shah *et al.* reported herniation of the anterior horn of the lateral ventricle in a frontoethmoidal encephalomeningocoele.[11]

The examination of choice is coronal CT, that is, performed axially with coronal reconstructions, and ideally with intrathecal contrast material delineating the basal cisterns.[12] Rojvachiranonda *et al.* suggested an alphanumerical system of frontoethmoidal encephalomeningocoele classification based on morphological findings on three-dimensional CT images, regarding facial deformities, external bony defects, herniation exit pathways and brain malformations.[13] This system emphasised the fact that frontoethmoidal encephalomeningocoele has a range of manifestations governed by dynamic interaction between structural defects and herniation.

- **Frontoethmoidal encephalomeningocoele is a rare congenital disease in which an intracranial mass protrudes through an anterior midline defect in the skull base**
- **It is relatively more common in South East Asian countries, where the incidence is 1 in 5000 births**
- **Early diagnosis and the involvement of a multidisciplinary team are of paramount importance in management**

Pre-operative investigation includes three-dimensional CT scanning of the facial skeleton. Surgical management is multidisciplinary in nature. The aim is to restore functional brain tissue to the cranial cavity, perform dural repair and correct bone lack before the deformity disturbs facial growth or alters stereoscopic vision. Combined craniofacial and neurosurgical intervention permits one-stage correction of the whole deformity, the associated hypertelorism being reduced by selective orbital translocation and fixation and the nasal deformity by rhinoplasty.[14] Used in place of the classical Tessier's operation, this technique gives good overall cosmetic outcome.[4] In their study, Mahatumarat *et al.* used the Chula technique, which involves the removal of mass, repair of bony defect and restoration of aesthetic facial appearance in a single-stage operation, without formal frontal craniotomy; they reported a satisfactory cure rate, safety and aesthetic outcome.[15] Boonvisut *et al.* reported one-stage closure of the skull defect with a medial orbital composite-unit translocation technique, which avoided augmentation rhinoplasty, allowed convenient resection of the mass and closure of the defect, restored normal interorbital and intercanthal distances, and eliminated the need for a transnasal medial canthopexy.[16] Holmes *et al.* also recommended a one-stage repair using both transcranial and external approaches; they emphasised that avoidance of a 'long-nose' deformity (often seen after repair) should be a priority for successful correction of frontoethmoidal encephalomeningocoele.[17] For a small bony defect, Mahatumarat *et al.* reported definitive clinical evidence of spontaneous closure, which proved that the skull defect and brain herniation were able to heal naturally; they thus suggested that conservative treatment may be considered in those with a small bony defect, with surgery considered later when required.[18] If frontoethmoidal encephalomeningocoele is associated with hydrocephalus, hydrocephalus should be dealt with

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first, followed by elective, single-stage reconstructive surgery.[19]

In selected paediatric patients, transnasal endoscopic repair can also be performed, which has less morbidity and fewer complications.[20] Prenatal diagnosis and information on prognosis and recurrence risk can be established from sonographic findings.[21] The prognosis depends largely on the presence of additional congenital anomalies of the brain.

The possibility of an intranasal encephalocoele must be considered when examining adult patients with nasal polyps. Nasopharyngeal endoscopy should also be performed, as large skull base lesions sometimes present only with a nasopharyngeal mass causing obstructive sleep apnoea.[22] An entirely atypical case of intranasal encephalocoele with no external swelling may sometimes present diagnostic difficulties.[23] Frontoethmoidal encephalomeningocoele must be differentiated from other midline nasofrontal masses, such as epidermal cysts, lipomas, haemangiomas and nasal gliomas.[24]

Conclusion

The current knowledge available in the medical literature illustrates the fact that frontoethmoidal encephalocoele is distinctive in its demographic distribution, in its effect on the growth of other facial structures and in the combined craniofacial approach required for treatment.

Despite many theories, the cause of congenital cephalocoele is still not known. Further investigation of the role of nutritional deficiencies, especially that of folic acid, in the pathogenesis of encephalomeningocoele is necessary to clarify the underlying mechanisms.

Frontoethmoidal encephalomeningocoeles occur with noteworthy frequency in South and South East Asia and require separate consideration for both genetic counselling and treatment.

This case report presents a case of frontoethmoidal encephalomeningocoele associated with gross intracerebral malformations of colpocephaly, with corpus callosal agenesis. Three-dimensional CT scanning well delineated the anatomy of the herniated mass and was helpful in planning treatment.

One-stage, combined craniofacial and neurosurgical intervention is the treatment of choice. Antenatal diagnosis is possible, and management of this condition should involve a multidisciplinary team approach. Early intervention allows normal growth forces to be re-established, thus achieving a good aesthetic and functional result.

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