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Case report

Transsellar trans-sphenoidal encephalocele with cleft lip, cleft palate and agenesis of corpus callosum ☆

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ABSTRACT

Basal encephalocele is a rare congenital malformation. Among basal encephaloceles, the transsellar, trans-sphenoidal encephalocele is the least common subtype. We present the case of a newborn female, who presented to us with cleft lip and cleft palate. Diagnostic neuroimaging revealed the presence of transsellar trans-sphenoidal encephalocele along with agenesis of the corpus callosum. There are very few case reports of trans-sphenoidal encephalocele with corpus callosum agenesis in a patient with midline cleft lip and palate. In this report, we discuss the clinico-radiological findings of this extremely rare condition and present a brief review of the literature.

Encephalocele is a congenital defect in the skull and the dura through which intracranial matter herniates outside. The transsellar, trans-sphenoidal variant is the least common subtype of encephalocele, and only a few cases have been previously reported. We report a case of transsellar, trans-sphenoidal encephalocele in a neonate with cleft lip and cleft palate. Additionally, there was agenesis of the corpus callosum. The combination of these rare congenital anomalies is seldom seen. There is a possible common link in the etio-

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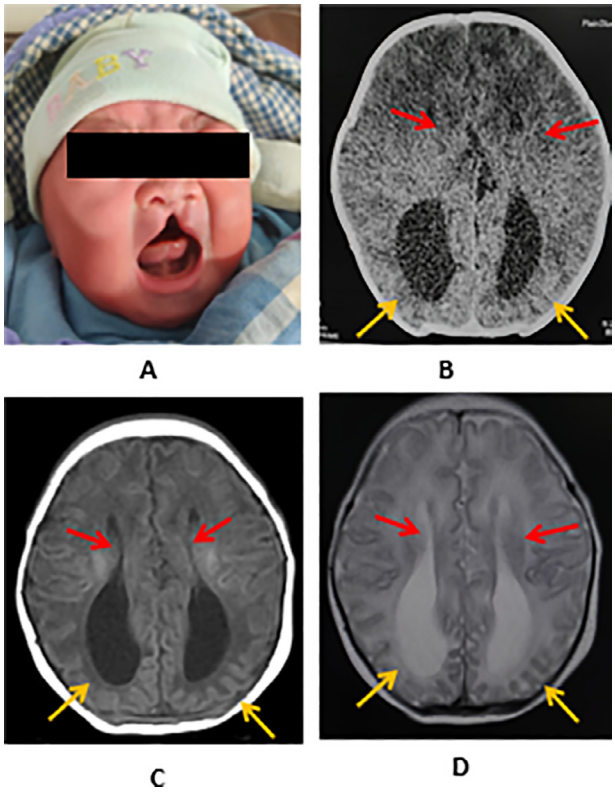

Fig. 1 – (A) Patient with a cleft lip. (B) Axial computed tomography and (C, D) Axial T-1 and T-2 weighted magnetic resonance imaging of the brain show non-converging lateral ventricles (red-arrows) with disproportionately enlarged occipital horns suggestive of colpocephaly (yellow-arrows) (color version of figure is available online.)

genesis of these craniofacial anomalies as the development of basal encephalocele, cleft lip, and cleft palate occur by around the 12th week of gestation.

Case report

A neonate born at full term was shifted to intensive care due to the presence of cleft lip and cleft palate. The baby was active and had good muscle tone and activity. She did not have any abnormal body movement, seizure, or vomiting. The nasal alar base was broad with midline cleft lip and cleft palate (Fig. 1A). Cranial ultrasound was done, which suggested the possibility of corpus callosum dysgenesis. Subsequently, a non-contrast magnetic resonance imaging (MRI) of the brain was performed, which revealed non-converging lateral ventricles with disproportionately enlarged occipital horns suggestive of colpocephaly (Fig. 1B). The rostrum, body, genu, and splenium of the corpus callosum were not visualized, suggesting complete agenesis of the corpus callosum (Fig. 1C).

Additionally, a midline defect was seen in the sphenoid bone with herniation of the meninges, cerebrospinal fluid, and the pituitary gland through the defect (Fig. 2), consistent with the diagnosis of transsellar trans-sphenoidal encephalocele.

After the radiologic diagnosis, neurosurgical and plastic surgical opinions were sought. As the newborn child was at high risk of a dysregulated hypothalamic-pituitary axis, endocrinological consultation was also done. Laboratory investigations for thyroid-stimulating hormone, T3, T4, prolactin, insulin-like growth factor-1, and cortisol were normal. She was planned to undergo combined repair of the cleft lip and palate at around 6 months of age, followed by endoscopic repair of the encephalocele at a later date.

Discussion

Encephaloceles are uncommon congenital malformations with an incidence of 1.64 cases per 10,000 live births [1]. Basal encephaloceles are even less common, with an estimated incidence of 1 in 35,000 live births [2]. Basal encephaloceles are classified into trans-ethmoidal, spheno-orbital, sphenomaxillary, and trans-sphenoidal subtypes. Trans-sphenoidal encephalocele is the rarest subtype of basal encephaloceles. Patients with trans-sphenoidal encephalocele usually present in childhood and may have associated craniofacial anomalies such as cleft lip, cleft palate, facial hypoplasia, ocular deformities, craniosynostosis, and hypertelorism. However, some patients are not diagnosed in childhood and may present with severe symptoms later in life, such as cerebrospinal fluid rhinorrhea, meningitis, endocrine dysfunction, and visual deficits.

There are several embryological theories to explain the formation of trans-sphenoidal encephaloceles. The most accepted theory proposes defective ossification of the body of the sphenoid bone with the persistence of the craniopharyngeal or trans-sphenoidal canal, resulting in a vertical midline skull base defect extending from the floor of the sella to the nasopharynx [3].

Neuroimaging is needed to confirm the diagnosis and to identify the content of the sac. A computed tomography scan with 3-D reconstruction is helpful to identify the bony skull defect. An MRI is useful to identify the content of the sac and other intracranial anomalies. An endocrine workup is important as hypothalamic-pituitary-adrenal axis dysfunction is often seen [4].

The prognosis of trans-sphenoidal encephalocele depends on the size, content of the encephalocele, and other intracranial and systemic anomalies. A multidisciplinary approach is required to manage these complex cases. Early neurosurgery provides definitive treatment in most cases, as it is technically easier to identify intracranial connections and to achieve a complete repair of the dural defect. Surgical repair can be done by transcranial, transpalatal, and trans-sphenoidal approaches. These approaches have their advantages and limitations. When there is a co-existing cleft palate, a transoral transpalatal approach is preferred for preserving and repositioning the content of the sac [5]. In this approach, there is relatively less risk of damaging functional tissue in the wall of the encephalocele, and it is easier to dissect the sac from the adjacent neural structures.

Hypogenesis or agenesis of the corpus callosum can be associated with basal encephalocele. Corpus callosum, which develops between the 12th to 20th weeks of gestation, is the

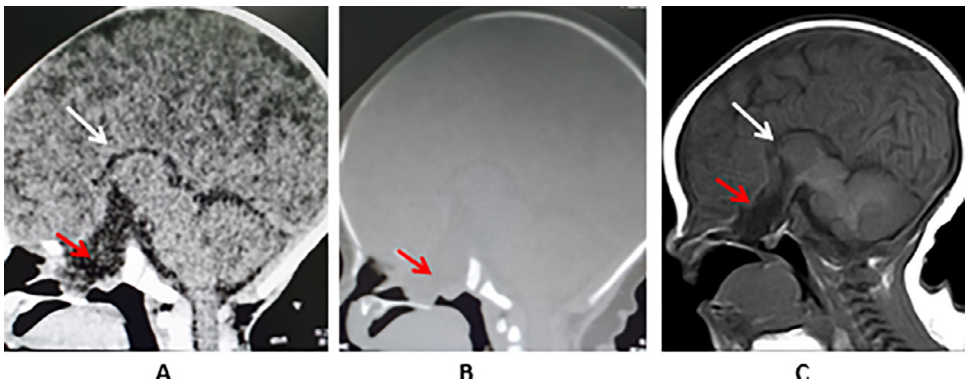

Fig. 2 – (A, B) Sagittal computed tomography soft tissue and bone window and (C) Sagittal T-1 weighted magnetic resonance imaging of the brain show complete absence of the corpus callosum (white-arrows) and midline sphenoid defect with herniation of the meninges and cerebrospinal fluid (red-arrows) (color version of figure is available online.)

largest cerebral commissure connecting neocortical areas [6]. In complete agenesis of the corpus callosum, common neuroimaging findings include widely separated non-converging lateral ventricles, disproportionately enlarged occipital horns suggestive of colpocephaly, and high-riding third ventricle. Gyri on the medial hemispheric surface typically radiate outward from the high-riding third ventricle. In partial agenesis of the corpus callosum, only the splenium and rostrum are generally absent.

Clefts of the lip and palate are heterogeneous disorders that occur either as isolated defects or are present together. The incidence of these defects is about 17 cases per 10,000 live births [7]. The basic morphology of the face is established between the 4th and 10th weeks of conception. The upper lip starts forming on the 24th-day post-conception and is complete by the 37th day [8]. Palatogenesis starts by the fifth week of gestation and the development is complete by the 12th week [8]. The etiology of the orofacial cleft is complex and multifactorial. It represents an interaction between the individual's genetics and the environment during a critical stage of intrauterine development. Several genes causing cleft lip and palate have also been discovered [9]. The basal encephalocele, cleft lip, and cleft palate are a spectrum of craniofacial abnormalities that occur by the 12th week of gestation. Hence, there can be a common link in their etiopathogenesis. However, the association of agenesis of the corpus callosum with cleft lip and cleft palate is not straightforward, as the corpus callosum develops later between the 12th to 20th weeks of gestation [10].

To conclude, in the presence of midline craniofacial anomalies, there should be a high index of suspicion for other intracranial lesions. Early diagnosis and treatment of intracranial anomalies, like basal encephalocele, are important for good clinical outcomes.

Patient consent

Written informed consent for publication was obtained from the patient's legal guardian.

Consent for publication

All authors expressed explicit consent for the publication of this manuscript.

Disclosures

None

REFERENCES

[1] Siffel C, Wong LY, Olney RS, Correa A. Survival of infants diagnosed with encephalocele in atlanta. 1979-98. Paediatr Perinat Epidemiol 2003;17:40–8.
[2] Smith DE, Murphy MJ, Hitchon PW, Babin RW, MM Abu-Yousef. Transsphenoidal encephaloceles. Surg Neurol. 1983;20:471–80.
[3] Naidich TP, Altman NR, Braffman BH, McLone DG, Zimmerman RA. Cephaloceles and related malformations. AJNR Am J Neuroradiol 1992;13:655–90.
[4] Rao RC, Larson SA, Dlouhy BJ. Growth hormone deficiency, basal encephalocele, and morning glory anomaly. Ophthalmology 2017;124:1531.
[5] Lewin ML. Sphenoethmoidal cephalocele with cleft palate: Transpalatal versus transcranial repair. Report of two cases. J Neurosurg 1983;58:924–31.
[6] Koshi R, Koshi T, Jeyaseelan L, Vettivel S. Morphology of the corpus callosum in human fetuses. Clin Anat 1997;10:22–6.
[7] Mossey PA, Little J, Munger RG, Dixon MJ, Shaw WC. Cleft lip and palate. Lancet 2009;374:1773–85.
[8] Levi B, Brugman S, Wong VW, Grova M, Longaker MT, Wan DC. Palatogenesis: Engineering, pathways and pathologies. Organogenesis 2011;7:242–54.
[9] Wong FK, Hagg U. An update on the aetiology of orofacial clefts. Hong Kong Med J 2004;10:331–6.
[10] Achiron R, Achiron A. Development of the human fetal corpus callosum: A high-resolution, cross-sectional sonographic study. Ultrasound Obstet Gynecol 2001;18:343–7.