

Binay Gurung¹
 Urmila Gurung²
 Laxman Shrestha¹

Department of Pediatrics, Institute of Medicine (IOM), Tribhuvan University Teaching Hospital (TUTH), Maharajgunj, Kathmandu, Nepal¹

Ganesh Man Singh Memorial Academy of ENT - Head & Neck Studies, TU Teaching Hospital, Kathmandu, Nepal.²

Corresponding Author:

Dr. Binay Gurung

Department of Pediatrics, Institute of Medicine (IOM), Tribhuvan University Teaching Hospital (TUTH), Maharajgunj, Kathmandu, Nepal

Email: binay_gg@hotmail.com

BASAL ENCEPHALOCELE – AN UNCOMMON CAUSE OF RESPIRATORY DISTRESS IN NEWBORN

Abstract:

Basal encephalocele is one of the important causes of respiratory distress in newborn, although uncommon and hence under emphasized. It is a form of neural tube defect that usually presents with symptoms of nasal obstruction. Delayed presentation with variable symptoms have been noted. Collaborative surgical intervention is the treatment modality with different approaches. However, intervention early in neonatal period is a challenge in our region with resource limitations. Prognosis is worse with associated malformations and chromosomal anomalies. We report two cases of newborn with basal encephalocele presenting as respiratory distress at birth.

Keywords: Encephalocele, Nasal obstruction, Neural tube defect, Respiratory distress in newborn

INTRODUCTION:

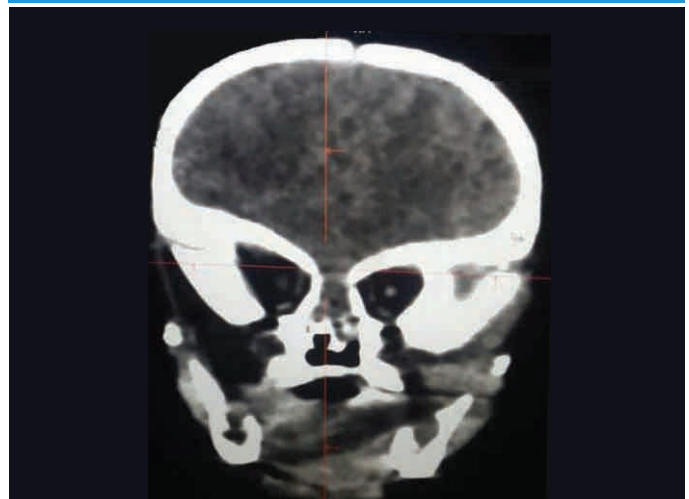
Respiratory distress in newborn results from a myriad of causes. Basal encephalocele is an uncommon cause which has an incidence of 1 in every 20,000 – 40,000 live births.¹ The brain tissue and meninges that herniate through a defect in the anterior skull base obstructs the nasal cavity. Initial blind interventions either to evaluate the patency of the nasal cavity or to secure a nasopharyngeal airway can rupture the encephalocele with inadvertent complications. Inadequate antenatal care and lack of prenatal diagnosis in resource-limited countries hinders prompt diagnosis and appropriate management. Surgical repair in early neonatal period is technically challenging. We report two cases of basal encephalocele presenting as respiratory distress in immediate newborn period, a rare cause unreported till date in Nepal.

CASE REPORTS:

Case 1

A vaginally delivered term (40 weeks of gestation) baby girl weighing 2750 grams developed respiratory distress at three hours of life and was referred to Tribhuvan University Teaching Hospital (TUTH), a tertiary referral center in Kathmandu, with a suspicion of bilateral choanal atresia. Although the antenatal care was inadequate, history review did not suggest maternal and obstetrical issues. Apart from respiratory distress, skull and spine was normal without features of dysmorphism. Attempts to negotiate nasogastric tube into both nose failed. Initial evaluation with nasopharyngeal laryngoscopy (NPL) suggested bilateral choanal atresia, however Computed Tomography (CT) scan head revealed basal encephalocele (Fig. 1). The baby succumbed following discontinuation of care on parental request.

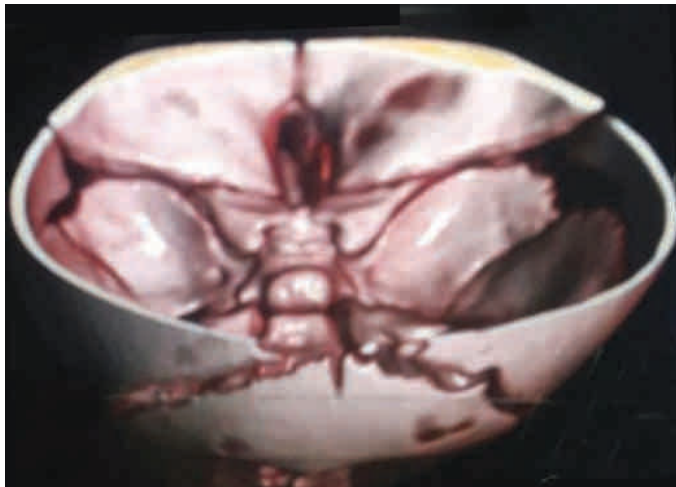
Fig: 1. Axial CT head showing basal encephalocele.



Case 2

A 3000 grams baby boy born vaginally in TUTH at 38 weeks of gestation, was evaluated for difficulty feeding few hours after birth. Nasal patency was dubious in view of mid-face hypoplasia with small nasal orifices and signs of upper airway obstruction on feeding. Although nasogastric tube could advance through both nose, further evaluation of nasopharyngeal area with imaging study showed basal encephalocele (Fig. 2). NPL excluded laryngomalacia. The course was complicated with hypoxic episodes and seizure while undergoing imaging studies. A multidisciplinary definitive therapeutic plan was devised. However, parental request for discontinuation of care was honoured.

Fig. 2. 3D reconstructed CT head image showing absence of cribriform plate which was not evident on standard sections.



DISCUSSION:

Newborns until initial two months of life are obligate nasal breathers. Therefore, any congenital cause of nasal obstruction results in respiratory distress. Choanal atresia is the most common congenital nasal anomaly. Beside other congenital causes like Nasal dermoid, Nasal Glioma and Pyriform aperture stenosis, Basal encephalocele is a rare cause with scarce literature.²

Basal encephalocele is a form of neural tube defect that results from failure of neural tube to close spontaneously between 3rd and 4th week of gestation. Herniation of meninges and brain matter through a defect in the anterior skull base forms basal encephalocele. The mass occupies the space in nasal cavity resulting in obstruction of upper airway and hence respiratory distress in newborn. Other associated midline anomalies such as hypertelorism, broad nasal root, cleft lip and palate may be present.³ Nasal encephaloceles can be divided into frontoethmoidal and basal encephalocele. Further, basal encephalocele is classified as transtethmoidal, sphenoethmoidal, transsphenoidal and frontosphenoidal types depending on the origin. The transtethmoidal is the type that herniates through the defect in cribriform plate.¹ Both forms of nasal encephaloceles are rare, but incidence of frontoethmoidal type is relatively high in Southeast Asia.⁴ Available literature on basal encephalocele depicts variable age of onset and symptom presentation. Upasani et al, reported a case of transtethmoidal encephalocele presenting as a mass protruding out of unilateral nostril at birth.⁵ Yokota et al, reported three cases of transsphenoidal encephalocele diagnosed in neonatal period with progressive obstruction of nasopharyngeal airway.⁶ A case report by Nayal et al, described a 4 year old boy with recurrent episodes of unilateral nasal block.⁷ Interestingly, cases of delayed presentation as spontaneous CSF rhinorrhea and meningitis at adulthood have been reported.⁸ Mahapatra AK, in his largest series of 133 cases over 40 year period on anterior encephalocele, also noted variable symptoms and age of presentation.⁴ Both cases mentioned in our report presented with symptoms of upper airway obstruction

at birth. The diagnosis can be confirmed by either CT (as in our case) or MRI (Magnetic Resonance Imaging). Collaborative surgical interventions performed by neurosurgeon, maxillofacial surgeon and ENT surgeon, as early as possible to reduce chances of infection and deformity is the treatment modality. Successful repair, at the earliest, on fifth day of life has been reported.⁵ Surgical approaches include transfrontal open craniotomy, endoscopic transnasal and transpalatal depending on feasibility and the risks involved. While endoscopic transnasal are evolving as first-line approaches for most lesions, open craniotomy is technically feasible in neonates. Successful transpalatal approach in neonate has also been reported by Hoff et al.⁹ Cases in our study could not undergo definitive treatment in view of technical limitations and clinical complications incurred during the hospital course. Postoperative complications of CSF leak, meningitis, convulsions and epiphora are reported in studies.^{1,3,4} Prognosis appears to be better for frontoethmoidal encephaloceles compared to occipital or parietal encephaloceles, and also depends on associated malformations and chromosomal anomalies.³ The longitudinal course of the previously described patients are unavailable, however, short term post-operative status seems favourable.

Fetal encephalocele is usually detected by antenatal ultrasonography (USG) and maternal alpha fetoprotein level. However, maternal serum and amniotic fluid levels of alpha fetoprotein are normal when lesions are completely epithelialized. Prenatal 2D USG detects approximately 80% of encephaloceles. 3D USG may contribute to early detection and better visual depiction. However, both forms of USG may not detect basal encephalocele which are clinically occult. Fetal MRI has several advantages over USG and is being increasingly used in countries with abundant resources, particularly in cases of abnormal prenatal USG.¹⁰ Encephalocele in our cases was not evident during antenatal period.

CONCLUSION:

Basal encephalocele is an uncommon cause of nasopharyngeal airway obstruction. It has to be considered while evaluating a newborn with respiratory distress. Prenatal diagnosis may not be evident if clinically occult. Multidisciplinary collaborative surgical interventions is the treatment modality. However, technical limitations in our regions create challenge on managing such patients.

REFERENCES:

1. Rahbar R, Resto VA, Robson CD, Perez-Atayde AR, Goumnerova LC, McGill TJ, et al. Nasal glioma and encephalocele: diagnosis and management. *Laryngoscope*. 2003 Dec;113(12):2069-77.
2. Zanetta A, Rodriguez H, Quiroga V, Cuestas G, Tiscornia C. Congenital nasal obstruction due to choanal atresia: case series. *Arch Argent Pediatr*. 2012 Apr;110(2):152-5.
3. Tirumandas M, Sharma A, Gbenimacho I, Shoja MM, Tubbs RS, Oakes Wj, Loukas M. Nasal encephaloceles: a review of etiology, pathophysiology, clinical presentations, diagnosis, treatment, and complications. *Childs Nerv Syst*. 2013 May;29(5):739-44.
4. Mahapatra AK. Anterior encephalocele – AIIMS experience a series of 133 patients. *J Pediatr Neurosci*. 2011 Oct;6 Suppl 1:S27-30.

5. Upasani AV, Patel DN, Chandna SB. A rare presentation of a transthemoidal encephalocele at birth. *Pediatr Neonatol*. 2014 Oct;55(5):404-6.
6. Yokota A, Matsukado Y, Fuwa I, Moroki K, Nagahiro S. Anterior basal encephalocele of the neonatal and infantile period. *Neurosurgery*. 1986 Sep;19(3):468-78.
7. Nayal B, Kudva R, Nanaiah A, Nair RP, Ramaswamy B. Nasal encephalocele – A case report. *International Journal of Advances In Case Reports*. 2015;2(12):736-38.
8. Naoyuki H, Masaaki N, Chikao M, Kosuke K, Hiroyuki M, Jun N, et al. Basal encephalocele in an adult patient presenting with minor anomalies: a case report. *J Med Case Rep*. 2014 Jan;8:24.
9. Hoff SR, Edwards MS, Bailey CM, Koltai PJ. The transpalatal approach to repair of congenital basal skull base cephaloceles. *J Neurol Surg B Skull Base*. 2014 Apr;75(2):96-103.
10. Mehdi K, Sana G, Hassine SA, Aida M, Mohamed BC. Antenatal Diagnosis of a Rare Neural Tube Defect: Sincipital Encephalocele. *Case Rep Obstet Gynecol*. 2015; 2015:613985.

