HK J Paediatr (new series) 2021;**26**:158-160

Case Report

An Extremely Rare Association of Cleft Lip and Palate, Basal Sphenoethmoidal Cephalocele and Corpus Callosal Agenesis in a Three-month Old Patient Presenting with Facial Cleft and Nasal Mass

RJDL ***URGEL***, GG ***VITERBO***

Abstract We present an infant who was born with a midline facial cleft and clinically apparent nasal mass. Non-enhanced facial computed tomography (CT) revealed the cleft lip and palate, and nasal mass - the latter was better defined as a sac containing meninges, cerebrospinal fluid and what appears to be dysplastic brain parenchyma. Further evaluation of the CT images revealed agenesis of the corpus callosum and colpocephaly. A non-enhanced magnetic resonance imaging was unable to be performed as patient was lost to follow-up. The need for neuroimaging may not always be implied in cases of cleft lip and palate. However, the presence of other findings such as a nasal mass may warrant investigation with cross-sectional imaging to determine its aetiology, define the extent of the midline facial cleft and look for other possible anomalies.

Key words *Agenesis of corpus callosum; Encephalocele; Neuroimaging; Orofacial cleft*

Case Report

A 3-month old male was born full-term to a then G3P2 mother via caesarean section. Due to the mother's poor health-seeking behaviour, only a late trimester ultrasound was done, which revealed the presence of a facial cleft. The family history was unremarkable, with no member of the traceable family lineage having a similar condition. A look into the maternal history did not reveal any episodes of infection or unwanted drug intake during the course of pregnancy. Upon delivery, a cleft lip and palate, and an intranasal mass were noted. Pertinent physical exam findings include hypertelorism, flattened nasal bridge, midline facial cleft and a soft mass in the oronasal cavity. There was no note of any abnormal back masses along the spine, sacral dimple or tuft of hair. Cross sectional imaging was requested and was performed to determine the possible aetiology of the nasal mass. However, there was poor compliance to medical advice as the family were not able to do further follow-up. No genetic testing was performed. The patient had an untimely death from unknown causes.

Radiographic Findings

Non-enhanced facial computed tomography (CT) scan showed a soft tissue defect in the upper lip, with associated discontinuity in the midline of the hard palate. The clinically apparent nasal mass was seen as a circumscribed focus with its base along the sphenoethmoidal region. Evaluation of its contents revealed a predominantly hypodense lesion, likely cerebrospinal fluid (CSF), which herniated through a 2.02 cm defect along the sphenoethmoidal region (Figure 1). The sella is not clearly delineated; the pituitary gland is likewise not appreciated in its entirety. The imaged portions of the brain showed absence of the corpus callosum with parallel configuration of the lateral ventricles, and radial arrangement of the parasagittal cerebral gyri. Disproportionate dilatation of the atrium and occipital horns of the lateral ventricles is noted, due to colpocephaly (Figure 2).

Department of Radiology, College of Medicine and Philippine General Hospital, University of the Philippines Manila, Philippines

RJDL **URGEL** *MD, FPCR*
GG **VITERBO** *MD*

Correspondence to: Dr RJDL **URGEL**
Email: urgelmd@gmail.com

Received April 3, 2020

Discussion

A failure in the proper coordination of complex in utero events related to cranial and facial development can lead to clefts.[1] Cleft lip and palate occurs when there is maldevelopment of the upper lip and roof of the mouth, respectively. These may be aesthetic problems; however, these are associated with more pressing concerns, such as feeding and speaking difficulties.[2,3] An interdisciplinary approach, including early repair and support from a speech pathologist, is important to ensure proper development of an infant, particularly its speech.[3]

Cephaloceles are sacs containing intracranial structures, extending beyond the skull, classified depending on the associated bone defect. A nasal cephalocele is a soft tissue focus detected in and around the nasal region, further classified into the frontoethmoidal and basal types – the latter being less common and rarely reported in literature.[4] Such finding must not be mistaken for a tumour, as its manipulation can be detrimental to the patient.

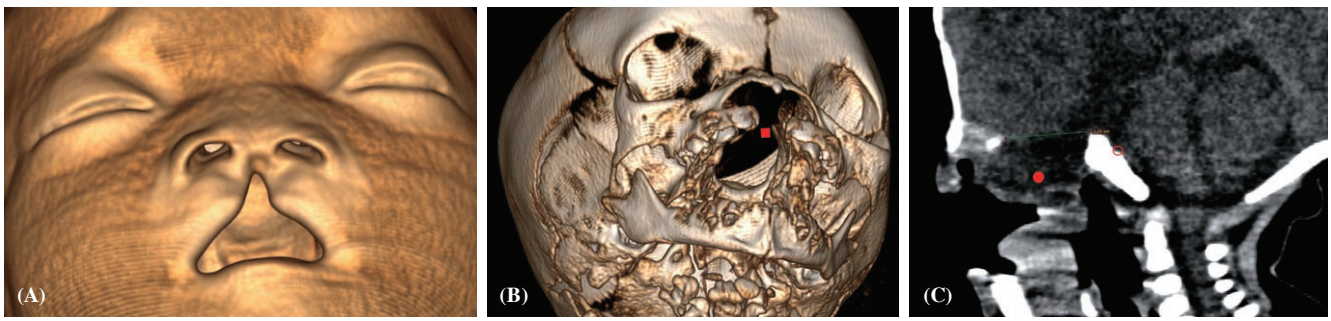

Figure 1 Volumetric rendered images showing the (A) soft tissue defect in the upper lip and associated (B) fusion abnormality of the hard palate and maxillary alveolus (red solid square). (C) Sagittal reconstruction of the facial CT showing the sphenoethmoidal defect. Radiographically, the clinically apparent nasal mass is seen as a sac containing herniated intracranial contents (red solid circle). Note the absent sella turcica with non-visualization of the pituitary gland (red hollow circle).

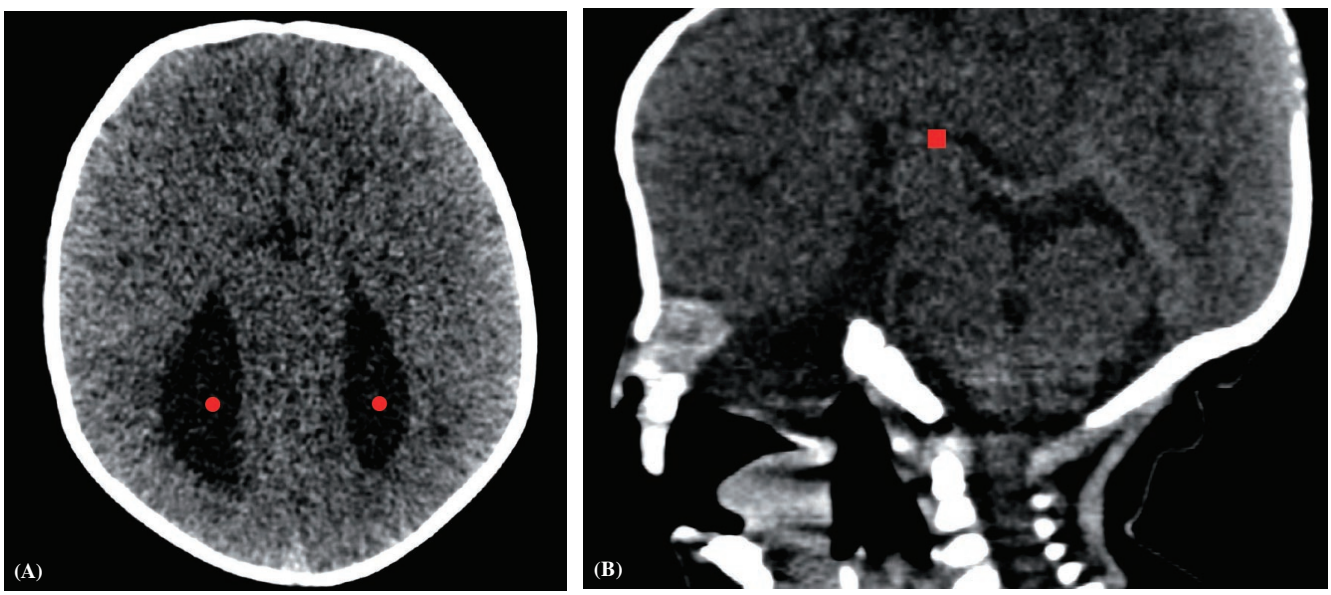

Figure 2 Facial CT, brain window: (A) Axial image showing parallel configuration of the lateral ventricles with disproportionate body and atrial dilatation (red solid circle). (B) Sagittal CT reconstruction showing the radial configuration of the gyri, with absence of the corpus callosum (red solid square)

Facial clefts and cephaloceles can occur simultaneously with high probability, as both are due to failure of structural fusion during embryogenesis. However, their association has not been adequately published in available literature. In our case, the incidentally seen rounded intranasal mass corresponded to a CSF-containing sac that herniated through a sphenoethmoidal defect. The rare incidence of basal cephaloceles and an even rarer association with cleft lip and palate was even made noteworthy by the absence of the corpus callosum. In agenesis of the corpus callosum, there is lack of the structure linking both cerebral hemispheres. It can be seen in isolation or as part of a syndrome and can be due to exogeneous factors during pregnancy or be genetically inherited.[5]

Radiologic imaging is not always needed in cases of cleft lip and palate. In our case, the presence of a nasal mass was the primary reason for performing a CT, as a neoplastic tumour in any site is not common for an infant. Its presence suggested a congenital abnormality or vascular pathology. With cross-sectional imaging, the exact aetiology of the nasal mass was better defined. Likewise, the presence of other congenital anomalies was detected with CT; magnetic resonance imaging may provide more information, but unfortunately was not done to poor patient compliance and follow-up. Ultrasound may also be beneficial despite its limitations in paediatric neuroimaging. Consent was obtained from the patient's mother for case publication.

Conclusion

Radiologic investigation of cases of cleft lip and palate is not always needed but may be considered in patients presenting with other cranial abnormalities. Performing additional imaging procedures in such cases will not only help in identifying associated pathologies not seen clinically but will also help the clinicians in choosing and planning the right management for patients.

Declaration of Interest

No conflict of interest to declare.

References

1. Monasterio FO, Taylor JA. Major craniofacial clefts: case series and treatment philosophy. Plastic and Reconstructive Surgery 2008; 122:534-43.
2. Goswami M, Jangra B, Bhushan U. Management of feeding problem in a patient with cleft lip/palate. Int J Clin Pediatr Dent 2016;9:143-5.
3. Nagarajan R, Savitha VH, Subramaniyan B. Communication disorders in individuals with cleft lip and palate: an overview. Indian J Plast Surg 2009;42(Suppl):S137-43.
4. Howing EW. Nasal encephaloceles. Childs Nerv Syst 2000;16: 702-6.
5. Schell-Apacik CC, Wagner K, Bihler M, Ertl-Wagner B, et al. Agenesis and dysgenesis of the corpus callosum: clinical, genetic and neuroimaging findings in a series of 41 patients. Am J Med Genet Part A 2008;146A:2501-11.