December 2018

Frontoethmoidal encephalocele presenting in concert with schizencephaly

Asra Tanwir
Aga Khan University, Asra.tanwir@aku.edu

Syed Sarmad Bukhari
Aga Khan University, sarmad.bukhari@aku.edu

Muhammad Shahzad Shamim
Aga Khan University, shahzad.shamim@aku.edu

SURGICAL NEUROLOGY INTERNATIONAL
AN OPEN ACCESS, INTERNATIONAL JOURNAL OF NEUROSURGERY AND NEUROSCIENCES
Home | Browse articles | Instructions | Submit articles

Surg Neurol Int. 2018; 9: 246.
Published online 2018 Dec 4. doi: 10.4103/sni.sni_242_18
PMCID: PMC6293601
PMID: 30603230

Frontoethmoidal encephalocele presenting in concert with schizencephaly

Asra Tanwir,* Sarmad Bukhari, and Muhammad Shahzad Shamim

Department of Neurosurgery, Aga Khan University Hospital, Karachi, Pakistan
Asra Tanwir: tanwirasra@gmail.com; Sarmad Bukhari: Sarmad.Bukhari@aku.com; Muhammad Shahzad Shamim: Shahzad.shamim@aku.com
*Corresponding author

Received 2018 Jul 14; Accepted 2018 Sep 25.

Abstract

Background:

Schizencephaly is a rare defect which is identified as clefts that are lined with grey matter extending from the ependyma of the cerebral ventricles to the pia mater. An encephalocele occurs due to failure of neural tube closure resulting in a gap through which cerebrospinal fluid and meninges can bulge into a pouch. There have been rare instances when these two defects have presented simultaneously.

Case Description:

We report a case of a 17-year-old child who was brought by his parents with complaint of swelling over his nose and forehead and aggressive behavior since birth. Magnetic resonance imaging findings were consistent with frontoethmoidal meningoencephalocele with schizencephaly. Lumbar drain was inserted and kept in place for 1 week followed by surgical correction of the defect. Our case is interesting because of delayed presentation as it is a rare entity and its association with schizencephaly.

Conclusion:

Encephalocele association with schizencephaly is rare.

Keywords: Frontoethmoidal, hypertelorism, meningoencephalocele, schizencephaly

INTRODUCTION

Meningoencephalocele results from failure of rostral neuropore closure during the fourth week of development or primary defect of mesoderm or ectoderm and involves overlying tissues such as meninges and calvarias. The causative factors are genetic, drugs, nutritional, and environmental factors. [1,3,6,7,10,11,12,13,14] Schizencephaly is a rare disorder of neuronal migration which is characterized by a cerebrospinal fluid (CSF)-filled cleft extending from the surface of the cerebral hemispheres (pial) to the ventricular surface (ependyma). Schizencephaly results from abnormal neuronal migration during the first few weeks after gestation.[1] Collagen type IV alpha 1 chain (COL4A1) is an important gene associated with schizencephaly, and hedgehog signaling pathway and ectoderm differentiation are among its related pathways/super pathways. Brain, spinal cord, and cortex and growth/size/body region and mortality aging are the related phenotypes.[14]

CLINICAL PRESENTATION

A 17-year-old male child was brought by his parents with complaints of swelling over his forehead and nasal bridge since birth. He underwent primary closure of the swelling at the age of 35 days. Postoperatively, he presented with discharge of clear fluid from the site of incision and discharge was resolved with daily dressing. He remained well for 2 months but swelling gradually started to reappear. The size of the swelling has remained unchanged since then and he had not sought further medical care for this swelling. He was delivered full term at a local hospital. There was no significant antenatal history of intrauterine infections or teratogenic drug use.

On examination, we found a well-behaved child with hypertelorism and a fluctuant swelling over his forehead and nasal bridge approximately 6 × 4.8 cm. The swelling had positive transillumination test and positive cough impulse. There were no other associated anomalies. Milestones were up-to-date. He never went to school because of cosmetic deformity, and as per the parents, the child was extremely aggressive. Neurological examination revealed the extraocular movements to be normal and cranial nerves were grossly intact. There was no pronator drift or distal extremity weakness. He had memory impairment and reduced IQ.

Magnetic resonance imaging (MRI) was consistent with frontoethmoidal meningoencephalocele with schizencephaly [Figures 1 and 2]. The patient was planned for surgery to resect the redundant protruding tissue and close the defect with help from the plastic surgery team. Intraoperatively, the patient was found to have an atrophied left cerebral hemisphere. No histopathology was sent. Dura was reconstructed by fascia taken from the pericranium and fat taken from the abdomen, reconstruction of the nasal bridge was done from bone graft from calvaria which was fixed with plate, redundant skin was excised, and medial canthus was repositioned to correct hypertelorism. Lumbar drain was placed for a week postoperatively to prevent chances of postoperative leak following dural closure. It was planned to perform a rhinoplasty at a later date for him.

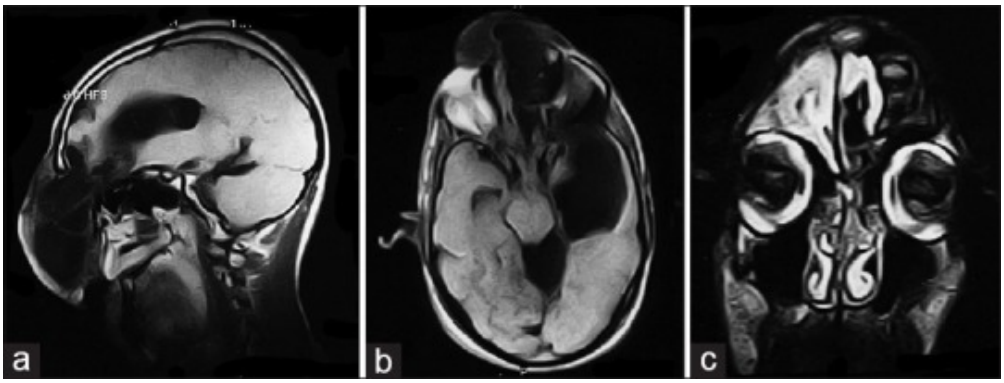

Figure 1

(a and b) Midline frontal cranial defect, more to the left with herniation of the meninges and brain tissue, representing frontoethmoidal encephalocele. The herniated brain tissue has hypotense T1 signals suggestive of gliosis. (c) The defect in coronal section

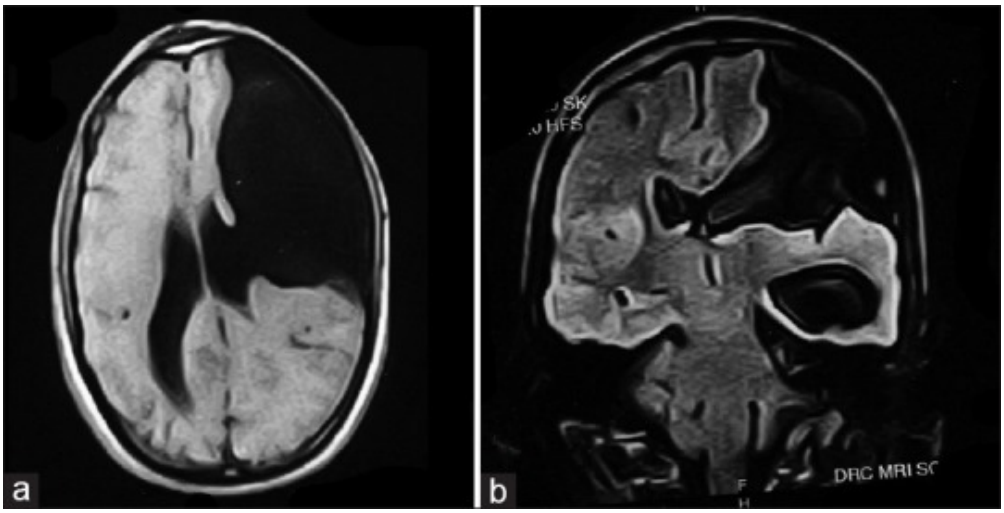

Figure 2

There is a grey matter lined cleft extending through the frontal region on the left down to the lateral ventricles, representing schizencephaly

DISCUSSION

The incidence of encephalocele globally is 1 per 35,000 births, but it is six times more common with 1 in every 6,000 births in South-East Asia.[7] The classification is based on the location – frontal, parietal, and occipital – and herniated contents such as meninges (meningeal) or meninges and parenchyma (meningoencephaloceles).[3] The most common cause is congenital defects secondary to improper closure of neural tube and it occurs in the midline; the cause can also be acquired or spontaneous occurring most commonly in cranial sutures.[3,10] Recent studies have shown a direct

role for collagen IV in rare genetic conditions such as cerebral hemorrhage and porencephaly in infants.[8] In our case, the congenital meningoencephalocele is hypothesized considering the midline position of the lesion, although no clear documentation of such is found.

The case presented is more interesting due to its association with schizencephaly, a rare birth defect with incidence in the United States estimated at 1.54/100,000 births per year.[13] Morphologically, schizencephaly can be divided into two types. Type I ("closed lips") is established when cerebral mantle has fused clefts with no relation to the ventricular system. Type II ("open lips") is established when there is connection of lateral ventricle with subarachnoid space filled with CSF.[13] A patient with schizencephaly clinically presents with epilepsy, hydrocephalus, hemiparesis, delayed milestones, and psychomotor retardation.[6] Depending on the extent of cerebral cortex involvement, the outcome is variable. In the case presented, the patient had nonprogressive swelling with absence of any other findings such as mental retardation, epilepsy, or hemiparesis.

A recently reported study showed relationship between schizencephaly and mutation of the procollagen alpha-1 (IV) (*COL4A1*) gene.[5] In mice, *COL4A1* mutation leads to ocular dysgenesis, cortical dysplasia, porencephaly, and myopathy;[4,9] approximately 20% of patients with schizencephaly have a *COL4A1* mutation.[5] Patients with the *COL4A1* mutation have shown to have an increased risk of cerebrovascular disease and intracerebral hemorrhage.[2] Studies have shown that meningoencephalocele has many contributing factors; in addition to genetic, environmental factor plays an important role.[9] However, there are two reported cases of familial recurrence of schizencephaly and meningoencephalocele, indicating that genetic factors are important to disease etiologies.[11]

Imaging studies help diagnosis of encephaloceles and schizencephaly. T1-weighted images reveal herniated parenchyma as hypotense and hyperintense on T2-weighted images.[3] Schizencephaly can be differentiated from porencephaly on imaging by the presence of gray matter lined cleft on MRI.[10] The clinical, radiographic, and pathologic findings of the patients confirm meningoencephalocele with schizencephaly. In fact, many of the findings such as patient's age, symptoms, and clinical course are very similar to a case reported in 2014.[11] However, two distinctions make this case notable. First, the patient's history of surgical correction of mass at age 35 days, and second the meningoencephalocele involvement of the frontal location. Management depends on the size and severity of the lesion leading to surgical correction by bone draft, and VP shunt is needed in cases where they are complicated by hydrocephalus.

CONCLUSION

Encephalocele association with schizencephaly is rare. While correcting large frontoethmoidal encephalocele, few important points should be considered such as slow decompression of CSF from the lesion and preservation of major veins.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Footnotes

http://surgicalneurologyint.com/Frontoethmoidal-encephalocele-presenting-in-concert-with-schizencephaly/

REFERENCES

1. Alexander RC, Patkar AA, Lapointe JS, Flynn SW. Schizencephaly associated with psychosis. J Neurol Neurosurg Psychiatry. 1997;63:373–5. [PMC free article] [PubMed]

2. de Vries LS, Mancini GM. Intracerebral hemorrhage and *COL4A1* and *COL4A2* mutations, from fetal life into adulthood. Ann Neurol. 2012;71:439–41. [PubMed]

3. Dobrin N, Mihaela B, Cost B, Tudorache C, Chiriac A, Poeat I. Acquired parietal intradiploic encephalocele. Case report and review of the literature. Romanian Neurosurg. 2011:18.

4. Gould DB, Phalan FC, Breedveld GJ, van Mil SE, Smith RS, Schimenti JC, et al. Mutations in Col4a1 cause perinatal cerebral hemorrhage and porencephaly. Science. 2005;308:1167–71. [PubMed]

5. Harada T, Uegaki T, Arata K, Tsunetou T, Taniguchi F. Schizencephaly and porencephaly due to fetal intracranial hemorrhage: A report of two cases. Yonago Acta Med. 2018;60:241–5. [PMC free article] [PubMed]

6. Hung PC, Wang HS, Chou ML. Schizencephaly in children: A single medical center retrospective study. Pediatr Neonatol. 2018;59:1–8. [PubMed]

7. Junaid M, Sobani ZA, Shamim AA, Kazi M, Khan MJ. Nasal encephaloceles presenting at later ages: Experience of Otorhinolaryngology Department at a tertiary care center in Karachi, Pakistan. J Pak Med Assoc. 2012;62:74–6. [PubMed]

8. Khoshnoodi J, Pedchenko V, Hudson BG. Mammalian collagen IV. Microsc Res Tech. 2008;71:357–70. [PMC free article] [PubMed]

9. Labelle-Dumais C, Dilworth DJ, Harrington EP, de Leau M, Lyons D, Kabaeva Z, et al. *COL4A1* mutations cause ocular dysgenesis, neuronal localization defects, and myopathy in mice and Walker-Warburg syndrome in humans. PLoS Genet. 2011;7:e1002062. [PMC free article] [PubMed]

10. Lotfinia I, Mahdkhah A. Intradiploic meningoencephalocele, case report and review of literature. J Clin Exp Neurosci. 2013;1:10.

11. Mishra SS, Senapati SB, Das S, Deo RC. Large vertex meningoencephalocele with schizencephaly: An interesting case with neurosurgical challenge. J Pediatr Neurosci. 2014;9:136–8. [PMC free article] [PubMed]

12. Pitkin, Roy M. Folate and neural tube defect. Am J Clin Nutr. 2007;85:285S–8S. [PubMed]

13. Stopa J, Kucharska-Miąsik I, Dziurzyńska-Białek E, Kostkiewicz A, Solińska A, Zając-Mnich M, et al. Diagnostic imaging and problems of schizencephaly. Pol J Radiol. 2014;79:444–9. [PMC free article] [PubMed]

14. Yoneda Y, Haginoya K, Kato M, Osaka H, Yokochi K, Arai H, et al. Phenotypic spectrum of COL4A1 mutations: Porencephaly to schizencephaly. Ann Neurol. 2013;73:48–57. [PubMed]