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Schizencephaly: An Incredibly A Rare Disease (Case Report and Review of Literature)

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Abstract

Schizencephaly is an extremely rare cortical malformation that manifest as a grey matter lined cleft extending from ependymal to the piamater .It is also known as split brain disease. It usually presents itself in childhood and in rare cases in adults. The most common mode of presentation is hemiparesis and recurrent seizure.

We present to you a case of 21 year old male who came to our Emergency department with recurrent seizure and the CT scan imaging showed Type 2 schizencephaly. This case highlights the importance of how uncommon diseases may have a common mode of presentation.

Keywords: Open lip schizencephaly, seizure, premature death, CT scan

INTRODUCTION

Schizencephaly as a disease is so rare that even today we don't know its exact incidence and prevalence. In a study carried out in California, USA prevalence of schizencephaly was estimated to be 1.5 per 100,000 live births in a population of 4 million live births from 1985 to 2001 [1]. The incidence is unknown. There are two types of Schizencephaly. Type 1 or closed Schizencephaly and Type 2 or open Schizencephaly. Type 2 is the more common form of Schizencephaly.

Yakovlev and Waldsworth coined the term Schizencephaly in 1946 and described that primitive normoblast fail to migrate in their normal mode thereby resulting into cerebral cleft.

CASE REPORT

A 22 year old male came to the Emergency department with 2 episodes of convulsion. The convulsion lasted for 2 minutes .There were total 3 episodes of convulsion with 5 min interval each. It was a complex partial seizure (CPS) with rolling eyes and froth in the mouth. It was precipitated by fall in the bathroom and relieved by Injection Eptoin 100mg loading and Injection Lopez. The last episode of seizure was took place 3 years back. The seizure was not associated with Fever, headache, dizziness, or photophobia. There is no history of tinnitus or hearing loss. There was no chest pain, palpitation or shortness of breath. There was no history of nausea, vomiting, and abdominal pain, urinary or fecal incontinence. There is no history of diarrhea. The there is no history of skin rash or joint pain. There is no history of travel. There is no history of night sweats, weight loss or fatigue. Sleep and appetite is normal. His mother gives a history of convulsion since the age of 5 which persisted till 12years of age. She describes it as aura of non-specific black out with dystonic posturing of both the hands lasting for 1- 2 minutes.

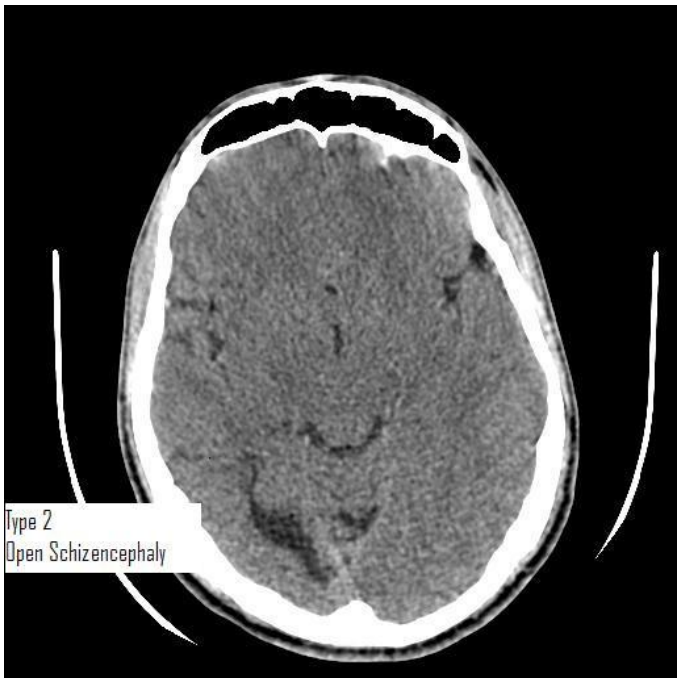
There was a honey moon period form the age 12 to age 19 where the patient did not experience any seizure hence the local doctor stopped his seizure medication. Patient has now started experiencing frequent seizure since last 3 yrs. There are 1 – 2 episodes per month of CPS and had experienced 5 to 6 episodes of GTC in last 3 year. The patient was started on Tab Decorate 250 mg once a day. He is non-compliant with his medication. The birth history was unremarkable. It was full term normal vaginal delivery with no birth related complication. All the milestones' were achieved. History of immunization was up-to-date. There is

no family history of seizure in parents or siblings. There is no history of Allergy. Patient is a non-alcoholic and non-smoker with no history of substance abuse.

On Physical Examination patient was oriented in time place and person. The vitals were stable. There was no pallor, icterus, cyanosis, clubbing, koilonychia or lymphadenopathy or peripheral edema. Fundoscopy revealed no papilledema. No tongue bite. Cranium and spinal examination was normal. Cranial Nerves 1–12 intact. Reflex 2+ on both sides. Motor strength 5/5 and power intact both sides. Babinski sign absent. Sensory examination was normal. Kernig's sign and Brudzinski's sign absent. No nuchal rigidity and memory loss. Gait normal. Romberg's sign negative. The CVS,RS , and per Abdominal examinations were unremarkable.

Complete Blood count, CRP, ESR, Liver Function test, Renal Function test Lipid profile, Random blood sugar, Serum electrolytes, Serum Calcium and Magnesium, Urine analysis were sent and came back normal. EKG, Chest X ray and 2 D echo was done and was unremarkable. CT scan of the brain was done and it showed Type 2 schizencephaly Fig.1.

Fig 1. Type2 Open Schizencephaly



MRI was offered but the patient refused due to financial limitation.

Sodium valproate was started and patient was counselled for proper compliance of the medication.

DISCUSSION:

Schizencephaly is an extremely rare congenital disorder characterized by infolding of cortical gray matter along a hemispheric cleft near the primary cerebral fissure. Yakovlev and Waldsworth referred to the localized failure of evagination and coined the term schizencephaly for it in 1946 [2,3]. They postulated in their theory that focal developmental defect in the wall of cerebral mantle was due to failure of migration of

primitive normoblast in their normal mode thereby resulting in cerebral cleft extending from subarachnoid space to ventricular system. The aetiology of it could be genetics or sporadic. Sporadic being the most common[1]. Risk factors include young mother, monozygotic twin (RR) 2.1, infection, toxins and environmental factors. Levine and Co-workers have attributed it to a destructive, possibly an ischemic insult during early gestation at a time when neuronal migration takes place. Brunelli et al have reported EMX2 gene (10q2.6) heterozygous mutation is associated with severe schizencephaly [4], although this theory has recently been disproved.

Tietjen et al (2007) sequenced the EMX2 gene in a cohort of 84 probands with schizencephaly, and found no pathologic mutations and suggested that EMX2 mutations are an uncommon cause of schizencephaly [5]. The role of CMV infection in the complex multi pathogenesis is also possible [6]. Schizencephaly is of two types. Type 1 or Closed Schizencephaly with cleft wall opposed causing obliteration of CSF space within the cleft. Type 2 or Open Schizencephaly with cleft wall separated and the CSF fills the cleft from Lateral ventricles to subarachnoid space. Type 2 or Open Schizencephaly is most common. Sometimes both the types of Schizencephaly can coexist in the same patient. The cleft can be unilateral or bilateral. The Neuropathological features include cleft line lined with gray matter, polymicroglia, subarachnoid cyst. Septum pellucidum is absent in 80 to 90 % of cases. It can also coexist with septic optic dysplasia.

Clinical presentation; It is seen in different age group but commonly seen in paediatric age group. It may have an adult onset or may be diagnosed late [7] . Clinical features depend upon the extent to which cerebral cortex in involved. Bilateral cleft is associated with the varied presentation from development delays to speech and language disorder.

Most patients have uncontrollable seizure which often mimics cerebral palsy, epilepsy etc.

Seizures are difficult to control in 1/3rd of cases. GTC, CPS, Partial seizure, sensory seizure are its various forms.

Scoliosis is an important part of Schizencephaly due to one side of brain is developed more than other side causing to child to compensate their usage on other side of body resulting in curvature of spine.

Optic nerve hypoplasia may result in blindness. It is characteristics in 70 % cases .

Motor deficit are predominant feature in Type 2 Schizencephaly, whereas Mental retardation is most common in bilateral closed lip variety of Schizencephaly. Premature death is common. Recently some association of schizencephaly is seen with patients of OCD and psychosis [8, 9].

The diagnosis of Schizencephaly can be made in utero by Ultrasound during the ANC check-up as early as 23 weeks or post-partum by cranial Ultrasound [12, 13] US shows defect in the cerebral mantle in the area of sylvian fissure and visualization of communication between enlarged lateral ventricle and subarachnoid space.

CT scan shows only slight out pouching at the ependymal surface and full thickness cleft may be

difficult to identify on CT scan. The degree of confidence is high when gray matter lining of the cleft can be identified.

MRI is the most sensitive test in diagnosing Schizencephaly and is the imaging modality of choice [10.11]. Type 1 is seen as nipple like out pouching at the ependymal surface whereas Type 2 shows heterotopic gray matter lined CSF cleft seen extending from ventricular to cortical surface. Identification of gray matter in the cleft is pathognomic of schizencephaly.

The treatment depends upon the clinical manifestation of schizencephaly. Physiotherapy (motor involvement), speech therapy, antiepileptic drugs or in case of recurrent seizure complicated with hydrocephalous ventriculoperitoneal shunt is done.

CONCLUSION

Recurrent seizure with unexplained aetiology must be investigated in detail and congenital development anomalies must be a part of differential diagnosis after ruling out common causes especially in young adults.

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REFERENCES

[1] Cynthia J. Curry1,*, Edward J. Lammer2, Verne Nelson3, Gary M. Shaw3 Schizencephaly: Heterogeneous etiologies in a population of 4 million California births 29 JUL 2005 DOI: 10.1002/ajmg.a.30862

[2] Yakovlev PI, Wadsworth RC. Schiz-encephalies: A study of the congenital clefts in the cerebral mantle. I. Clefts with fused lips. J Neuropathol Exp Neuro 1946; 5: 116-130.[3] Yakovlev PI, Wadsworth RC. Schiz-encephalies: A study of the congenital clefts in the cerebral mantle. II Clefts with hydro-cephalus and lips separated. J Neuropathol Exp Neurol 1946: 5: 169-206.

[4] Brunelli S, Faiella A, Capra V, Nigro V, Simeone A, Cama A. Germline mutations in the homebox gene EMX2 in patients with severe schizencephaly. Nat Genet 1996; 12: 94-96.[5]Tietjin I, Bodell A Ann J Med , Genetics A , 2007

[6]Ianneti P, NiagroG, Spatice A Faella A,Boncenelli E, Ann Neuro 1998 Jan;43(1) :123-7

[7] Sitniko AR clinical cas of the late diagnosis of Type 2 schizencephaly Rural and Remote Health 7:661.(online),2007[8] Matias Carvalho Aguiar Melo, Saulo Giovanni Castor Albuquerque, Jose Henrique Sousa Luz and Alexandre Basto Lima Case Reports in Medicine Volume 2013 , Article ID 210868

[9] Erica E.Coates , MA; Jason I .Chen BA Eric A . Storch, Phd, MS A case of Schizencephaly associated with obsessive compulsive disorder. Annals of Clinical Psychiatry 2013;25 (2) : 151 - 152

[10] Chamberlain MC, Press GA, Bejar RF. Neonatal schizencephaly; Comparison of brain imaging. Pediatr Neurol 1990; 6: 382-387.