CASE REPORT

UNILATERAL OPEN LIP SCHIZENCEPHALY ASSOCIATED WITH PARTIAL AGENESIS OF CORPUS CALLOSUM.

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ABSTRACT: Schizencephaly is a rare developmental disorder of congenital cerebral malformation. We bring to your attention the case study of a 5 year old female child reported with focal seizures and right hemiparesis since 5 months of her age. Her parents gave history of birth asphyxia and global developmental delay. On examination decreased muscle bulk, increased muscle tone, decreased muscle power, exaggerated tendon reflexes on right upper and lower limbs, and up going plantars on right lower limb with hemiplegic gait was observed. Magnetic resonance imaging (MRI) scan revealed open lip schizencephaly of left parietal cortex and partial agenesis of corpus callosum.

KEY WORDS: Schizencephaly, seizures, hemiparesis, corpus callosum.

MeSH TERMS: Schizencephaly, Agenesis of corpus callosum, cerebral cortex, Female, Seizure, Paresis, Reflex, MRI scans, cortical dysplasia.

INTRODUCTION: Schizencephaly is an uncommon congenital cerebral malformation that involves cerebral mantle and consists of a cleft that extends through the entire cerebral hemisphere from the lateral ventricle to the cerebral cortex. Schizencephaly has a prevalence of 1.48/100000 births (1). It is a disorder with heterogeneous origin many of which are vascular disruptive in origin (2). They may present with intractable seizures and variable developmental delay (3). A large number of patients have associated brain abnormalities which may account for the severity of some cases. These include agenesis of the septum pellucidum, focal cortical dysplasia, and dysgenesis of the corpus callosum (4).

The corpus callosum is a wide, flat bundle of neural fibers beneath the cortex which connects the left and right cerebral hemispheres and facilitates interhemispheric communication. It is the largest white matter structure in the brain, consisting of 200–250 million contra lateral axonal projections. Agenesis of the corpus callosum is a rare congenital disorder. It occurs when the corpus callosum fails to develop normally, typically during pregnancy. This can be complete (absence of all components) or partial (presence of a short remnant) (5). Partial agenesis of the corpus callosum results from an arrest of growth which occurs between 12 and 18 weeks of gestation (6).

CASE REPORT: A 5 year old female child was brought to our hospital by her father. The father reported that his daughter has been experiencing weakness in her right hand and leg since 5

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months of her age. For the past 7 days, she has seizures in her right hand, each lasting for about five minutes and around 3 to 5 episodes per day. There is no history of loss of her consciousness during seizure episodes. She had been taken to local hospital and treated as a case of epilepsy. Child was referred to our hospital for further investigation. After prescribing antiepileptics to the patient seizures subsided. In the past similar episodes of seizures were noticed by her parents. The birth history consisted of delivery at full term gestation by vaginal route at a tertiary care hospital. The child did not cry soon after birth and was admitted in NICU as a case of birth asphyxia. There is no history of consanguineous marriage. On physical examination, the child had no dysmorphism of the face, the anterior fontanelle and sutures were normal. There was a global delay in developmental milestones. Anthropometric measurements and vitals were within normal limits. Examination revealed decreased muscle bulk, increased tone, decreased power, exaggerated tendon reflexes in right upper and lower limbs, and up going plantars in right lower limb with a hemiplegic gait. Routine hematological and urine examination were unremarkable. MRI scan revealed morphology in favor of open lip schizencephaly of left parietal cortex (Figure 1) and partial agenesis of corpus callosum (Figure 2).

DISCUSSION: Schizencephaly appears as a cleft filled with cerebrospinal fluid and extending medially from the subarachnoid space into the lateral ventricle. The wall of the cleft is lined with dysmorphic gray matter. The gray matter sometimes extends into the lateral ventricle in the form of subependymal heterotopias. The cleft may be small or large, unilateral or bilateral. The anomaly may be of the open-lip or closed-lip type. Closed-lip schizencephaly is characterized by gray matter-lined lips, which are in contact with each other (type 1). Open-lip schizencephaly has separated lips and a cleft of CSF extending to the underlying ventricles (type 2) (7). Toxic exposure, viral infections, genetic factors, vascular, familial, or metabolic involvement have been suggested to explain the complex cause of schizencephaly (8). Loss-of-function mutations in *EMX2* have been described in individuals with the radiological diagnosis of schizencephaly (9). Clinical manifestations may be variable i.e. asymptomatic to those with epilepsy and neurodevelopment problems. MRI is the imaging modality of choice because of its superior differentiation of gray matter. Identification of gray matter lining the cleft is the pathognomonic finding (10). Treatment consists of commencing antiepileptic therapy and physiotherapy. Surgical treatment has been reported in cases of medically intractable epilepsy, but it is rarely considered (11).

Agenesis of the corpus callosum is one of the most frequent malformations in brain with a reported incidence ranging between 0.5 and 70 in 10,000 (12). Complete and partial agenesis of corpus callosum can result from genetic, infectious, vascular, or toxic causes (13). Clinical symptoms of corpus callosum agenesis are quite different, the most common being macrocephaly, facial dysmorphism, in combination with different levels of mental retardation, epileptic seizures, vision and other nerve impairments (14).

We intend to report this rare case because of the complex clinical presentation with a combination of congenital malformations (schizencephaly, and dysgenesis of corpus callosum) in the same child.

CONCLUSION: Schizencephaly is a rare congenital disorder of cerebral cortical development which mainly manifests as two types i.e. open type and closed type. Most of the cases present with seizures and developmental delay. It may be associated with malformations like cortical

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dysplasia and dysgenesis of corpus callosum. These malformations can be clearly visualized on the MRI scans.

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Figure 1: MRI scan showing open lip Schizencephaly of left parietal cortex.

Figure 2: MRI scan showing partial agenesis of corpus callosum.