A RARE CASE OF CONGENITAL INTRA-CRANIAL MALFORMATION (OPEN LIP SCHIZENZEPHALY)

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12 years old Hindu male child was brought to the hospital by the parents with the complaints of focal convulsions of right side of the body with right sided facial deviation during convulsions since 2 years. It was occurring once in every 1-2 months. This was lasting initially for about 5-10 minutes then gradually the time duration increased, since last 4-5 episodes patient was having convulsion lasting for more than 3-4 hours followed by unconsciousness lasting for 5-6 hours. The Child was treated by local doctors in the form of tab. Sodium valproate and Tab. Gardenal but with this regimen, these convulsions were not controlled.

The patient is the second born child to nonconsanguinously married parents. Other sibs are 3 brothers and 2 sisters & both are keeping good health and no similar history in any other family members. The patient was born in home and was breast fed for 2-2½ years. It was interesting to note that he was only child who was left handed in whole of the family.

On examination, the patient had right sided weakness hypertonia of all limbs more so on right side and ankle clonus was present on the right side. He had bilateral brisk deep tendon reflexes & extensor Planters on both sides. The patient was well oriented to the time, place and person & was mentally normal.

On Investigations on MRI imaging the patient had open lip schizencephaly on left side of the cerebrum with absent septum pallidum.

SCHIZENZEPHALY

Schizencephaly is a rare developmental disorder of brain characterized by abnormal continuity of histologic grey matter tissue extending from the ependyma lining of the cerebral ventricles to the pial surface of the cerebral hemisphere surface. Type I Schizencephaly has a cord of grey matter tissue, either with no fluid cleft or with ventricular or cortical lips closing one end of an abnormal fluid cleft through the hemisphere. Type II Schizencephaly shows a cerebrospinal fluid-filled cleft of varying size and shape extending through the hemisphere from the ependyma centrally to the pia peripherally.

It can be distinguished from porencephaly by the fact that in schizencephaly the fluid-filled component, if present, is entirely lined by heterotopic grey matter while a porencephalic cyst is lined mostly by white matter.

Schizencephaly is probably a disorder in normal neuron migration during the second trimester of intra-uterine development, when primitive neuron pre-ursors (germinal matrix) migrate from just beneath the ventricular ependyma to the peripheral hemispheres where they form the cortical grey matter.

Grey matter contains neuronal cell bodies and dendrites whereas white matter contains axons, which are coated in myelin (i.e., a fatty tissue that aids in the speed of action potentials down the axon). Individuals with clefts in both hemispheres, or bilateral clefts, are often developmentally delayed and have delayed speech and language skills and corticospinal dysfunction. Individuals with smaller, unilateral clefts (clefts in one hemisphere) may be weak on one side of the body and may have average or near-average intelligence.

Patients with schizencephaly may also have varying degrees of microcephaly, mental retardation, hemiparesis (weakness or paralysis affecting one side of the body), or quadriplegia (weakness or paralysis affecting all four extremities), and may have reduced muscle tone (hypotonia). Most patients have seizures and some may have hydrocephalus.

ETIOLOGY

In schizencephaly, the neurons border the edge of the cleft, implying a very early disruption of the usual grey matter migration during embryogenesis. The cause of the disruption is not known, but likely the cause may be either genetic or a physical insult, such as infection, infarction, hemorrhage, toxin or mutation.
So far there have been only few documented case histories. The majority seem only sporadic, but one case with two brothers has been described. Another instance occurs in identical twins, one was diagnosed at seven months while the other is not affected. The inheritance is autosomal dominant or recessive inheritance, the locus is 10q26.1.

Causes of Schizencephaly may be caused by environmental exposures during pregnancy such as medication taken by the mother, sickness during pregnancy (such as Cytomegalovirus), exposure to toxins, or a vascular insult. Often there are additional associated heterotopias (isolated islands of neurons) which indicate a failure of migration of the neurons to their final position in the brain.

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**PATHOLOGY**

The **septum pellucidum** (also called the **septum lucidum**) is a thin, triangular, vertical membrane separating the anterior horns of the left and right lateral ventricles of the brain. It runs as a sheet from the corpus callosum down to the fornix.

The septum pellucidum is located in the midline of the brain, between the two cerebral hemispheres. It is attached inferior to the corpus callosum, the large collection of nerve fibers that connect the two cerebral hemispheres. It is attached to the anterior part of the fornix, and on either side of the structure are the two lateral ventricles.

Absence of the septum pellucidum or corpus callosum, caused by mutations in the **HESX1** gene, is associated with septo-optic dysplasia. This may result in hypothalamic dysfunction and hypopituitarism, as well as problems of vision, coordination, and intelligence, as well as other unusual symptoms.

**Septo-optic dysplasia** (SOD), also known as **de Morsier syndrome** is a congenital malformation syndrome made manifest by hypoplasia (underdevelopment) of the optic nerve and absence of the septum pellucidum (a midline part of the brain).

Neuroradiologically, intracranial malformations associated with septo-optic dysplasia include agenesis of the septum pellucidum, schizencephaly, and lobar holoprosencephaly.

The prognosis for individuals with schizencephaly varies depending on the size of the clefts and the degree of neurological deficit.