Case Report

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A case of schizencephaly

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Abstract

Schizencephaly is a rare congenital disorder of cerebral cortical development. It is a neuronal migration anomaly, caused by insults to migrating neuroblasts during 3rd to 5th gestational months. We encountered schizencephaly in the cranial magnetic resonance imaging (MRI) of a 4 month-old male baby. MRI demonstrated wide clefts occupying parietal regions bilaterally and the right occipital region partly. These areas were connected with lateral ventricles and also filled with cerebrospinal fluid. Although prevalence of this disorder is quite low and its incidence is unknown and also there may be no clear symptoms as in our case, we emphasize that it should not be overlooked in differential diagnosis.

Key words: cranial; MRI; neurodevelopment anomaly; schizencephaly

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Introduction

Schizencephaly is a rare congenital disorder of cerebral cortical development. It is a neuronal migration anomaly, caused by insults to migrating neuroblasts during 3rd to 5th gestational months.[1] Other anomalies in this category include agyria, pachygyria, polimicrogyria, unilateral megalencephaly, gray matter heterotopias, lissencephaly and agenesis of corpus callosum.[2,3] Basically, schizencephaly is characterized by a cleft traversing cerebral hemisphere, infolding of cortical gray matter along the cleft, an abnormal ventricular system and other associated cerebral anomalies.[4] Congenital cleft of the cerebral mantle extends from the pial surface to the lateral ventricle. Schizencephalic cleft occurs more often in the anterior than in posterior neocortex. It is commonly observed in the posterior frontal and parietal regions of the anterior neocortex.[5] Clefts most commonly involve the parasylvian region and have been divided in two subtypes. Closed-lip schizencephaly (Type 1), with narrow clefts and lips fused in certain areas (pial-ependymal seam); and open-lip schizencephaly (Type 2) with separated walls encompassing an excessive cerebrospinal fluid space.[6] There are four categories defined radiologically i.e. unilateral open lip (36.2%), unilateral closed lip (25.6%), bilateral open lip (25.6%) and bilateral closed lip (12.8%). Closed lip schizencephaly usually presents with hemiparesis or motor delay, whereas patients with open lip schizencephaly is present with hydrocephalus.[7]

Case Report

We encountered schizencephaly in the cranial MRI of a 4 month-old male baby. There was no definite clinical history except weakness of suckling. Both of the parietal lobes were not seen in the MRI (**Figures 1** and **2**). MRI also demonstrated areas which occupied parietal regions bilaterally and right occipital region partly (**Figure 3**). These areas were connected with lateral ventricles and also filled with cerebrospinal fluid (CSF). This appearance was consistent with schizencephaly type II (open lips).

MRI imaging was performed under general anesthesia. However, considering the age and condition of the patient, unwanted movements of the patient caused artifacts in MR images.

This study has been presented as a poster at the 10th Congress of European Association of Clinical Anatomy (EACA) in Istanbul, Turkey in 2009.

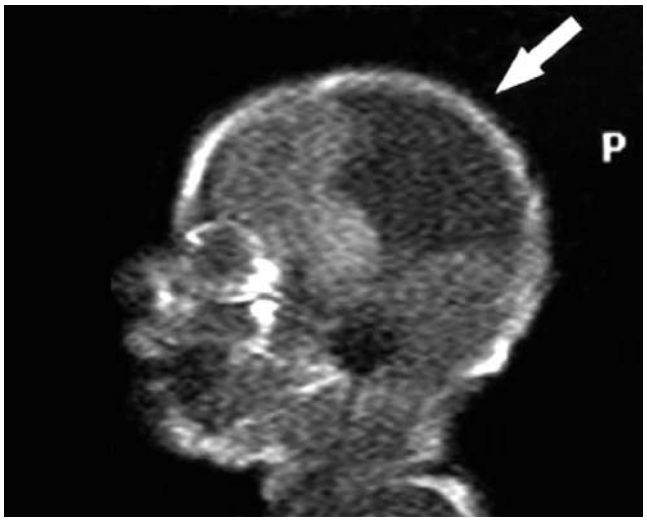

Figure 1. A wide CSF cleft is seen occupying the parietal lobe (**arrow**) in the T1 weighted sagittal MRI image.

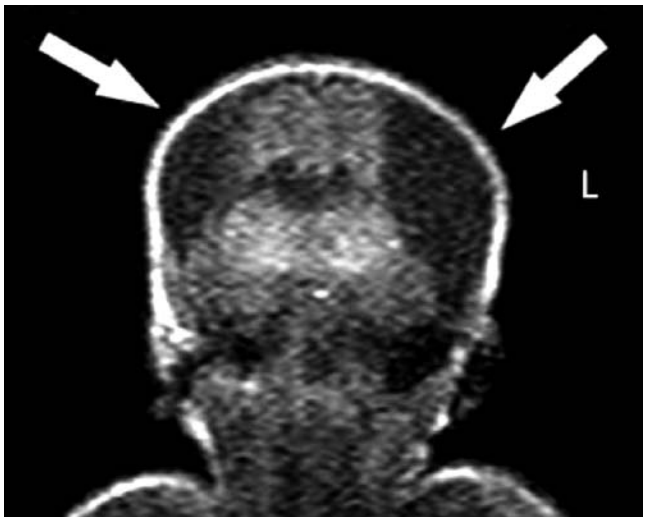

Figure 2. Bilateral parietal clefts (**arrows**) are demonstrated in the T1 weighted coronal MRI image.

Discussion

Schizencephaly may occur due to various causes. It may be due to localized ischemia in periventricular germinal matrix, or it may be an extreme variant of cortical dysplasia, in which the infolding of cortex extends all the way into the lateral ventricle. A report of schizencephally in two affected sibs suggested genetic basis and indicated the possibility of autosomal recessive form of transmission. Schizencephaly has recently been associated with a germline mutation in homeobox gene EMX2 in 70% of patients. CMV infection, postnatally, has also been blamed in its complex multifactorial pathogenesis. Intoxicants, radiation and intrauterine exposure to warfarin have also been associated with this anomaly.[1]

The schizencephalic cleft in our case was extending to lateral ventricle. This is consistent with the definition of schizencephaly, however, there are number of case reports of schizencephaly involving the third ventricle.

Age at detection ranges from 8 months to 30 years. Clinical manifestations vary from mild to severe and include developmental delay and retardation, microcephaly, language dysfunction, focal or generalized motor abnormalities and seizures.[4] However, in our case the only distinct clinical finding was developmental delay which was manifested as weakness of suckling.

Cerebrospinal fluid pulsation from the lateral ventricles with resultant pressure effects on the inner table of the skull vault is a well-described feature of open lip anomaly.[8] This may emphasize the importance of physical examination in infants with open lip schizencephaly as in our case.

Neurodevelopmental outcome is generally poor with 51% showing severe deficits and 32% moderate impairment. However, 17% have mild or no problem. In our case, data was not available about the neurodevelopmental outcome of the patient. Open lip schizencephaly is more likely to be associated with worse outcome than closed lip schizencephaly. Thus presentation and outcome vary but are related to extent of cortical involvement. Mental retardation is common with bilateral clefts.[9] Although prevalence of this disorder is quite low and its incidence is unknown and also there may be no clear symptoms, as in our case, we emphasize that it should not be overlooked in differential diagnosis.

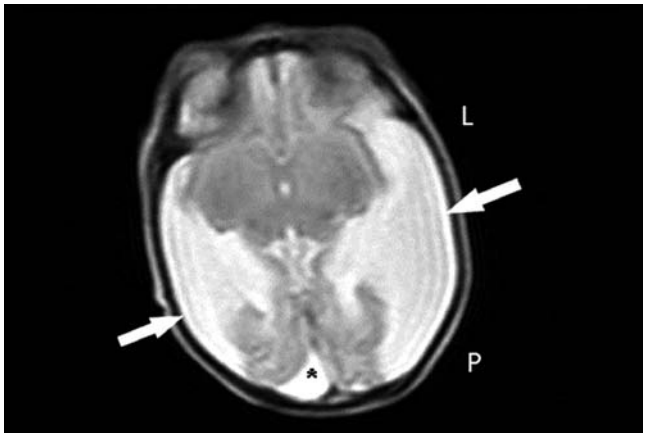

Figure 3. T2 weighted transverse MRI demonstrating wide clefts occupying parietal regions bilaterally (**arrows**) and right occipital region partly (**asterisk**). These areas are connected with lateral ventricles and also filled with CSF.

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Conflict of interest statement: No conflicts declared.