Case Report

Unilateral Open-lip Schizencephaly with Tonsillar Herniation in a Preterm Infant

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Schizencephaly is a rare type of neuronal migration disorder characterized by the presence of a cerebral hemispheric cleft that extends from lateral ventricles to the cortical surface of the brain. We report a rare case of prenatally diagnosed unilateral schizencephaly in a late preterm infant who manifested with rapidly progressive hydrocephalus with massive enlargement of posterior cerebrospinal fluid spaces with tonsillar herniation that was successfully treated with placement of a ventriculoperitoneal shunt.

Keywords: Hydrocephalus, preterm infant, schizencephaly, ventriculoperitoneal shunt

Introduction

Schizencephaly is a rare type of congenital brain malformation characterized by a cleft in the cerebral hemisphere, extending from the cortical surface of the lateral ventricles.[1] Broadly, two forms of schizencephaly have been described: type 1 or closed-lip schizencephaly, characterized by the absence of open communication with the cerebrospinal fluid (CSF) space, and type 2 or open-lip schizencephaly, in which the cleft communicates with the ventricular system and subarachnoid CSF space.[2,3] Schizencephaly is a neuronal migration disorder with the presence of heterotopic gray matter along the cleft.[4] Usually, the presence of schizencephaly is suspected based on the postnatal abnormal neurological findings and psychomotor developmental impairments.[1] Very rarely, prenatal diagnosis based on the findings of prenatal ultrasound findings and early postnatal recognition based on the presence of microcephaly or macrocephaly have been described.[4,5] In this report, we describe a rare type of unilateral open-lip schizencephaly in a late-preterm infant, manifesting with rapidly progressive hydrocephalus with massively enlarged posterior fossa CSF space and resultant tonsillar herniation that was successfully treated with the placement of ventriculoperitoneal (VP) shunt.

Case Report

A Caucasian male infant was born by cesarean delivery to a 33-year-old mother at 34 weeks of gestational age. The pregnancy was complicated by the presence of insulin-dependent gestational diabetes and the prenatal diagnosis of bilateral ventriculomegaly. Further prenatal evaluation with fetal brain magnetic resonance imaging (MRI) study revealed the presence of right open-lip schizencephaly with bilateral ventriculomegaly. Furthermore, her last pregnancy was complicated by shoulder dystocia, and given this history, along with the macrocephaly noted in this pregnancy with concerns for cephalopelvic disproportion, a decision was made to proceed with cesarean delivery. The cesarean delivery was uncomplicated, and the infant needed routine care with no resuscitation. On postnatal physical examination, the infant was noted to have macrocephaly with a head circumference measuring 36 cm, which was at >99th percentile for the gestational age. Anterior fontanel was very wide open. In addition, significant sutural diastases were also noted on physical examination. Even though the infant appeared non-encephalopathic, the presence of generalized axial and appendicular hypotonia was noted. During the first postnatal week, rapid head
growth with tense and bulging anterior fontanel along with worsening sutural diastases was noted. Distended scalp veins were visible. Postnatal MRI performed on postnatal day four revealed the presence of open-lip schizencephaly located in the right occipitoparietal region of the cerebral hemisphere, communicating with the right lateral ventricle and extra-axial CSF space. Posterior fossa CSF space, which was in continuity with extra-axial CSF space [Figure 1], was markedly dilated causing a mass effect on the cerebellum and tonsillar herniation by approximately 1 cm [Figure 2]. Both lateral ventricles were asymmetrically dilated, with right lateral ventricular dilatation more marked than the left. Also, a pronounced right lateral ventricular dilatation caused a leftward midline shift. Syndromic evaluation, evaluation for intrauterine infection, and evaluation for prenatal teratogenic exposure evaluation through detailed history yielded negative results. The pediatric neurosurgical evaluation followed by right VP shunt placement was performed. The infant had an uneventful recovery following the surgery. Parental consent was obtained for reporting this case.

**Discussion**

In this case study, we have reported a rare case of unilateral open-lip schizencephaly in a preterm infant, which was detected during the prenatal period due to the presence of bilateral ventriculomegaly and hydrocephalus. In the index case, both communicative and obstructive components of CSF flow contributed to the occurrence of hydrocephalus. Moreover, this case was also unique for showing a rapid progression of hydrocephalus with both early leftward central herniation and early tonsillar herniation. Early intervention with VP shunt prevented the further progression of hydrocephalus and herniation.

The worldwide prevalence of schizencephaly is not very well known, but the estimated prevalence in the United States is 1/100,000 births. Although the etiologies of schizencephaly are largely unknown, there are few potential causes including prenatal teratogenic exposures, prenatal viral infection, intrauterine fetal stroke, and genetic mutations. Other known risk factors of schizencephaly include young maternal age and the illicit use of alcohol and narcotic substances.

The diagnosis of schizencephaly is often delayed. The most common presentations that lead to the diagnosis, include psychomotor impairment and neurocognitive dysfunction. However, in our patient, the presence of ventriculomegaly with hydrocephalus led to an early prenatal diagnosis. Other early postnatal manifestations, including epileptic seizures, were not seen in our patient. Surgical intervention with VP shunt is reserved for those with ventriculomegaly with...
hydrocephalus.[8] We feel that VP shunt placement was performed at a very crucial point, which prevented the further progression of hydrocephalus and herniation. In addition, as our index case was also at risk of developing a varying degree of learning disabilities, speech impairment, and motor dysfunction, including cerebral palsy, we initiated early intervention with physical and occupational therapy. Early interventions initiated during the early postnatal period are very vital for improving long-term neurological outcomes.[8,9]

Our index case highlights the fact that having a prenatal diagnosis of schizencephaly is very useful in the optimal postnatal management of neonates with schizencephaly with hydrocephalus. In addition, very close monitoring of head circumference and a meticulous physical examination are vital for early detection of rapidly progressive hydrocephalus, which can be effectively managed to prevent intracranial complications and to improve neurological outcomes.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

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