Prenatal diagnosis of schizencephaly - case report

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Objective

Schizencephaly is a rare with an estimated incidence of ~1.5:100,000 live births malformation of cortical development characterized by congenital clefts extending from the pial surface to the lateral ventricle that are lined by heterotopic gray matter. The clinical presentation is variable and can include motor or cognitive impairment and epilepsy. The causes of schizencephaly are heterogeneous and can include teratogens, prenatal infection, or maternal trauma. Unilateral schizencephaly is more frequent (60%) than bilateral clefts; the most frequent anatomic localization is in the frontal and parietal lobes (in up to 70%).

Results

24-year old patient was admitted to our Unit due in 28 wks of gestation to pyelonephritis. During a routine ultrasonographic scan a bilateral schizencephaly in place of temporal lobes was present with no other anomalies. Patient was treated with antibiotics due to infection and discharged from our unit after a week. Patient delivered in 37wks of gestation a baby girl 2960 grams. Baby was later consulted in Pediatric Neurologic Unit were MRI confirmed diagnosis. Additionally agenesis of corpus callosum was diagnosed. During first months of life patient suffered from epilepsy, but after drug administration the number attacks decreased.

Discussion

Schizencephaly is usually associated with other malformations of the central nervous system, such as, ventriculomegaly, polymicrogyria, pachygyria, heterotopias, and lissencephaly. It may also be associated with the absence of the cavum septum pellucidum, agenesis of the corpus callosum, and optic nerve hypoplasia. The clinical manifestations of schizencephaly include different levels of retardation of neuropsychomotor development and seizures, and is usually associated with the extent of the lesions. Therefore, an accurate prenatal diagnosis is important to enable counseling of parents. The differential diagnosis is done to distinguish this disorder from holoprosencephaly, hydranencephaly, and bilateral arachnoid cysts. The difficulty is to diagnose this condition at the beginning of second trimester of pregnancy. MRI and computed tomography have been used postnatally to prove the suspected obstetric ultrasonographic diagnosis.