Open-lip Schizencephaly: A case report

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Objective:

Schizencephaly is a rare congenital disorder of the brain, characterized by a cleft in the gray matter, filled by cerebrospinal fluid (CSF), connecting the lateral ventricles and the external subarachnoid space and it can be unilateral ou bilateral. It can be classified in Type I, with a trans-mantle column of abnormal gray matter on the cleft; Type II (closed-lip), with CSF on the cleft, with lining lips adjacent of abdnormal grey matter opposed; Type III (open-lip), with clefts containing CSF in the cerebral mantle, without lining lips adjacent to the abnormal grey matter and with associated ventriculomegaly. The exact etiopathogy is not clearly understood, but it is believed that exposure to teratogenic agents, viral infections, stroke in utero, gene mutations and young maternal age are associated with the abnormal neuronal migration in the first weeks of pregnancy.

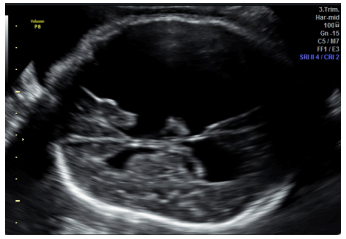

Figure 1

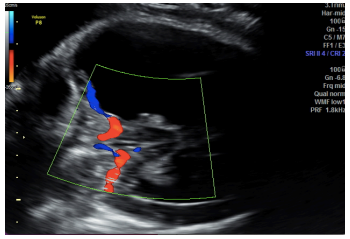

Figure 2

Methods:

A 20-year-old primiparous, pregnant of 22 weeks, was referred to our center due to fetal brain anomaly found in the second trimester scan. The fetus was diagnosed with right open-lip schizencephaly (Fig. 1 and 2) and the mother was referred to high-risk prenatal care. A multidisciplinary follow-up was carried out with obstetricians, psychologists and neonatal care specialists. Fetal MRI was performed to exclude Septo-Optic Dysplasia, however the optic nerve could not be properly evaluated. Obstetric ultrasound was performed every four weeks to monitor the cephalic perimeter.

Results:

High risk prenatal care was taken up to term. The patient presented premature rupture of membranes with 37^{+5} weeks and a C-section was performed due to macrocephaly (HC > 95^{th} centile). The baby was born with APGAR score 9/10, weighing 3.460g and cephalic perimeter of 38,5 cm (> 99^{th} centile). Breastfeeding was possible without major difficulties and a slight motor reflex deficiency on the right side was identified by a pediatric neurologist. Postnatal MRI was performed showing thinning of chiasm and optic nerves and may correspond to septo-optic dysplasia. (Fig.3)

Conclusion:

Genetic counseling should be made, as chances of recurrence are low and it is not associated with chromosomal abnormalities or genetic syndromes. Prognosis is poor and multidisciplinary care should be offered. Fetal MRI should always be performed, if possible, to confirm the diagnosis as it better evaluates septo-optic nerve and differentiates schizencephaly of porencephaly.

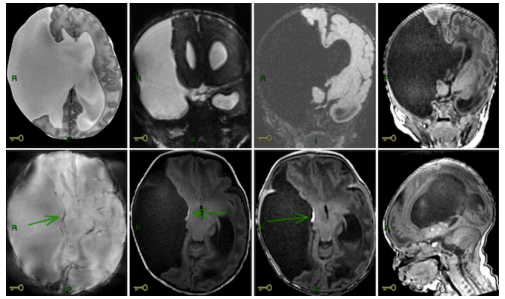

Figure 3

References:

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