A case of occipital encephalocele and ventriculomegaly evaluated by 2D US and MRI

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Objective
To report the case of a fetus with occipital encephalocele associated with ventriculomegaly assessed by two-dimensional ultrasound (US) and magnetic resonance imaging (MRI).

Methods
Case report.

Results
We report the case of a 19 year-old primigravida. No family history of genetic conditions or congenital malformations was reported. She denied drug abuse and no folic acid supplementation had been undertaken. She had no first trimester screening. At 25 weeks of pregnancy the US evaluation performed at our institution also showed a bone defect in the right occipital region with protrusion of brain tissue and cerebrospinal fluid measuring 5.1 cm, all surrounded by membrane. A diagnosis of occipital encephalocele was then made. Bilateral ventriculomegaly (right 2.4 cm, left 1.2 cm) was also found. Fetal MRI at 29 weeks of pregnancy confirmed US diagnosis of occipital encephalocele by describing two cystic images measuring approximately 2.6 cm x 1.5 cm, adjacent to the hole the first one and another larger cystic cavity to the right of midline measuring 4.8 cm x 2.3 cm. The child was born by cesarean section at 38 weeks gestation, weighing 3895g and Apgar scores of 7 and 9. She was not dysmorphic and underwent surgery for correction of occipital encephalocele on the second day of life. At the moment, the neonate is waiting for a ventriculo-peritoneal shunt placement.

Conclusion
Encephalocele is one of the main types of neural tube defects and is characterized by the protrusion of the brain and/or meninges through a skull defect, closed or covered by skin. Occipital encephalocele is usually obvious at birth but the big majority are diagnosed during the prenatal period through US, as observed in our case. The prognosis usually depends on the extension of the herniated cerebral tissue and the presence of associated anomalies. Therefore, this report highlights the importance of a more detailed assessment of fetal anatomy using additional tests, such as MRI, when this diagnosis is made.