Encephalocele: 1st trimester sonographic spectrum of findings
Sepulveda W, Andreeva E, Odegova N, Martinez-Ten P, Meagher S
Clinica Las Condes, Santiago, Chile

Objective
To describe the sonographic features of fetal cephalocele diagnosed at the time of the first-trimester ultrasound screening for aneuploidy.

Methods
Retrospective review of cases diagnosed at four fetal medicine referral centers.

Results
A total of 34 cases were identified; 32 were from singleton pregnancies and two from a twin pregnancy in which the other fetus was unaffected. 12 cases (35%) were classified as cranial meningocele and 22 (65%) as encephalocele. The bone defect was occipital in 26 (76%), frontal in 3 (9%), parietal in 3 (9%), and non-classifiable in 2 (6%). Regarding the size, 12 (35%) were subjectively considered small, 11 (32%) medium, and 11 (32%) large. There were no reported cases of aneuploidy; however, 3 (9%) cases were associated with Meckel-Gruber syndrome and 2 (6%) with disruptive syndromes. All cases of large encephalocele were associated with a smaller than expected biparietal diameter. 8 (24%) pregnancies were lost to follow up and in 20 of the 26 remaining cases the parents opted for termination of the pregnancy. Of the 6 ongoing pregnancies, three fetuses miscarried or died in utero in the second trimester, one was a monoamniotic twin pregnancy in which both fetuses died at 26 weeks, one died shortly after delivery, and one underwent neonatal surgery for an isolated cranial meningocele and currently is doing well.

Conclusion
First-trimester sonographic diagnosis of cephalocele is easily accomplished with a detailed examination of the skull contour. The diagnosis is facilitated by the routine assessment of the axial view of the head for biparietal diameter measurement and examination of the occipital area for nuchal translucency measurement. However, the sonographic features are very variable. A small proportion of cases are associated with disruptive syndromes or genetic syndromes. A major limitation of this study is the lack of information regarding antenatal course and associated findings due to the high rate of termination of pregnancy and early intrauterine demise. The prognosis therefore remains dismal.