A case of encephalocele

Inan C, Sayin NC, Gurkan H, Erzincan SG, Uzun I, Sutcu H, Atli EI, Varol FG
Trakya University, Faculty of Medicine, Departments of Obstetrics & Gynecology and Medical Genetics, Edirne, Turkey, Edirne, Turkey

Objective

Posterior encephalocele may coexist with cortical dysplasia, agenesis of the corpus callosum, hydrocephalus, microcephaly and craniofacial abnormalities. Our aim to report a case of encephalocele.

Methods

We report a case of a huge posterior encephalocele in a fetus coexistent with unexpected major abnormalities including transposition of the great arteries, cerebellar atrophy and severe ventriculomegaly.

Results

As 18-year-old, gravida 1, woman was referred to our clinic at 20 weeks and 4 days of gestation due to fetal malformation. Obstetric ultrasonography (USG) revealed a posterior encephalocele (28x24mm), cerebellar atrophy, together with transposition of the great arteries and muscular type ventricular septal defect (Figure 1a, b). The atrial width of the lateral ventricle was measured 15mm. The options of amniocentesis and termination of the pregnancy were discussed with the family, but they kindly declined any invasive procedure. At 33 weeks of gestation a follow-up USG was performed. Posterior encephaloce sac was increased to 85x77mm, and atrial width of the lateral ventricle was increased to 21,5mm (Figure 2a, b). She was reevaluated at 36 weeks of gestation. A cesarean section was performed due to fetal malpresentation and preterm premature rupture of the membranes. A 3265g, 50 cm male infant was delivered (Figure 3a, b, c). Respective Apgar scores were 9 and 10 at the 1st and 5th minutes, respectively. Blood sample from the umbilical cord was taken for genetic analysis. Huge posterior encephalocele sac was observed in the newborn which was operated for posterior encephalocele and transposition of the great arteries respectively with two weeks interval. The fetal karyotype, fluorescence in situ hybridization (FISH) and array-comparative genomic hybridization (array-ACG) analyzes were normal. Altghough karyotype analysis with the addition of array-CGH did not bring out any chromosomal defect, proceeding with whole exome sequencing could not be performed because of its high cost.

Conclusion

Encephalocele is a neural tube defect characterized by protrusion of the cerebral or cerebellar structures through the skull within a sac. It is usually seen in the occipital region and the only treatment modality is surgery. The success of the treatment depends on its size and location. Although several malformations associate with encephalocele, to the best of our knowledge this is the first case of huge posterior encephalocele coexisting with transposition of the great arteries.

Figure 2a. Ultrasonographic imaging of a huge posterior encephalocele at 33 weeks of gestation. b. Severe ventriculomegaly imaging in the ultrasonography examination.
Figure 3a,b. Macroscopic views of the posterior encephalocele in the newborn. c. Magnetic resonance imaging of the posterior encephalocele in the newborn. Asterix: The posterior encephalocele sac.