Subependymal cysts in the fetal brain: prenatal diagnosis of 9 cases

De Keersmaecker B, Jansen K, Naulaers G, De Catte L
University of Leuven, Leuven, Belgium

Objective

Subependymal cysts are usuallly located in the wall of the caudate nucleus or in the caudothalamic groove. They are found in up 1 to 5% of all neonates. When isolated, they regress spontaneously and their prognosis is good. In association with vascular disorders (infarction, hemorrhage), infection (Cytomegalovirus, toxoplasmosis, Rubella) or chromosomal and metabolic problems their neurological outcome is poor. The objective of this study was to determinethe sonographic criteria for the diagnosis of ependymal cysts, the role of MRI and the clinical implications and fetal outcome in a case series of 9 fetuses.

Methods

Retrospective review of all our cases of antenatally diagnosed subependymal cysts from 2007 to 2015. All cases were offered prenatal MRI. Our cases were divided into 2 groups: isolated cysts and subependymal cysts associated with other cerebral disorders.

Results

In 2 cases of isolated subependymal cysts on ultrasound and MRI: In one patient 2 small cysts were diagnosed at 25 weeks of gestation; In the other one a single cyst was found at 33 weeks. In the remaining 7 cases, 3 patients had a primary CMV seroconversion in the first trimester with a positive amniotic fluid PCR in all cases. In 2 /3 patients the fetal MRI confirmed the ultrasound diagnosis and a termination of pregnancy (TOP) was performed at 23 and 30 weeks respectively. The remaining patient declined MRI and this pregnancy is still ongoing. In 1 patient a huge cyst (47x38x49 mm) was found at 36 weeks of pregnancy. Postnatally the diagnosis of plexus papilloma grade 2 was made. This child underwent surgery and chemotherapy and is doing well at the age of 6 years. The 2 patients with multiple subependymal cysts were associated with progressive ventriculomegaly. MRI confirmed the ultrasound findings. The first child died day 5 postnatally and metabolic investigation is still ongoing. The other baby died at the age of 8 months. The diagnosis of repiratory long chain deficiency was made due to a mutation in the mitochondrial DNA: ND1. Finally, multiple subependymal cysts were found at 26 weeks in a dichorionic diamniotic twin pregnancy. The infectious serology was negative and there were no associated anomalies. A selective feticide was carried out at 31 weeks and investigation revealed a molybdeen cofactor deficiency /sulphate oxidase deficiency.

Conclusion

Although already described in the first trimester of pregnancy, our series of subependymal cysts were detected in the second and third trimester. Fetal MRI may be of help to rule out other brain abnormalities and TORCH screening to rule out infectious origin. Subependymal cysts with associated anomalies often result in adverse outcome.