19th World Congress in Fetal Medicine

A case of cardiac rhabdomyoma

J. Paunovic, A. Music
Clinical center of Montenegro, Clinic for Gyneacology and Obstetrics, Podgorica, Montenegro

Objective

TSC (tuberous sclerosis complex) is a rare, multi-system genetic disease that causes benign tumors in the heart, the brain and other vital organs such as the kidneys, eyes, lungs, and skin. TSC is caused by mutations on TSC1 and TSC2 genes. The TSC1 is on chromosome 9 and produces a protein called hamartin. The TSC2 is on chromosome 16 and produces tuberin. These proteins inhibit the activation of a protein mTOR acting like a growth suppressors. Loss of mTOR regulation leads to abnormal differentiation and development, to the generation of enlarged cells, as are seen in TSC brain lesions. Cardiac rhabdomyomas are often found on prenatal fetus ultrasound exams and in the hearts of infants and young children with TSC, causing atrial or ventricular arrhythmias, sinus node dysfunction, obstruction of the ventricular outwlow tracts, secondary cardiogenic shock. Despite the potentially favourable cardiac evolution of patients with cardiac rhabdomyomas, their presence suggests a tuberous sclerosis with a neurological prognosis that is not related to the number and the dimensions of rhabdomyomas.

Methods

For the diagnosis of cardiac rhambdomyomas we used the two dimensional ultrasound and 3-D echocardiography.

Results

A 26- year-old, pregnant women, multigravida, primi- para, at her 29. th gestational week with previous history of hypertensive sy in first pregnancy and cesarean cection and no remarkable family history was reffered to us for sonographic morphological examination of a cardiac malformation diagnosed by routine sonographic study. Ultrasonographic examination of the fetus demonstrated multiple solid masses consistent with rhabdomyoma in the left ventricular wall (11 mm), interventriculare septume (8 mm) and mitral and tricuspid valve. These masses presented as a hyperechogenic, homogeneous, avaskular aspect and were diagnosed as cardiac rhabdomyomas (Fig. 1, Fig. 2). Cardiac size was normal without any associated cardiac anomaly. During the examination cardiac arrhytmia in the form of premature atrial contractions was detected followed by pericardial effusion and discreet fetal hydrops. Fetal cranial sonographic examination revealed borderline ventriculomegaly (11-12 mm). No other anomalies could be deteceted. Feticid followed by parva cection – hysterotomy was performed in 29 weeks. Umbilical cord blood was taken for genetic testing for tuberous sclerosis. The autopsy findings confirmed the diagnosis.

Conclusion

Fetal echocardiography enables early diagnosis of tuberous sclerosis through prenatal detection of cardiac rhabdomyoma and facilitates genetic councelling of familiies at risk. When fetal cardiac rhabdomyoma is diagnosed, control of their development and precisse evaluation of other fetal structures should be performed to search for signs of TSC. The significance of it's diagnosis is exemplified by the neurodevelopmental complications in patients, showing epilepsy, delayed development.