FETAL CARDIAC RHABDOMYOMA A CASE OF MATERNAL AND FETAL TUBEROUS SCLEROSIS COMPLEX

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Objective

• Rhabdomyomas are the most common fetal cardiac tumors. They may be located in all myocardial areas but are usually detected in the septum or ventricles and are seen as non-vascular homogenous hyper echogenic masses originating from the myocardium. They are usually detected antenataly in the second trimester and are most often associated with tuberous sclerosis complex. TSC (tuberous sclerosis complex) is an autosomal dominant syndrome with variable penetration, characterized by benign tumors in numerous organs including the heart, kidneys, brain, skin. Two genes implicated in tuberous sclerosis have been identified: the tuberous sclerosis 1 (TSC1) hamartin gene on chromosome 9 (9q34) and the tuberous sclerosis 2 (TSC2) tuberin gene on chromosome 16 (16p13.3)

Methods

• The fetal ultrasonographic imaging was performed using a 5 MHz transabdominal transducer from Voluson E8 ultrasonography device

Results

• We present a case of 32-year-old patient, gravida four, parity three. At her 36th gestational week she was referred for sonographic morphological examination of a cardiac malformation diagnosed by routine sonographic study. She had three previous births, two with cesarean section and one spontaneous birth. She had one healthy child, one had died 21 days after birth and one child was diagnosed with tuberous sclerosis complex and was receiving therapy. Medical history revealed that the patient had a mosaic form of deletion of TSC2/PKD1 gens. The genetic form that she was caring was recognized to be asymptomatic or with modest symptoms accompanied with polycystic kidneys disease. General physical examination and systemic examination findings were normal except for the polycystic kidneys. She didn't receive any therapy. Obstetric history showed that in her second pregnancy the fetus was diagnosed with cardiac rhabdomioma in 36 the gestational week and was delivered in 38 th gestational week. Genetic testing was done after delivery and showed heterozygotic deletion of exons 3, 31-42 of the TSC2 gene and exons 35-46 of PKD1 gene. The child died 21 days after birth. The mother was the carrier of the novo mutations that were transferred to the fetus. In her third pregnancy genetic testing was not done until after delivery. The neonate had the same mutations, heterozigotic deletion of exons 3, 31-42 of the TSC2 gene and exons 35-46 of PKD1 gene and had polycystic kidneys disease and was receiving proper therapy. Obstetric history of her current fourth pregnancy showed that she had only one prenatal visit in 33 th week of pregnancy. At her 36 th gestational week she was referred to our Clinic for evaluation of cardiac malformation. Obstetric ultrasound showed multiple hyperehogenic, avascular, homogenous and solid masses in the wright ventricle (8 mm), interventricular septum (11x6 mm) and left ventricle (11x10 mm and 10x9 mm). These were identified as fetal cardiac rhabdomiomas . Cardiac size was normal without any associated cardiac anomaly. Cystic hypoechoic structure with diameter of 4 cm was identified on the left kidney. Amniocentesis and genetic testing was done. The results showed heterozygotes deletion of exons 3, 31-42 of TSC2 gene and exons 35-46 of PKD1 gene. The patient was delivered in 39 th week of gestation by cesarean section. Live female neonate weight 2680 grams and 51 cm long. Apgar scores were 6 in first minute and 7 In fifth minute. The neonate was transferred to specialized cardiovascular intensive care unit. Neonatal cardiosonografy showed tuberous formation in the left ventricle (8x11 mm), apex of the heart (6x2 mm), interventricular septum (12x3 mm) and wright atrium (3 mm). The great vessels of the heart had normal configuration. The tuberous formations didn't cause any obstruction. Sonography of the neonatal brain showed no pathological findings. Sonography of the kidneys confirmed the prenatal findings of cyst on the left kidney. The baby was treated for neonatal infection and was later discharged.

Conclusion

• Fetal cardiac rhabdomyioma is a rare condition but it is the most common cardiac tumor in fetal life. Early prenatal diagnosis of cardiac rhabdomyomas is important for perinatal follow-up and a multidisciplinary approach to treatment. Fetal cardiac rhabdomyomas are most often associated with tuberous sclerosis complex and it should prompt further fetal evaluation and genetic testing. Genetic counseling is recommended for couples who have a family history of tuberous sclerosis and who want to have children. Prenatal diagnosis is available for families with a known gene mutation or history of this condition. A pregnancy complicated by maternal or fetal tuberous sclerosis deserves careful observation considering the multisystem nature of the condition and the neurologic impairment that comes with it.