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Prenatal diagnosis of cardiac rhabdomyoma case report

Abstract

We describe a case of a cardiac tumor diagnosed by ultrasound in a fetus of 20 weeks of gestation with presumptive diagnosis of cardiac rhabdomyoma. The pregnancy was finished by cesarean section at 38 weeks of gestation, because previous cesarean section. In the assessment of the newborn, the diagnosis of cardiac rhabdomyoma was confirmed on echocardiography. We report case of prenatal rhabdomyoma associated with maternal clinical diagnosis of Tuberous Sclerosis (TE).

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Introduction

Tuberous sclerosis (OMIM #191100; #613254), is a genetic, autosomal dominant, multisystem disease; characterized by the presence of hamartomas in the brain, skin, heart, kidneys and lung and by neuropsychiatric manifestations. It has a prevalence of 1:10000. Caused by the mutation in one of the genes TSC1, which is located in the long arm of chromosome 9 or TSC2 (located in the short arm of chromosome 16). It has variable expression and can occur at any stage of life.

Case report

A 35-year-old patient, G2Para0 (C-Section), without hereditary data since the patient is adopted. Presents a daughter of 5 years referred healthy at the time of consultation. As a personal history, she presented resection of nasal vascular fibroids in 2008, electro coagulation of facial maculopapular lesions and embolization of 3cm and 4 cm renal angiolipomas in 2015.[1]

Figure 1 shows the evolution of the pregnancy was uneventful, with negative first-trimester markers for trisomies 21, 18 and 13. She came to monthly prenatal control and a fetal scan was performed at week 20 of pregnancy. The biometric parameters were found in normal ranges for gestational age, with male fetus growing in 75º percentile, normal fluid liquid index (FLI).[2] Heart with four chambers and three vessels view, with hyper echogenic homogeneous focus in the left ventricle, apical region, measuring 5 x 4 mm with growth towards the ventricular cavity, which could correspond to rhabdomyoma (Figure 2). At week 25, new ultrasound control that shows persistence of hyper echoic image at left ventricular apex, currently measuring 11x9 mm, A new echographic control was performed at week 32 with a hyper echoic image persisting and measuring 20 x 11 mm. Control in week 36 evidence: fetus growing in 25º percentile, normal FLI, with hyper echoic mass of 22 x 16 mm. Fetal Doppler ultrasound was normal with preserved arterial and venous flows, with no signs of redistribution of flow, with preserved resistance and pulsatility index Mlczoch.[3] Fetal echocardiography was performed in week

36º, showing a homogenous tumor, isolated in a left ventricle cavity compatible with rhabdomyoma. Laminar flow in the entrance and exit tracts. FCF 130 beats per minute, regular rhythm. Left and right ventricular function preserved. Abdominal cesarean section was scheduled for 38 weeks of gestational age due to a previous caesarean section. A male new born was delivered, estimated gestational age 38 weeks, 2600 grs, Apgar 8/9. The diagnosis of rhabdomyoma was corroborated by echocardiography in the newborn, which reported: large tumor mass in the left ventricle of 27 mm Chung,[4] preserved ventricular function. Normal ultrasound of abdomen and brain. A Holter was performed on the 2º day of life, which reported preserved sinus rhythm, without AV blocks or depression of sinus automatism.
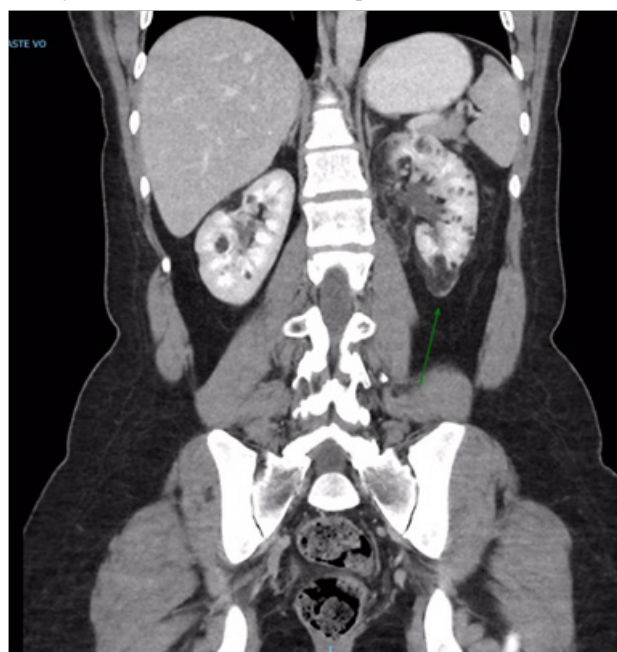

Figure I TAC of the abdomen demonstrates angiolipomas in the left kidney.

Figure 2 Fetal scan with a four-chamber section demonstrating cardiac tumor at the apex of the left ventricle.

Discussion

Among the most common clinical manifestations of TE are: pigmentary and proliferative alterations of the skin, cerebral malformations(subependymal nodules, tubers), cardiac rhabdomyomas, renal angiomyolipomas and pulmonary lymphangioleiomyomatosis. The highest morbidity and mortality is determined by the involvement of brain functions and a high prevalence of refractory epilepsy. Renal lesions, usually angiomyolipomas, can cause clinical problems secondary to hemorrhage or compression and replacement of healthy renal tissue, which can cause renal failure. Patients can also develop renal cysts and renal cell carcinomas. Skin lesions include melanotic macules, facial angiofibromas, and connective tissue nevi patches. Approximately 10-30% of cases of tuberous sclerosis are due to mutations in the TSC1 gene: the frequency of cases due to mutations in the TSC2 gene is 69%. TSC2 mutations are associated with a disease with a more severe presentation. The diagnosis is based on internationally diagnostic clinical criteria (last revision in 2012), which include the most frequent manifestations of the disease. It is confirmed molecularly with the identification of a mutation in TSC1 or TSC2, detectable in approximately 80% of cases. The remaining 15% of cases is probably due to a mosaicism. Regarding treatment, the different manifestations of the disease require a multidisciplinary management, from the treatment of epilepsy to the eventual renal or respiratory failure. The use of everolimus, an inhibitor drug of the mTOR pathway, has recently been approved to slow down the growth of subependymal giant brain cell tumors and renal angiomyolipomas. It is expected that in the future its use could be extended to treat other manifestations of the disease.

Conclusion

The early diagnosis of prenatal cardiac rhabdomyoma is of great importance for the monitoring and prognosis of pregnancy and the TEsuspicion, since patients with cardiac rhabdomyoma have a 75 to 80% risk of being affected by TE. In the case of clinical and / or molecular postnatal diagnosis, strict follow-up must be carried out with dermatological, dental and ophthalmological control, control of blood pressure, perform brain NMRI, EEG, ECG, pulmonary function tests, chest and abdominal CT and control of renal function. Performing an early diagnosis of TE in pediatric patients before the onset of seizures has a less severe epilepsy and a better outcome of development. Therefore it is essential to advise pregnant patients with TE.

Acknowledgments

Conflicts of interest

Author declares that there are no conflicts of interest.

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