A CASE OF MASSIVE FETAL CARDIAC RHABDOMYOMA: ULTRASOUND FEATURES AND MANAGEMENT

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Introduction:

Primary heart tumors are an extremely rare finding during prenatal examination. Fetal cardiac tumors are usually benign, with variable clinical manifestations. We report the case of a massive fetal cardiac rhabdomyoma.

Case report:

A 31-year-old woman was referred to our clinic at 23 weeks of gestational age because of the finding of a large cardiac mass during the second trimester screening ultrasound, raising the suspicion of cardiac fibroma. Non-invasive prenatal testing and nuchal translucency thickness assessed during the first trimester aneuploidies screening were within normal limits. Echocardiographic examination showed a hyperechogenic homogeneous area of 26x15 mm at the atrio-ventricular junction (Figure 1). It was extending throughout the whole cardiac diameter and moved synchronically with cardiac chambers. Pulsed wave and Color Doppler assessment of atrioventricular and semilunar valves showed normal waves form. A modest pericardial effusion was shown but cardiac frequency was normal. A fetal echoencephalography was performed, showing no visible pathological lesions. Fetal magnetic resonance confirmed the presence of an expansive cardiac neoformation of 29x13x12 mm.

The woman underwent an amniocentesis, resulting in a normal female karyotype 46 XX and normal array CHG. However, molecular analysis of genes TSC1 and TSC2 revealed an heterozygotic missense variant of gene TSC2 associated with Tuberous Sclerosis (TS). The same mutation was later found in the asymptomatic mother as well. So the mass was most likely referrable to a single large rhabdomyoma of progressively increasing dimensions. At 37 weeks of gestational age, the mass had reached the remarkable dimension of 48x42 mm, raising the fear of fetal heart failure and malignant arrythmias. At 39 weeks and 5 days of gestational age, the patient underwent a cesarean section for failed induction and delivered a newborn of 2980 grams, with normal umbilical cord gas analysis and Apgar score. The baby was promptly transferred to Neonatal Intensive Care Unit for cardiac monitoring. At ECG, a ventricular pre-excitation was found, with a marked and diffused ST down-sloping. Echocardiogram confirmed the presence of an intrapericardial wide mass of 42x28 mm strictly connected to the right ventricle, the right atrium and the posterior wall of the left ventricle without obstruction. Postnatal therapies with Everolimus and Flecainide were started.

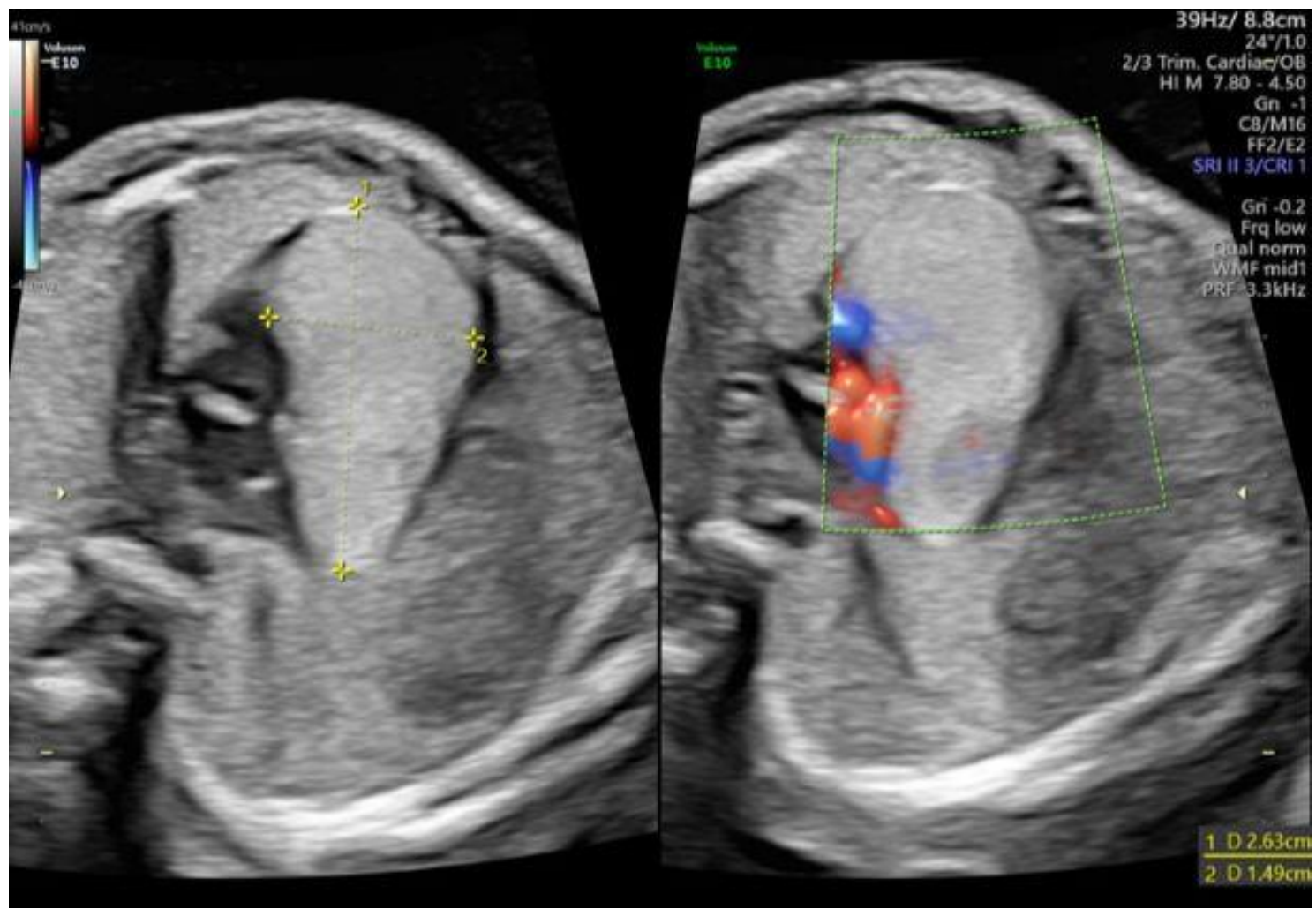

Figure 1. Echocardiographic examination showed a hyperechogenic homogeneous area of 26x15 mm at the atrio-ventricular junction

Conclusions:

Among cardiac tumors, rhabdomyomas are the most common. Even though symptoms and prognosis are determined by their size, location and rhythm or flow impairment, they are mostly asymptomatic and are often diagnosed as incidental findings during second or third trimester of pregnancy. Rhabdomyomas might increase in size in utero, with possible arrythmias or hemodynamic impairment in case of large tumors. The association with TS is known so the detection of a rhabdomyoma should be considered a warning sign for TS. Rhabdomyomas are quite often multiple and located along the intraventricular sept. In this case, it was a single large tumor, developing externally along the ventricular wall, making differential diagnosis and management more challenging. New treatments with mTOR inhibitors such as Sirolimus or Everolimus have been proposed to stimulate involution of rhabdomyomas and reduce the necessity of intervention.