

An Unusual Case of Large, Multifocal Cardiac Rhabdomyomas in Suspected Fetal Tuberous Sclerosis

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[Supplemental Video](#)

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Introduction:

Cardiac rhabdomyomas are the most prevalent form of fetal intracardiac tumors. Such tumors may be detectable by echocardiography as early as 22 weeks gestation. Cardiac rhabdomyomas often but do not always spontaneously decrease in size postpartum. While many are not hemodynamically significant, some rhabdomyomas may cause arrhythmias, obstruction of flow, or heart failure. Fetal cardiac rhabdomyomas are usually discovered as small, solitary masses and may be the earliest sign of tuberous sclerosis, especially in cases of multifocality. This case report presents a rather rare finding of multiple large intracardiac rhabdomyomas during the prenatal period.

Case presentation:

A patient presenting at 36 weeks gestation was referred for a fetal echocardiogram due to concern for fetal tuberous sclerosis given maternal history. Transabdominal fetal echocardiogram was notable for 2 nodular masses, one in the interventricular septum and one near the mitral valve apparatus (Figure 1a). Despite the unusually large size of the multiple rhabdomyomas, bilateral ventricular inflow and outflow was unobstructed. An additional 2 masses were also noted near the moderator band (Figure 1b). Cardiac rhythm was notable for repeated premature beats likely classified as premature atrial contractions. Postnatal echocardiography confirmed the prenatal findings but also revealed an additional two masses.

Working Diagnosis:

The multifocal cardiac rhabdomyomas served to confirm the likelihood of fetal tuberous sclerosis.

Follow-up and discussion:

Postnatal echocardiography confirmed the prenatal findings but also revealed an additional two masses. For one, the case illustrates how fetal echocardiography can serve as a cost-effective and non-invasive method of detecting tuberous sclerosis. However, as displayed in this case, prenatal echocardiography may be somewhat limited and can miss even large masses in significant regions. As such, longitudinal postnatal follow-up of cardiac masses and function is essential. Moreover, the case emphasizes the importance of early prenatal echocardiography especially in instances of known maternal tuberous sclerosis. While the ubiquitous multifocality of cardiac tumors in this case was not hemodynamically significant, the early application of fetal echocardiography can allow early detection and treatment in cases where the tumor burden does affect cardiac function. In sum, this report presents an unusual instance of heavily multifocal cardiac rhabdomyomas without hemodynamic effect and highlights the utility and limitation of echocardiography in the prenatal workup of tuberous sclerosis.

Learning Objectives

- 1) Identify early signs of tuberous sclerosis in utero and associated abnormalities.
- 2) Identify cost-effective and safe strategies to detect tuberous sclerosis in cases of high suspicion.

3) Identify guidelines and indications for medical treatment in cases of cardiac rhabdomyomas.



Figure 1a) Prenatal echocardiography revealing a cardiac rhabdomyoma near the mitral valve apparatus.

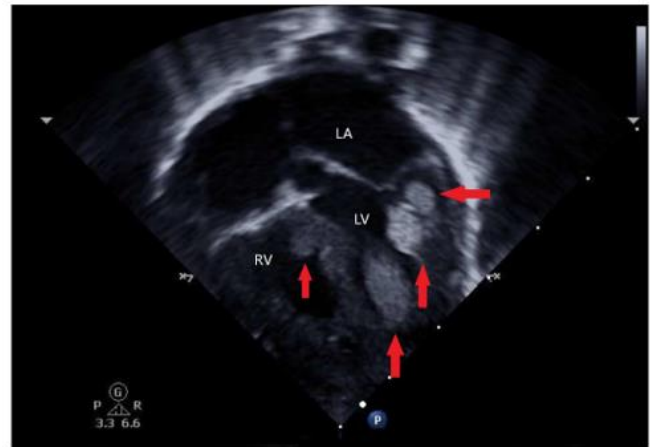


Figure 1b) Postnatal echocardiography revealing multiple cardiac rhabdomyomas, two of which were not visible during prenatal examinations.