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Case Report

A Rare Presentation of Fetal Cardiac Rhabdomyoma: Prenatal Diagnosis and Perinatal Management

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Abstract

Fetal cardiac rhabdomyoma is an uncommon benign cardiac tumour and is the most frequently encountered primary cardiac tumour during fetal life. Its prenatal recognition is clinically important because cardiac rhabdomyomas are strongly associated with tuberous sclerosis complex (TSC), particularly when lesions are multiple. We report a 28-year-old primigravida referred at 27 weeks after an echogenic cardiac mass was identified at the routine anomaly scan at 20 weeks. Targeted ultrasonography and fetal echocardiography at 28 weeks demonstrated multiple homogeneous echogenic masses involving both ventricles and the interventricular septum, the largest measuring approximately 2 cm, with mild restriction of left-sided atrioventricular flow. No extracardiac abnormalities or hydrops were identified. The pregnancy was followed with fortnightly surveillance for hydrops, cardiac dysfunction and arrhythmia, with multidisciplinary counselling involving maternal-fetal medicine, neonatology, paediatric cardiology and genetics. Elective caesarean delivery at 38 weeks resulted in a 3-kg male neonate with normal cardiac function and rhythm at birth. Postnatal echocardiography confirmed significant biventricular rhabdomyomas without outflow obstruction. This case highlights the importance of detailed fetal echocardiography, surveillance for haemodynamic compromise, and evaluation for TSC after prenatal detection of cardiac rhabdomyoma. Recent literature supports an expectant approach in haemodynamically stable fetuses, while selected severe cases with progressive obstruction or cardiac compromise may be considered for mTOR-inhibitor therapy in specialist centres.

Introduction

Case presentation

We report a 28-year-old primigravida referred at 27 weeks of gestation after an echogenic cardiac mass was identified at the routine anomaly scan at approximately 20 weeks. Targeted ultrasonography and fetal echocardiography at 28 weeks demonstrated multiple homogeneous echogenic intramyocardial masses involving both ventricles and the interventricular septum, the largest measuring approximately 2 cm. Mild restriction of left-sided atrioventricular inflow was observed, without significant outflow tract obstruction, ventricular dysfunction, arrhythmia, or hydrops. No extracardiac structural abnormalities were identified. The pregnancy was followed with fortnightly fetal echocardiographic surveillance. An elective caesarean delivery at 38 weeks resulted in a 3-kg male neonate with normal cardiac rhythm and preserved cardiac function. Postnatal echocardiography confirmed significant biventricular rhabdomyomas without outflow obstruction. Neonatal cranial ultrasonography was normal, postnatal TSC1/TSC2 molecular testing was negative, and there was no relevant family history. Serial follow-up for 2 years demonstrated progressive spontaneous regression, with complete resolution of the cardiac rhabdomyomas by the end of the first year of life without medical, surgical, or interventional treatment.

Discussion

The principal clinical importance of a prenatal diagnosis of cardiac rhabdomyoma extends beyond the cardiac lesion itself. Rhabdomyoma is a major prenatal marker of TSC, an

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autosomal-dominant disorder caused by pathogenic variants in TSC1 or TSC2 and associated with dysregulation of the mTOR pathway. Contemporary series continue to demonstrate a substantial proportion of fetuses with cardiac rhabdomyoma having TSC, with the association being particularly strong in the presence of multiple lesions.

In the present case, the presence of multiple tumours involving both ventricles and the interventricular septum made the association with TSC clinically important. However, there were no reported extracardiac abnormalities on prenatal ultrasound. The absence of extracardiac findings does not exclude TSC, and postnatal genetic and clinical assessment remains appropriate.

The differential diagnosis of a fetal cardiac mass includes fibroma, teratoma, haemangioma and other rare cardiac lesions. Rhabdomyomas are characteristically homogeneous and hyperechoic and are often intramyocardial, with a predilection for the ventricles and septum. Fibromas tend to be solitary, arise from the ventricular myocardium and may be associated with calcification. Teratomas are usually extracardiac or pericardial and may be heterogeneous with cystic components and pericardial effusion. Fetal echocardiography is therefore central to defining the number, size, location and haemodynamic effect of the lesions.

Clinical outcome depends less on the mere presence of a rhabdomyoma than on its size, number, location, growth pattern and effect on cardiac physiology. Potential prenatal complications include ventricular inflow or outflow obstruction, impaired ventricular function, arrhythmia, pericardial effusion, hydrops and fetal demise. Large lesions, tumour progression, hydrops and fetal dysrhythmia have been associated with adverse perinatal outcomes in published series.

The present fetus remained haemodynamically stable despite multiple lesions and a maximum diameter of approximately 2 cm. Serial surveillance without prenatal intervention was therefore appropriate in the reported course. The absence of hydrops, significant obstruction or arrhythmia allowed continuation to term.

Review of literature

Recent evidence reinforces the generally benign natural history of many fetal cardiac rhabdomyomas but also identifies a clinically important subgroup requiring intensive surveillance. A 2024 systematic review and meta-analysis including 400 fetuses reported prenatal arrhythmia in approximately 13%, outflow tract obstruction in 16% and hydrops in 10%; fetal demise occurred in approximately 12%. TSC was diagnosed in approximately 60% of reported cases. Tumour reduction was documented in a minority before delivery and in a substantially larger proportion after birth.

A 2024 single-centre study similarly found that multiple lesions were strongly associated with TSC and that most surviving infants demonstrated diminution or disappearance of cardiac rhabdomyomas during follow-up. Among infants with TSC, neurological morbidity remained an important long-term consideration, emphasising that cardiac findings may represent the first opportunity for early TSC diagnosis.

Prenatal brain MRI has increasingly been used when TSC is suspected. Published cohorts demonstrate that MRI can identify subependymal nodules and cortical abnormalities that may be occult on ultrasound. Genetic testing, particularly analysis of TSC1 and TSC2, provides an additional route to diagnosis and is especially valuable when the prenatal phenotype is suggestive but not diagnostic.

The role of prenatal mTOR inhibition is evolving. A 2024 systematic review and meta-analysis identified a small number of reported fetuses with TSC treated with prenatal mTOR inhibitors, mainly sirolimus, with reported reduction in tumour size and relief of obstruction. However, the evidence remains based predominantly on case reports and small series rather than randomized trials. Recent reviews suggest considering such treatment only in highly selected pregnancies with progressive tumour growth and severe haemodynamic compromise, within an experienced multidisciplinary fetal medicine and paediatric cardiology setting.

A 2026 systematic review of 859 reported cases identified hydrops as the strongest predictor of mortality, while tumour progression, fetal heart failure and fetal growth restriction were also associated with adverse outcome. Multiple tumours were associated with a higher likelihood of TSC. These findings support the surveillance strategy used in the present case, with particular attention to haemodynamic deterioration rather than tumour presence alone (Figures 1,2).

Prenatal management considerations

Confirm the diagnosis with detailed fetal echocardiography and document tumour number, size, location and relationship to valves and outflow tracts.



Figure 1: Prenatal fetal echocardiographic images demonstrating multiple echogenic cardiac masses involving both ventricles and the interventricular septum. Colour Doppler imaging illustrates the relationship of the masses to intracardiac blood flow. LV: Left ventricle; RV: Right ventricle; LA: Left atrium; RA: Right atrium.

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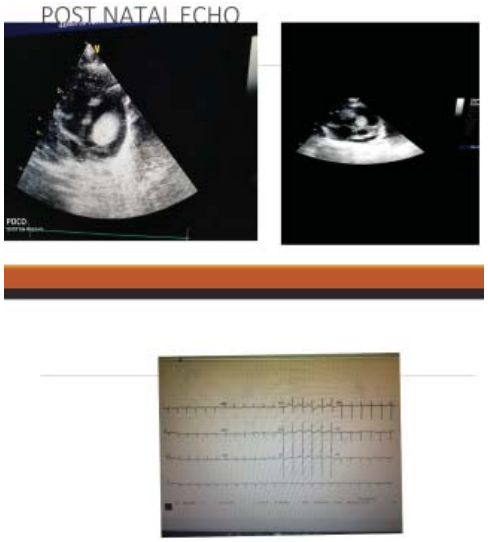


Figure 2: Postnatal echocardiographic images demonstrating cardiac rhabdomyomas, with representative views showing the intracardiac masses following delivery.

Assess cardiac rhythm, ventricular function, inflow and outflow velocities and evidence of obstruction or pericardial effusion.

Screen carefully for extracardiac features suggestive of TSC, particularly intracranial abnormalities.

Consider fetal brain MRI and genetic counselling/testing for TSC1/TSC2 when clinically appropriate.

Use serial surveillance to detect tumour progression, arrhythmia, ventricular dysfunction, hydrops or fetal growth restriction.

Plan delivery according to obstetric indications and the fetal cardiac condition; stable cases may often continue to term.

Arrange postnatal echocardiography and structured evaluation for TSC, with paediatric cardiology and genetics follow-up.

Reserve invasive or pharmacological intervention for selected cases with severe, progressive haemodynamic compromise and manage such cases in specialist multidisciplinary centres [1-14].

Conclusion

Prenatal fetal cardiac rhabdomyoma is an important diagnosis because it combines a usually benign, potentially regressing cardiac lesion with a significant association with TSC. In a haemodynamically stable fetus, careful serial fetal echocardiographic surveillance and multidisciplinary counselling can permit continuation to term, as demonstrated in this case. Multiple cardiac rhabdomyomas should prompt consideration of TSC even when no extracardiac abnormalities are apparent on ultrasound. Postnatal echocardiographic assessment, genetic evaluation and longitudinal neurological and developmental surveillance are important. Emerging evidence regarding prenatal mTOR inhibition is promising but remains limited and should not replace individualized multidisciplinary assessment.

Primary fetal cardiac tumours are rare, and rhabdomyoma is the most common tumour identified during fetal life. On ultrasound, rhabdomyomas typically appear as well-circumscribed, homogeneous, hyperechoic, non-vascular masses arising within the myocardium, frequently involving the ventricular walls or interventricular septum. Lesions may be solitary or multiple. Multiple rhabdomyomas have a particularly strong association with TSC and should prompt a structured evaluation for this multisystem genetic disorder. Although many lesions remain clinically silent and regress spontaneously, haemodynamically significant tumours can cause inflow or outflow obstruction, arrhythmia, ventricular dysfunction, hydrops and, rarely, fetal or neonatal death.

Declarations

Informed Consent: Written informed consent was obtained from the patient's parent(s) for publication of the clinical details and anonymised prenatal and postnatal echocardiographic images included in this case report.

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