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Prenatal cardiac rhabdomyomas as the first manifestation of tuberous sclerosis complex: A case report

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ABSTRACT

Fetal cardiac masses are rare findings, and rhabdomyomas represent the most frequent type, with a strong association with tuberous sclerosis complex (TSC). Their prenatal detection is therefore an important indicator requiring targeted postnatal assessment. We report a case in which routine antenatal ultrasound revealed multiple cardiac rhabdomyomas, confirmed on fetal MRI. After term delivery and a normal initial examination, postnatal neuroimaging identified subependymal nodules and cortical tubers consistent with TSC, later confirmed by a pathogenic TSC2 mutation. This case illustrates the value of recognizing fetal cardiac rhabdomyomas as an early marker of TSC and the importance of timely multidisciplinary follow-up.

KEYWORDS :

Fetal cardiac tumor, Cardiac rhabdomyoma, Prenatal diagnosis, Tuberous sclerosis complex.

MAIN ARTICLE

INTRODUCTION

Cardiac rhabdomyoma represents the most frequently identified cardiac tumor in the prenatal period, accounting for approximately half of fetal cardiac neoplasms [1]. These lesions are benign and often multifocal, and many fetuses remain asymptomatic even when the masses are sizable or numerous [1]. More importantly, their prenatal identification is strongly associated with tuberous sclerosis complex (TSC), a rare autosomal dominant multisystem disorder characterized by hamartomatous lesions in the brain, heart, kidneys, and eyes [2]. As one of the earliest and major diagnostic criteria of TSC, antenatal detection of cardiac rhabdomyoma assists in postnatal assessment, genetic counseling, and early multidisciplinary care [2]. We report a prenatally diagnosed case of multiple cardiac rhabdomyomas that enabled early confirmation of TSC.

CASE REPORT

A 31-year-old woman, gravida 4 para 3, was referred for routine third-trimester ultrasound at 30 weeks of gestation. The examination revealed three well-defined, oval, hyperechoic cardiac masses (Figure 1). The largest lesion was located within the left ventricle. No pericardial effusion or signs of cardiac dysfunction were noted.

Fetal cardiac MRI (Figure 2) was subsequently performed, which confirmed the prenatal findings. It demonstrated well-circumscribed, oval lesions with regular contours and intermediate signal intensity on T2-weighted sequences. Their locations included one within the left ventricle and another extending between the left atrium and the left ventricle. No obstruction of inflow or outflow tracts was observed. Based on the multiplicity of the lesions and their typical imaging features, the diagnosis of cardiac rhabdomyomas was strongly suggested.

The pregnancy progressed uneventfully, and delivery occurred at 37 weeks of gestation. The newborn had reassuring Apgar scores, exhibited no respiratory distress, and initial clinical examination was otherwise normal. Anthropometric parameters were within expected ranges, and no dysmorphic features or cutaneous stigmata were identified. Cardiovascular examination confirmed stable hemodynamics without murmurs or arrhythmias.

Because cardiac rhabdomyomas are frequently associated with TSC, a systematic search for extracardiac manifestations was initiated postnatally. Cranial ultrasound revealed multiple subependymal nodules. Cerebral CT (Figure 3) confirmed rounded, spontaneously hyperdense subependymal nodules, along with triangular cortico-subcortical lesions in the

right frontal and left occipital regions, consistent with cortical tubers.

Systemic evaluation was comprehensive and showed no abnormalities. Renal ultrasound did not reveal angiomyolipomas or cystic lesions, ophthalmologic examination found no retinal hamartomas, and dermatological assessment identified no cutaneous stigmata of tuberous sclerosis.

The diagnosis of tuberous sclerosis complex was established based on the coexistence of cardiac rhabdomyomas and cerebral lesions. Genetic testing identified a pathogenic mutation in the TSC2 gene, thereby confirming the diagnosis.

During follow-up, echocardiography demonstrated a reduction in the size of the cardiac masses, with preserved ventricular function and no hemodynamic compromise. Clinically, the infant remained stable, with no seizures, developmental delay, or feeding difficulties in the early neonatal period. Neurological monitoring and multidisciplinary follow-up were initiated to anticipate potential complications.

DISCUSSION

Primary cardiac tumors are exceptionally rare in the pediatric population, with an estimated incidence of about 0.2% [1]. Among them, cardiac rhabdomyomas are by far the most frequent, accounting for more than 60% of primary cardiac tumors identified in fetuses and children [2]. Although uncommon overall, their prenatal incidence is estimated around 0.17%, and they are typically detected between 20 and 30 weeks of gestation [1,3]. Rhabdomyomas tend to be multiple and usually arise within the left ventricle or the interventricular septum [4]. Despite their benign nature and spontaneous regression in early childhood, these tumors may lead to clinically relevant complications depending on their size and location [1]. Approximately 10% can produce outflow tract obstruction, potentially progressing to cardiac failure or hydrops [2]. Lesions situated near the conduction system may also trigger arrhythmias or hemodynamic instability, though many fetuses remain asymptomatic even when several masses are present [2].

Cardiac rhabdomyomas have a strong association with TSC and often represent its earliest antenatal manifestation [5]. TSC is an autosomal dominant multisystem disorder caused by pathogenic variants in TSC1 or TSC2, genes widely expressed in neural, renal, epithelial, and cardiac tissues [1,5]. The prenatal discovery of multiple cardiac rhabdomyomas carries a 75–80% likelihood of underlying TSC [1].

Prenatal imaging plays a central role in evaluation. On fetal echocardiography, rhabdomyomas typically appear as homogeneous, hyperechoic, well-defined intramyocardial nodules [6]. Assessment focuses on detecting any hemodynamically significant inflow or

outflow obstruction and on monitoring fetal rhythm for associated arrhythmias [6]. Fetal MRI shows these tumors as homogeneous lesions, typically isointense to mildly hyperintense on T1-weighted images and hyperintense on T2-weighted images relative to myocardium, and it is particularly useful for mapping the extent of larger masses [7]. Given their strong association with tuberous sclerosis complex, MRI also enables systematic assessment of the brain and kidneys, where subependymal nodules, cortical or subcortical tubers, subependymal giant cell astrocytomas, megalencephaly, hemimegalencephaly, or renal cysts may be detected, especially from the second trimester onward [6, 8].

After diagnosis, fetal follow-up aims to identify those at risk of hemodynamic compromise. At least one third-trimester evaluation is usually performed [6]. Maternal treatment with sirolimus has recently emerged as a therapeutic option for selected symptomatic cases, requiring close echocardiographic monitoring [6].

Postnatally, most rhabdomyomas regress spontaneously during the first years of life [6]. Asymptomatic infants undergo periodic echocardiography until regression is confirmed [9]. In children with larger lesions affecting valve function or causing outflow tract obstruction, the follow-up schedule is adapted to the degree of impairment [6, 9]. In patients with confirmed TSC, mTOR inhibitors may accelerate tumor regression [6]. Surgical intervention remains exceptional and is reserved for significant obstruction, valve dysfunction, or refractory arrhythmias [10].

This case highlights the value of antenatal imaging in detecting fetal cardiac rhabdomyomas and evaluating their potential hemodynamic impact. Early identification is particularly relevant because of the strong association with tuberous sclerosis complex, as the prenatal discovery of multiple rhabdomyomas frequently represents its earliest manifestation. Making the diagnosis in utero allows targeted screening for extracardiac features of TSC, supports appropriate genetic counseling, and helps plan both perinatal care and postnatal follow-up.

CONCLUSION

The antenatal discovery of multiple cardiac rhabdomyomas is a key marker for tuberous sclerosis complex. This case illustrates how prenatal imaging can direct focused postnatal assessment, leading to early confirmation of the diagnosis through characteristic brain lesions and genetic testing. Early recognition provides an opportunity to organize appropriate follow-up and anticipate potential neurological complications.

FIGURES

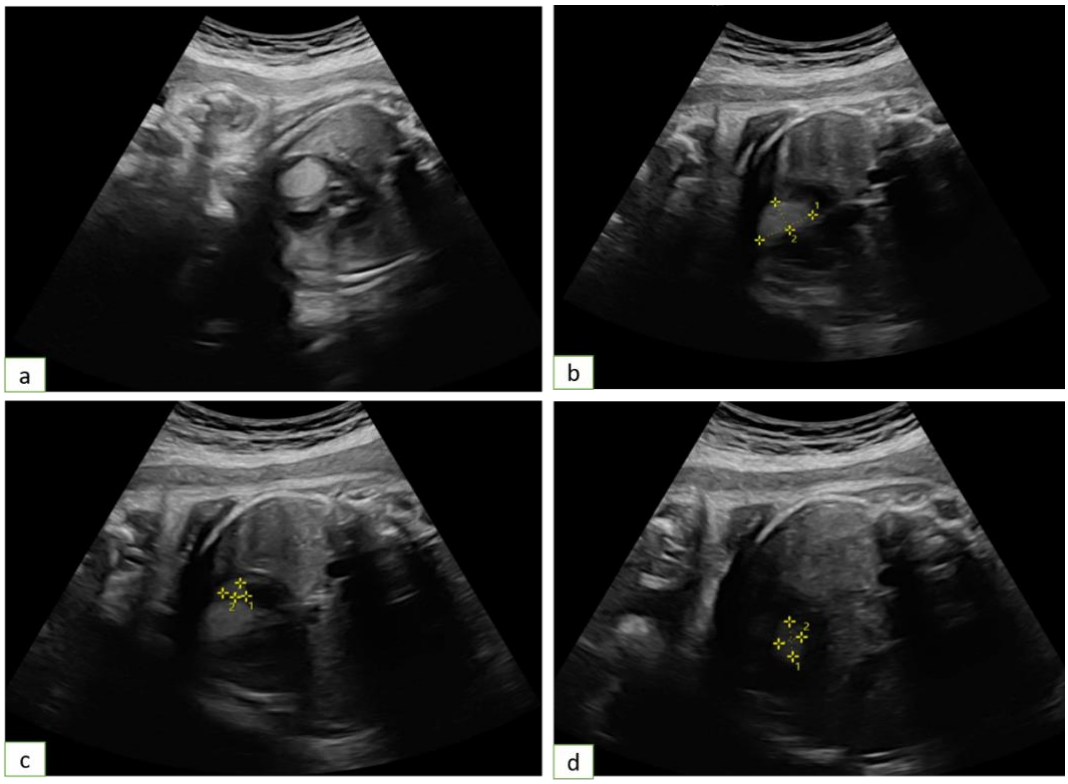

Figure 1: *Prenatal echocardiography images (a, b, c, d) demonstrating three well-defined, hyperechoic intracardiac masses.*

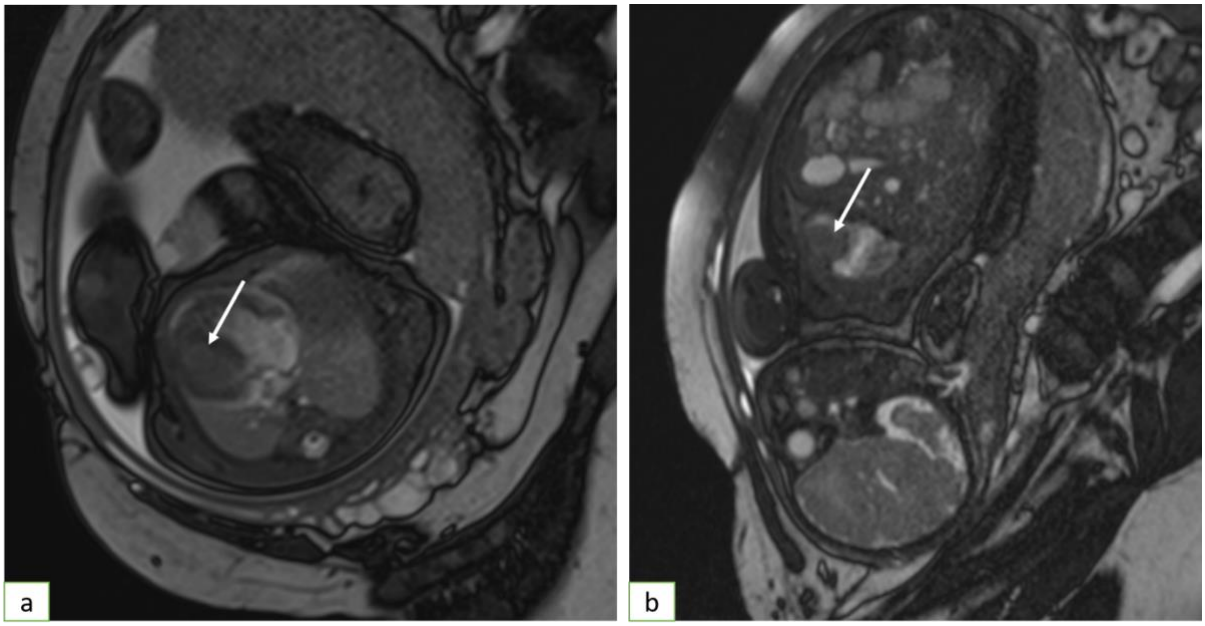

Figure 2 *: Axial (a) and coronal (b) T2-weighted fetal MRI images showing multiple well-defined, oval intracardiac lesions with regular borders and intermediate signal intensity (arrows), suggestive of rhabdomyomas.*

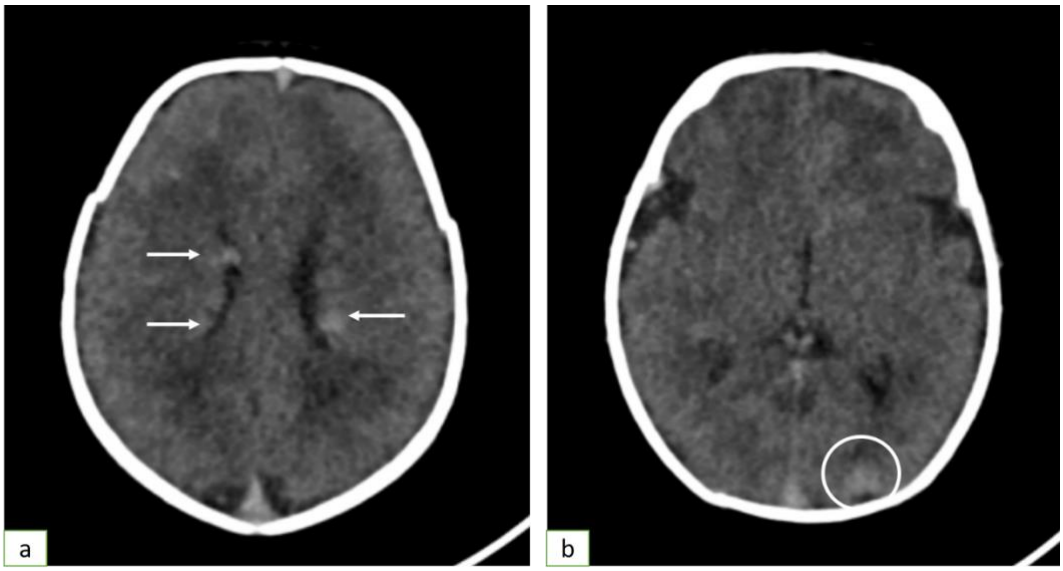

***Figure 3**: Axial cerebral CT images (a, b) showing hyperdense subependymal nodules (arrows) and triangular cortico-subcortical lesions consistent with cortical tubers (circle).*

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The authors declare that they have no conflicts of interest.

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