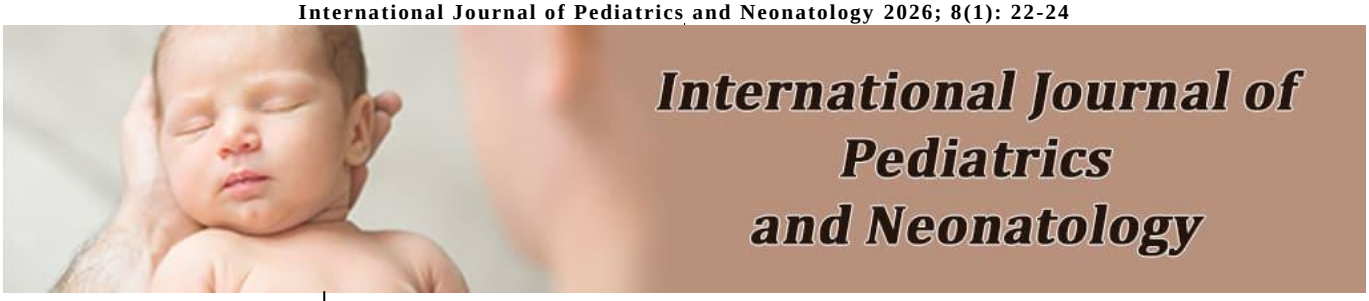

ISSN Print: 2664-8350
ISSN Online: 2664-8369
Impact Factor: RJIF 6.02
IJPN 2026; 8(1): 22-24
www.pediatricsjournal.net
Received: 15-10-2025
Accepted: 16-11-2025

Dr. Hillori Vithalani
Fellow in Neonatology, Department of Neonatology, Setu Newborn Care, Ahmedabad, Gujarat, India

Dr. Arpit Parashar
Senior Resident, Department of Neonatology, All India Medical Science, Bhopal, Madhya Pradesh, India

Corresponding Author:
Dr. Hillori Vithalani
Fellow in Neonatology, Department of Neonatology, Setu Newborn Care, Ahmedabad, Gujarat, India

Cardiac rhabdomyoma in preterm twins: One diagnosis, two different journeys

Hillori Vithalani and Arpit Parashar

DOI: https://www.doi.org/10.33545/26648350.2026.v8.i1a.180

Abstract
Cardiac rhabdomyoma is the most common primary cardiac tumor in the neonatal period. Although generally benign and often regressing spontaneously, these tumors may be associated with significant hemodynamic compromise or underlying genetic syndromes, most notably tuberous sclerosis complex (TSC). We report a rare case of preterm twins with antenatally detected cardiac masses, one of whom was later genetically confirmed to have tuberous sclerosis. The contrasting outcomes highlight the variable clinical spectrum of this condition.
Conclusion: Antenatal detection of cardiac masses, particularly multiple rhabdomyomas, should alert clinicians to the possibility of tuberous sclerosis. Genetic confirmation aids in counselling and long-term management. The divergent outcomes in these twins—one surviving with neurodevelopmental impairment and the other succumbing to cardiac obstruction—illustrate the unpredictable nature and variable severity of tuberous sclerosis. A multidisciplinary approach involving neonatology, cardiology, neurology, and genetics is essential for optimal care and family guidance.

Keywords: Cardiac rhabdomyoma, tuberous sclerosis complex, preterm twins, Molecular analysis, echocardiography, autosomal dominant, counselling

Introduction
Fetal cardiac rhabdomyoma is a benign, smooth muscle tumor [1] of the myocardium consisting of embryonal myoblasts. About 60 to 90% of cardiac rhabdomyomas [2, 3] are associated with or have a positive family history of tuberous sclerosis which is an autosomal-dominant hamartomatous condition. The remaining cases can present as isolated conditions. Unlike other tumor histologic types, rhabdomyomas are unique in that they exhibit high rates of partial or complete spontaneous regression with approximately 50% of these tumors decreasing in size over time [7]. As such, it is widely believed that surgical resection of cardiac rhabdomyomas should be reserved for patients who have hemodynamically significant sequelae directly from their tumors, usually occurring in the form of outflow tract obstruction [8].

Case Report
A twin pregnancy at 28 + 2 weeks of gestation was complicated by the antenatal detection of cardiac masses in both fetuses. Following preterm delivery, both twins required active resuscitation, endotracheal intubation, and mechanical ventilation. Twin 1, a female infant with a birth weight of 1.26 kg, was stabilized post administration of surfactant and inotropic support. Two-dimensional echocardiography (Figure A and B) revealed multiple intracardiac masses involving the left ventricular myocardium, interventricular septum, and tricuspid valve, consistent with cardiac rhabdomyomas. Cranial sonography (Figure C and D) demonstrated cortical and subependymal tubers. Genetic testing confirmed the diagnosis of tuberous sclerosis complex. The infant was later discharged in stable condition. At present, she is 1 year and 10 months of corrected age, on regular follow-up. Developmental assessment shows global delay, and she developed infantile spasms at 8 months of age, for which she is on vigabatrin therapy. Molecular analysis revealed no deletion or duplication of exons in the TSC1 gene at 9q34.13 chromosomal region; however, this does not exclude TSC, as mutations in TSC2 are more common.

The infant had a heterozygous deletion involving exon 12 in the TSC2 gene located at 16p13 chromosomal region, confirming the genetic diagnosis. Twin 2, a male infant with a birth weight of 1.5 kg, required intubation and mechanical ventilation at birth. Echocardiography (Figure A and B) demonstrated a large pericardial mass causing severe obstruction to cardiac outflow with resultant lung compression. Despite intensive neonatal care and inotropic support, the infant succumbed in the early neonatal period. Autopsy was not performed, but based on antenatal and echocardiographic findings, cardiac rhabdomyoma associated with tuberous sclerosis was strongly suspected.

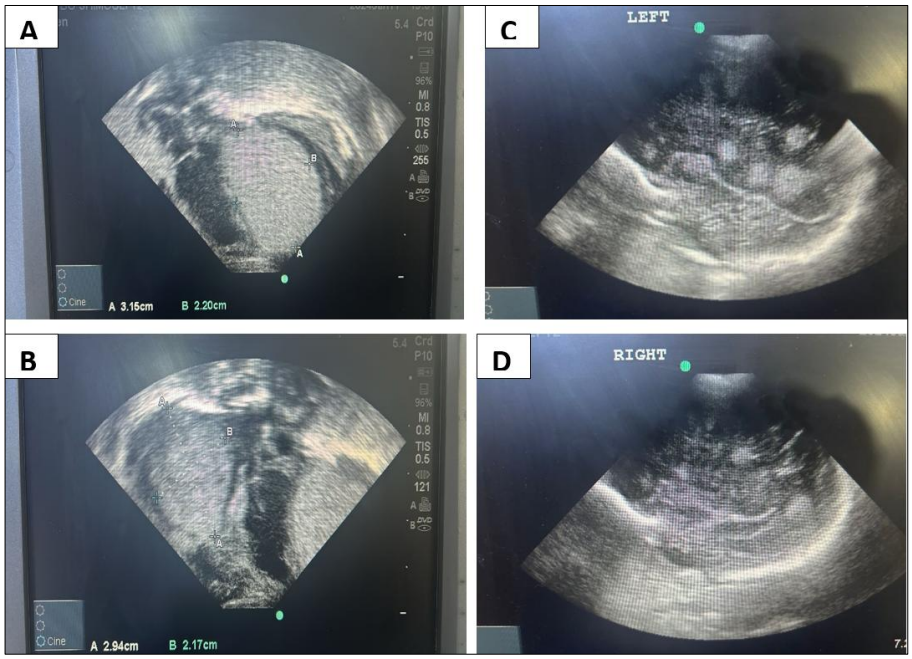

Fig 1: Figure A and B shows two-dimensional echocardiography revealed intracardiac masses Figure C and D shows cranial sonography revealed cortical and subependymal tubers

Discussion

This case underscores the phenotypic variability of tuberous sclerosis even among twins. While smaller, multiple lesions often regress spontaneously, large obstructive tumours can lead to significant hemodynamic compromise and early neonatal death. Cardiac rhabdomyomas may be the earliest and sometimes the only antenatal manifestation of TSC. In a case series by Fesslova V *et al* [4], six out of eleven cases showed decrease in tumour size. Three cases initially showed growth in size of tumour causing partial obstruction but no case required surgery. All the cases regressed to smaller size within time. 81% of the cases were associated with TSC. In a case series by Li Yeng Lg. *et al* [5], mTORi treatment showed decrease in tumour size in four out of 5 patients. In a case by Benyounes *et al* [6], there was concomitant occurrence of rhabdomyoma and giant cell astrocytoma. The coexistence of cardiac rhabdomyomas and intracranial tubers should prompt evaluation for TSC. Early identification facilitates anticipatory management of neurological complications such as infantile spasms and developmental delay.

Conclusion

Antenatal detection of cardiac masses, particularly multiple rhabdomyomas, should alert clinicians to the possibility of tuberous sclerosis. Genetic confirmation aids in counselling and long-term management. The divergent outcomes in these twins—one surviving with neurodevelopmental impairment and the other succumbing to cardiac obstruction—illustrate the unpredictable nature and variable severity of tuberous sclerosis. A multidisciplinary approach involving neonatology, cardiology, neurology, and genetics is essential for optimal care and family guidance.

Acknowledgements

Authors would like to thanks the immense help received from the scholars whose articles are cited and included in references of this manuscript. They are also grateful to authors/editors/publishers of those articles, journals and books from where the literature for this article has been reviewed and discussed.

Funding: No funding sources

Conflict of Interest: None declared

Ethical approval: The study was approved by the Institutional Ethics Committee.

References

1. Isaacs H Jr. Fetal and neonatal cardiac tumors. Pediatr Cardiol. 2004;25(3):252-273. PMID: 15360117.
2. Bader RS, Chitayat D, Kelly E, Ryan G, Smallhorn JF, Toi A, Hornberger LK. Fetal rhabdomyoma: prenatal diagnosis, clinical outcome, and incidence of associated tuberous sclerosis complex. J Pediatr. 2003;143(5):620-624.
3. Chao AS, Chao A, Wang TH, Chang YC, Chang YL, Hsieh CC, Lien R, *et al*. Outcome of antenatally diagnosed cardiac rhabdomyoma: case series and a meta-analysis. Ultrasound Obstet Gynecol. 2008;31(3):289-295.
4. Fesslova V, Villa L, Rizzuti T, Mastrangelo M, Mosca F. Natural history and long-term outcome of cardiac rhabdomyomas detected prenatally. Prenat Diagn. 2004;24(4):241-248.

5. Ng LY, McGuinness J, Prendiville T, Franklin O, Walsh M, Kenny D, Nolke L, McMahon CJ. Cardiac rhabdomyomas presenting with critical cardiac obstruction in neonates and infants: treatment strategies and outcome—a single-center experience. Pediatr Cardiol. 2024;45(5):1132-1141.
6. Benyounes N, Fohlen M, Devys JM, Delalande O, Moures JM, Cohen A. Cardiac rhabdomyomas in tuberous sclerosis patients: a case report and review of the literature. Arch Cardiovasc Dis. 2012;105(8-9):442-445.
7. Günther T, Schreiber C, Noebauer C, Eicken A, Lange R, *et al*. Treatment strategies for pediatric patients with primary cardiac and pericardial tumors: a 30-year review. Pediatr Cardiol. 2008;29:1071-1076.
8. Jordan CP, Costello JP, Endicott KM, *et al*. Intracardiac tumor causing left ventricular outflow tract obstruction in a newborn. J Saudi Heart Assoc. 2016;28:170-172.

How to Cite This Article
Vithalani H and Parashar A. Cardiac Rhabdomyoma in preterm twins: One diagnosis, two different journeys. International Journal of Pediatrics and Neonatology. 2026;8(1):22-24.