

Spontaneous Postnatal Resolution of a Large Foetal Right Ventricular Aneurysm Associated with 9q21.1 Microduplication

Nur Izyani Zainal Abidin¹, Putri Yubbu³, Vairavan L. Velayudham², Nuzulfatiha Sufhia Zalazfian¹, Renuka Rajendran²
1. Obstetric & Gynaecology, Kementerian Kesihatan Malaysia, Putrajaya, Putrajaya, Malaysia. 2. Maternal Fetal Unit, Hospital Sultan Idris Shah Serdang, Kajang, Selangor, Malaysia.3. 3. Paediatric Department, Fakulti Perubatan dan Sains Kesihatan ,Universiti Putra Malaysia

Background

Congenital fetal ventricular aneurysm (VA) is a rare cardiac abnormality with a clinical spectrum ranging from asymptomatic presentation to life-threatening complications such as arrhythmia, rupture, or cardiac failure. Because of its rarity, the natural history and optimal management remain poorly defined.

Case Presentation

A 29-year-old primigravida was referred at 33 weeks' gestation for cardiomegaly. Foetal echocardiography showed a cardiothoracic area ratio of 0.45 and a large aneurysmal outpouching measuring 27 × 22 mm arising from the RV apex, displacing the tricuspid valve superiorly without tricuspid regurgitation, pericardial effusion, or intracardiac thrombus. Chromosomal microarray identified a 4.3 Mb microduplication involving chromosome 9q21.1. Fetal MRI at 31 weeks' gestation supported the diagnosis of an apical RV aneurysm. The pregnancy was closely monitored for hydrops and thrombus formation. At 38 weeks' gestation, an emergency Caesarean section was performed for foetal distress and a baby girl was delivered with good Apgar scores.

Postnatal Course

Postnatal echocardiography confirmed a thinned and hypokinetic RV apical segment measuring 21 × 29 mm, consistent with an aneurysm. The neonate remained haemodynamically stable and asymptomatic. Prophylactic anticoagulation was initiated and she was discharged after a short observation period.

Serial Follow-up

Serial echocardiography at 2 and 4 months demonstrated progressive reduction and eventual complete resolution of the aneurysm, with normal RV size and systolic function. Anticoagulation was discontinued and developmental milestones were age appropriate.

Discussion:

The pathophysiology of congenital ventricular aneurysm (VA) remains uncertain. Although the pathogenic significance of the 9q21.1 microduplication in relation to VA is unclear, this chromosomal region contains COL15A1, which encodes collagen XV, an extracellular matrix protein involved in maintaining myocardial structural integrity and cardiovascular homeostasis. We hypothesize that localized myocardial weakness, together with the relatively high-pressure fetal RV circulation, contributed to aneurysm formation. After birth, the physiological reduction in pulmonary vascular resistance likely decreased RV afterload, facilitating stabilization and myocardial remodelling.

Conclusion

This case demonstrates that even large foetal RV aneurysms may resolve spontaneously postnatally, highlighting the importance of careful surveillance and multidisciplinary perinatal planning before considering early intervention.

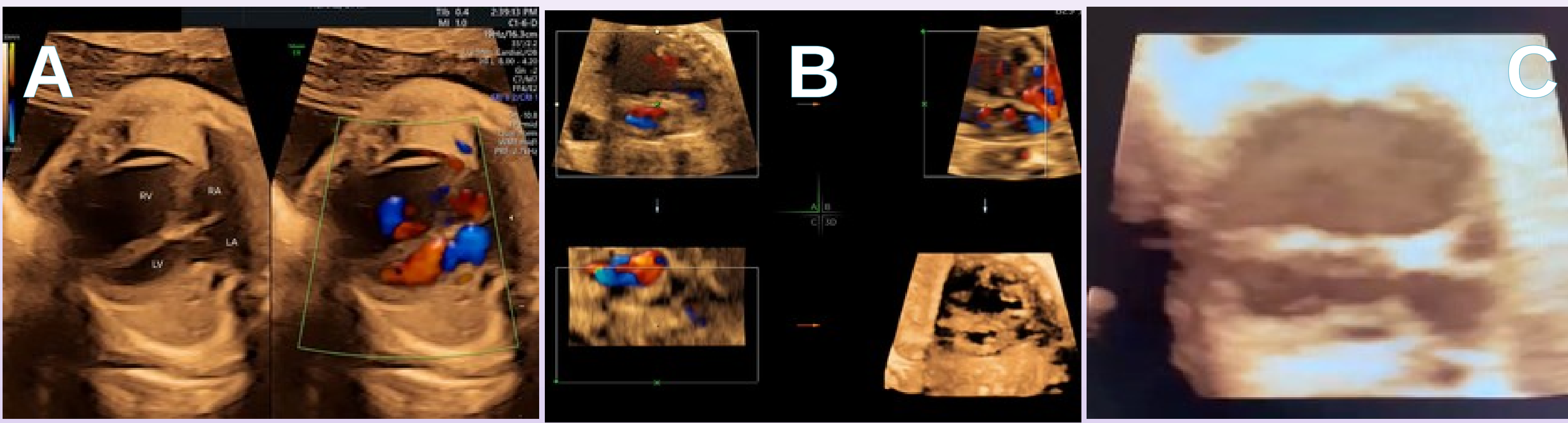


Figure 1. Fetal echocardiographic assessment
A. Four-chamber view demonstrating a large aneurysm arising from the RV apex.
B. Sagittal view confirming the RV apical aneurysm.
C. Three-dimensional echocardiography demonstrating the morphology of the RV apical aneurysm

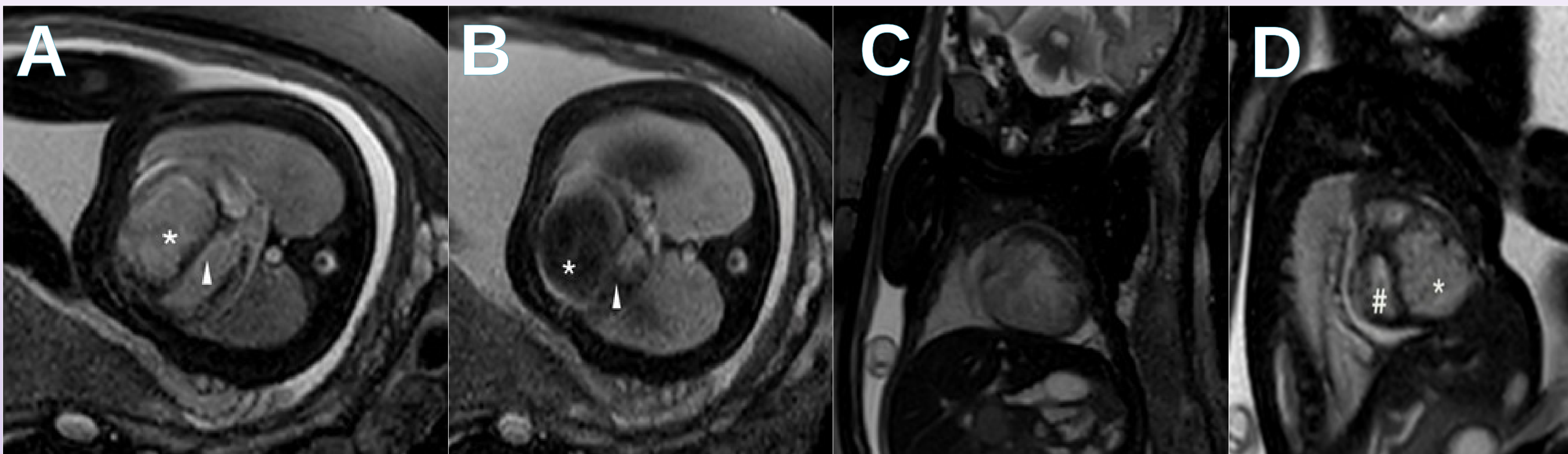


Figure 2. Fetal cardiac MRI assessment.
A. Axial balanced turbo field echo (BTFE) image demonstrating marked dilatation of RV (*) compared to RV (▲).
B. Axial BTFE image demonstrating dilated RV (*) beyond the apex (▲), indicating aneurysm
C. Coronal BTFE image demonstrating dilated RV.
D. Sagittal BTFE image demonstrating the dilated RV (*) extending across the LV (#), compressing and displacing it posteriorly.

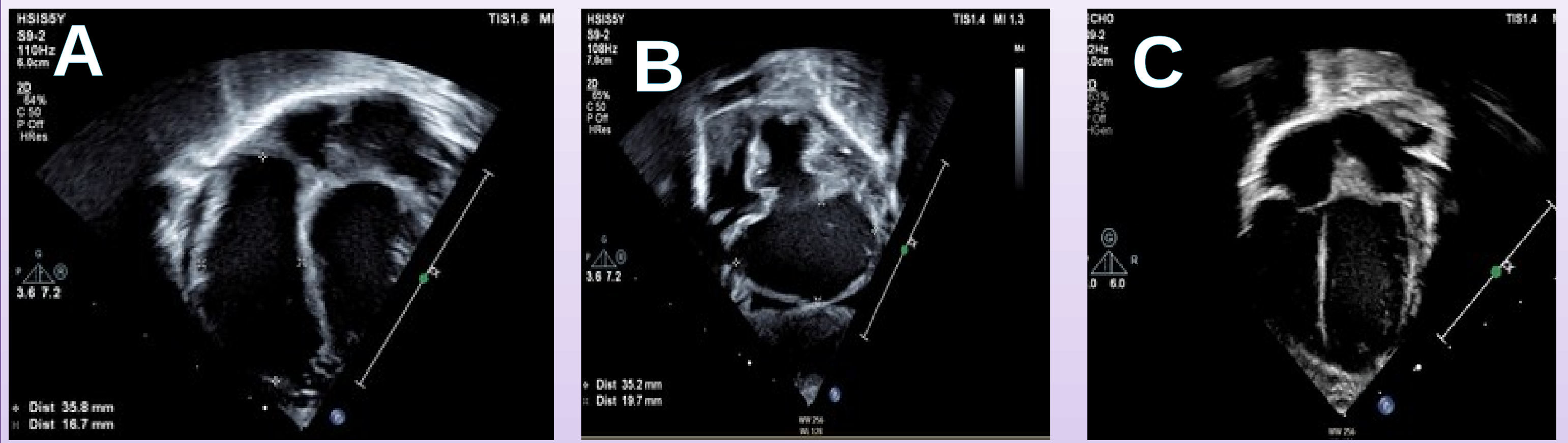


Figure 3. Postnatal echocardiographic assessment. **A.** Four-chamber view demonstrating a large right ventricular (RV) apical aneurysm measuring 35 × 16 mm, occupying the inferior and posterior aspect of the RV apex with superior displacement of the tricuspid valve. The affected RV apical segment was hypokinetic, with no evidence of intracardiac thrombus. **B.** Subcostal view confirming the RV apical aneurysm **C.** Four-chamber view at 2 months demonstrating complete spontaneous resolution of the RV aneurysm with restoration of normal RV morphology