Prenatal diagnosis and lifelong follow-up of a patient with right atrial appendage aneurysm

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Introduction

Congenital right atrial appendage aneurysm (RAAA) is an extremely rare malformation of unknown etiology. Patients may be asymptomatic or present with arrhythmia or recurrent pulmonary embolism. There are no guidelines for treating this condition; however, surgical resection of RAAA has been performed successfully. The data available in the literature is scarce, and therefore, the management and timing of surgery for this condition remain controversial. We present our experience managing a patient with fetal RAAA diagnosis that has continued to be followed through the extent of his life at Unidad de Cirugía Cardiovascular de Guatemala (UNICAR).

Case description

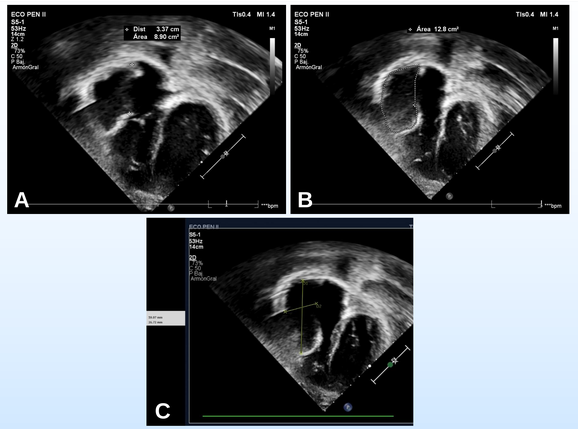
Male patient with fetal echocardiogram diagnosis of right atrial and aortic root dilation. On follow-up, newborn echocardiogram diagnosed RAAA. One-year-of-age echocardiogram reported 20cm^2 area RAAA and 21mm (z+3.6) aortic-root dilation. Patient was asymptomatic otherwise. Patient continues follow-up with yearly echocardiogram and electrocardiogram. Progressive enlargement of RAAA has been documented with last RAAA area 26cm2, and aortic-root 27mm (z+2). Cardiac MRI reported similar findings. No thrombosis nor arrhythmias have ever been documented (four normal Holter monitoring studies). Comprehensive gene analysis and whole-exome-sequencing ruled out monogenic association. Patient is currently 9-years-old and complains of occasional chest pain and palpitations, taking atenolol and aspirin.

Genetic testing at 7 years of age

Test	Result
Sequence analysis and deletion/duplication of 27 genes known to be associated with aortopathies, aneurysms, and connective tissue disorders	Negative
Exome, Proband-Only testing	Positive identification of pathogenic heterozygous CHEK2 gene variant c.417C>A (p.Tyr139*) which is associated with autosomal dominant predisposition to breast, colon, thyroid and prostate cancer.

Echocardiographic and Cardiac MRI images

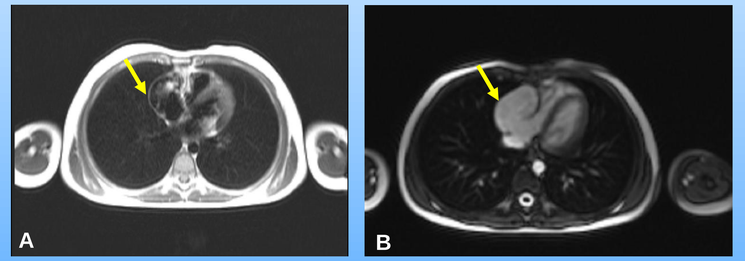
Transthoracic echocardiogram at 8 years of age



Description: Apical four chamber view

A) Shows an aneurysmatic dilation of the RAA with the base of the appendage measuring 33mm
B) Shows the RAA area of 12.8cm^2
C) Shows the RAA of 60mm height by 27mm width

Cardiac MRI at 8 years of age



Description: Axial view

A) Shows right atrium of 27x28mm (area 6.68cm^2 excluding RAA) and RAA of 46.07x27.81mm (yellow arrow)
B) Demonstrates absence of thrombus within the RAA (yellow arrow)

Discussion

We present a patient that was diagnosed as a fetus with RAAA and has had close monitoring since birth. RAAA presents as aneurysmal dilation with no focal pericardial defect. RAAA is most commonly diagnosed with echocardiogram during the third decade of life, and the most common symptom is palpitation caused by atrial tachyarrhythmia due to atrial dilation. However, patients can be diagnosed as a fetus or during the neonatal period. Occasionally, RAAA is an incidental finding after a thrombotic event or on a chest imaging study. Potentially fatal complications of RAAA include aneurysm rupture and sudden death. This is due to progressive thinning of the right atrial wall and is why our patient has periodic echocardiographic evaluation. Atrial tachyarrhythmia originating from RAAA is usually resistant to antiarrhythmic medication therapy, and radiofrequency catheter ablation is risky, with a low success rate and high recurrence rate. Surgical resection of RAAA is a safe, effective, and minimally-invasive option. Therefore, if atrial arrythmia or thrombotic symptoms ensued, we may be prompted to consider surgery. Our patient has only occasional mild symptoms at 9 years of age, and we will continue with close follow-up with periodic echocardiogram, electrocardiogram, and thrombosis monitoring.

Conclusions

- Treatment of patients with RAAA continues to be challenging as data available is scarce. Only 34 patients with RAAA have been reported in the literature.
- Asymptomatic RAAA should be managed conservatively and include anticoagulation therapy to prevent thrombus formation and close follow-up.
- Surgical intervention should be considered for those with symptoms or enlarging aneurysm.
- Further studies are needed to determine the best management approach for these patients.

References

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