Article published online: 2024-06-04

Prenatal Diagnosis of a Right Atrial Appendage Aneurysm: Case Report and Review of the Literature

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AJP Rep 2024;14:e170–e176.

Abstract

Introduction Congenital malformations of the right atrium are rare heart defects with only a few cases described prenatally. Early diagnosis of these anomalies is becoming increasingly important for proper follow-up and due to the possibility of serious complications such as supraventricular arrhythmia, thromboembolic events, and sudden death.

Objective The atrial appendage aneurysm (AAA) is a dilatation of the atrial appendage. It is considered an extremely rare congenital anomaly. However, this condition is clinically significant because it leads to atrial arrhythmias, recurrent emboli, heart failure, and chest pain. In addition, it is possible to recognize AAA prenatally with fetal echocardiography, even if it rarely happens. However, few fetal AAA cases have been reported in the literature.

Study Design We report a case of a fetal AAA; diagnosed prenatally and with postnatal confirmation. We undertook a systematic review of studies on fetal AAA to synthesize available knowledge on diagnosing and managing this rare condition.

Results A total of eight studies describing 24 patients were identified and analyzed.

Conclusion Despite their rarity, fetal atrial appendage aneurysms necessitate early detect on due to associated severe complications. Our findings emphasize the importance of prenatal diagnosis through fetal echocardiography and highlight the need for further research to optimize management strategies and improve outcomes for affected individuals.

Keywords
- fetal atrial appendage aneurysm
- fetal echocardiography
- prenatal diagnosis
- cardiac heart disease

Introduction

Right atrial appendage (RAA) aneurysm is considered an extremely rare congenital anomaly, consisting of abnormal dysplasia of the *musculi pectinati* of the atrial that would impair the contraction of the appendage during atrial systole.[1,2] The true frequency and the natural course of RAA aneurysm remain unclear.[2] It was first described by Parmley

received
January 26, 2024
accepted after revision
March 27, 2024

DOI https://doi.org/10.1055/s-0044-1787066.
ISSN 2157-6998.

Thieme Medical Publishers, Inc., 333 Seventh Avenue, 18th Floor, New York, NY 10001, USA

in 1962 in an 11-year-old boy with atrial arrhythmia and episodes of syncope.[3] It is found in all age groups including fetuses but most cases are diagnosed in the third or fourth decades of life.[2]

This anomaly is often revealed as an incidental finding during cardiac imaging, autopsy, or cardiac surgery performed for other reasons.[2] Acquired aneurysms are generally accompanied by atrial enlargement secondary to mitral valve disease that chronically increases atrial pressure or weakens the atrial wall, such as rheumatic mitral valve disease, tuberculosis, and syphilitic myocarditis.[4]

With advancing ultrasound technologies, cases of RAA aneurysm diagnosis in children are increasing. However, cases of prenatal diagnosis of this pathology still remain the exception.[2] Ultrasonographic signs of RAA are a large cystic structure linked to the atrium with blood flow between them that can be confused with an aneurysm of the left atrial wall, enlarged coronary sinus, bronchogenic cyst, mediastinal mass, and pericardial effusion.[5,6] Surgical treatment has been reported to have good outcomes, and it is recommended to manage RAA. However, because of the rarity of this entity, a direct comparison of medical treatment with anticoagulation and antiarrhythmic therapy is not available in the current literature.[7] Herein, we present a case of a prenatal diagnosis of a congenital atrial appendage aneurysm (AAA) and review the literature on this rare subject.

Materials and Methods

We performed a systematic search of literature indexed on PubMed, Scopus, Institute for Scientific Information Web of Science, and Cochrane, using a combination of keywords represented by "fetal," "atrial," and "appendage," and "aneurysm" (►**Appendix 1**). Two reviewers independently screened titles and abstracts of the records that were recuperated through the database searches. We also performed a manual search to include additional relevant articles using the reference lists of key articles. Full texts of records recommended by at least one reviewer were screened independently by the same two reviewers and assessed for inclusion in the systematic review. Disagreements between reviewers were solved by consensus. The systematic review was conducted and reported according to the Preferred Reporting Items for Systematic reviews and Meta-Analyses guidelines. Data selection and extraction were conducted following the Population, Intervention, Comparison, and Outcome study type using a piloted form specifically designed for capturing information on the study and its characteristics. Data were extracted independently by two authors to ensure accuracy and consistency.

Results

Case report

A 33-year-old primigravid patient was referred to the maternal-fetal medicine office at 22 weeks' gestation after a second-trimester anatomical examination after a mediastinal mass was noted on prenatal ultrasonography. The patient was asymptomatic and her medical history was not relevant. She denied exposure to toxins, smoking, or recreational drug use. At 25 weeks of gestation, a fetal echocardiogram demonstrated a large cystic structure adjacent to the right atrium and ventricle (►**Fig. 1**). She had a structurally normal heart and cardiac function, with suspicion of RAA aneurysm. However, there was no evidence of fetal heart failure or hydrops during ultrasound evaluation, the anatomy of the heart chambers, atrioventricular valves, and great arteries was normal without signs of intracardiac thrombus. Therefore, expectant surveillance and management were planned until the end of the pregnancy. The baby was born at term at 37 weeks and 3 days of gestation, by cesarean section due to arrest of dilation and descent, Apgar scores of 8/1 minute and 9/5 minutes, with admission to the intensive care unit for prenatal diagnostic surveillance, a color Doppler echocardiogram was performed that revealed a 12-mm thin-walled aneurysm, no thrombi were seen inside, no cardiac malformations, foramen ovale of 2 mm without repercussion, tricuspid with a 10-mm ring with mild tricuspid regurgitation, right ventricle of normal diameters, and normal biventricular systo-diastolic function.

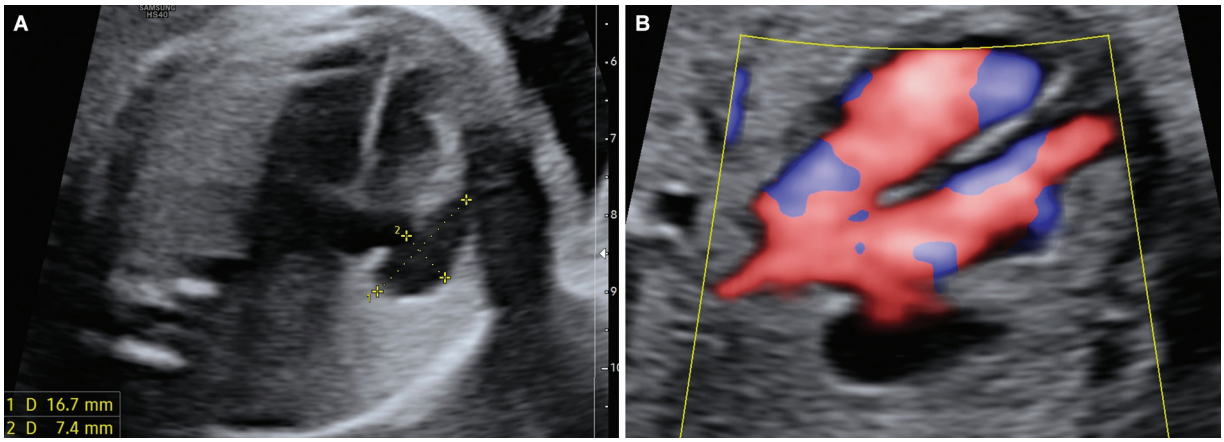

Fig. 1 Fetal ultrasound at 22 weeks' gestation. Right atrial appendage aneurysm (*) on different cardiac views. (**A**) Note the thin wall of the aneurysm (yellow lines) and the size of the abnormal chamber compared with the atria and ventricles. (**B**) Using color Doppler, the same cystic structure with blood flow is observed.

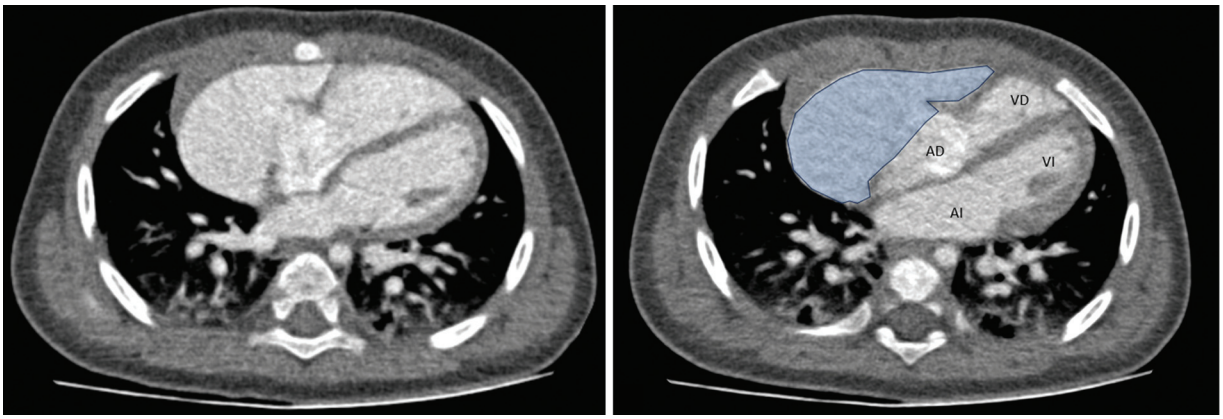

Fig. 2 Computed tomography angiography of the newborn. Axial scan. Right atrial appendage aneurysm (blue).

It is evaluated by a pediatric cardiologist who considers a patient with a diagnosis of right atrial aneurysm, stable without signs of heart failure, without evidence of cardiac anomaly, managed with acetylsalicylic acid. At 7 months of life, a computed tomography angiography of the coronary arteries was performed (►**Fig. 2**), which reported the right and left atrium of normal sizes, with an aneurysmal RAA, without masses or perfusion defects, in direct contact with the superior vena cava, without generating extrinsic compression of the latter, closed ductus arteriosus, without coarctation of the aorta, with which we can confirm the diagnosis of postnatal right atrial aneurysm. A three-dimensional reconstruction of the anterior chest wall is also performed (►**Fig. 3**) where normal cardiac structures are delimited as well as the aneurysm of the RAA.

Since then, he has been under observation by the cardiologist, remains asymptomatic, has not required surgery, or pharmacological treatment.

Systematic Review

The electronic database search provided a total of 37 results. After duplicate exclusion, there were 13 citations left. Two studies were excluded for languages other than English (Chinese). One paper was added through reference list searching. Finally, 12 studies were considered for full-text assessment. Hence, eight studies met the inclusion criteria and were incorporated into the systematic review.[8–15] All the included studies were case reports, and one retrospective study was published from 2009 to 2021, describing 24 patients. The main characteristics of these studies are listed in ►**Table 1**. Seven studies included were case reports, [8–14] and only one was a retrospective study.[15] The maternal age was reported only in four studies,[8,10,11,15] as shown in ►**Table 1**. The mean gestational age at diagnosis was 27.8 ± 4.3 weeks. Among all cases, 13 were RAA aneurysm and 11 were left AAA.

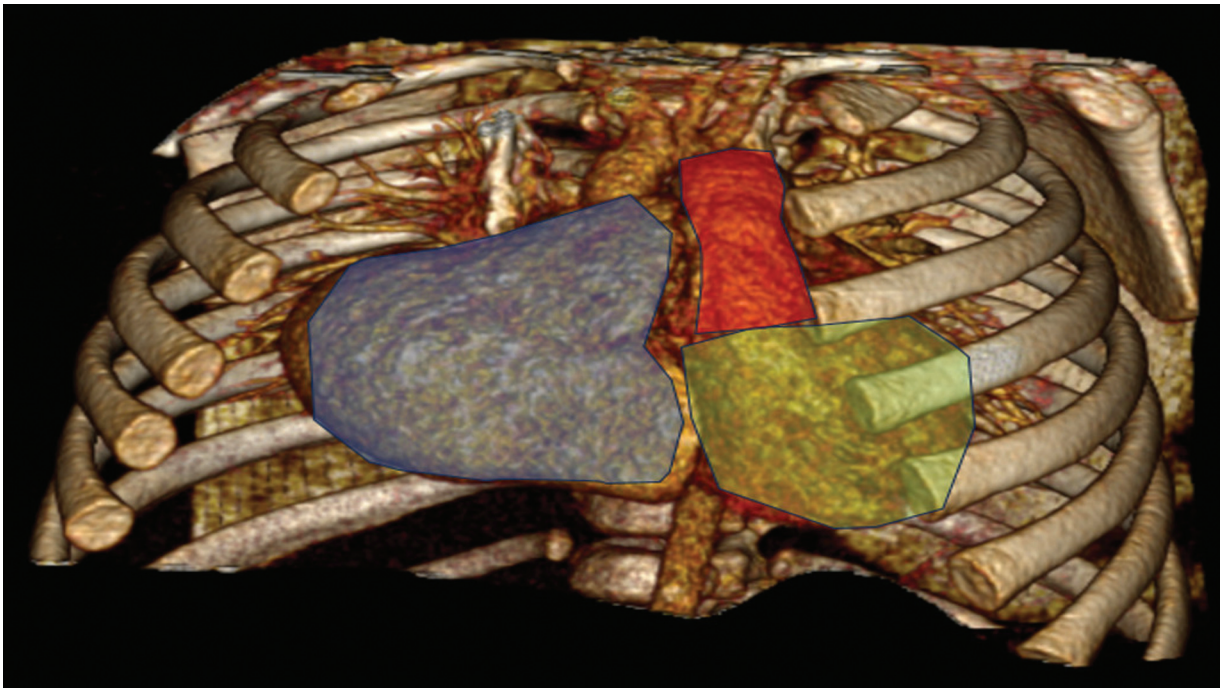

Fig. 3 Three-dimensional (3D) reconstruction: Anterior view of the thorax. We can see in blue: right appendage, green: right ventricle, red: pulmonary artery.

Table 1 List of publications included in the literature review, including clinical characteristics, sonographic characteristics of the atrial aneurysms, perinatal outcomes and postnatal management

Ref	First author	Year	Country	Study	Mothers age	Weeks	Side	Dimension mm	Fetal hydrops	Fetal heart failure	Delivery -weeks -mode of delivery	Weigh g	Postpartum findings	Postnatal management	Outcome
[8]	Su	2021	China	Case report	33	26	Left	20 × 9	No	No	-39 + 3 -Cesarean	3,000	-Patent ductus arteriosus (PDA) detected -No intracardiac thrombus identified	Elective surgery at 12 months	6 months postsurgery full recovered
[9]	Sasaki	2021	Japan	Case report	Not reported	31	Left	16 × 14	Not reported	Not reported	-38 -Not reported	2,500	-No sign of compression -No intracardiac thrombus identified	Elective surgery at 7 months	5 years postsurgery full recovered
[10]	Oztunc	2016	Turkey	Case report	30	20	Right	15 × 7.5	Yes	Pericardial effusion	Intrauterine fetal death accrued at 32 week	/	/	/	/
[11]	Ishii	2012	Japan	Case report	43	38	Right	34 × 17	No	No	-41 -Vaginal	3,038	No findings at cardiorespiratory examination after birth	Medical follow-up	Medical follow-up
[12]	Cho	2010	Korea	Case report	Not reported	28	Left	22 × 11	No	No	At term	- At term - Not reported	-Mild respiratory distress -Moderate to severe mitral regurgitation without structural abnormality -No intracardiac thrombus identified	Urgent surgery at 7 days	8 months postsurgery full recovered
[13]	Bornaun	2016	Turkey	Case report	Not reported	21	Right	Not reported	No	No	Not reported	Not reported	Asymptomatic	Medical follow-up	Medical follow-up
[14]	Tunks	2015	USA	Case report	Not reported	28	Right	Not reported	No	No	-36 -Vaginal	Not reported	Increase in the size of the aneurysm with evidence of right ventricular impingement	Elective surgery at 5 weeks	4 months postsurgery full recovered
[15]	Wang	2021	China	Case series (17)	30.0 ± 4.4	28.0 ± 3.7	8 Left 9 Right			-Cardiac enlargement ($n = 4$) -CHD ($n = 4$) -Extracardiac abnormalities ($n = 3$) -Fetal arrhythmia ($n = 1$)	-3 cases selectively terminated -14 cases born		-3 cases appendage aneurysm disappeared -11 cases aneurysm persist	-11 cases in follow-up: 1 surgery for concomitant atrial septal defect, 1 case of persistent atrial tachyarrhythmia	Follow-up

Abbreviation: CHD, cardiac heart disease.

Regarding the size of the aneurysm, due to the different gestational ages at diagnosis, it is not possible to report an average size at diagnosis.

In one case, the AAA was associated with fetal hydrops, and fetal death occurred at 32 weeks of gestation.[10] Other fetal findings were cardiac enlargement ($n = 4$), cardiac heart disease (CHD) ($n = 4$), extracardiac abnormalities ($n = 3$), and fetal arrhythmia ($n = 1$).[15] Three complex CHD cases were selectively terminated.[15] The gestational age and delivery route are extremely heterogeneous due to the small size of the sample and the variety of data collected. After birth, 18 newborns were asymptomatic,[8,9,11,13,15] one newborn had mild respiratory distress and moderate to severe mitral regurgitation without structural abnormality,[12] another one had an increase in the size of the aneurysm with evidence of right ventricular impingement,[14] In three cases, the aneurysm disappeared.[15]

Regarding postpartum management, four babies had an elective surgery with full recovery at follow-up,[8,9,14,15] one had an urgent surgery due to the respiratory distress, and moderate to severe mitral regurgitation described above,[14] 12 patients are still in medical or observational follow-up.[11,15]

Discussion

Congenital AAA is an extremely rare abnormality that may require urgent and life-saving interventions. Because of its rarity, we only have case reports and one retrospective study reported in the literature regarding fetal AAA. Only one study reported the detection rate of the prenatal diagnosis of AAA, which was 0.35%.[15] Timely prenatal diagnosis aids patient management including the place for birth, mode of delivery, and the appropriate medical care for the newborn. It is possible in the early second trimester; however, half of the right atrial aneurysm cases are detected during a third trimester scan.[10]

This malformation is associated with significant morbidity; diagnostic imaging such as echocardiography and magnetic resonance imaging play a fundamental role in diagnosis, management, and follow-up.[16] The treatment is controversial and there is still no established consensus, however surgical resection is preferred in patients with symptoms or progressive enlargement in order to prevent complications such as thromboembolism, atrial arrhythmia, heart failure, and sudden death due to rupture. Conservative management with anticoagulants reduces the risk of thromboembolic events and is recommended in patients for whom surgery is contraindicated.[16] The AAA should form part of the differential diagnoses in cases where the abnormal chamber is adjacent to the atrium.[7] In addition, another differential diagnosis should be made in the case of fetal hydrops. In the past, the most frequent cause of hydrops was due to Rh immunization, while recent data show that the most frequent cause of nonimmune hydrops is cardiac pathologies seen in 20 to 25% of cases.[17] The diagnosis is most frequently made during the third decade of life, of which two-fifths of the cases of AAA are congenital.[18–20]

Even if asymptomatic after birth, the AAA may grow over several years and become symptomatic with an increased risk of thromboembolism.[21] Thus, early surgical intervention is advised even in asymptomatic patients to prevent the occurrence of myocardial dysfunction, atrial fibrillation, and systemic embolism, as described above.[7,21]

Conclusion

To our knowledge, this is the first systematic review to focus on fetal AAAs. This is, of course, the strength of this work, along with the robust methodology. On the other hand, a weakness is due to the rarity of the condition. We relied mainly on case reports and only one retrospective study. In addition, we present a recent fetal AAA in which an accurate prenatal and early diagnosis was made, which was confirmed in postnatal life with angio-tomography. The prenatal diagnosis of fetal appendage aneurysm, due to its rarity, is difficult but essential for proper follow-up and to avoid catastrophic consequences and complications at birth.

Ethical Approval

This work has been approved by the Local Ethics Committee of the Women's Clinic, Cartagena, Colombia.

Authors' Contributions

J.M. contributed with ultrasound data, wrote the first draft of the manuscript, reviewed and analyzed the literature, and wrote the final version. N.A. contributed with postnatal images and analysis of the literature. Furthermore, D.V. contributed with ultrasound data, article writing, and data analysis and B.C. contributed with postnatal images and analysis of the literature. All authors reviewed and approved the final version of the manuscript.

Funding

None.

Conflict of Interest

None declared.

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Appendix 1 Search strategy

	Fetal atrial + appendage + aneurysm
#1	((Fetal atrial) in Any Field) and ((appendage) in Any Field) and ((aneurysm) in Any Field)
	Fetal + atrial + appendage + aneurysm
#2	((Fetal) in Any Field) and ((atrial) in Any Field) and ((appendage) in Any Field) and ((aneurysm) in Any Field)
	Fetal appendage + aneurysm
#3	((Fetal) in Any Field) and ((appendage) in Any Field) and ((aneurysm) in Any Field)

