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Case Report

Familial anomalous aortic origin of the right coronary artery: Implications for screening and clinical decision-making

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Abstract

Anomalous Aortic Origin of Coronary Arteries (AAOCA) occurs in approximately 0.64% of the population. It represents the second leading cause of Sudden Cardiac Death (SCD) in adolescents, following hypertrophic cardiomyopathy. The risk of SCD is estimated to be around 6.3% from an Anomalous Aortic Origin of the Left Coronary Artery (AAOLCA) and 0.2% from an anomalous Aortic Origin of the Right Coronary Artery (AAORCA).

Current strategies - including activity restriction, surgical intervention, and surveillance - are largely guided by expert consensus, with limited data to inform risk stratification. Consequently, optimal management of AAORCA remains controversial. This case series highlights this persistent uncertainty in clinical decision making for AAORCA management, particularly in the absence of ischemia on functional testing.

Additionally, this case provides the first documented example of phenotypically similar AAOCA in multiple first-degree relatives, strengthening the evidence for a potential genetic predisposition and supporting consideration of familial screening.

Case

Sibling A was initially evaluated at age 7 for episodes of non-exertional chest pain. At that time, a 12-lead Electrocardiogram (ECG) and echocardiogram were unremarkable – with no identified abnormalities in the origin or course of the coronary arteries. Nine years later, Sibling B presented at age 14, with exertional chest pain and dyspnea. Cardiac evaluation revealed a normal ECG, without electrocardiographic evidence of ischemia, and an echocardiogram demonstrated normal ventricular function but identified an Anomalous Aortic Origin of the Right Coronary Artery (AAORCA). This finding was confirmed with Computed Tomography Angiography (CTA), which also demonstrated a 12 mm intramural and intraarterial course (Figure 1), consistent with an acute takeoff angle [1]. Given this diagnosis, Sibling A - who was now asymptomatic and active in multiple competitive sports, including basketball and dance, underwent repeat cardiac evaluation that similarly revealed AAORCA. Both siblings underwent exercise stress testing with myocardial perfusion imaging which showed no evidence of inducible ischemia or regional wall motion abnormalities.

Both parents underwent cardiac evaluation with echocardiogram. The mother had an unremarkable echocardiogram. The father's echocardiogram was unable to adequately visualize the origins of the coronary arteries. He subsequently got a coronary CTA which identified the same anomaly as his two sons. The imaging additionally revealed 70-80% stenosis of the proximal right coronary artery; however, subsequent stress testing showed no evidence of inducible ischemia. There was no proximal stenosis noted on imaging of either of the siblings.

Whole exome sequencing was performed in Sibling A, with both parents and Sibling B included as comparators. Exome was negative for any known pathogenic mutation or variant of uncertain significance. A three-generation pedigree revealed several notable events on the paternal side, including an

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aunt who was stillborn, (attributed to a nuchal cord). Father had a paternal half-brother who drowned at 17 despite being a strong swimmer and another paternal half-brother who died at approximately 35 years of age following complications after being assaulted. Additionally, Father had a paternal half-sister whose son was stillborn, attributed to maternal fibroids. These half-siblings shared the same mother. The paternal grandfather died at 77 from a stroke. The mother also had an uncle who died age 45 from a myocardial infarction. While these events are collectively of uncertain etiology, the clustering of premature or unexplained deaths, depicted in the pedigree below (Figure 2), raises the possibility of an underlying inherited predisposition.

At the time of referral, Sibling A, Sibling B and their father were all asymptomatic without prior intervention. The father was beyond the age range associated with the highest reported risk of SCD [2]. Through shared decision-making, including detailed counseling about the risk and benefits of surgical intervention, activity restriction, and conservative surveillance, the family elected for conservative management with yearly monitoring. Both siblings elected to remain active in competitive athletics.

Table 1: Summary of findings of father and two siblings, showing near identical clinical and anatomical features.

Name	Father	Sibling A	Sibling B
Age at Discovery	47	16	14
Diagnosis	AAORCA from the left sinus of Valsalva	AAORCA from the left sinus of Valsalva	AAORCA from the left Sinus of Valsalva
Symptoms at time of discovery (+/-)	-	+	-
Results of Stress Test	-	-	-
Acute Take off Angle	+	+	+
Intramural Course	12 mm	12 mm	12 mm

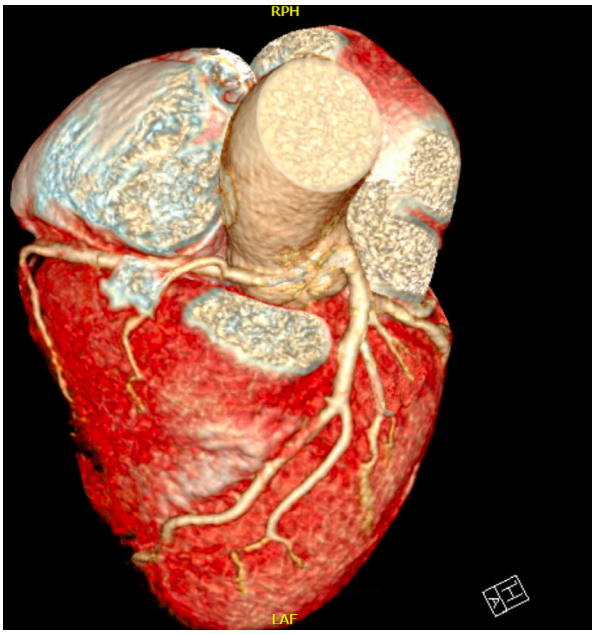

Figure 1: 3D reconstruction from CTA demonstrating the ostia of right and left main coronary arteries are closely spaced with R coronary arising just left ward of the right/left commissure. There is an approximately 12 mm intramural and intraarterial course.

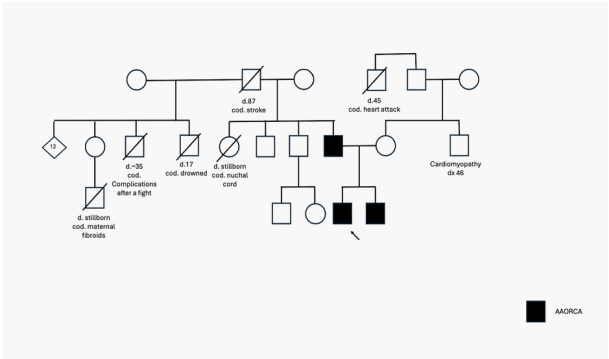

Figure 2: A pedigree depicting the familial cluster of phenotypically similar AAORCA as well as multiple close relatives who passed at a young age as well as another with a history of heart disease. The arrow indicates Sibling A.

Discussion/Conclusion

This case underscores the persistent challenges in the management of AAORCA, particularly in patients with minimal or no symptoms and non-ischemic functional testing. In such scenarios, contemporary expert recommendations remain inconsistent. All three individuals in this series demonstrated AAORCA with high-risk anatomic features, including an acute takeoff angle inferred from the intramural course length. Proposed management strategies in similar patients range from surgical approaches vbbb - such as coronary translocation, reimplantation, unroofing or osteoplasty - to conservative management [1]. The current American Heart Association (AHA) and American College of Cardiology (ACC) guidelines regarding activity eligibility suggest that no intervention or restriction may be necessary in the absence of ischemia [3].

A central issue highlighted by this case is the absence of standardized, evidence-based criteria for risk stratification in AAORCA. Functional testing, including exercise stress imaging, has limited sensitivity for detecting ischemia in this population - diminishing its utility in guiding clinical decision-making, and so recommendations to patients are typically based on anatomic features alone [2]. As a result, management often relies predominantly on anatomic features, such as length of intramural course or diameter and morphology of ostia, yet validated thresholds of these high-risk features are lacking. This gap creates substantial uncertainty for both patients and providers with a difficult decision to make. In the present case, despite reassuring functional evaluation and absence of exertional symptoms, the presence of high-risk anatomic features raises concern that SCD could represent the initial cardiac manifestation, underscoring the limitations of current risk assessment paradigms.

No definitive genetic determinants of AAOCA have yet to be identified [4]. However, as genomic technologies such as whole genome and whole exome sequencing become increasingly accessible, the potential for uncovering heritable contributions to this condition is likely to expand. To our knowledge, this case represents the first documented example of phenotypically similar AAOCA in multiple first-degree relatives, providing compelling support for a possible genetic predisposition [5-

9]. While causality cannot be established, these findings strengthen the rational for further investigation into the genetic basis of AAOCA. Continued systematic genomic evaluation and aggregation of familial cases may ultimately enable more precise risk stratification and inform screening strategies in affected families.

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