

Anomalous Aortic Origin of a Coronary Artery With an Interarterial Course

Should Family Screening Be Routine?

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- Objectives** We sought to present cases of familial occurrence of anomalous aortic origin of a coronary artery with an interarterial course (AAOCA) to determine if it would alter our current screening and management recommendations.
- Background** Anomalous aortic origin of a coronary artery with an interarterial course is a rare congenital anomaly that carries an increased risk of sudden death in children and young adults. There are no reports in the literature of familial AAOCA in the pediatric population.
- Methods** In preparation for a multi-institutional prospective study evaluating patient management and surgical outcomes in children and young adults with AAOCA, a questionnaire was sent to multiple pediatric institutions in North and South America. Several respondents indicated caring for families with more than 1 member with AAOCA. These patients were identified and charts were retrospectively reviewed.
- Results** We identified 5 families in which a child was diagnosed with AAOCA and another family member was subsequently identified through screening with echocardiography. The odds of this occurring are significantly greater than what would be expected by chance. All identified by screening were asymptomatic and had anomalous right coronary artery despite 2 of the 5 index cases having anomalous left coronary artery.
- Conclusions** It is possible that there is a genetic link for AAOCA. Future research into this is warranted. Due to the potential risk of myocardial ischemia and sudden death associated with AAOCA, screening first-degree relatives for AAOCA using transthoracic echocardiography would be the prudent approach to potentially prevent a sudden catastrophic event. (J Am Coll Cardiol 2008;51:2062-4) © 2008 by the American College of Cardiology Foundation

Anomalous aortic origin of a coronary artery with an interarterial course (AAOCA) is a rare congenital anomaly consisting of the left main coronary arising from the right sinus of Valsalva (ALCA) or the right arising from the left sinus (ARCA). It is associated with an increased risk of sudden death, notably in otherwise healthy children and young adults (1–6). The diagnosis can be challenging because patients are often asymptomatic (7–10) and have a normal physical examination. The diagnosis is usually made by transthoracic echocardiography (TTE) (9). Prevalence estimates in the general population range from 0.1% to 0.3% (7,8,11).

The authors are unaware of any reports in the literature of familial AAOCA in the pediatric population. We now present our findings from 5 families in which a child or young adult was diagnosed with AAOCA and another family member was subsequently identified through screening TTE.

Methods

As preparation for a multi-institutional prospective study evaluating patient management and surgical outcomes in children and young adults diagnosed with AAOCA between 1998 and 2007, a questionnaire was sent to partici-

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pating Congenital Heart Surgeon Society (CHSS) institutions for cardiologists, cardiothoracic surgeons, and nurse practitioners to complete. The CHSS is a consortium of approximately 85 surgeons from 60 university-based hospi-

tals in the U.S., Canada, and South America. We received responses from several members stating that they were aware of families in which more than 1 person had been diagnosed with AAOCA. Once these families were identified, each patient's chart was retrospectively reviewed.

Results

Family I. A 15-year-old African-American male was playing basketball when he collapsed and was unable to be resuscitated. It is unclear if he had symptoms prior to death. The autopsy found ARCA. At his uncle's request, the patient's first cousins were screened with TTE and his 5-year-old female cousin was diagnosed with ARCA. She was asymptomatic. She underwent the unroofing procedure and has done well 4 years post-operatively.

Family II. A 30-year-old Caucasian woman presented to her primary care physician with nonspecific chest pain. Cardiac catheterization revealed ARCA. It was recommended she take oral beta-blockers but she has been noncompliant due to side effects. Her 1-year-old daughter had TTE and was found to have ARCA. She is asymptomatic and has had no interventions to date.

Family III. A 32-month-old asymptomatic Caucasian male was evaluated for a murmur. A TTE suspected ARCA and transesophageal echocardiography confirmed the diagnosis. The TTE performed on his asymptomatic 6-month-old brother showed ARCA. Neither child has had any interventions to date.

Family IV. A 6-year-old Caucasian male collapsed after running. He was unable to be resuscitated despite emergency measures. Autopsy revealed ALCA. The child's siblings were screened with TTE and the 2-year-old asymptomatic brother was diagnosed with ARCA. He has not had any interventions to date.

Family V. A 10-year-old African-American male collapsed while playing basketball. He was resuscitated with extracorporeal membrane oxygenation and subsequently diagnosed with ALCA. He underwent coronary artery stenting but ultimately succumbed to intracerebral hemorrhage. His siblings had screening TTE and his 7-year-old brother was diagnosed with ARCA. He has not had any interventions to date.

Discussion

We present the first case series of familial AAOCA in the pediatric population. The odds of having 5 families with this rare anomaly are significantly greater than what would be expected to occur by chance. All unexpected cases were infants and young children with ARCA despite 2 of the 5 index cases having ALCA. This finding is not surprising given that ARCA occurs more frequently than ALCA (11) and is often found serendipitously (7-10). All cases identified through TTE screening were asymptomatic with all but 1 remaining free of medical or surgical intervention. Extensive literature search revealed only 1 other case report of

familial clustering of AAOCA but this was in 2 adults (12). Since screening relatives has not been routine, the actual number of familial AAOCA may be significantly greater than the 5 families of which we are aware.

Our findings raise the question of a genetic link with AAOCA. There are several known genetic causes of sudden death in the young for which screening first-degree relatives is recommended to identify other family members at risk for sudden death (13). There has been limited research examining the inheritance pattern of congenital coronary anomalies. Angiographic studies have suggested genetic differences between races and ethnicities as the reason for finding a larger number of 1 coronary anomaly in certain populations compared to others (14,15). Indeed, the only way to develop a genetic pedigree for AAOCA is to have multiple family members from more than 1 generation tested.

As the second leading cardiovascular cause of sudden death in young athletes (16), it is not unreasonable to recommend TTE screening for family members as part of a complete evaluation in selected populations. The identification of patients with AAOCA by history and physical examination alone is nearly impossible since these individuals are often asymptomatic (7-10) and typically have normal examinations and electrocardiograms. Therefore, TTE would be necessary to adequately screen for this anomaly. In those identified with AAOCA, we would repair those with ALCA but treatment of asymptomatic ARCA patients remains controversial (17). Presently, screening first-degree relatives of children and young adults using TTE seems to be the prudent approach to potentially identify these defects and possibly prevent sudden catastrophic events.

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Abbreviations and Acronyms

AAOCA = anomalous aortic origin of a coronary artery with an interarterial course

ALCA = anomalous origin of left main coronary artery

ARCA = anomalous origin of right coronary artery

CHSS = Congenital Heart Surgeon Society

TTE = transthoracic echocardiography

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