Pediatric Sudden Cardiac Death

Sep 9th, 2023

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OUTLINE

- Epidemiology
- Pre-participation Physicals
- Causes of Sudden Cardiac Death

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- Causes of Sudden Cardiac Death

Epidemiology

The incidence of SCA in the young is difficult to ascertain:

- No mandatory reporting system for juvenile sudden death

- Most data for pediatric SCD are collected postmortem, such that many cases of SCA or aborted SCD are often not reported

~ 0.5 to 20 per 100,000 person-years

Meyer et al, Circulation. 2012;126:1363-1372

Epidemiology

- ~ 2000 patients younger than 25 years will die from a sudden cardiac event each year in the United States

- The incidence is higher in athletes, with a relative risk of sudden death of 2.1 compared with nonathletes

Pediatric Emergency Care • Volume 32, Number 9, September 2016

Epidemiology

- The prevalence of sudden unexpected death, in general, and SCD, in particular, is age dependent:

- Higher risk in early infancy (SIDS)
- Falls in early childhood
- Rise again in adolescence

Sudden Cardiac Death in the Young. Ackerman et al. Circulation. 2016;133:1006-1026

Epidemiology

One large study identifying 114 cases of SCD in children ages 1 to 18 in Denmark

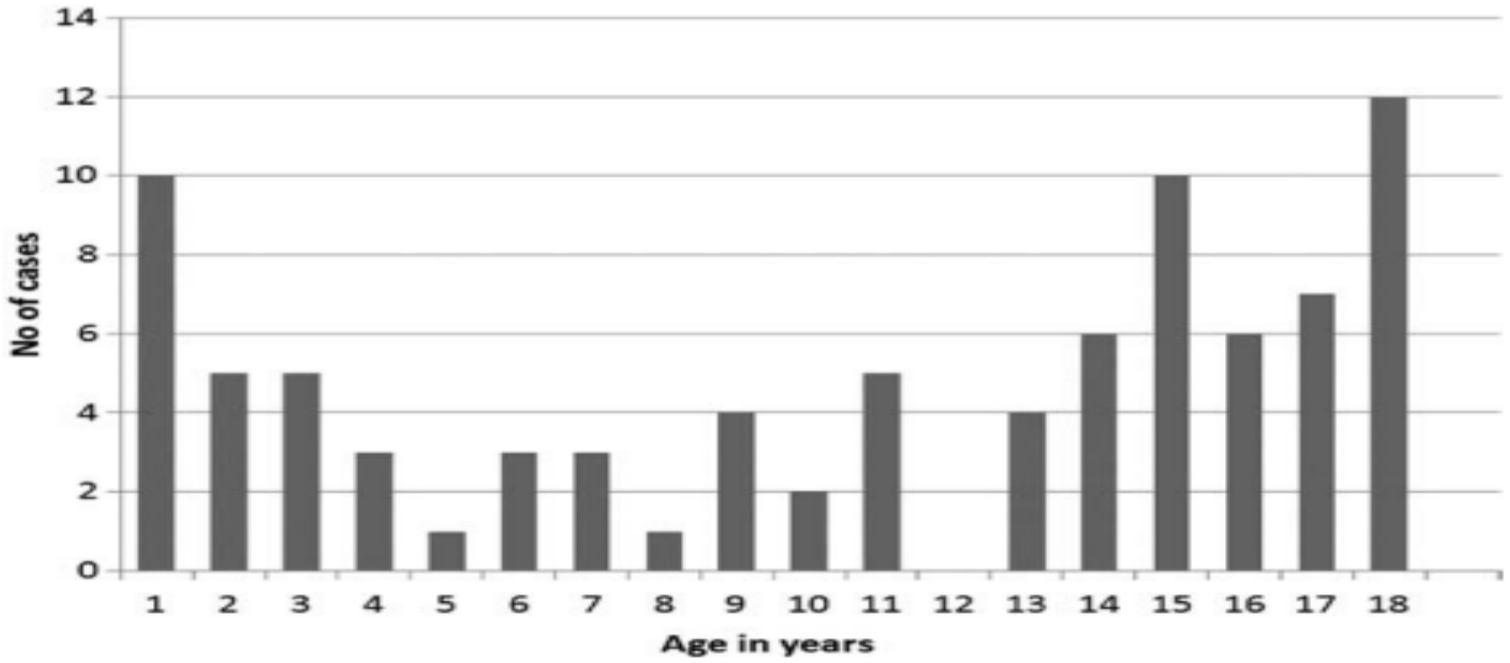

Figure 2. Age distribution of SCD in the Danish population, 2000 to 2006. SCD indicates sudden cardiac death. Reprinted from Winkel et al[33] with permission of the publisher. Copyright © 2014, European Society of Cardiology.

Sudden Cardiac Death in the Young. Ackerman et al. *Circulation*. 2016;133:1006-1026

Epidemiology

- Sudden cardiac death (SCD) is statistically uncommon in the young, but its dramatic presentation and cascading effects in the family and community make it a newsworthy event

- High degree of concern regarding the risk of SCD in children and teenagers among the lay public, in comparison with other much more prevalent deadly risks of childhood, such as accident, injury, suicide, and violence

Sudden Cardiac Death in the Young. Ackerman et al. Circulation. 2016;133:1006-1026

Epidemiology

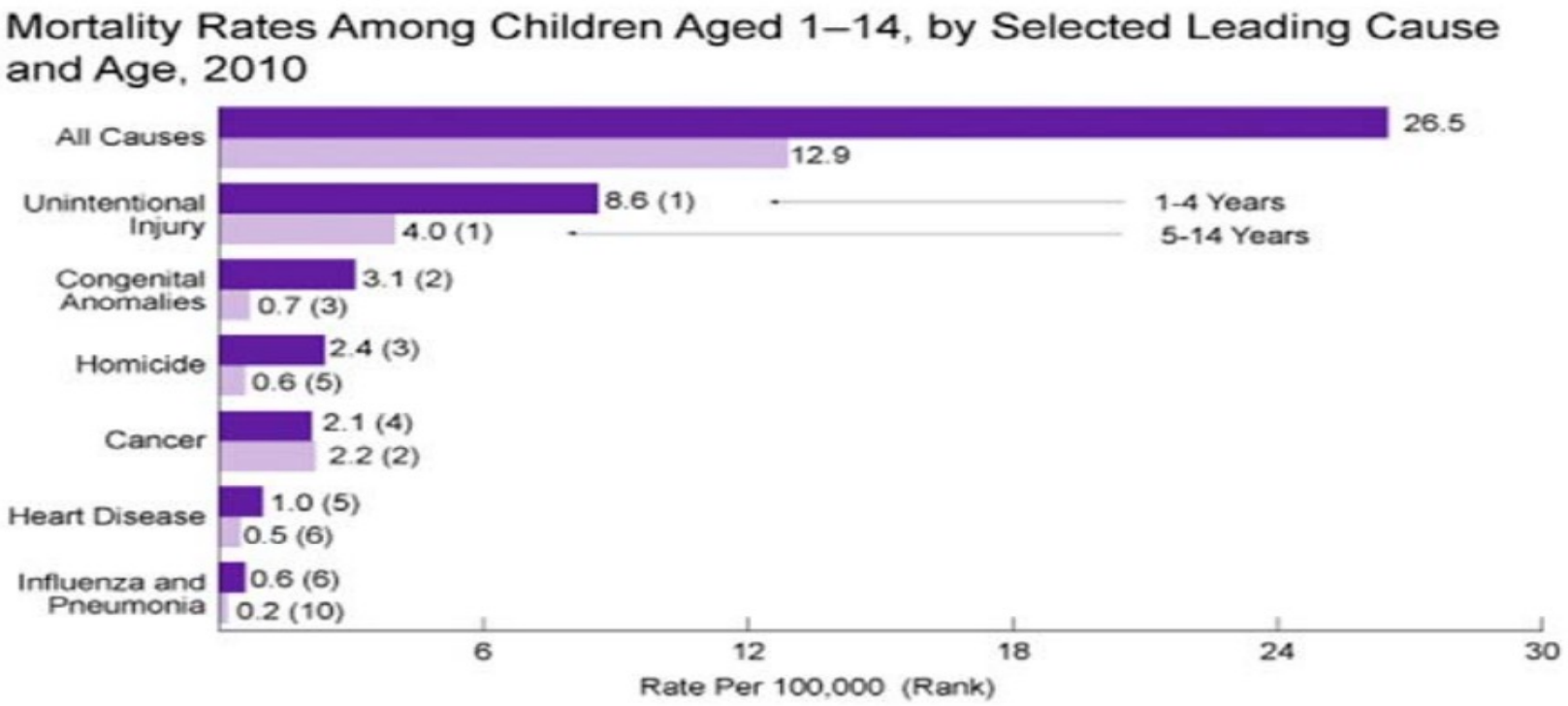

Figure 1. Causes of death in American children. Mortality rates among children 1 to 14 years of age, by selected leading cause and age, 2010.

Sudden Cardiac Death in the Young. Ackerman et al. (Circulation. 2016;133:1006-1026)

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Epidemiology

Circulation
JOURNAL OF THE AMERICAN HEART ASSOCIATION

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Sudden Deaths in Young Competitive Athletes : Analysis of 1866 Deaths in the United States, 1980 –2006

Barry J. Maron, Joseph J. Doerer, Tammy S. Haas, David M. Tierney and Frederick O. Mueller

Circulation 2009, 119:1085-1092: originally published online February 16, 2009
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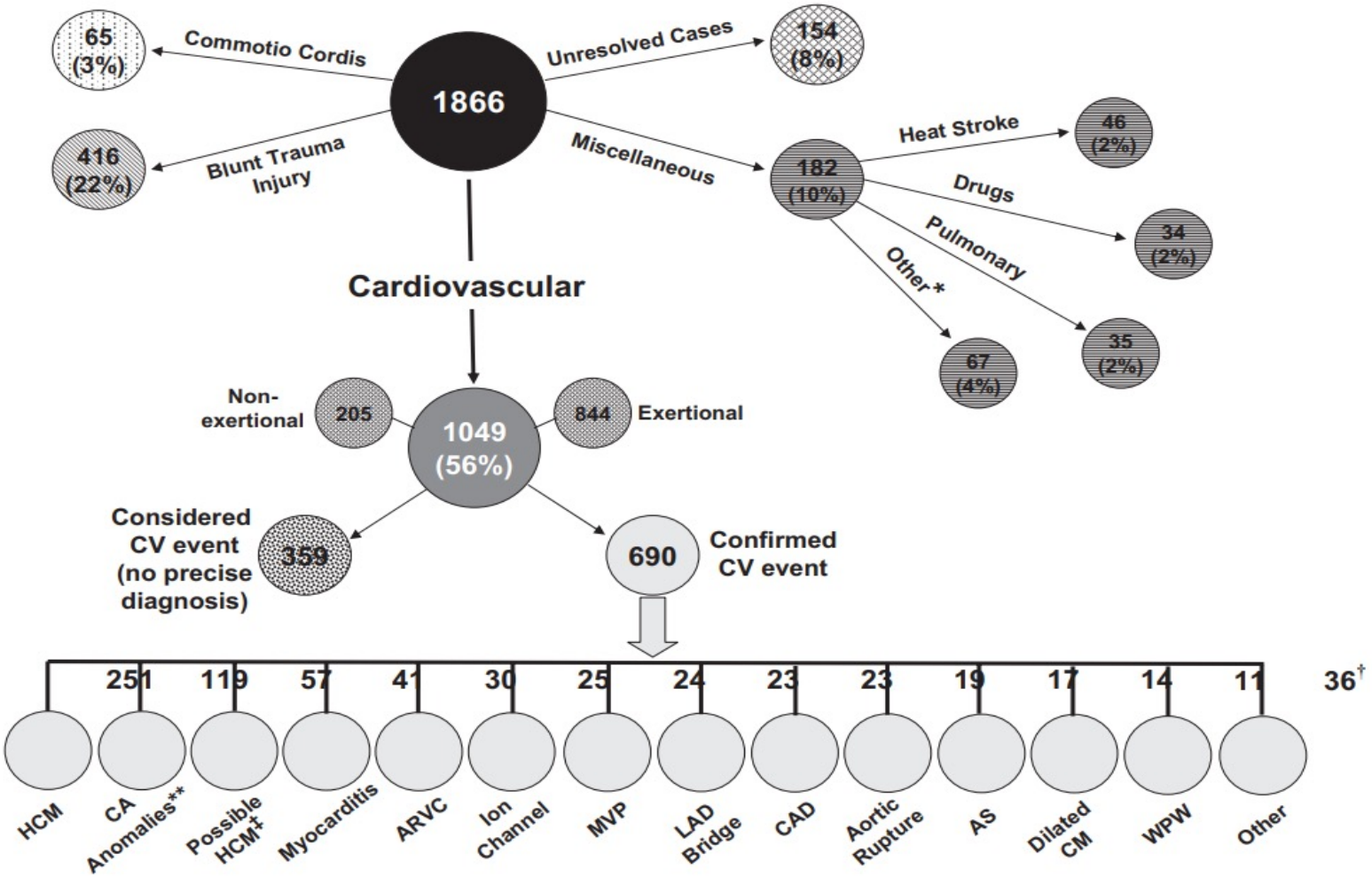
Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology

- To estimate the absolute number of sudden deaths in US competitive athletes, they assembled a large registry over a 27-year period
- A total of 1866 athletes who died suddenly (or survived cardiac arrest), were identified throughout the United States from 1980 to 2006 in 38 diverse sports
- Sudden deaths were predominantly due to cardiovascular disease (1049 [56%]), but causes also included blunt trauma that caused structural damage (416 [22%]), commotio cordis (65 [3%]), and heat stroke (46 [2%])
- The most common cardiovascular causes were hypertrophic cardiomyopathy (36%) and congenital coronary artery anomalies (17%)

Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology



Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

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Epidemiology

❖ **Demographics - Age and Gender:**

- These athletes ranged in age from 8 to 39 years at the time of death or cardiac arrest, mean 18±5 years
- 677 (65%) were 17 years old, 300 (29%) were 18 to 25 years old, and 72 (7%) were ≥ 26 years old
- A total of 937 athletes were male (89%), and only 112 were female (11%)
- The proportion of cardiovascular deaths reported in female athletes has increased over time (P0.023; 95% CI 1.05 to 1.92), reaching 12% in 2000 to 2006.

Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology

❖ **Demographics - Race:**

- Deaths due to cardiovascular disease were more common in nonwhite than white athletes (64% versus 51%, P0.001)

- White and nonwhite athletes did not differ significantly with respect to age (18±5 versus 18±4 years, P0.4) or gender (87% versus 93% males, P0.5)

Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology

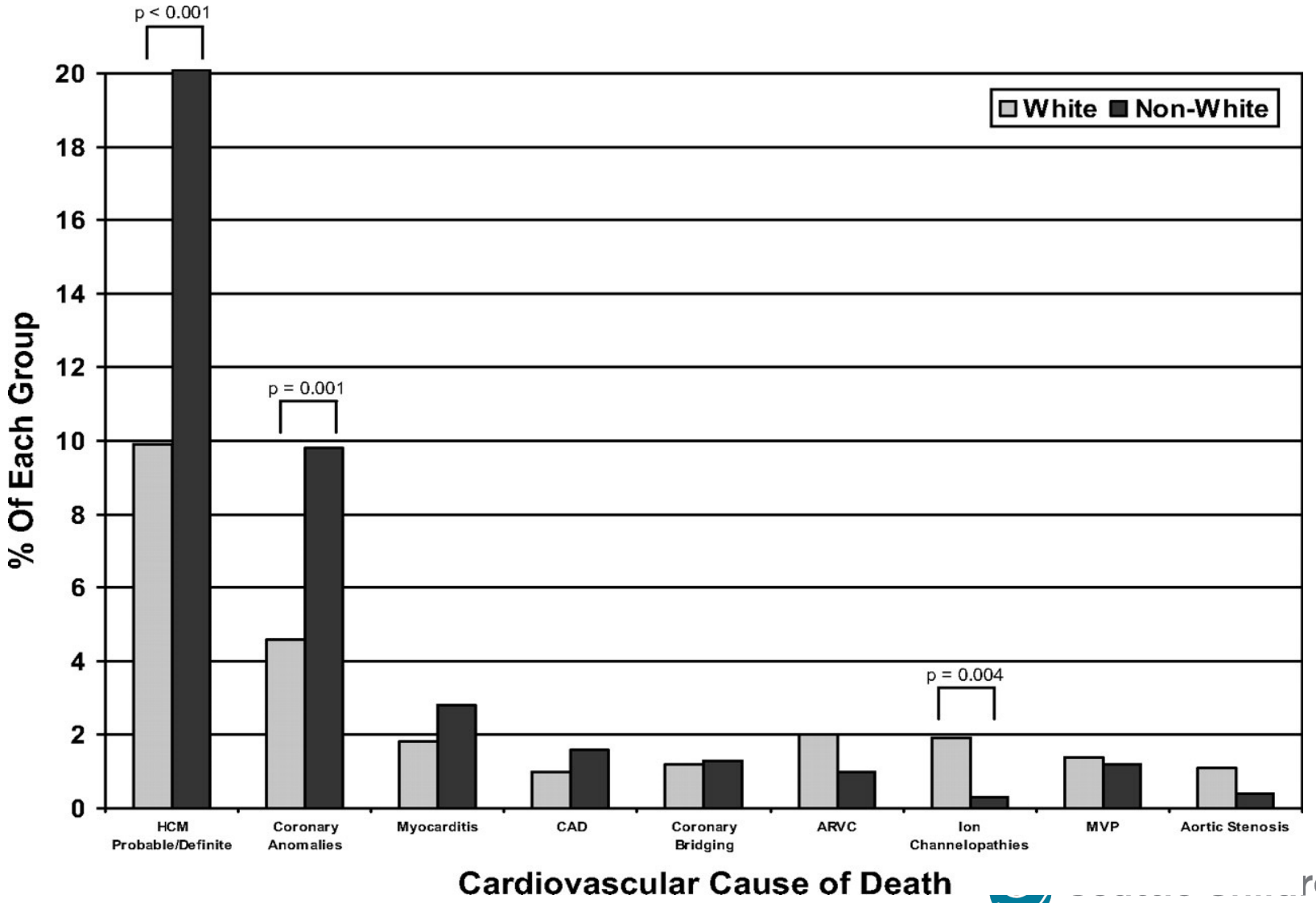
❖ **Demographics – Race, Cont.:**

- The fraction of reported deaths attributable to hypertrophic cardiomyopathy and congenital coronary anomalies was higher among nonwhites (predominantly blacks) than whites: 136/676 (20%) versus 112/1135 (10%; P0.001) for hypertrophic cardiomyopathy and 66/676 (10%) versus 52/1135 (5%; P0.001) for coronary anomalies

- Conversely, the fraction of reported deaths attributable to ion channelopathies was higher among whites than nonwhites: 22/1135 (2%) versus 2/676 (0.3%, P0.004).

Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology



Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology

❖ **Demographics – Sports and Level of Participation:**

- Athletes participated in a wide variety of 38 competitive sports, most commonly basketball (n349 [33%]) and football (n281 [25%])

- Most athletes who died of cardiovascular-related causes were engaged in competitive high school (n623 [59%]), middle school (120 [11%]), or youth (26 [2%]) sports

- Seventy-two athletes (7%) were considered elite by virtue of achieving professional status or a national level of excellence in amateur sports.

Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

Epidemiology

❖ **Demographics – Circumstances:**

- Sudden cardiac death occurred most commonly during or just after physical exertion, while the athlete was engaged in practice sessions, organized competition, or other sports activities (844 [80%])

- Another 205 trained athletes (20%) died suddenly in circumstances unassociated with sports, during routine daily activities or while sedentary or asleep

- In 16 athletes, sudden death events occurred while submerged in water (ie, swimming pool, lake, or ocean)

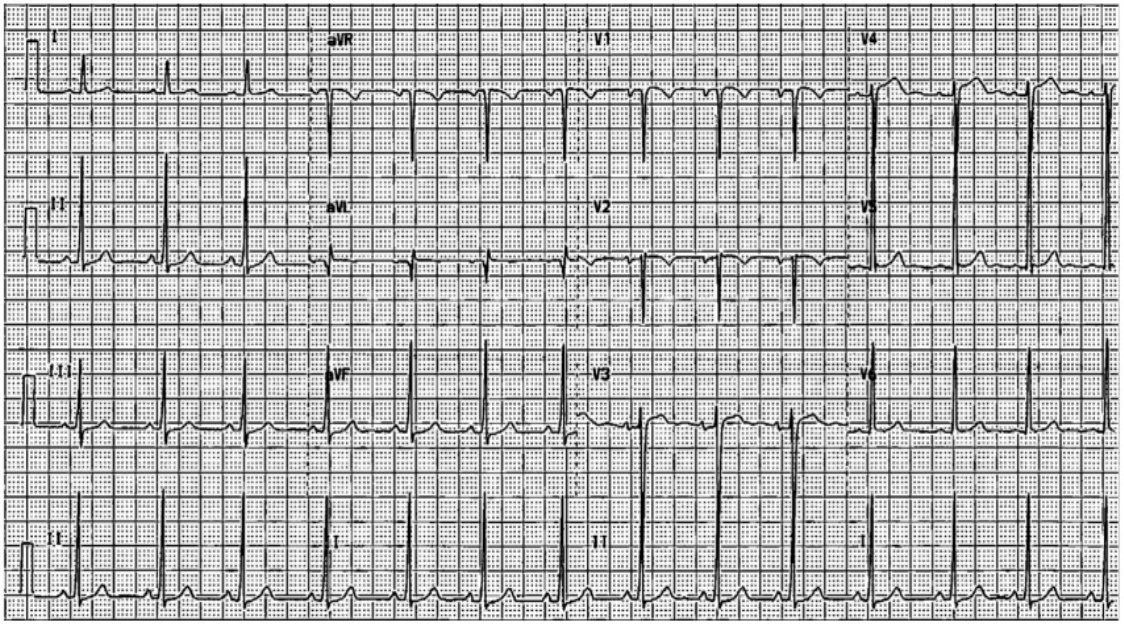
Sudden Deaths in Young Competitive Athletes. Maron et al. Circulation 2009, 119:1085-1092

OUTLINE

- Epidemiology
- Pre-participation Physicals
- Causes of Sudden Cardiac Death

Pre-Participation Physicals
Case 1

- An 11-year-old boy presents for sports clearance to play high school basketball. He has a family medical history of hypertrophic cardiomyopathy (HCM) in his father. His father has not had genetic testing. What do you do next?



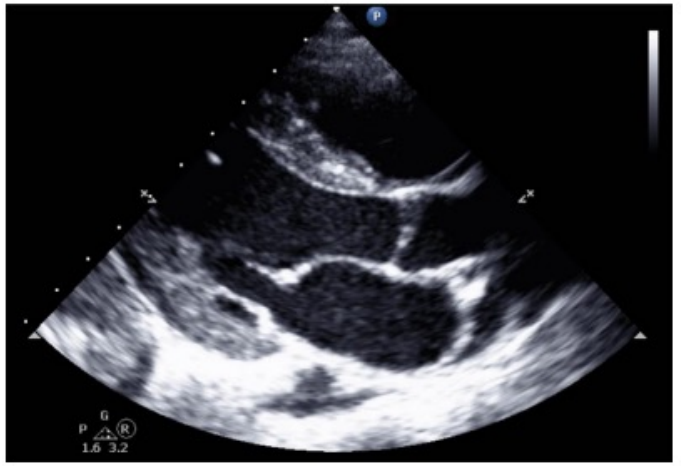


Fig. 2. Initial echocardiogram of an 11-year-old boy from Case 1 with a family history of HCM. This is a parasternal long axis image in 2D showing normal left ventricular and interventricular septal dimensions.

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Pre-Participation Physicals
Case 2

- A 10-year-old boy presents for clearance before youth league soccer. When asked about a history of syncope or unresponsive spells, his mother reports he had a recent syncopal episode. She reports that he was running to the school bus with his siblings when she noticed that he started to lag behind his siblings, fell to his knees, screamed, and then fell on his face. He was noted to be unconscious for a period of about 15 seconds and did not require any resuscitative measures. He was noted to have significant abrasions on his face. When he recovered, he apologized for the fact that “his legs stopped working.” His mother recounts that 2 months earlier she received a call from school after he fell while running on the track during gym class. This event was also associated facial abrasions. His mother attributed the inability to catch himself during his fall to prevent facial injury to what she described as general clumsiness and his history of mild gross motor delay. What do you do next?

Reittinger et al. Cardiol Clin 41 (2023) 1–14

Pre-Participation Physicals

POLICY STATEMENT Organizational Principles to Guide and Define the Child Health Care System and/or Improve the Health of all Children

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

Sudden Death in the Young: Information for the Primary Care Provider

Christopher C. Erickson, MD, FAAP,[a,b] Jack C. Salerno, MD,[c] Stuart Berger, MD, FAAP,[d] Robert Campbell, MD, FAAP,[e] Bryan Cannon, MD,[f] James Christiansen, MD,[c] Kody Moffatt, MD, MS, FAAP,[a] Andreas Pflaumer, MD,[g] Christopher S. Snyder, MD, FAAP,[h] Chandra Srinivasan, MD,[i] Santiago O. Valdes, MD, FAAP,[j] Victoria L. Vetter, MD, MPH, FAAP,[k] Frank Zimmerman, MD,[l] SECTION ON CARDIOLOGY AND CARDIAC SURGERY, PEDIATRIC AND CONGENITAL ELECTROPHYSIOLOGY SOCIETY (PACES) TASK FORCE ON PREVENTION OF SUDDEN DEATH IN THE YOUNG

Pre-Participation Physicals

The Sports Preparticipation Evaluation

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*Departments of Orthopaedics and Pediatrics, UT Southwestern, and Texas Scottish Rite Hospital for Children Sports Medicine Center, Dallas, TX

†Departments of Pediatrics and Orthopaedics/Rehabilitation, University of Iowa, Iowa City, IA

Preparticipation Cardiac Evaluation from the Pediatric Perspective

Andrew M. Reittinger, MD[a], Lanier B. Jackson, MD[b], Peter N. Dean, MD[a,*]

Pre-Participation Physicals

1. WHAT IS THE PURPOSE OF PREPARTICIPATION EVALUATION?
2. WHO SHOULD BE EVALUATED AND WHEN SHOULD IT START?
3. WHERE DO PREPARTICIPATION EVALUATIONS OCCUR?
4. WHAT ARE THE COMPONENTS OF THE PREPARTICIPATION EVALUATION?

Pre-Participation Physicals

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Pre-Participation Physicals

1. **WHAT IS THE PURPOSE OF PREPARTICIPATION EVALUATION?**

- To identify individuals who may be at risk for adverse health effects such as illness or injury secondary to sports participation

- To catch underlying cardiac diseases that predispose individuals to SCA and SCD

Pre-Participation Physicals

Table 1
Possible differences between pediatric and young adult/adult preparticipation cardiac screening

Pediatric Specific Screening	Young Adult and Adult Screening
More likely to have vague or unclear symptoms	Better able to communicate symptoms
Likely requires repeat screening as adolescent and/or young adult	More likely to have already expressed the phenotype of specific cardiac disease
Parents too young to demonstrate phenotype	Older parents with more detail about family history
Pediatric providers are less experienced in ECGs	Family physicians and adult providers more experienced interpreting ECGs
More likely to have undiagnosed congenital heart disease	Less likely to have undiagnosed congenital heart disease
More likely to have history of Kawasaki disease	
Suboptimal criteria for ECG abnormalities	

Reittinger et al. Cardiol Clin 41 (2023) 1–14

Pre-Participation Physicals

- Need to stress to patients and families:

- A “normal cardiac screen” means that there is no evidence of cardiac disease at that specific point in time

- New cardiac symptoms or concerns should be reevaluated

Pre-Participation Physicals

- The rate of SCD in 11,168 adolescent soccer players was still 6.8 per 100,000 despite extensive cardiac screening (history, examination, ECG, and echocardiogram) before participation
- The reason for the relatively high number of SCD was related to cardiomyopathies that were not detected by screening
- The mean age of screening was 16.4 ± 1.2 years, and the time between the screening and the episode of SCD was 6.8 years

Malhotra A, Dhutia H, Finocchiaro G, et al. Outcomes of cardiac screening in adolescent soccer players. N Engl J Med 2018;379(6):524–34.

Pre-Participation Physicals

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Pre-Participation Physicals

2. WHO SHOULD BE EVALUATED AND WHEN SHOULD IT START?

American Academy of Pediatrics (AAP) policy statement on the topic of screening for sudden death:

- Recommend screening patients every 2- 3 years without any differentiation between athlete and nonathlete

- It also suggests that screening should be performed starting at age six

Miller SM, Peterson AR. The sports preparticipation evaluation practice gaps. Available at: http://pedsinreview.aappublications.org

Pre-Participation Physicals

2. WHO SHOULD BE EVALUATED AND WHEN SHOULD IT START?

- Athletes should be encouraged to complete their PPE at least 6 weeks before the first practice

- The beginning of summer or near the end of the previous school year is an ideal time

- A comprehensive PPE with a complete personal and family history and thorough physical examination should be performed at least every 2 years, with an annual review of the patient's history and a problem-focused examination as needed

Miller SM, Peterson AR. The sports preparticipation evaluation practice gaps. Pedsinreview. 2019

Pre-Participation Physicals

2. WHO SHOULD BE EVALUATED AND WHEN SHOULD IT START?

- Providers need to ask questions about physical activity, cardiac symptoms with exertion, and family history regardless of whether there is an official “school preparticipation form” to sign

Miller SM, Peterson AR. The sports preparticipation evaluation practice gaps. Available at: http://pedsinreview.aappublications.org

Pre-Participation Physicals

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Pre-Participation Physicals

3. WHERE DO PREPARTICIPATION EVALUATIONS OCCUR?

- Ideally, these visits would occur with the patient's primary care provider, but there are other settings where evaluations occur, including mass screening events or urgent care offices

- The preferred provider is the patient's primary care provider
  - This allows for centralization of a patient's care, access to patient's past medical records, better understanding of the patient's medical, family, and social histories, and the ability to have consistent longitudinal follow-up

  - It also provides appropriate follow-up of referrals to ensure they are performed, and recommendations are followed

Miller SM, Peterson AR. The sports preparticipation evaluation practice gaps. Available at: http://pedsinreview.aappublications.org

Pre-Participation Physicals

- **Mass screening events:** time efficient and allow for improved access to medical providers, but:

- Difficulty maintaining patient privacy and a safe space for patient to bring concerns to physician
- Suboptimal environment for physical examination
- Lack of access to patient's medical records
- Lack of time and space for appropriate counseling when an abnormality or suspected abnormality is found
- Difficulty ensuring appropriate follow-up/referrals when abnormalities are found.

Pre-Participation Physicals

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Pre-Participation Physicals

❖ **WHAT ARE THE COMPONENTS OF THE PREPARTICIPATION EVALUATION?**

- Typically, PPEs are based on the PPE monograph that has been developed and endorsed by the AAP, American Academy of Family Physicians, the American College of Sports Medicine, and American Medical Society for Sports Medicine for the past 40 years (AMSSM)

- American Heart Association (AHA) 14-point questionnaire also has a similar format and questions

- The PPE has three, and possibly four, main parts: history, family history, and examination and sometimes an ECG

Pre-Participation Physicals

WHAT ARE THE COMPONENTS OF THE PREPARTICIPATION EVALUATION?

1. History:

- Past medical history
- Cardiac symptoms with exercise
- Odd or suspicious symptoms: seizures, syncope
- Any known or previously diagnosed cardiac pathology, including congenital heart disease, cardiac arrhythmia or channelopathies, cardiomyopathy, history of myocarditis, or coronary artery anomalies, including those caused by KD, should be evaluated by a cardiologist before clearance.

- Studies have shown that complete agreement between athlete and parental reports of medical history on the PPE is less than 20%. This highlights the importance of involving parents or primary caregivers in the history

Pre-Participation Physicals

The AAP Policy statement recommended 4 questions directed toward SCA and SCD detection:

1. Have you ever fainted, passed out, or had an unexplained seizure suddenly and without warning, especially during exercise or in response to sudden loud noises, such as doorbells, alarm clocks, and ringing telephones?

2. Have you ever had exercise-related chest pain or shortness of breath?

3. Has anyone in your immediate family (parents, grandparents, siblings) or other, more distant relatives (aunts, uncles, cousins) died of heart problems or had an unexpected sudden death before age 50? This would include unexpected drownings, unexplained auto crashes in which the relative was driving, or sudden infant death syndrome (SIDS).

4. Are you related to anyone with HCM or hypertrophic obstructive cardiomyopathy, Marfan syndrome, arrhythmogenic cardiomyopathy, LQTS, short QT syndrome, Brugada Syndrome, or catecholaminergic polymorphic ventricular tachycardia or anyone younger than 50 years with a pacemaker or implantable defibrillator?

Miller SM, Peterson AR. The sports preparticipation evaluation practice gaps.

Pre-Participation Physicals

TABLE 1. The American Heart Association 14-Element Cardiovascular Screening Checklist for Congenital and Genetic Heart Disease

MEDICAL HISTORY[a]
Personal history
1. Chest pain/discomfort/tightness/pressure related to exertion
2. Unexplained syncope/near-syncope[b]
3. Excessive and unexplained dyspnea/fatigue or palpitations associated with exercise
4. Previous recognition of a heart murmur
5. Elevated systemic blood pressure
6. Previous restriction from participation in sports
7. Previous testing for the heart, ordered by a physician

Pre-Participation Physicals

TABLE 1. The American Heart Association 14-Element Cardiovascular Screening Checklist for Congenital and Genetic Heart Disease

Family history
8. Premature death (sudden and unexpected or otherwise) before 50 y of age attributable to heart disease in ≥1 relative
9. Disability from heart disease in a close relative <50 y of age
10. Hypertrophic or dilated cardiomyopathy, long-QT syndrome, or other ion channelopathies, Marfan syndrome, or clinically significant arrhythmias; specific knowledge of genetic cardiac conditions in family members

Pre-Participation Physicals

WHAT ARE THE COMPONENTS OF THE PREPARTICIPATION EVALUATION?

2. Physical Examination:

Vital signs:

- Height, weight, and BMI

- Blood pressure

- Elevated blood pressure is common. It can be a sign of underlying cardiac disease (eg, coarctation of the aorta) or undiagnosed hypertension

- If a patient has upper extremity hypertension, careful attention should be paid to femoral pulses and blood pressure gradient between right arm and leg.

Pre-Participation Physicals

2. Physical Examination:

General:

General examination is important to identify features that could be suspicious for certain syndromes:

- Abnormalities of spinal curvature, pectus deformities, hyperextensible joints, arm span to height ratio, myopia, and other characteristic facial features: Marfan syndrome

- Neck webbing, short stature, a low hairline, or low-set ears: Turner syndrome

Pre-Participation Physicals

2. Physical Examination:

Cardiovascular exam:

- There should be a focus on identifying pathologic heart murmurs, abnormal or extra heart sounds, or rhythm irregularity

- Benign murmurs are common in pediatrics, and it can be a challenge to differentiate between benign and pathologic

Pre-Participation Physicals

TABLE 1. The American Heart Association 14-Element Cardiovascular Screening Checklist for Congenital and Genetic Heart Disease

Physical examination
11. Heart murmur[c]
12. Femoral pulses to exclude aortic coarctation
13. Physical stigmata of Marfan syndrome
14. Brachial artery blood pressure (sitting position)[d]

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Pre-Participation Physicals

❖ **Electrocardiogram Screening:**

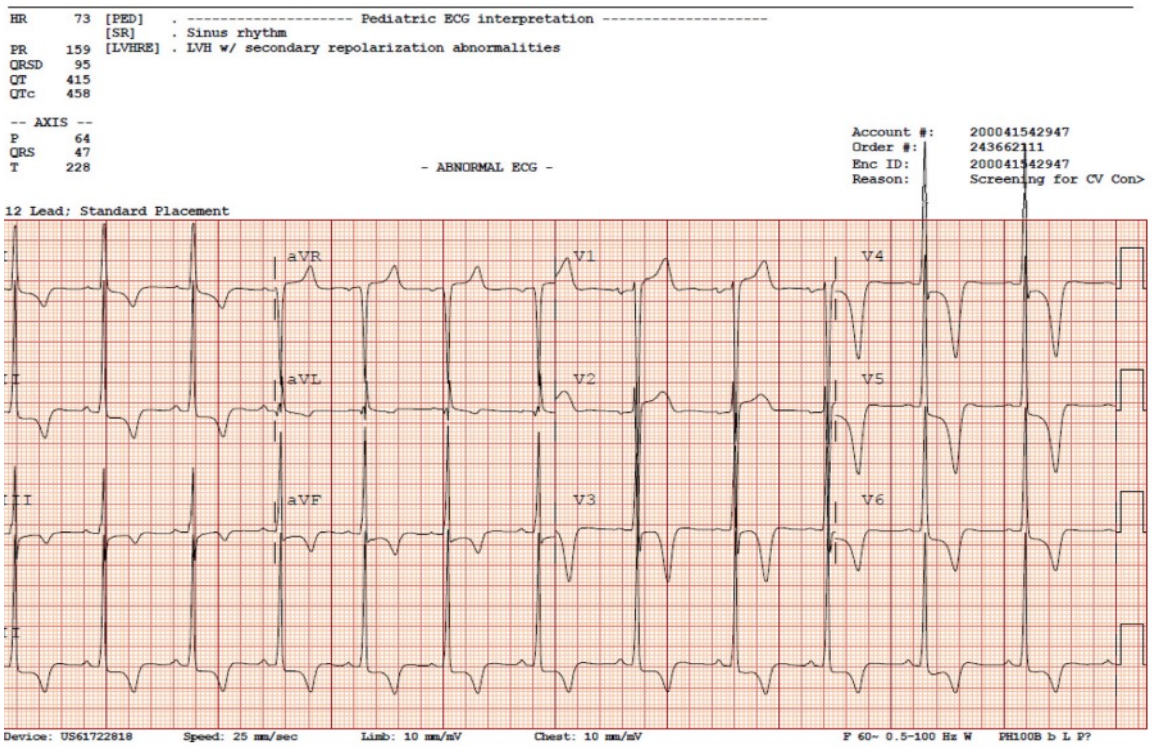
- There continues to be debate regarding whether or not ECGs should be added to the typical PPE

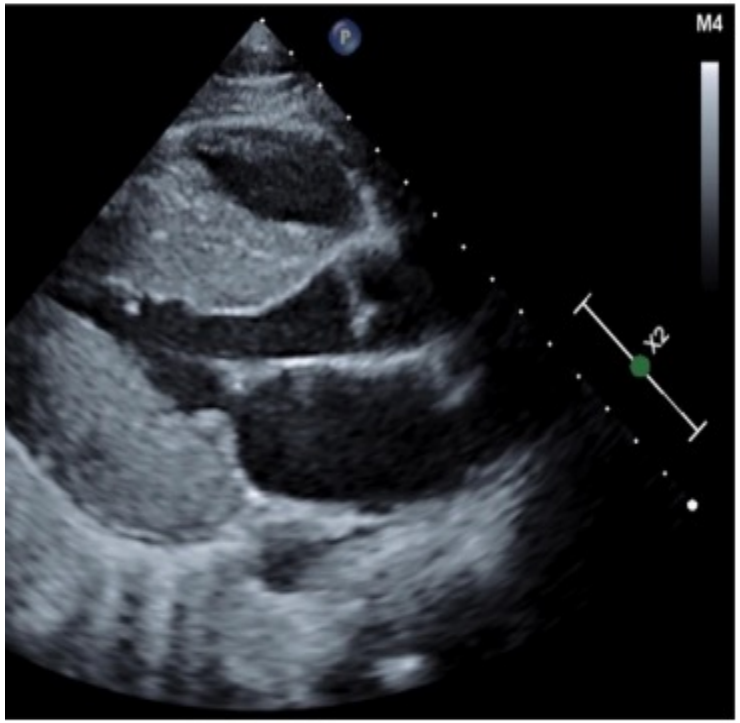
- The European Society for Cardiology recommends including ECG for competitive athletes and some countries mandate ECGs at various ages

- The most recent guidelines from the American College of Cardiology (ACC), AHA, and AMSSM suggest that ECG may be performed but did not recommend universal ECG screening

Pre-Participation Physicals
Case 1

As the patient was 11 year old at the time of his initial normal cardiac evaluation, it was recommended that he return to clinic in 4 years for repeat evaluation. At that visit, he reports that he has been participating in competitive sports without problems and has no cardiac symptoms. His cardiac examination was significant for a heart murmur.





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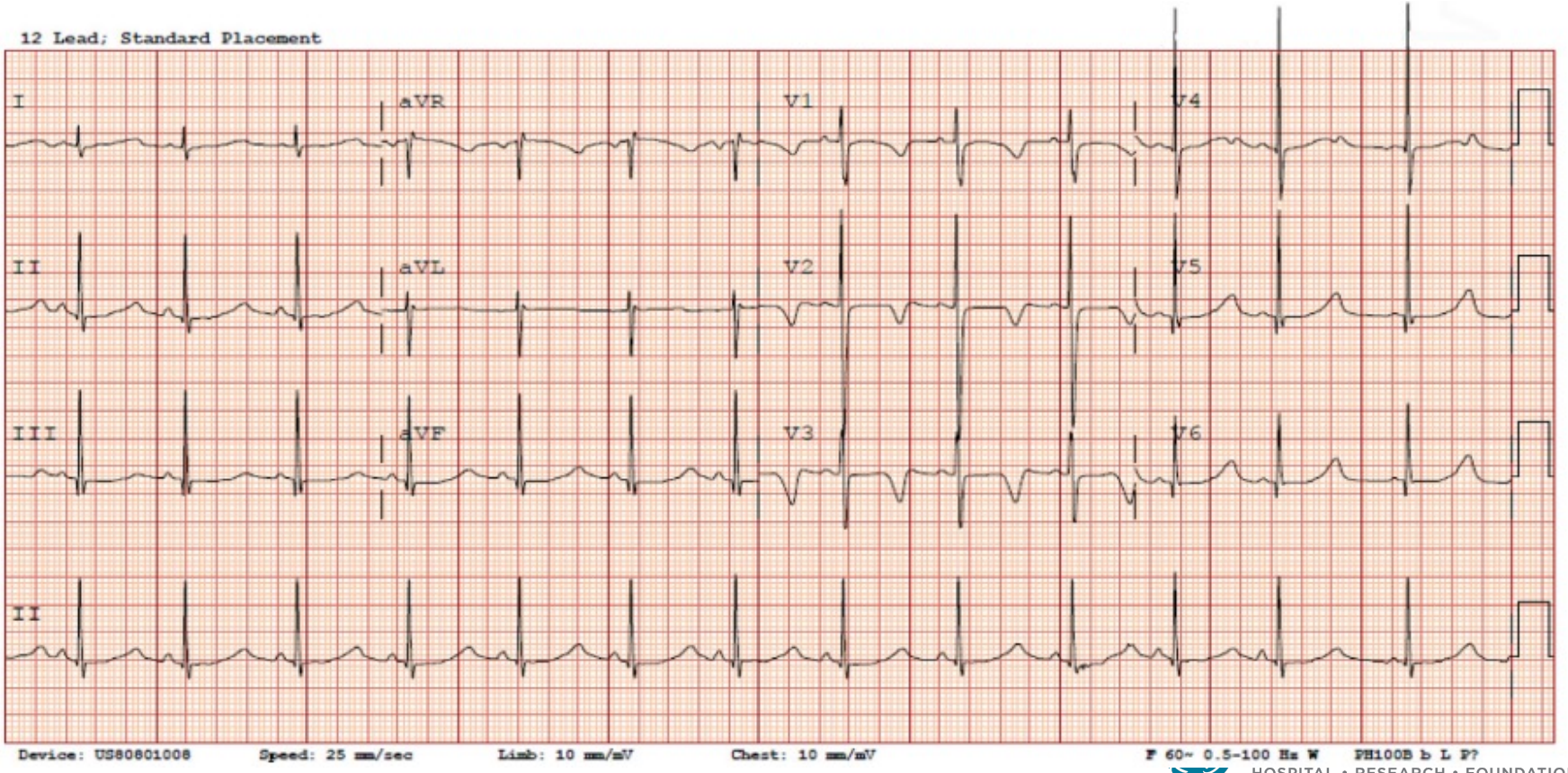
Pre-Participation Physicals
Case 2

- The 10-year-old patient with syncope underwent further cardiac evaluation due to the history of atypical nature of the syncope and the association with exercise. An ECG (Fig. 6) is notable for a correct QT duration of 597 ms. Laboratory testing does not reveal a secondary cause of prolonged QT



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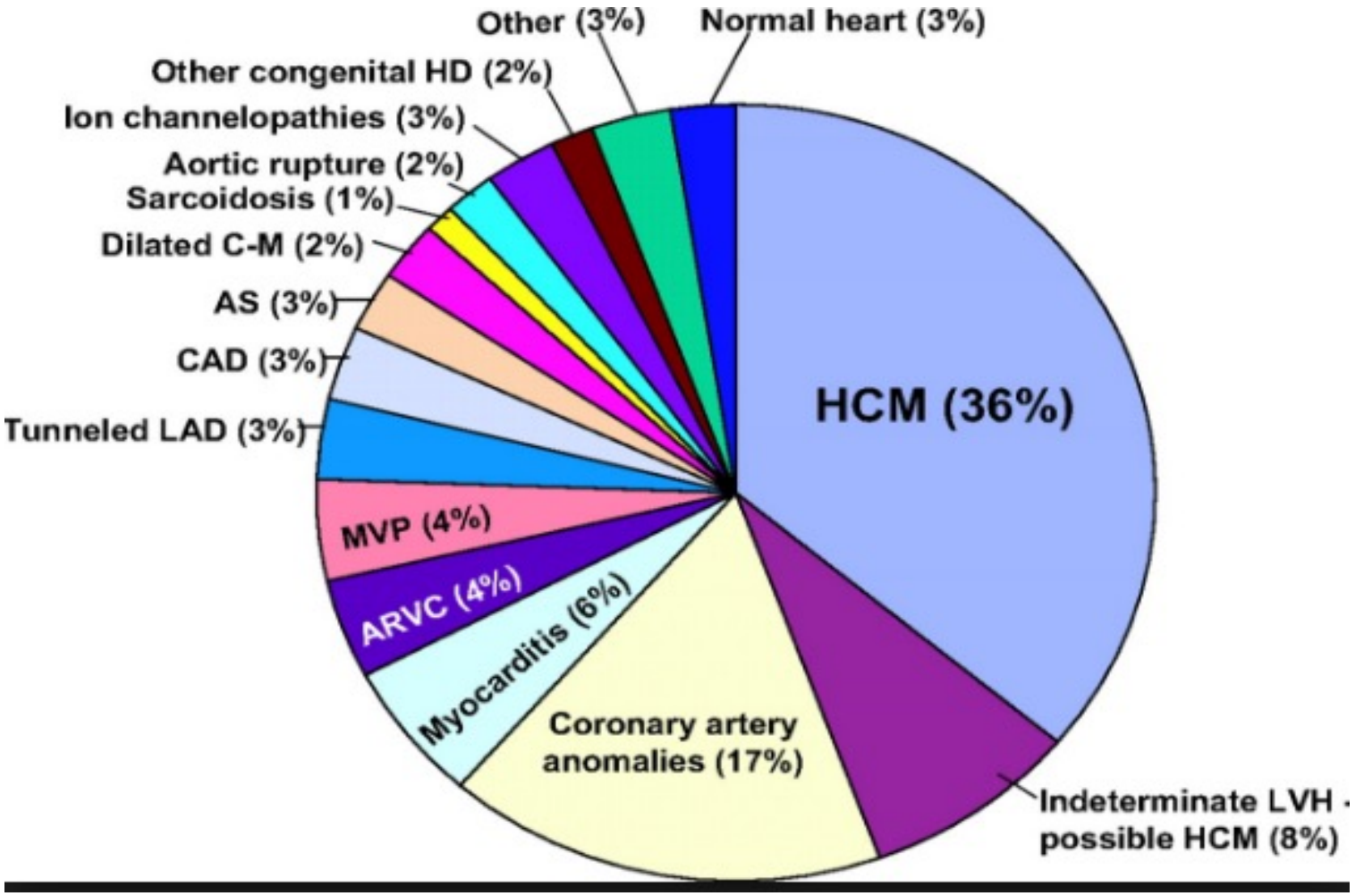
OUTLINE

- Epidemiology
- Pre-participation Physicals
- Causes of Sudden Cardiac Death

Etiology based on largest US data set



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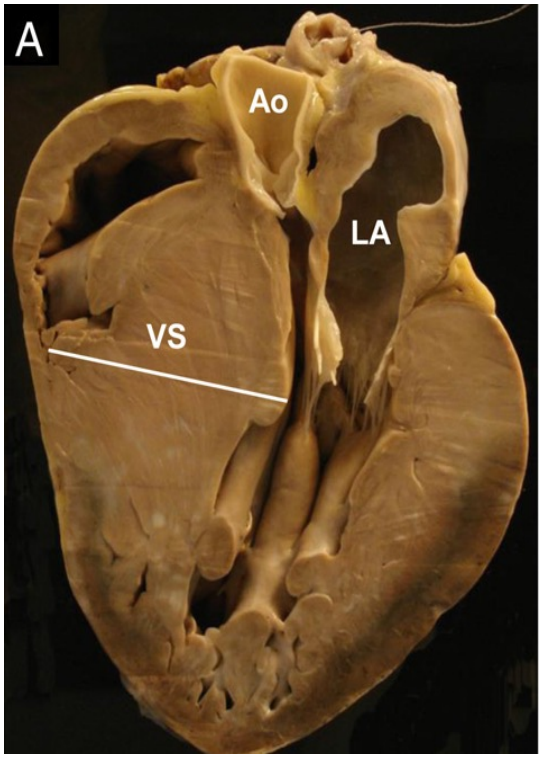
Causes of Sudden Death

- **Hypertrophic Cardiomyopathy**
- Coronary artery anomalies

Hypertrophic Cardiomyopathy

"A disease state characterized by unexplained LV hypertrophy associated with non-dilated ventricular chambers, in the absence of another cardiac or systemic disease that could produce this hypertrophy"



Hypertrophic Cardiomyopathy - Epidemiology

- Global disease
  - reported in over 50 countries on all continents, all ethnic/racial groups, both genders
- Prevalence of 0.2% (1 in 500)

Scope of the Disease

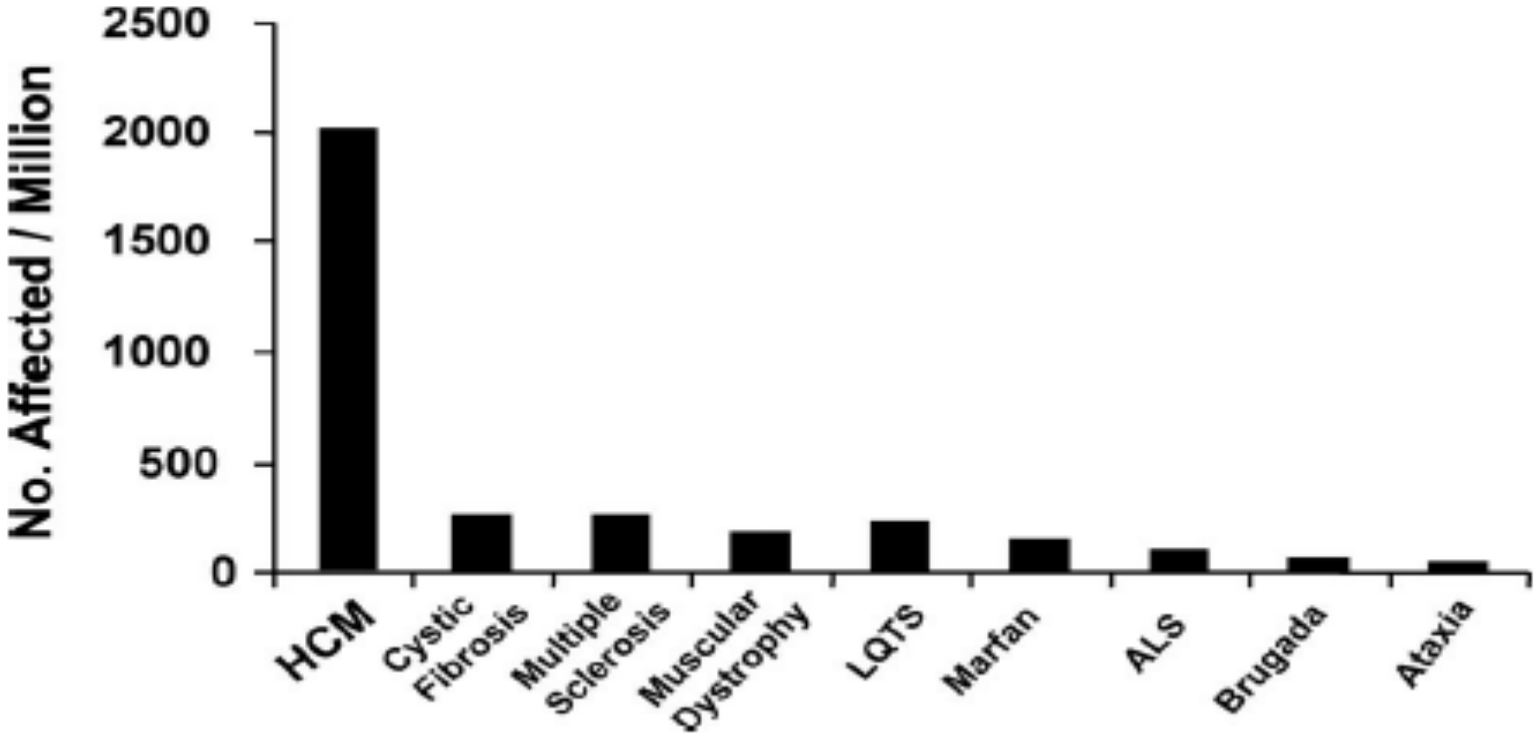


Figure 2. The prevalence of hypertrophic cardiomyopathy in the general population substantially exceeds that of several other cardiac and noncardiac diseases, which paradoxically have achieved greater recognition in the public sphere.

Genetics

- Transmission is autosomal dominant, 50% chance of inheriting
- De novo /sporadic mutations do arise
- Phenotypic heterogeneity is present within and between families
- Most will demonstrate LVH by early adulthood, accelerated during adolescent growth spurt

Genetics

TABLE 1. Distribution of Genes in HCM-Genotyped Index Cases According to Familial or Sporadic Cases

Gene	Total*	Familial HCM	Sporadic	Mutations (Novel)
Total	n=124	n=109	n=15	97 (60)
MYBPC3	52 (42%)	45 (41%)	7 (47%)	39 (25)
MYH7	50 (40%)	45 (41%)	5 (33%)	40 (24)
TNNT2	8 (6.5%)	5 (4.5%)	3 (20%)	7 (2)
TNNI3	8 (6.5%)	8 (7%)	0	7 (6)
MYL2	5 (4%)	5 (4.5%)	0	4 (2)
MYL3	1 (<1%)	1 (<1%)		1 (1)

*There were 120 initial index cases, but 2 different mutations within the same family were identified in 4 families. The distribution was therefore performed on 124 index cases.

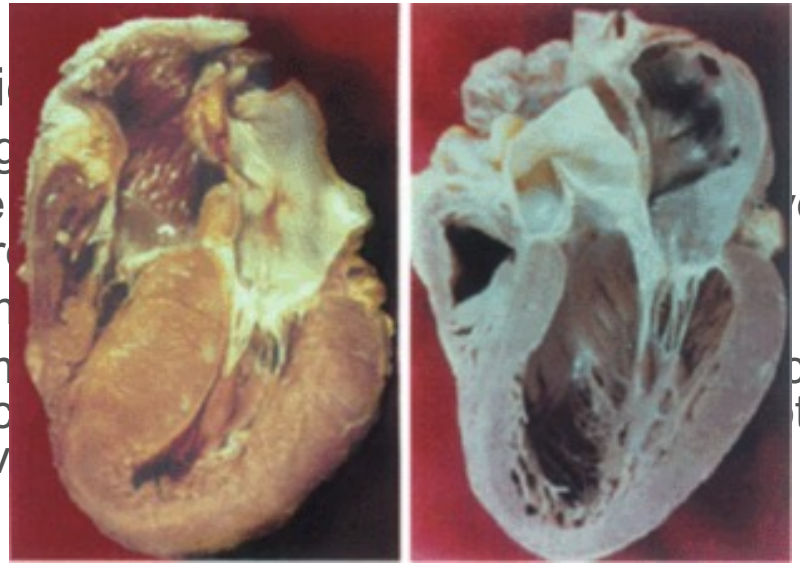
Phenotype and Morphologic Features

- Left ventricular hypertrophy
  - No single pattern
  - Diffuse disease more common, but 1/3rd have localized hypertrophy
  - Asymmetric- anterior septum predominant
  - Other markers: hypercontractile LV, dynamic subaortic obstruction produced by systolic anterior motion of the mitral valve (SAM)

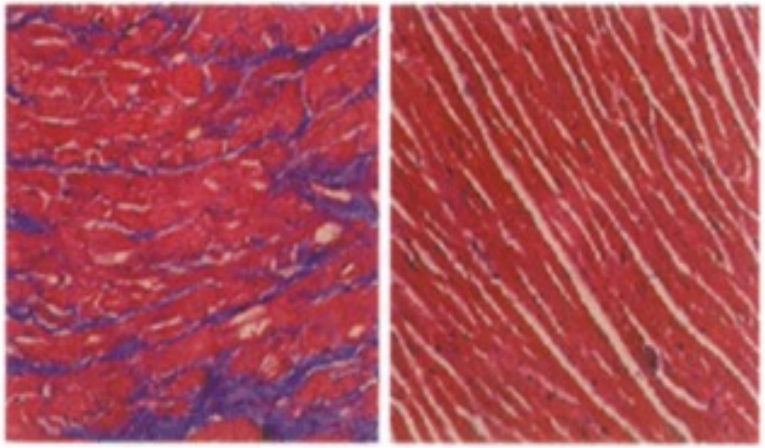
- Cellular components
  - LV myocardial architecture is disorganized
  - Hypertrophied cardiac muscle cells with bizarre shapes, intercellular connections in chaotic alignment
  - Degree of cellular disarray variable- more extensive in patients who die young
  - Abnormal intramural coronary arteries

Phenotype and Morphologic Features

- Left ventri
  - No sing
  - Diffuse … e localized hypertr
  - Asymm
  - Other m … c subaortic obstruc … tion of the mitral v



- Cellular cc
  - LV myc
  - Hypertr … irre shapes, intercel … t
  - Degree … xtensive in patients
  - Abnorm



HCM Normal

Pathophysiology

- LVOT obstruction
- Diastolic dysfunction
- Myocardial ischemia
- Autonomic dysfunction
- Mitral regurgitation

Pathophysiology

- **LVOT obstruction**
- Diastolic dysfunction
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Pathophysiology

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Pathophysiology

- LVOT obstruction
- Diastolic dysfunction
- **Myocardial ischemia**
- Autonomic dysfunction
- Mitral regurgitation

Pathophysiology

❖ Myocardial Ischemia

- Demand Supply mismatch
- Myocardial bridging

Demand

Hypertrophy
Afterload

Supply

Abnormal CAs

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Pathophysiology

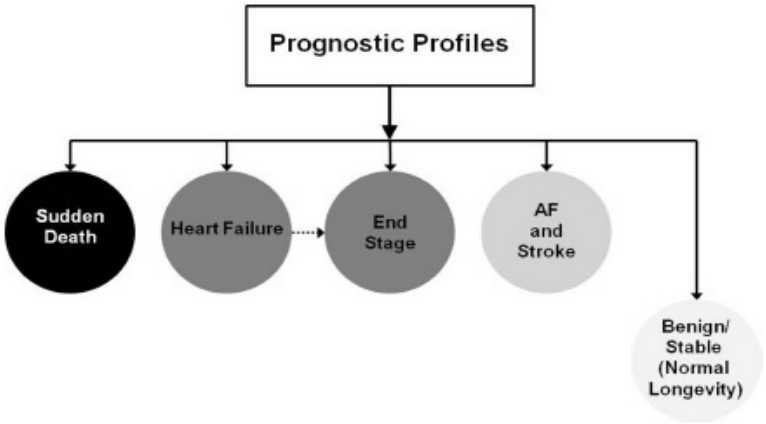
- LVOT obstruction
- Diastolic dysfunction
- Myocardial ischemia
- **Autonomic dysfunction**
- Mitral regurgitation

Pathophysiology

- LVOT obstruction
- Diastolic dysfunction
- Myocardial ischemia
- Autonomic dysfunction
- **Mitral regurgitation**

Clinical Course/Natural history

- Presented with murmur, family history, abnormal EKG and arrhythmia
- From asymptomatic to obstructive disease to aborted sudden cardiac death
- Mortality rate 1% per year, much lower than older studies



Asymptomatic/mildly Progressive HF
Sudden death
Atrial fibrillation and embolic stroke

Sudden Death

- Most devastating and unpredictable consequence
- Can be the presenting manifestation
- Most common cause of sudden death in young adults
- Most frequent cause of SCD in US competitive athletes
- Sudden death risk decreases with increasing age
- Mechanism: Primary VT and Vfib; substrate:
  - abnormal cellular architecture
  - myocardial fibrosis from silent ischemia

Diagnosis: EKG

- As screening for sports physical/murmur/family history
- 12 lead EKG is abnormal in %75-95 of the patients with HCM
- Falsely negative in 10%
- Evidence of WPW
- Holter: evidence of VT

TABLE 1. *Frequency of Common Electrocardiographic Abnormalities in Patients with Hypertrophic Cardiomyopathy*

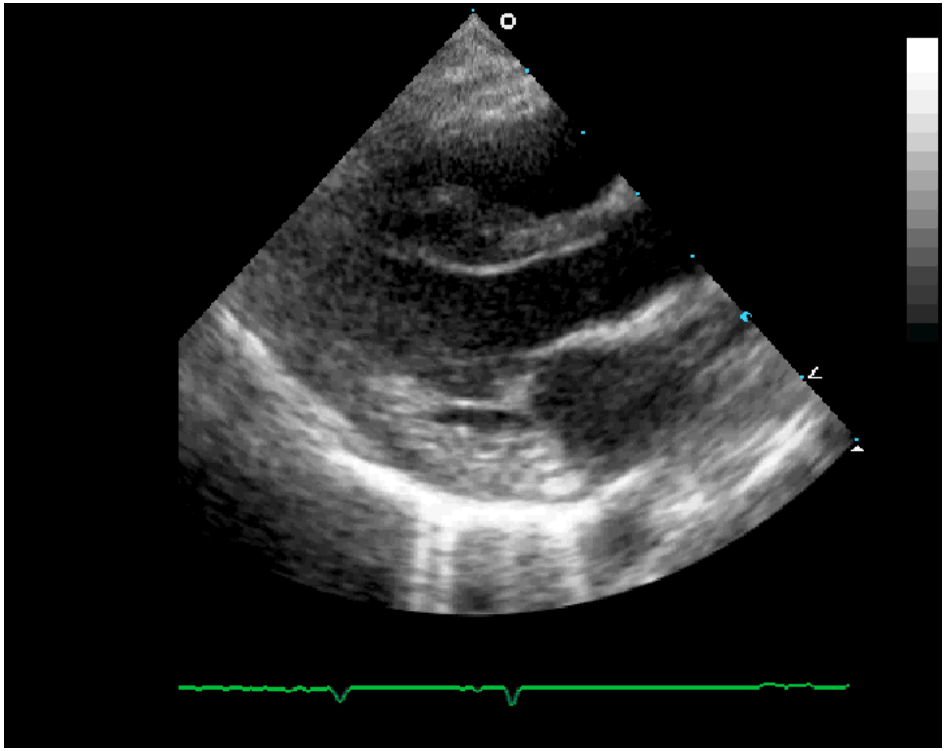
ECG Finding	Obstructed		Nonobstructed	
	Symptomatic	Asymptomatic	Symptomatic	Asymptomatic
Abnormal ECG	54/55 (98)	13/13 (100)	37/40 (93)	19/26 (73)*
Repolarization abnormalities	48/55 (87)	11/13 (85)	34/40 (85)	15/26 (58)†
Left ventricular hypertrophy	31/38 (82)	8/10 (80)	19/33 (58)	7/21 (33)‡
Left atrial abnormality	37/50 (74)	5/12 (42)	14/38 (37)*	5/26 (19)*
Left axis deviation (0 to −90°)	25/53 (47)	8/13 (62)	12/40 (30)	5/26 (19)‡
Abnormal Q waves	23/55 (42)	5/13 (38)	8/40 (20)	8/26 (31)
Right atrial enlargement	9/50 (18)	3/13 (23)	3/38 (8)	1/26 (4)
Right atrial enlargement and left ventricular hypertrophy	7/38 (18)	2/10 (20)	3/33 (9)	0/21 (0)
P-R interval >0.20 sec	5/50 (10)	2/13 (15)	4/38 (11)	1/26 (4)

Diagnosis- Imaging: TTE

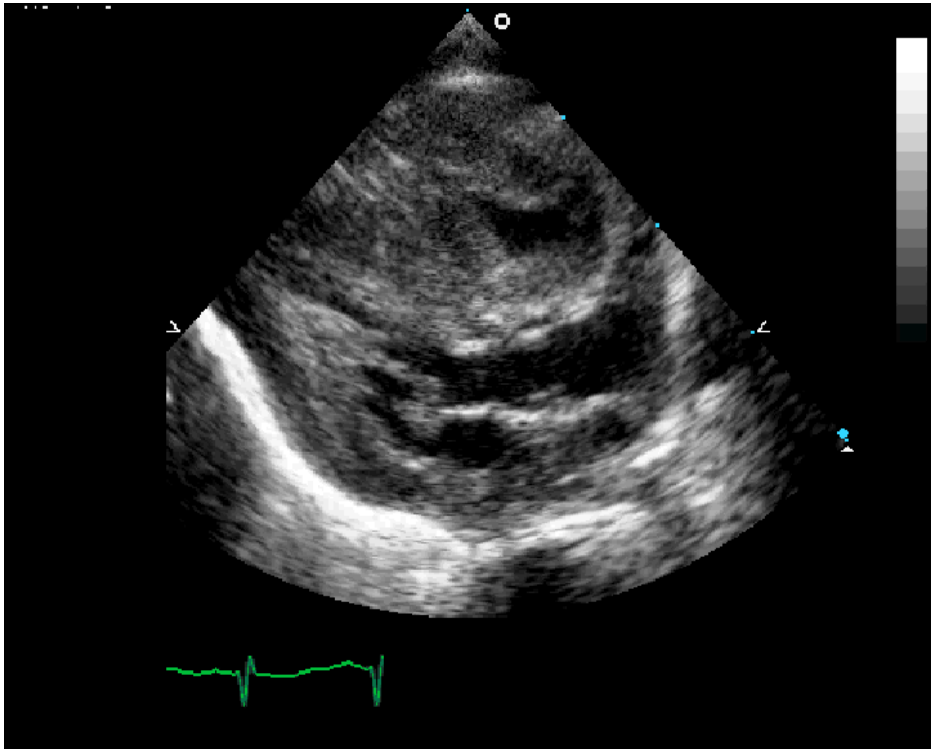
- Suspicion arises with a heart murmur or abnormal EKG
- Diagnosis confirmed with 2D echocardiography
- Echocardiogram:
  - LV/RV wall thickness
  - LV mass
  - Symmetric/asymmetric
  - Systolic anterior motion of the MV
  - Hyperdynamic LV
  - Subaortic obstruction and degree (CW Doppler)
  - LV function- systolic and diastolic

Echocardiogram

Normal



HCM



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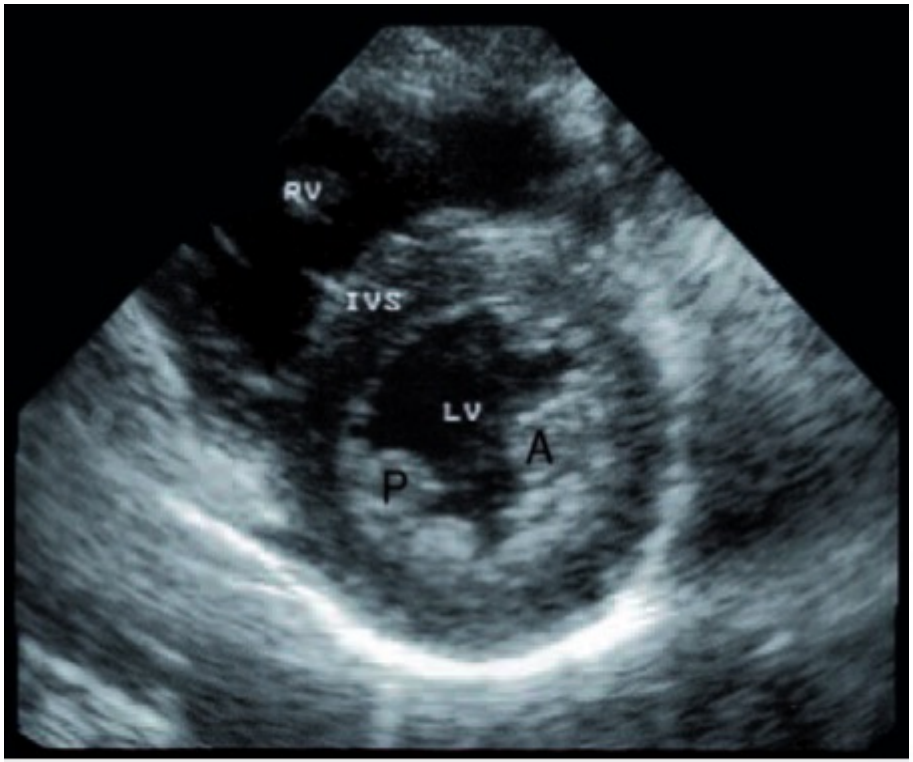
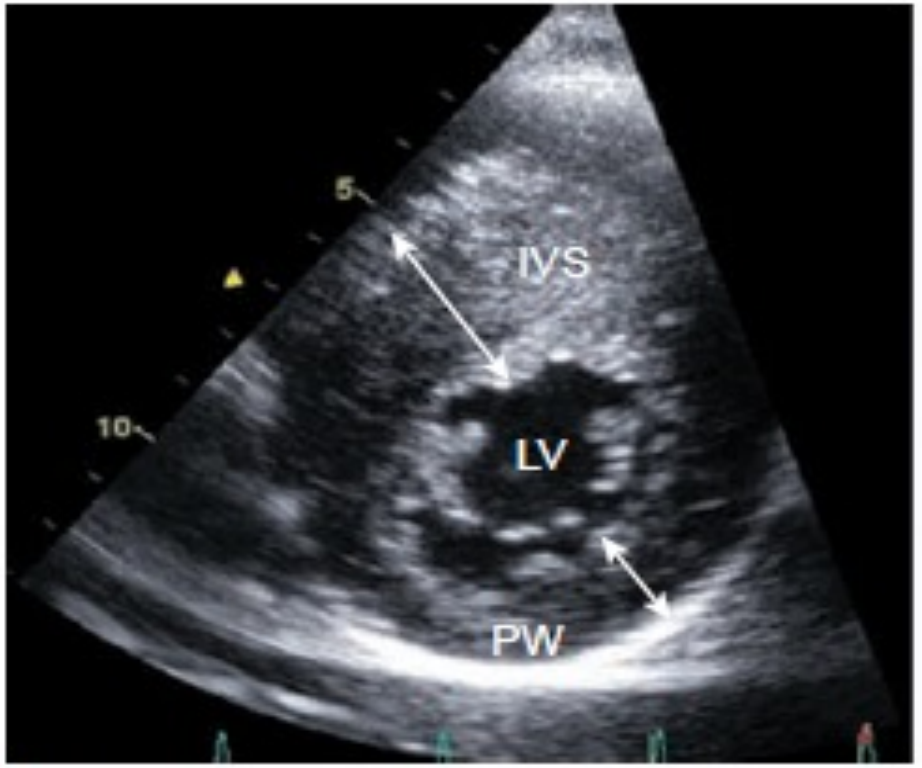
Echocardiogram



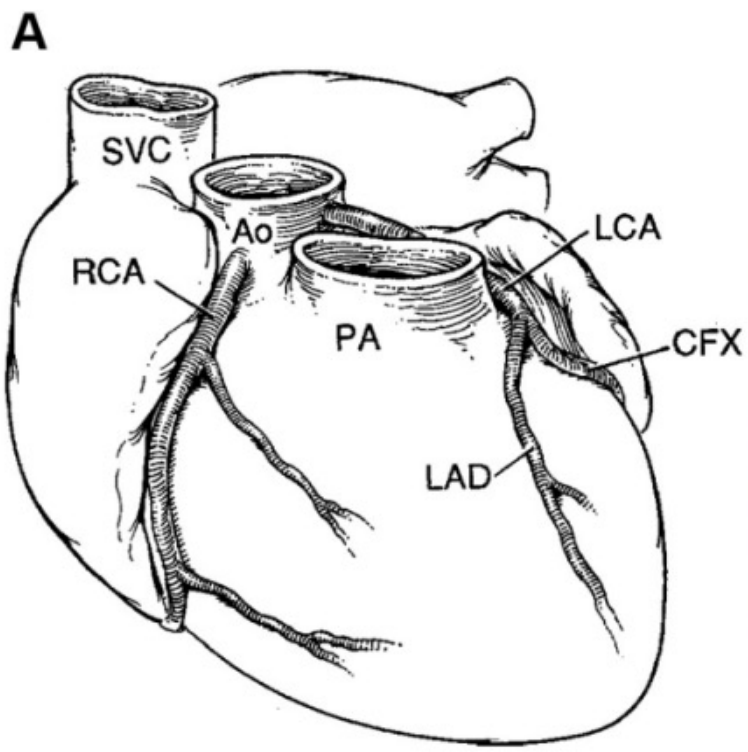


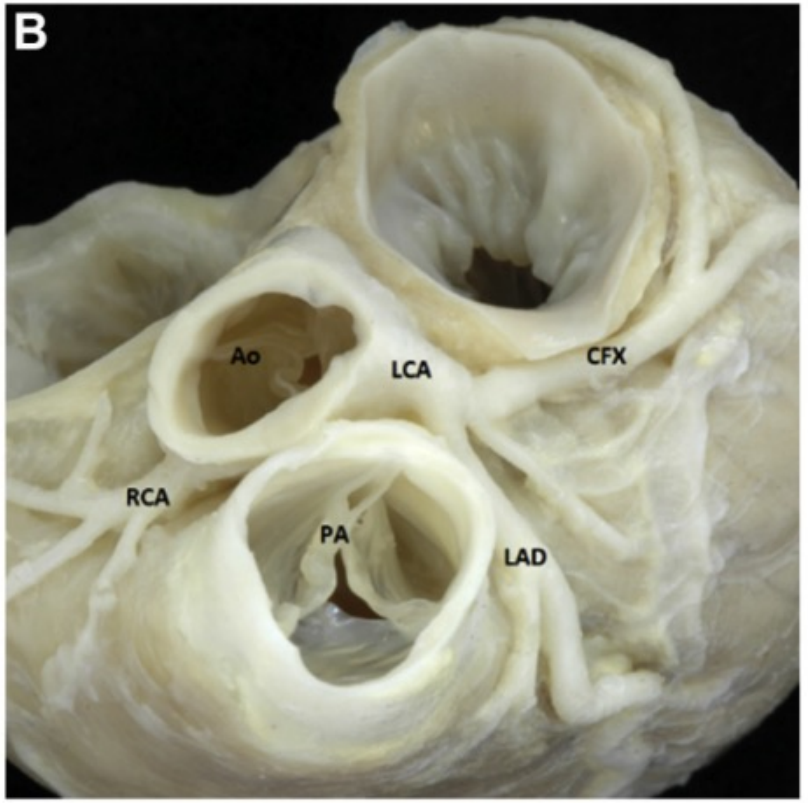
(a)

Causes of Sudden Death

- Hypertrophic Cardiomyopathy
- Coronary artery anomalies

Coronary artery abnormalities





Frommelt et al. Journal of the American Society of Echocardiography. March 2020

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Coronary artery abnormalities

Table 1. Simplified Nomenclature of CAAs

Type of anomaly	Variant	Subvariants
Anomalies of origin	Anomalous pulmonary origin of the coronaries	Origin of left main coronary artery from the pulmonary artery Origin of right coronary artery from the pulmonary artery Origin of circumflex coronary artery from the pulmonary artery Origin of left and right coronary arteries from the pulmonary artery
	Anomalous aortic origin of the coronaries	Origin of left main coronary artery from the right aortic sinus of Valsalva Origin of right coronary artery from the left aortic sinus of Valsalva Origin of left anterior descending coronary artery from the right aortic sinus of Valsalva Origin of left anterior descending coronary artery from the right coronary artery Origin of circumflex coronary artery from the right aortic sinus of Valsalva Origin of circumflex coronary artery from the right coronary artery Single coronary artery Inverted coronary arteries Others
	Congenital atresia of the left main artery	
Anomalies of course	Myocardial (or coronary) bridging	Symptomatic Asymptomatic
	Coronary aneurysm	Congenital Acquired
Anomalies of termination	Coronary arteriovenous fistula	Congenital Acquired
	Coronary stenosis	Congenital Acquired

CAA indicates coronary artery anomaly.

Coronary Artery Anomalies. Gentile et al. Circulation. 2021;144:983–996

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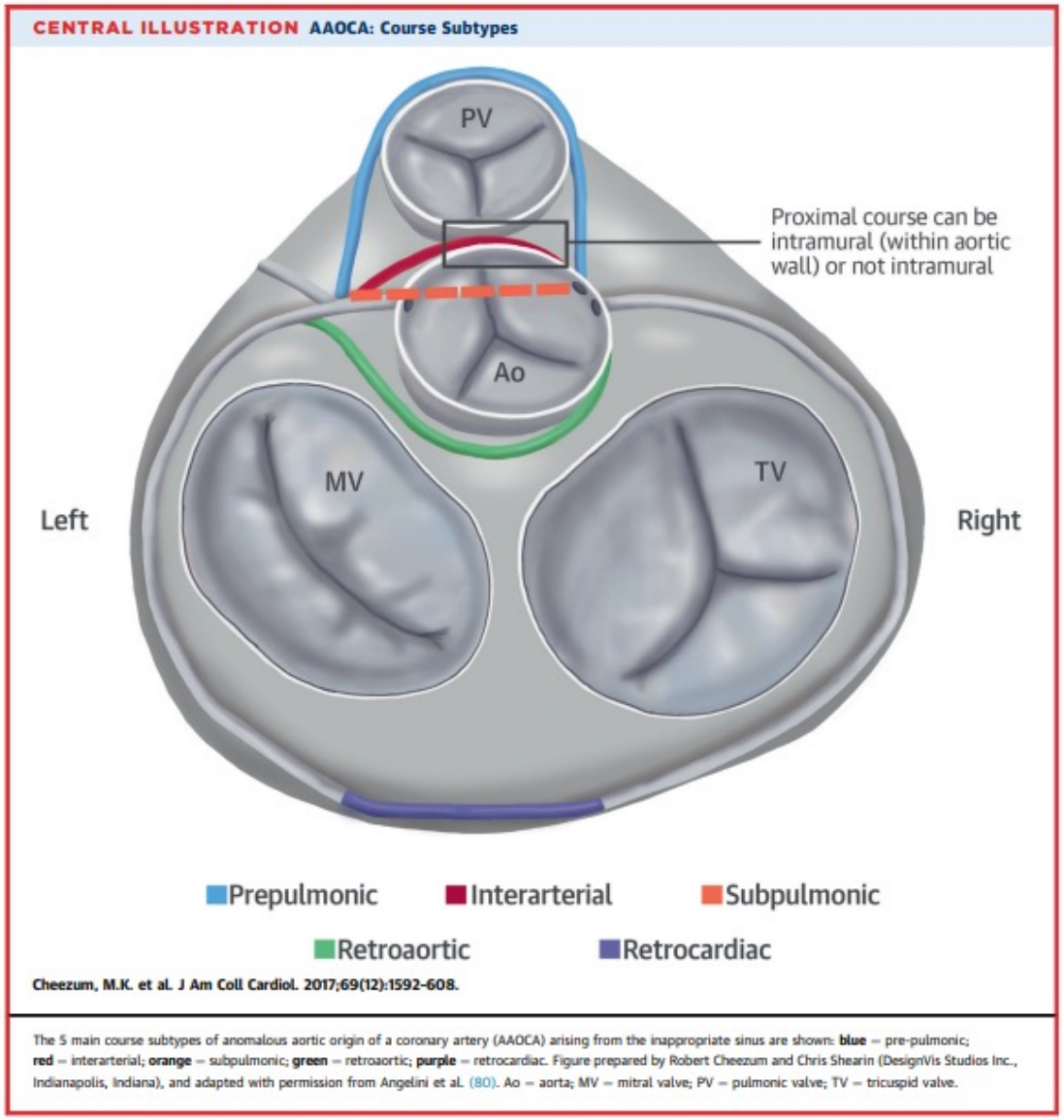
AAOCA Variations

- Origin: at or above the inappropriate sinus of Valsalva

- Ostium Morphology: Slit-like, ovale, round

- Course:
  - Interarterial
  - pre-pulmonic (0.04%)
  - sub-pulmonic (0.07%)
  - Retrocardaic (0.03%)
  - Retroaortic (0.28%)

AAOCA Variations



Cheezum et al. JACC 2017

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Anomalous Aortic Origin of a Coronary Artery (AAOCA)

- Which coronary artery has the anomalous origin?
- What is its course?
- What are the patient's symptoms if any?
- What is the management?

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Anomalous Aortic Origin of a Coronary Artery (AAOCA)

- Estimated Prevalence:
  - Interarterial AAOLCA: 0.03%
  - Interarterial AAORCA: 0.23 %

AAOCA – Sudden Cardiac Death

- Cumulative risk of death with AAOCA (15-35 years) participating in competitive sports:
  - 6.3% AAOLCA
  - 0.2% AAORCA

Case reports of SCD with AAOCA at rest/recreational activities

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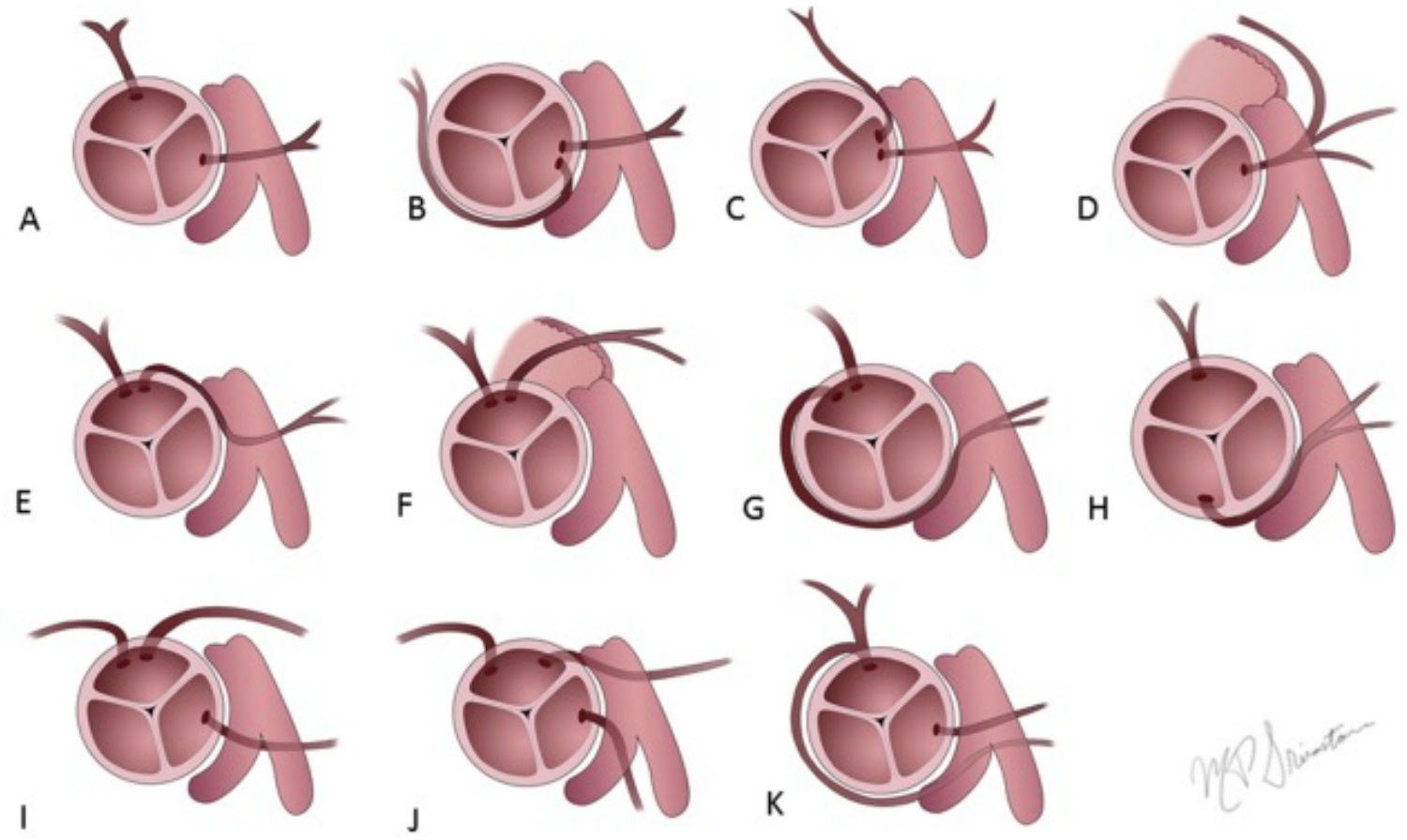
AAOCA Variations



Frommelt et a. Journal of the American Society of Echocardiography March 2020

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Anomalous Aortic Origin of a Coronary Artery (AAOCA)

Clinical presentation:

- Asymptomatic
- Exertional chest pain
- Exertional syncope
- Sudden death

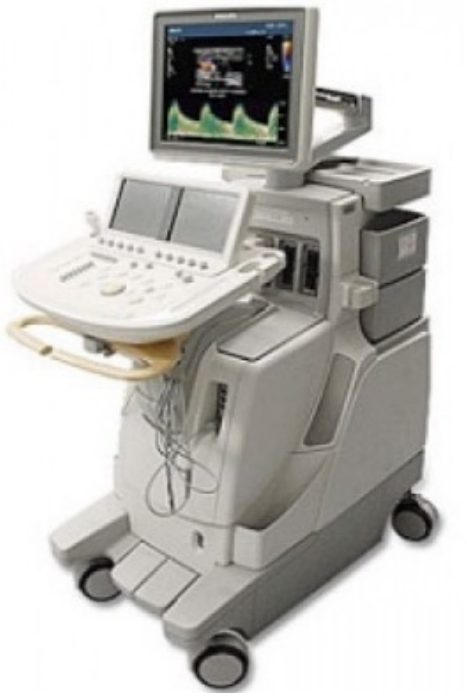
Pathophysiology of SCD with AAOCA

Possible mechanisms of sudden death

- Compression of intramural segment
- Compression of the interarterial segment
- Acute angle at take-off
- Ostial stenosis
- Coronary spasm

Anomalous Aortic Origin of a Coronary Artery (AAOCA)
Imaging modalities: Echo

- Pros:
  - available
  - affordable
  - Portable
  - Noninvasive nature

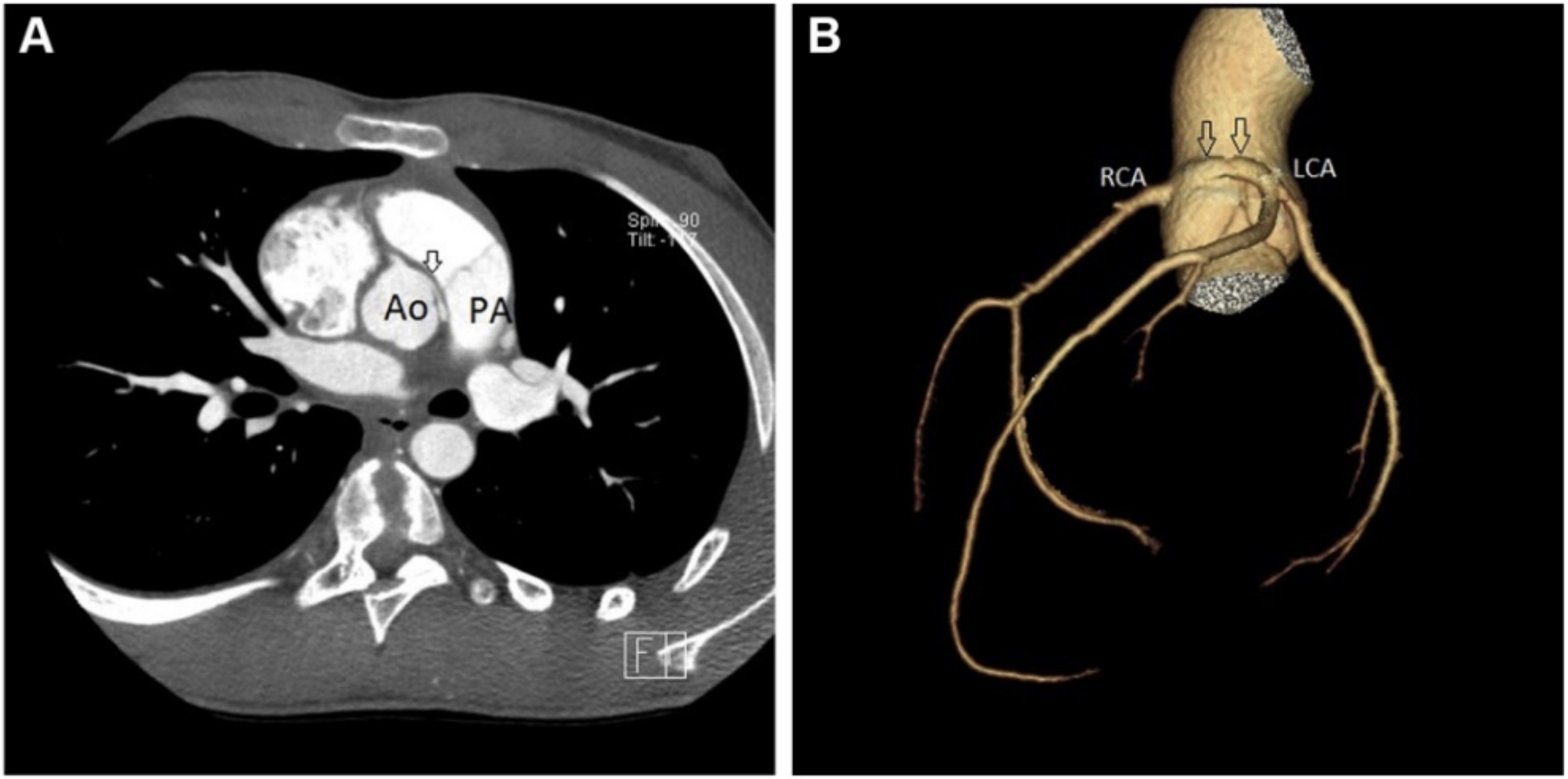


Cons:

- Spatial resolution
- Body habitus and sonographer experience

Anomalous Aortic Origin of a Coronary Artery (AAOCA)
Imaging modalities: Coronary CTA

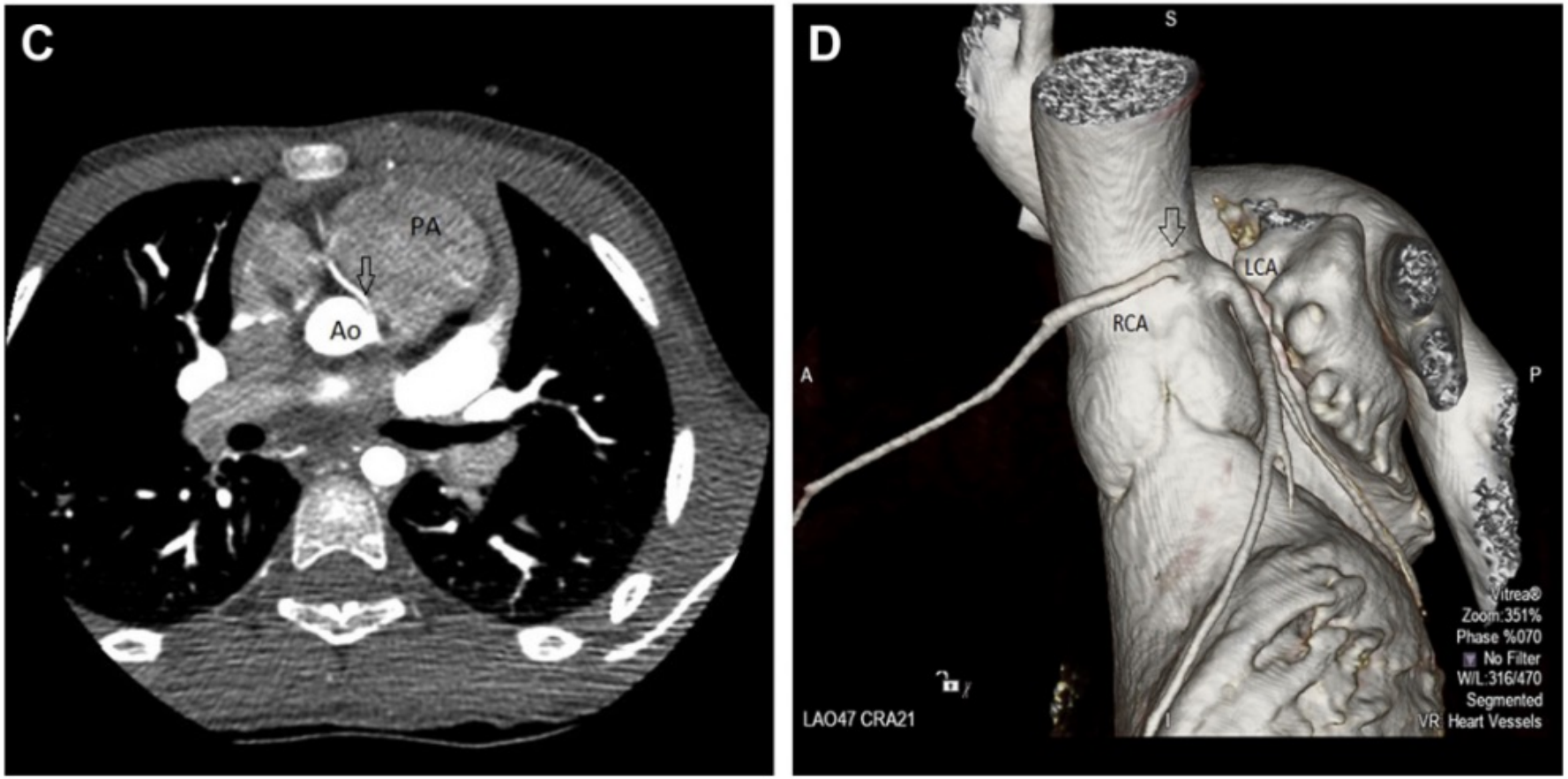
AAOLCA with an interarterial, intramural course



Frommelt et al. Journal of the American Society of Echocardiography. March 2020

Anomalous Aortic Origin of a Coronary Artery (AAOCA)
Imaging modalities: Coronary CTA

AAORCA with an interarterial, intramural course



Frommelt et al. Journal of the American Society of Echocardiography. March 2020

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Anomalous Aortic Origin of a Coronary Artery (AAOCA)

- Which coronary artery has the anomalous origin?
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Anomalous Aortic Origin of a Coronary Artery (AAOCA)

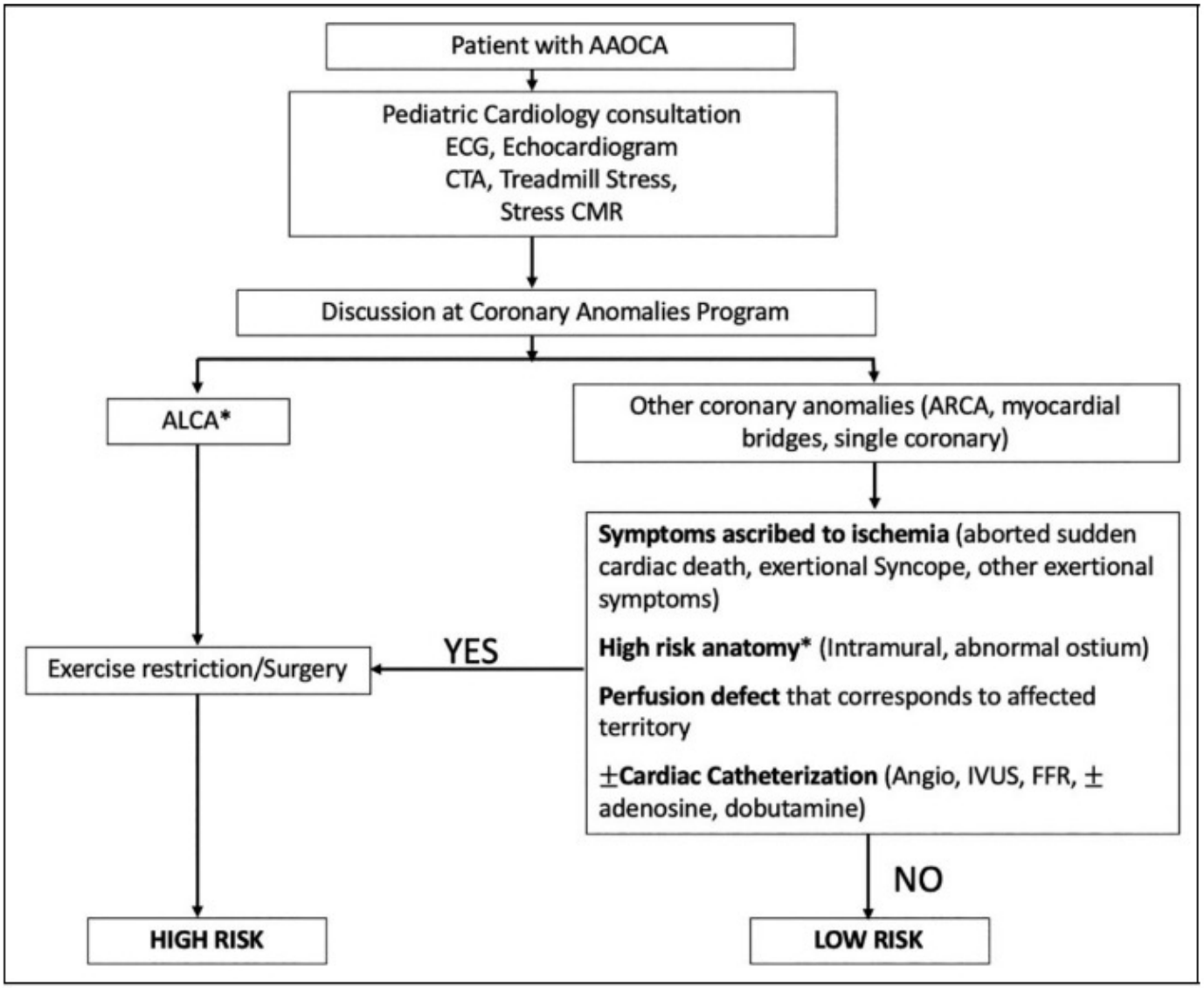
❖ **Treatment**:

- The indications for intervention in asymptomatic patients with AAOCA with an intramural course are debated, especially with AAOCA RCA from the left sinus of Valsalva

• **Surgical intervention is recommended:**

- Signs or symptoms of myocardial ischemia (eg, true angina, findings on provocative testing, aborted sudden cardiac death or arrest, or non-vagally-mediated arrhythmia)
- Anomalous origin of the left coronary artery from the right sinus with an interarterial/intramural course, even in the absence of symptoms (due to the higher calculated risk of sudden cardiac death)
- Asymptomatic patients with an intramural RCA from the left sinus of Valsalva (AAORCA), provocative testing is recommended

Anomalous Aortic Origin of a Coronary Artery (AAOCA)



Agrawal et al. World Journal for Pediatric and Congenital Heart Surgery

THANK YOU!