Should athletes be screened for genetic heart diseases?

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5th February 2017 16:00 – 17:00 Inherited Arrhythmia Syndromes VII: Exercise, Athletes, Screening and Recommendations

Topics

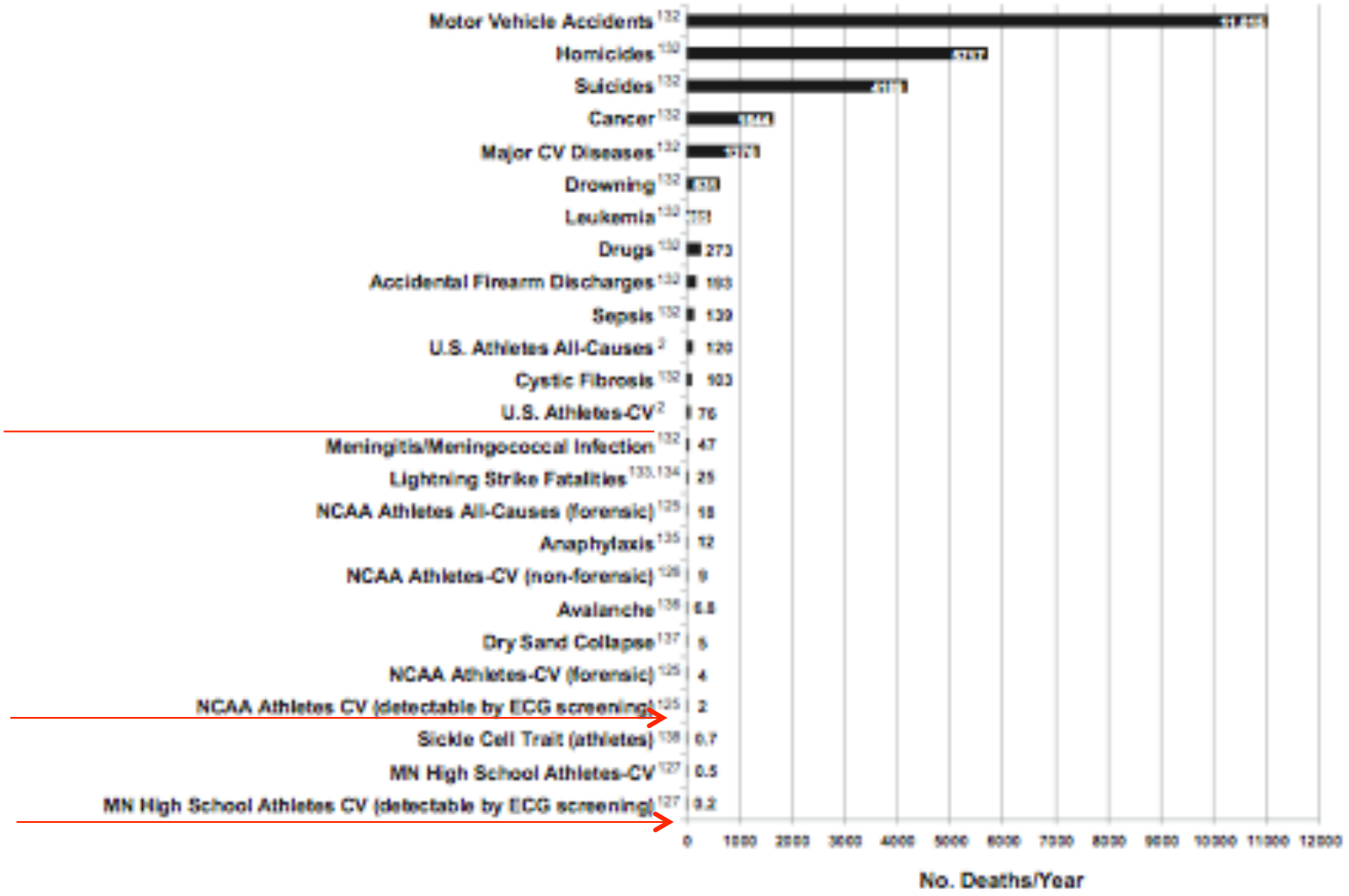
I. Why athlets ?

II. Why genetic heart disease ?

III. Is screening feasible ?

II. Why athlets?

Comparative frequencies of death attributable to all causes in young individuals aged <25 years



I. Why athlets ?

Although incidence is low
SCA/SCD in athletes - remarkably visible

I. Why athlets ?

general health benefit from regular exercise

activity increases the risk of SCA/SCD

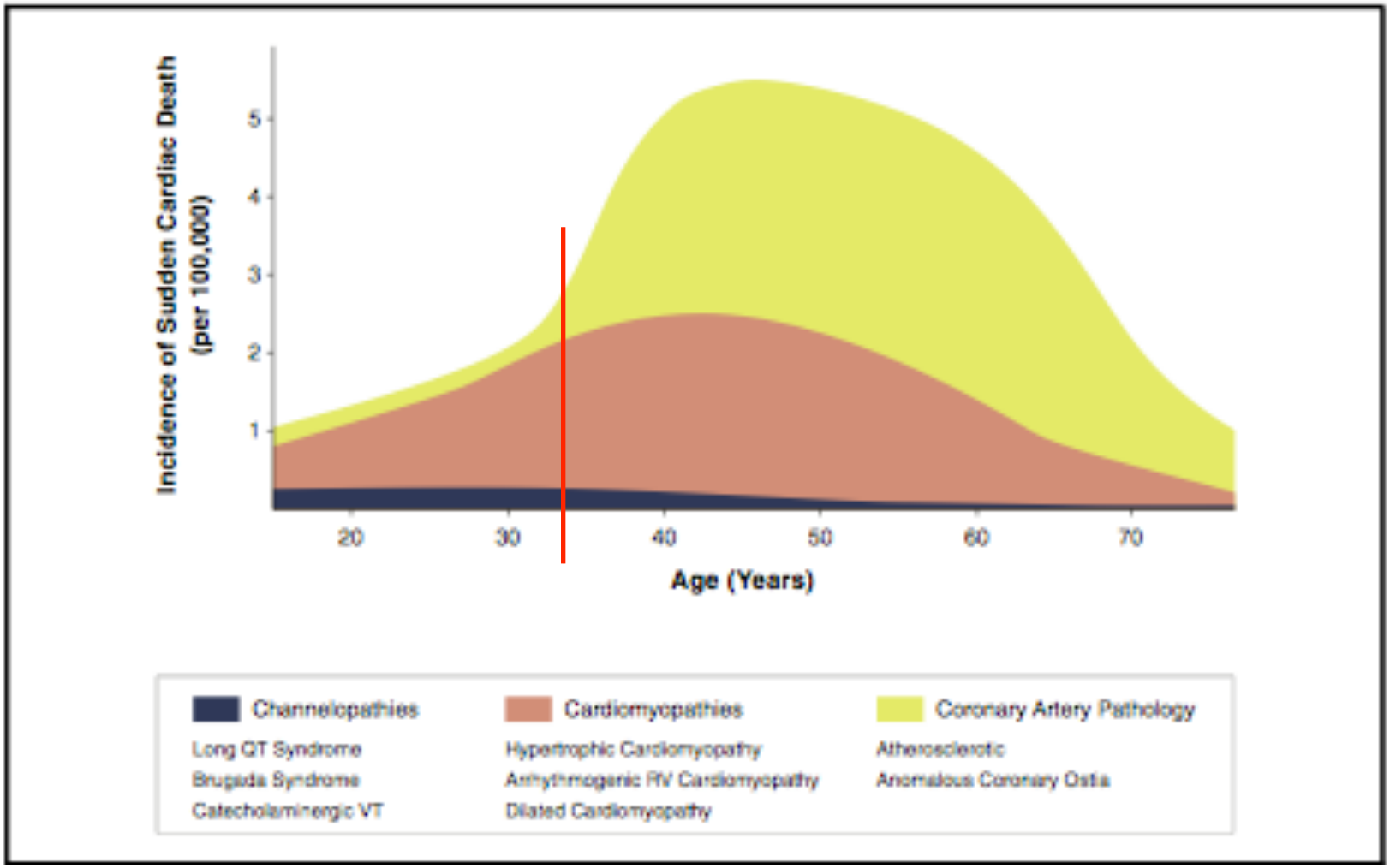
…,but !

- Screening only athletes will miss millions of children who are extremely active and competitive
- SCA may occur without activity
- Screening only those with symptoms is problematic
  - 50% antecedent symptoms 16% FHx

Liberthson N Eng J Med 1996

II. Why genetic heart diseases ?

Age-Dependent Changes in Incidence and Etiology of SCD



La Gerche et al. JACC, 2013:993–1007

II. Why genetic heart disease ?

Etiology of SCA in children

Structural cardiac abnormalities

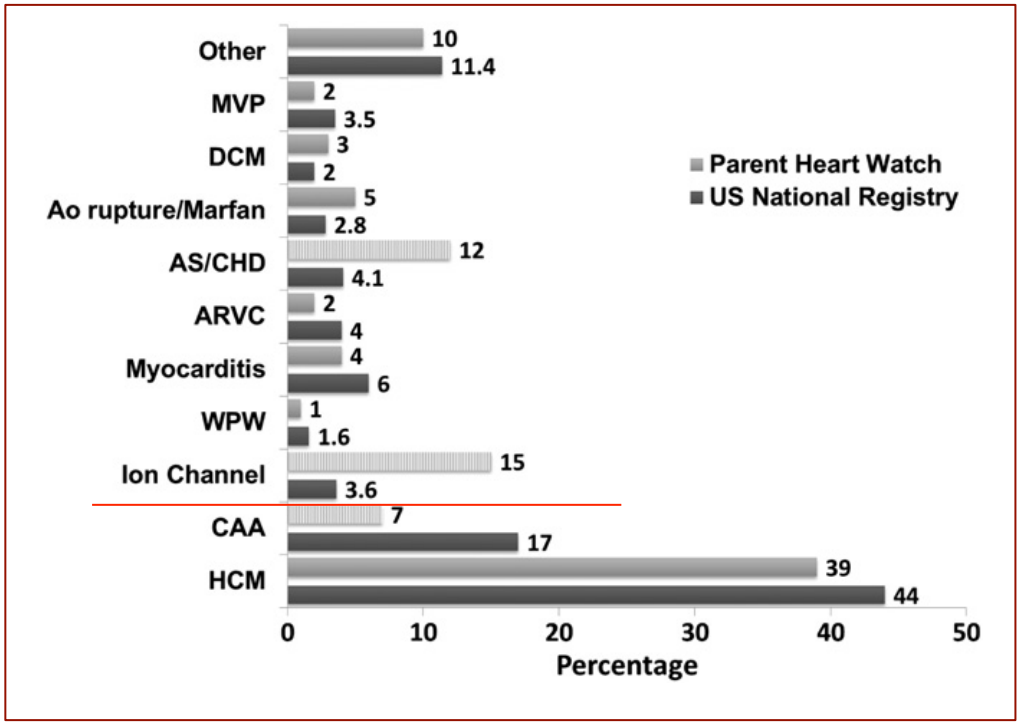
- HCM
- ARVD
- DCM
- Other CM
- Coronary artery anomalies
- Congenital heart defects

Electrical cardiac conditions

- LQTS/SQTS
- Brugada sy
- CPVT
- WPW

Others

- Myocarditis
- Marfan sy.
- drugs
- commotio cordis



V.L. Vetter / Journal of Electrocardiology 48 (2015) 316–323

II. Why genetic heart disease ?

Exercise and inherited arrhythmias

LQTS,CPVT

ventricular arrhythmias

HCM

hemodynamic/ischaemic changes

ventricular arrhythmia

ARVC

underlying disease process is accelerated by exercise

Brugada

Hyperthermia after streneous exercise ? High vagal tone ? during recovery

- *Interassociation Consensus Statement on Cardiovascular Care of College Student-Athlets, JACC 2016*
- *CC Cheung/Canadian Jpornal of Cardiology 32(2016) 452-458)*

II. Why genetic heart disease ?

- majority of SCA associated conditions have a genetic basis
- In those with a positive family history, family genetic or cascade screening can found additional affected individuals

Tan, Circulation 2005

- effectiveness of genetic screening is still a complicated issue

III. Screening ?

Purpose of screening

- to identify those potentially at risk for SCD and not to prevent SCD
- this identification allows the application of clinical guidelines and surveillance for changes in symptoms or clinical status
- disease specific prevention has to be applied to prevent the disease either from occurring or from resulting in death

III. Screening ?
Obstacles of screening SCD

- SCD is not a single disease but a potential outcome of many cardiac conditions

- SCD often occurs without warning signs or symptoms
- the first symptom in up to 50% of children will be the SCA

Corrado J Am Coll Cardiol 1997, Maron JAMA 1996

ECG detectable CV diseases

Table 4 ECG Features of cardiac diseases detectable at pre-participation screening in young competitive athletes

Disease	QTc interval	P wave	PR interval	QRS complex	ST interval	T wave	Arrhythmias
HCM	Normal	(left atrial enlargement)	Normal	Increased voltages in mid-left precordial leads; abnormal Q waves in inferior and/or lateral leads; (LAD, LBBB); (delta wave)	Down-sloping (up-sloping)	Inverted in mid-left precordial leads; (giant and negative in the apical variant)	(Atrial fibrillation); (PVB); (VT)
Arrhythmogenic right ventricular cardiomyopathy/ dysplasia	Normal	Normal	Normal	Prolonged >110 ms in right precordial leads; epsilon wave in right precordial leads; reduced voltages ≤0.5 mV in frontal leads; (RBBB)	(Up-sloping in right precordial leads)	Inverted in right precordial leads	PVB with a LBBB pattern; (VT with a LBBB pattern)
Dilated cardiomyopathy	Normal	(Left atrial enlargement)	(Prolonged ≥0.21 s)	LBBB	Down-sloping (up-sloping)	Inverted in inferior and/or lateral leads	PVB; (VT)
Long QT syndrome	Prolonged >440 ms in males >460 ms in females	Normal	Normal	Normal	Normal	Bifid or biphasic in all leads	(PVB); (torsade de pointes)
Brugada syndrome	Normal		Prolonged ≥0.21 s	S1S2S3 pattern; (RBBB/LAD)	Up-sloping coved-type in right precordial leads	Inverted in right precordial leads	(Polymorphic VT); (atrial fibrillation) (sinus bradycardia)
Lenègre disease	Normal	Normal	Prolonged ≥0.21 s	RBBB; RBBB/LAD; LBBB	Normal	Secondary changes	(2nd or 3rd degree AV block)
Short QT syndrome	Shortened <300 ms	Normal	Normal	Normal	Normal	Normal	Atrial fibrillation (polymorphic VT);
Pre-excitation syndrome (WPW)	Normal	Normal	Shortened <0.12 s	Delta wave	Secondary changes	Secondary changes	Supraventricular tachycardia; (atrial fibrillation)
Coronary artery diseases[a]	(Prolonged)	Normal	Normal	(Abnormal Q waves)[b]	(Down- or up-sloping)	Inverted in ≥2 leads	PVB; (VT);

Less common or uncommon ECG findings are reported in brackets.

QTc: QT interval corrected for heart rate by Bazett's formula. LBBB: left bundle branch block. RBBB: right bundle branch block. LAD: left axis deviation of −30° or more. PVB: either single or coupled premature ventricular beats. VT: either non-sustained or sustained ventricular tachycardia.

[a]Coronary artery diseases: either premature coronary atherosclerosis or congenital coronary anomalies.

[b]Abnormal Q waves (see Table 3).

European Heart Journal (2005) 26, 516–524

III. Screening ?
Screening methods

- H&P ± ECG ???

debate

- optimal content of the pre-participation evaluation, particularly regarding the inclusion of routine screening electrocardiograms

decisions

- regarding approaches to prevention of sudden death in athletes will be dictated largely by region-specific financial, political, and cultural factors

How to stratify the risk of sudden death in athletes/ ***in general population*** ?

AHA/ACC Scientific Statement

Assessment of the 12-Lead ECG as a Screening Test for Detection of Cardiovascular Disease in Healthy General Populations of Young People (12–25 Years of Age)

A Scientific Statement From the American Heart Association and the American College of Cardiology

Endorsed by the Pediatric and Congenital Electrophysiology Society and American College of Sports Medicine

- ECG has the capability of raising suspicion for or identifying certain genetic cardiovascular disease as true positive results, including ion chanellopathies and HCM

Circulation. 2014;130:1303-1334.)

III. Screening
12-Lead ECG as a population screening test ?

General considrations:

- Positive predictive value of a test *(proportion of true positive)* is necessarily low for disorders of low prevalence
  - *a test with nearly perfect specificity will have more false positive than true-positive responses when prevalence of disease in the population is < 10%*
  - Incidence of SCD is lower than prevalence of the disease
- Positive predictive value for disease is less dependent on test sensitivity than on its specificity
  - Standard ECG criteria should be improved
- When prevalence is low as 0,1% the negative predictive value of the test nevertheless remains high irrespective of test sensitivity although its “statistical” usefulness is questionable

Best practices for ECG screening in children

Table 2
Screening concerns, barriers, and solutions.

Concern or barrier	Solutions
Low prevalence	• Conditions are more common than appreciated but need improved identification methods.
Large numbers of youth in US	• ECG screen can be performed periodically, not annually.
Resource and manpower issues	• Multiple physician specialists and physician extenders can be utilized. • Computer technology and remote readings can be performed. • *Current computer readings are not always correct.*
Lack of infrastructure for screening	• Screening can be incorporated into current athletic preparticipation screening and well child visits. • Both a clinic-based and a community-based model can be used.
Differences in US and European populations	• Differences in prevalence of various conditions in Europe and US may relate to differences in identification. • ECG can identify many of the common conditions.
ECG sensitivity and specificity, false positives	• New ECG standards including age, gender, race and ethnicity should improve sensitivity and specificity • The ECG is a screening tool, not a definitive diagnostic tool as most screening tests require confirmation and further evaluation. • Prompt clarification of ambiguous diagnoses will relieve associated anxiety.
Disqualification	• Long-term disqualifications are uncommon. • The presence of AEDs and personnel who can perform bystander CPR helps to ensure safe playing fields which can allow some type of athletic participation for most youth.
Liability	• Families should be told the scope of screening when performed and educated that one screen is not a forever guarantee.
Costs	• Variable results have been noted, but several studies suggest cost-effectiveness per life year saved.

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III. Screening
12-Lead ECG as a population screening test ?

Screening can be valid if,

- clinically useful
- cost-effective methods can separate true positive from false positive
- society accept the costs

identification of a children with increased risk of SCD can save more than one life

(genetic or cascade screening for additional affected individuals)

Should we screen ?