

Text Size: ☒ S ☐ M ☐

Home » Information for Researchers » Workshops, Meetings, and Scientific Reports » Heart

Friday, June 18

NHLBI Working Group Screening for Sudden Cardiac Death in the Young

Executive Summary

The National Heart, Lung, and Blood Institute (NHLBI) convened a Working Group meeting on April 12-13, 2010 in Bethesda, MD to develop a research agenda identify resources to evaluate whether screening for sudden cardiac death (SCD) in the young would effectively reduce SCD and add health care value. The Working Group consisted of experts in pediatric cardiology and electrophysiology, adult cardiology, epidemiology, biostatistics, sports medicine, child psychiatry, health economics, ethics, oncology and newborn screening. The Working Group's charge is responsive to the [NHLBI Strategic Plan Goals 1, 2, and 3](#).

Discussion

Sudden cardiac death in the young has a devastating impact on families, care providers, and the community and attracts significant public and media attention. most common diagnoses that increase risk for SCD in the young include hypertrophic cardiomyopathy, coronary artery anomalies of wrong sinus origin, arrhythmogenic right ventricular cardiomyopathy, and ion channelopathies. These diagnoses have prevalence rates of 1:500 or less and are typically undetected prior to the SCD event. SCD related to these diagnoses has been documented in infancy (approximately 10% of SIDS cases may be due to ion channelopathies during competitive athletics. SCD has also been postulated to increase with stimulant use for treatment of attention deficit disorder. Some experts advocate for institution of screening programs, usually using an electrocardiogram (ECG) with history and physical examination, to identify at-risk individuals and prevent SCD in these various settings. Critics of this approach cite lack of evidence to support effectiveness or feasibility; cost implications; and the potential clinical, financial, emotional, and other consequences of the high rates of false positive screening test results.

The Working Group was unanimous in stating the importance of preventing SCD. However, there was also unanimity that more research is required to determine the best approach to achieve this goal.

The U.S. Preventive Services Task Force outlines an analytic framework that represents the chain of logic that evidence must support to connect a screening program with improved health outcomes. Currently, there is no evidence in a U.S. population that an ECG or any other screening program will reduce the incidence of SCD in any of the patient populations discussed. Further, there are limited objective data supporting the various links along the chain of logic. Specifically, gaps in knowledge exist regarding the:

- Descriptive epidemiology and etiology of SCD
- Performance of the screening methodology in the target population
- Optimal management of asymptomatic heart disease that will be discovered by an ECG screening program
- Impact of an ECG screening program on the individual, the family, the community, and society

Recommendations

The Working Group made several recommendations to address the identified knowledge gaps.

- **Epidemiology and etiology of SCD**

To prospectively define the incidence of SCD in infants, children, and young adults, the Working Group recommended a SCD registry with comprehensive ascertainment in a defined geographic region(s). Death scene evaluation, medical record review, uniform autopsy protocols, and molecular autopsy of cases with “undetermined” etiology would help define causes of SCD. A case-control study using registry defined cases and various controls (accident victims, community/school controls, and family members) would facilitate comparisons of epidemiologic, anatomic, and genetic variables. The Working Group recommended leveraging the CDC/National Center for Child Death Review infrastructure and possibly working with the National Association of Medical Examiners.

- **Performance of the screening methodology in the target population**

To test the characteristics of the ECG in target populations, the Working Group recommended a pilot ECG screening study. An adequately sized cohort would undergo ECG testing and gold standard testing for disease(s) of interest (e.g., echocardiogram for hypertrophic cardiomyopathy). Compared to the gold standard, sensitivity, specificity, and positive predictive value would be determined for the ECG. Age-specific issues would need to be addressed, including normal changes with growth and development, and changes in the ECG over time in children with target disorders. A pilot ECG screening study would also evaluate feasibility, resource requirements, reliability, reproducibility, and potential harmful effects of screening. It might also define parameters for the design of a larger clinical trial. The Working Group also recommended comparative effectiveness studies to determine the incremental value of various screening methodologies, including history and physical exam, ECG, echocardiogram, and possibly genetic testing.

- **Management of asymptomatic heart disease discovered by ECG screening**

Identification of asymptomatic heart disease would be an important byproduct of a screening program. The Working Group recommended the development of evidence-based management strategies for these asymptomatic patients. The Working Group recognized the need for novel study designs (e.g., observational studies using large HMO databases) and innovative recruitment strategies (e.g., internet-based, direct-to-consumer recruitment) for these low prevalence diseases. Potential studies might include a trial of an agent to slow the progression of mild hypertrophic cardiomyopathy or a comparative study of management strategies for asymptomatic Wolff-Parkinson-White syndrome in the adolescent.

- **Impact of an ECG screening program**

In the absence of a definitive clinical trial, decision modeling can be used to synthesize multiple data sources to evaluate overall effectiveness of a screening program. Such modeling can also determine cost-effectiveness, can project resource requirements and downstream implications, and, with the use of information analysis, can identify areas of uncertainty and prioritize the use of limited research resources. Any decision modeling, however, requires good estimates of sudden death incidence in various populations to yield useful results. The Working Group also recognized the need for research to assess the impact of a screening program on the individual and family.

Many of the studies that will be necessary to sort out the epidemiology of SCD and the efficacy of screening programs raise complex ethical and regulatory issues. Legal and ethical analysis of the acceptability of such study designs, and recommendations for the Office of Human Research Protections and Institutional Review Boards will be necessary for such studies to go forward successfully.

Publication Plans

The Working Group committee will develop a report of the meeting for publication in an appropriate professional journal.

Participating Division

Division of Cardiovascular Science

Staff Contact

Jonathan Kaltman, MD
kaltmanj@nhlbi.nih.gov

301-435-0528

Working Group Members

Co-Chairs

- Richard Friedman, MD, Texas Children's Hospital
- Paul D. Thompson, MD, Hartford Hospital, Hartford, CT

Members

- Charles Berul, MD, Children's National Medical Center
- Jeffrey Botkin, MD, University of Utah
- Joshua Cohen, PhD, Tufts University
- Nancy Cook, PhD, Harvard University
- Adolfo Correa, MD, MPH, PhD, Centers for Disease Control and Prevention
- Domenico Corrado, MD, PhD, University of Padova
- Peter Denchev, PhD, National Institute of Mental Health
- Jonathan Drezner, MD, University of Washington
- Kevin Frick, PhD, Johns Hopkins University
- Stuart Goldman, MD, Harvard University
- Darryl Gray, MD, ScD, Agency for Healthcare Research and Quality
- Mark Hlatky, MD, Stanford University
- Prince Kannankeril, MD, Vanderbilt University
- Barnett Kramer, MD, MPH, National Institutes of Health
- John Lantos, MD, University of Missouri-Kansas City
- Laurel Leslie, MD, MPH, Tufts University
- Silvia Priori, MD, PhD, New York University
- Phil Saul, MD, Medical University of South Carolina
- Michael Schoenbaum, PhD, National Institute of Mental Health
- Carrie Shapiro-Mendoza, PhD, MPH, Centers for Disease Control and Prevention
- David Siscovick, MD, MPH, University of Washington
- Victoria Vetter, MD, MPH, University of Pennsylvania
- Ben Vitiello, MD, National Institute of Mental Health

NHLBI Staff

- Diane Bild, MD, MPH
- Robin Boineau, MD
- Kristin Burns, MD
- Jonathan Kaltman, MD
- David Lathrop, PhD
- Xin Tian, PhD

Last Updated: June 2010

[HOME](#) | [SEARCH](#) | [ACCESSIBILITY](#) | [SITE INDEX](#) | [OTHER SITES](#) | [PRIVACY STATEMENT](#) | [FOIA](#) | [CONTACT US](#)



National Heart, Lung, and Blood Institute



National Institutes of Health



Department of Health and Human Services