Education and Training Subcommittee Report

Don Bailey, Chair
Beth Tarini, Co-Chair

DACHDNC MEETING
SEPTEMBER 20, 2013

Subcommittee Charge

- Review existing educational and training resources, identify gaps, and make recommendations regarding five groups:
  - Parents and the public
    - Parents
    - The public
  - Health professionals
    - Health professionals
    - Screening program staff
    - Hospital/birthing facility staff

Education and Training Subcommittee Members

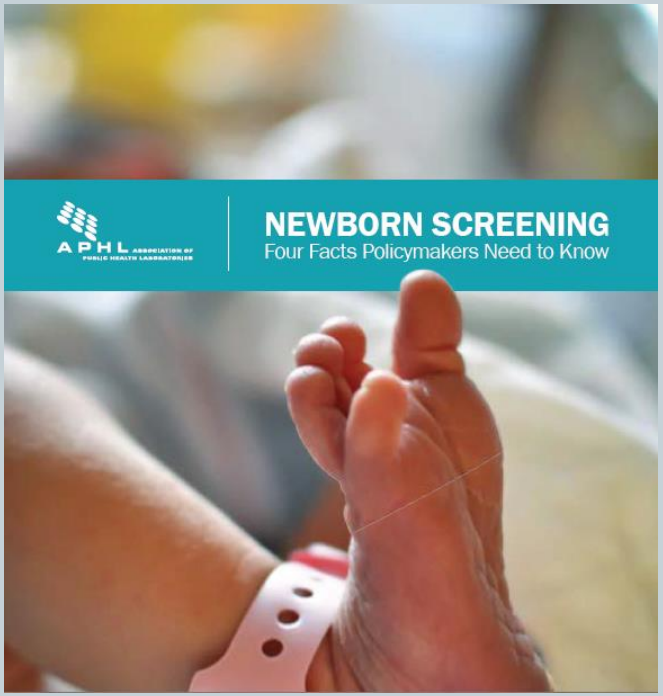
- **SACHDNC Members**
  - Don Bailey (chair)
  - Stephen McDonough
  - Joe Bocchini
  - Catherine Wicklund
  - Jeffrey Botkin
- **Organization Representatives to SACHDNC**
  - Frederick Chen (AAFP)
  - Beth Tarini (co-chair) (AAP)
  - Nancy Rose (ACOG)
  - Cate Vockley (NSGC)
  - Adam Kanis (DoD)
  - Natasha Bonhomme (GA)
  - Lisa Bujno (AMCHP)
- **Federally-Funded Grantees**
  - Joyce Hooker (Regional Collaboratives)
- **Consultant Members**
  - Emily Drake (birthing facility)
  - Jeremy Penn (parent)
  - Jacque Waggoner (parent)
  - Joan Scott (professional training)
  - Deborah Rodriquez (state lab)

Priority: Promote newborn screening awareness among the public and professionals

- **Current activities**
  - Support and provide input on the 2013 Newborn Screening Awareness Campaign plans and activities
  - Identify ongoing strategies for NBS awareness after 2013

Campaign Activities

- NBS Exhibits
- 2013 NBSGT/ISNS Meeting – May 5-10
- Website/ PSAs
- Coffee table and e-book
- Educational brochures
- Media coverage
- DC Reception and Awards Ceremony
- Social media outreach



QUESTION: What should be the focus of our post-campaign awareness activities?

- Our focus thus far has been on promoting awareness among the general public and professionals
- What is the most pressing awareness need in the next few years?

Priority: Provide better guidance for advocacy groups and others regarding the nomination and review process

- **Original Project**
  - Develop public-friendly summaries of previously conducted evidence reviews as well as evidence review nominations that have not gone forward
- **Problem**
  - The nomination and review process has evolved since the committee was first formed, and the lessons learned from earlier failures might not be as helpful as a forward looking document
- **Revised Project**
  - Prepare a public-friendly summary of the nomination and review process
  - Goal: Support future nominators in preparing successful application packages

Guidance Document Timeline

- **Original Timeline**
  - Summer, 2012 Activity proposed and framed
  - Fall-Spring, 2013 Draft documents prepared by Atlas Research
  - Summer, 2013 CRW and E&T document revision
  - September, 2013 Draft document to DACHDNC

Revised Activity

- Interview experts closely associated with the committee and familiar with the review process
- Review existing framework and guidance documents
- Prepare “snapshot” summary document based on this review and the interviews

Experts Interviewed

- Joseph Bocchini, MD, Committee Chair
- Rodney R. Howell, MD, former Committee Chair
- Don Bailey, PhD, Committee Member and E&T Chair
- Natasha Bonhomme, E&T Subcommittee member and Committee organizational representative from Genetic Alliance
- Susan Tanksley, PhD, Condition Review Workgroup member and Committee organizational representative from APHL
- Beth Tarini, MD, Committee organizational representative from AAP and E&T Subcommittee Co-Chair
- Alex Kemper, MD, Condition Review Workgroup Chair
- Nancy Green, MD, Nomination & Prioritization Workgroup and Condition Review Workgroup member
- Lisa Prosser, PhD, Condition Review Workgroup member
- Jelili Ojodu, MPH, Condition Review Workgroup member

Focus of Interviews

- Factors and/or priorities guiding the Committee;
- The importance of personal stories;
- The importance of the nomination package;
- The decision matrix;
- The condition review process;
- The importance of screening tests and how the Committee evaluates State screening capabilities;
- The importance of sufficient, high quality data;
- Understanding what the definition of “treatment” is;
- The importance of multidisciplinary teams and advocacy organizations;
- Resource recommendations

Guidance Document Timeline

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  - September, 2013 Draft document to DACHDNC
- **Revised Timeline**
  - Summer, 2013 Atlas interviews and document preparation
  - September, 2013 Review of draft document
  - September, 2013 Advocate and professional interviews
  - Fall, 2013 E&T review and re-write
  - September, 2014 Draft document to DACHDNC

Priority: Track, provide input on, and facilitate integration of national education & training initiatives

- **Project**
  - Identify one heritable condition that is not part of the RUSP and for which screening and treatment most likely would occur at a later point in child development
  - In partnership with professional and parent organizations, identify major education and training needs for that condition

Childhood Screening Prototype Review Timeline

- January, 2013 — Three exemplar conditions selected
  - -- fragile X syndrome
  - -- long QT syndrome
  - -- Wilson's disease
- May 2013 — Fragile X syndrome
- September, 2013 — Long QT syndrome
- January, 2014 — Wilson's disease
- May,, 2014 — Report to Committee

Six Questions for Each Condition

- What is the typical pattern of identification of children with this condition?
- What problems exist with the current pattern of identification, problems that could be ameliorated to some extent by earlier identification?
- Would population screening outside of the newborn period be at all feasible or desirable?
- In the absence of population screening, what could be the likely best case scenario for earlier identification?
- What level of effort would be required to substantially change the current paradigm – minimal, moderate, substantial, or heroic?
- Which stakeholder groups would need to be engaged in any discussions about altering current practice?

What is Hereditary Long QT Syndrome (LQTS)

- Inherited/genetic channelopathy
- Identified by abnormal QT interval prolongation on ECG
- Causes increased propensity to syncope, polymorphous ventricular tachycardia (torsades de pointes), and sudden arrhythmic death
- 5 genes make up the classic forms of LQTS
  - LQT1, LQT2, LQT3, LQT5, and LQT6
  - over 300 different LQTS-related mutations have been identified on these genes

Goldenberg I, Moss AJ. Long QT syndrome. J Am Coll Cardiol. 2008 Jun 17;51(24):2291-300.

Hereditary Long QT Syndrome (LQTS)

- Estimated prevalence about 1:5,000
  - Italian study of neonates cites prevalence of about 1:2,500
- Variable presentation
  - Influenced by age, genotype, gender, environmental factors, therapy, and possibly other modifier genes
  - Clinical risk in LQTS is age specific

How is LQTS Treated?

- Beta-blockers
  - First-line prophylactic therapy
  - Initiation of treatment dependent upon clinical risk
- Implantable cardioverter-defibrillator (ICD)
  - Secondary prevention
  - Primary prevention in high-risk patients

What is the typical pattern of identification?

- ECG and clinical history
- Scoring system can be used in difficult cases
- Genetic testing used largely for research, not clinical identification
  - Current genetic test identifies about 75% of individuals with symptomatic LQTS = decent specificity
  - Negative genetic test in a subject with symptomatic LQTS does not diagnosis = poor sensitivity

Hereditary Long QT Syndrome (LQTS)

- Possible presentations
  - Evaluation triggered by a syncopal event in the absence of acquired causes of QT prolongation
  - Unexplained sudden death in a young individual
  - An asymptomatic individual identified from ECG obtained for another reason
  - Positive family history
    - Identification of a family member
    - Suspicious family history

What problems exist with current pattern of identification?

- First presentation of LQTS can be sudden death

Hereditary Long QT Syndrome (LQTS)

Would population screening outside of the newborn period be at all feasible or desirable?

- Yes IF diagnosis predictive of clinical severity

Hereditary Long QT Syndrome (LQTS)

In the absence of population screening, what could be the likely best case scenario for earlier identification?

- Screening for symptoms
- Assessing family history

Hereditary Long QT Syndrome (LQTS)

What level of effort would be required to substantially change the current paradigm – minimal, moderate, substantial, or heroic?

Heroic

Hereditary Long QT Syndrome (LQTS)

Which stakeholder groups would need to be engaged in any discussions about altering current practice?

- Cardiologists
- Geneticists
- Primary care physicians
- Patients and families

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