



Education and Training Subcommittee Report



SACHDNC Advisory Committee Meeting
February 1, 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



Current E&T Subcommittee Members

▶ SACHDNC Members

- ▶ Don Bailey (chair) Catherine Wicklund
- ▶ Stephen McDonough Jeffrey Botkin
- ▶ Joe Bocchini

▶ Organization Representatives to SACHDNC

- ▶ Frederick Chen (AAFP) (DoD)
- ▶ Beth Tarini (co-chair) (AAP) Natasha Bonhomme (GA)
- ▶ Nancy Rose (ACOG) Lisa Bujno (AMCHP)
- ▶ Cate Vockley (NSGC)

▶ Federally-Funded Grantees

- ▶ Joyce Hooker (Regional Collaboratives)

▶ Consultant Members

- ▶ Emily Drake (birthing facility) Joan Scott (professional training)
- ▶ Jeremy Penn (parent) Deborah Rodriguez (state lab)
- ▶ Jacque Waggoner (parent)



Goals for January 31, 2013 meeting

- ▶ Finalize our Prototype Review statement and discuss the status of recommendations for a prototype condition
- ▶ Hear updates on CDC/APHL NBS awareness activities; provide feedback on draft NBS brochure
- ▶ Review materials prepared to date on plain language summaries of conditions already reviewed but not approved for the RUSP



Priority A: Track, provide input on, and facilitate integration of national education and 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

▶ Rationale

- ▶ The SACHDNC is charged with advising the Secretary about aspects of newborn and childhood screening....
 - ▶ Exploratory work is needed to understand the challenges inherent in future attempts to make national recommendations regarding childhood screening
 - ▶ The E&T subcommittee agreed to begin this exploration
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Approach

- ▶ Purpose: Use a “best case example” or “exemplar condition” approach as a strategy for exploring the issues, challenges, and opportunities that will arise if the committee were to consider policies or priorities for screening for a condition at some point after the newborn period.
- ▶ Not the purpose: This activity is not intended to create a policy recommendation for the exemplar condition, but rather to use it as a case study to determine whether and how the committee should approach education and training needs for childhood screening in the future.



Timeline for Childhood Screening Prototype Review

- ▶ Summer, 2012 Nominations for exemplar conditions
- ▶ September, 2012 SACHDNC feedback
- ▶ Fall, 2012 Informal rating of conditions
- ▶ January, 2013 Three exemplar conditions selected
 - fragile X syndrome
 - long QT syndrome
 - Wilson's disease
- ▶ Spring-Fall, 2013 Input from stakeholders
- ▶ Winter, 2014 Report to SACHDNC regarding lessons learned and possible next steps



Fragile X Syndrome

- ▶ Most common inherited form of intellectual disability (1:4000 – 1:5000) – single gene disorder
- ▶ Average age of diagnosis is 36 months
- ▶ Late identification means that children miss early intervention, and 25-30% of families have a second child with FXS before the first is identified
- ▶ DNA based test would identify carriers
- ▶ High rate of carriers (@1:200 females, 1:400 males)
- ▶ Carriers are at risk for late-onset disorders and perhaps other cognitive and emotional problems
- ▶ Professional organizations recommend that any child diagnosed with a developmental delay or autism be referred for FX testing, but this is not happening in any consistent way
- ▶ Research on core mechanisms have led to new generation of targeted treatments. Clinical trials currently underway for adolescents and adults, some pediatric (5 years or older)
- ▶ Newborn and infant trials will take a while to justify and initiate

Long QT Syndrome

- ▶ Disorder of heart's electrical activity, related to ion channel malfunction or deficiency
- ▶ Prevalence @ 1/3000
- ▶ Can cause sudden, uncontrollable, dangerous arrhythmia in response to exercise or stress
- ▶ >10 types, many with gene associations, plus other unknown origin
- ▶ >50% of people with untreated, inherited LQTS die within 10 years
- ▶ Treatments include lifestyle changes, dietary adjustments selective medication avoidance, beta blockers, pacemaker
- ▶ Current identification (in the absence of family history) depends on clinical symptoms
- ▶ Gene test is about 75% likely to identify mutation in patients meeting clinical criteria
- ▶ Most identified patients have an affected parent, yet not all have been recognized prior to a cardiac event in child
- ▶ Urgent treatment during the infant and newborn period probably not justified, but early identification at some point in early childhood could be important for prevention



Wilson's Disease

- ▶ Inherited autosomal recessive single-gene (ATP7B) disorder
- ▶ Well characterized molecularly
- ▶ Prevalence 1/30,000
- ▶ Copper is not eliminated properly, leading to build-up in body tissues, damaging liver and nervous system
- ▶ Symptoms begin to show by age 4, but sometimes later
- ▶ Fatal or severely disabling without treatment
- ▶ Lifelong treatment with chelation (plus possible dietary and exercise routines) can substantially alter outcomes
- ▶ Symptom-based diagnosis is challenging, long diagnostic odyssey
 - ▶ Can be difficult to differentiate from other liver diseases
 - ▶ Symptoms may gradually evolve over time



Priority B: Promote newborn screening awareness among the public and professionals

▶ **Project**

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



Newborn Screening: 50 Years of Saving Babies Lives



NBS: 50 Years of Saving Babies Lives

Purpose

- ▶ A national awareness campaign to celebrate a half-century of achievements and raise awareness about the scope and importance of newborn screening
- ▶ Sponsored by APHL, CDC and other partners



NBS: 50 Years of Saving Babies Lives

▶ AUDIENCES

- ▶ Expectant parents and their families
- ▶ HCPs—OBs, Peds, NPs, Hospital staff, Midwives, PH Clinics
- ▶ Policy Makers
- ▶ Scientists
- ▶ State and National Media



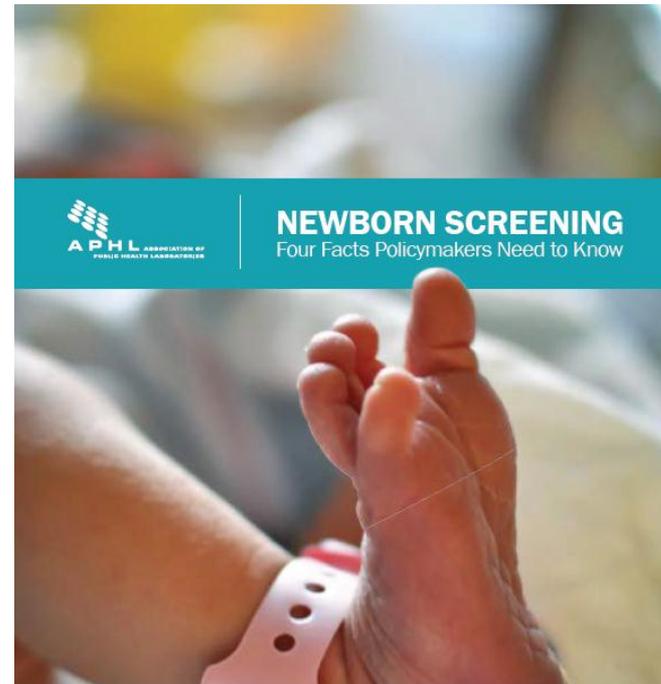
Campaign Messages

- ▶ More than 12,000 lives are saved or improved by newborn screening every year in the U.S.
- ▶ Newborn screening is perhaps the fastest, safest way to protect your baby against certain diseases and medical conditions.
- ▶ Follow up with the hospital and health care provider for your baby's newborn screening results.
- ▶ If you receive a call that your baby's results are positive, take it seriously and get appropriate follow up immediately.



Campaign Activities

- ▶ NBS Exhibits
- ▶ 2013 NBSGT/ISNS [Meeting](#)
- ▶ [Website/ PSAs](#)
- ▶ Coffee table and e-book
- ▶ Educational [brochures](#)
- ▶ Media coverage
- ▶ DC Reception and Awards Ceremony
- ▶ Social media outreach



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

▶ **Project**

- ▶ Collaborate with the Condition Review Group to develop public-friendly summaries of previously conducted evidence reviews as well as evidence review nominations that have not gone forward



Collaboration with Condition Review Group

▶ **Problems to be solved**

- ▶ Increase public transparency for what we do and the rationale for decisions made
- ▶ Support future nominators in preparing successful application packages

▶ **Activities**

- ▶ Create short, plain language summaries of evidence reviews
- ▶ Provide “blueprint” for future nominators
- ▶ Improve information on SACHDNC website
- ▶ Create a “lessons learned” case study book for future nominators



- - Report Being Developed - -

SACHDNC Decisions on Conditions Nominated for Inclusion in the RUSP

- ▶ Final version estimated to be 30+ pages and for a broad audience; contains graphics of committee processes, tables and other visuals
- ▶ Provides an overview of SACHDNC and its purpose and functions
- ▶ Details the RUSP and how conditions are reviewed for inclusion
- ▶ Presents detailed explanations of nominated conditions (2 pages per condition) that were determined by the Committee to be NOT ready for formal evidence review or the RUSP
- ▶ Descriptions are intended to educate stakeholders in how SACHDNC makes its decisions and the many reasons why a condition does NOT move forward; helping stakeholders to improve their own condition nominations
- ▶ After review by the Education Subcommittee, and other Members who were involved in reviewing a particular condition, the final document will be forwarded to the full Committee in April for approval at the May meeting