Education and Training Subcommittee

Don Bailey, Chair
Catherine Wicklund, Chair-elect
Beth Tarini, Co-Chair

SACHDNC MEETING

MAY 30, 2014

AGENDA

- Introductions and “2-minute updates” from committee members
- Final summary and next steps regarding Priorities

Priority A

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

Priority A: Project Review

- Identify heritable conditions that are not part of the RUSP and for which screening and treatment most likely would occur at a later point in child development
  - Heritable conditions were chosen to represent a variety of clinical characteristics (age of presentation, age of diagnosis, clinical morbidity, etc)
- In partnership with professional and parent organizations, identify major education and training needs for each condition

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?

Priority A: Final Steps

- Summarize major issues/themes that have emerged from this work

What is the typical pattern of identification of children with this condition?

Fragile X Syndrome	Long QT	Wilson's Disease

• Identification after clinical symptoms
  • Developmental delay
	• Incidental
	• Affected family member
	• Population screening
		• Identification after clinical symptoms
		  • Jaundice
		  • Neurological symptoms in adolescents

What problems exist with the current pattern of identification?

Fragile X Syndrome	Long QT	Wilson's Disease
• Not all children at risk are tested • Missed opportunity for evaluation • Future affected children born before index child identified	• Death before identification • Challenge with predicting clinical severity	• Variable and non-specific symptom presentation • Clinical progression and morbidity (e.g., liver damage)

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

Fragile X Syndrome	Long QT	Wilson's Disease

• Yes
• Challenge:
  • Education of clinicians
	• Yes
	• Challenge:
	  • Determination of clinical severity
		• Yes
		• Challenge:
		  • Education of clinicians
		  • Genetic testing

In the absence of population screening, what is the best case scenario for early identification?

Fragile X Syndrome	Long QT	Wilson's Disease
• Increase awareness/education about risk factors or clinical symptoms that should trigger evaluation • Panel testing after identification of clinical symptoms (e.g., developmental delay)	• Increase awareness/education about risk factors or clinical symptoms that should trigger evaluation	• Increase awareness/education about risk factors or clinical symptoms that should trigger evaluation

What effort would be required to substantially change the current paradigm?

Fragile X Syndrome	Long QT	Wilson's Disease

• Substantial
  • Education
  • Clinical access
	• Substantial
	  • Identification of clinical severity
		• Substantial
		  • Education
		  • Clinical access
		  • Genetic testing

Which stakeholders would need to be engaged in discussions about altering current practice?

Fragile X Syndrome	Long QT	Wilson's Disease
• Primary care providers • Specialists • Public health (e.g., Early On) • Patients and families	• Primary care providers • Specialists • Public health (e.g., Early On) • Patients and families	• Primary care providers • Specialists • Public health (e.g., Early On) • Patients and families

Priority B Completed

Promote newborn screening awareness among the public and professionals

Priority C

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

Priority C: Past efforts

- Revision of SACHDNC website
- Public-friendly summary document of SACHDNC process
  - Drafts reviewed
  - Interview of advocates to identify important issues
  - Next steps: continued development TBD

Priority C: Current Effort

- Development of a glossary of terms to be incorporated into SACHDNC website

Priority C: Next Steps

- Revise the glossary to appropriate reading level
- Work on implementation logistics
  - Identify appropriate location for website (e.g., SACHDNC website or Clearinghouse)

Next Steps

- Priority objectives have been completed
- Await guidance from Committee