Population-Based Carrier Screening Work Group
Interim Report

Meredith Weaver, PhD, ScM, CGC
ACMG
September 14, 2012

The NCC is funded by U22MC03957, awarded as a cooperative agreement between the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, and the American College of Medical Genetics.
Population-based carrier screening work group

- **Charge from SACHDNC, 2010 (supported by SACGHS)**
  - To engage a multidisciplinary stakeholder group using the modified Delphi process to collect and document perspectives on public health, personal health, and healthcare system readiness and needs for expanded population-based carrier screening for genetic conditions

- **End product**
  - Outline of recommendations
  - Roadmap of considerations needed prior to implementation of population-based carrier screening
May, 2010 SACHDNC approved project:
- Examine carrier screening issues and put forth guidelines for test selection and possible implementation strategies for screening.

September, 2012 reality:
- Points to consider when screening for a condition have been identified.
  - Both general to the screening process and condition specific.
  - Not currently intended to be used as a list of which conditions to screen for and when to screen.
Population-based carrier screening work group

Parameters used

- Four criteria for each issue: desirability, feasibility, importance, confidence in judgment
- Five components
  - Social issues
  - Economic issues
  - Psychological issues
  - Education and communication issues
  - Test issues
- Consensus = <20% disagree ("super majority")
- Non-consensus = >20% disagree with the majority
Results
(30,000 foot view)

Consensus

(1) Social issues:
• Consensus around the desirability to consider certain issues, sometimes consensus around the desirability and feasibility of issues
  • Including:
    • Level of detail of informed consent
    • Determine whether disparities exist in insurance coverage
    • Disclosure of conflicts of interest
(2) **Economic issues:**

- Consensus around the desirability to consider certain issues
  - Including:
    - Consider the cost of screening to the individual
    - Consider the cost to follow-up service(s)
    - Consider the cost effectiveness of the screening to the healthcare delivery system
(3) Psychological issues:

- Consensus around the desirability to consider certain issues
  - Including:
    - Whether psychological support is available
    - Understanding the psychological implications of carrier identification
    - Potential harms and benefits
(4) **Education and communication issues:**

- Consensus around the desirability to consider certain issues (to accompany screening)
  - Including:
    - Educating the public and healthcare professionals about carrier screening
    - Providing comprehensive genetic counseling
    - Engaging in shared decision making
    - Performing outreach activities
(5) **Test issues:**

- Consensus around the desirability *and* importance to consider certain issues
  - Including:
    - Robustness
    - Wide availability
    - Reducing the cost
  - Preferred timing for screening is preconception
  - Understanding the natural history of the disease
  - Frequency of a mutation (from which population?)
Non-consensus

- In general, non-consensus was found when querying the feasibility of certain issues
  - Including:
    - Determining individual perceptions of risk
    - Providing comprehensive genetic counseling
    - Non-exclusive licensing
    - Return, ownership, access and storage of carrier test results
    - Determining burden carrier screening puts on healthcare system
    - Re-testing when new information about a test or condition becomes available
Summary

- The results from the modified Policy Delphi are consistent with popular discourse on population-based carrier screening → similar issues and red flags
  - Can be related to carrier screening in general or specific to individual conditions
- General agreement existed for the desirability and (sometimes) importance of issues
- Conversely, there was little agreement regarding the feasibility of (assessing, determining, considering, etc.) an issue
Population-based carrier screening work group

Anticipated SACHDNC actions:

- Report with recommendations about carrier screening in general and criteria for specific conditions will be circulated prior to January, 2013 meeting → please review and comment
- During January, 2013 meeting there will be a vote to support the report with recommendations as a product of SACHDNC
- Determine final disposition of report
- Use the report to inform subsequent discussions
Work Group Members (29 people):

- Don Bailey
- Jeff Botkin
- Amy Brower
- Alan Fleischman
- Michelle Fox
- Mary Frederickson
- Elena Gates
- Jonathan Gitlin
- Aaron Goldenberg
- Susan Gross
- Scott Grosse
- Kathy Hassell
- Deborah Heine
- Rod Howell
- Lanetta Jordan
- Steve Keiles
- Gabriel Lazarin
- Elisa Levin
- Thomas Musci
- Shivani Nazareth
- Margaret Piper
- Tom Prior
- Mark Rothstein
- Larry Sernovitz
- Elaine Sugarman
- Judith Tsipis
- Tiina Urv
- Mike Watson
- Andrea Williams

Co-leaders: Sara Copeland (HRSA) & Meredith Weaver (ACMG)
Population-based carrier screening work group

Questions?

Meredith Weaver, PhD, ScM, CGC
mweaver@acmg.net