Update & Highlights

from the

Population-based Carrier Screening Work Group

Meredith Weaver, PhD, ScM, CGC
ACMG
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Charge from SACHDNC (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 health care system readiness and needs for expanded population-based carrier screening for genetic conditions

End product
- Report to SACHDNC will include:
  - outline of recommendations
  - roadmap of considerations needed prior to implementation of population-based carrier screening
Population-based carrier screening work group

Work group composition:

- Ethicists
- Legal experts
- Providers
- Advocates
- Consumers
- Representatives from commercial labs
Population-based carrier screening work group

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

<table>
<thead>
<tr>
<th>September 2010</th>
<th>Jan 2011</th>
<th>Feb 2011</th>
<th>Mar 2011</th>
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<tbody>
<tr>
<td>Charge given by SACHDNC</td>
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<td>In-person meeting</td>
<td>Task force organized into work group</td>
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<td>Delphi survey pilot tested</td>
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### Population-based carrier screening work group

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<tbody>
<tr>
<td>First round of Delphi survey</td>
<td>Second round of Delphi survey</td>
<td>Work group divided into breakout groups to review primary data</td>
<td>Review of primary data</td>
<td>Conference call to discuss progress and next steps</td>
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<tr>
<td>Breakout groups summarize findings</td>
<td>Breakout groups summarize members’ recommendations</td>
<td>Summaries revised/collated into recommendations</td>
<td>Report revisions</td>
<td>Report submitted to SACHDNC for comments</td>
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# Population-based carrier screening work group

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<tr>
<td>Report out for public comment</td>
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<tr>
<td>Report revised and finalized</td>
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<td>Final Report presented to SACHDNC</td>
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Population-based carrier screening work group

Progress:

- Literature review
  - Sept – Dec 2010
  - Updated by Don Bailey (Sept 2011)
- Modified Policy Delphi
  - Proven mechanism to develop reports
  - Discern areas of non-consensus
  - Two rounds: April – June 2011
    - Several areas of non-consensus
Population-based carrier screening work group

Progress:

- Examples of areas of non-consensus:
  - Social issues:
    - Issues of carrier test results including release, ownership, access, and storage
  - Psychological issues:
    - Feasibility of considering psychological implications and individual life experiences
  - Economic issues:
    - Cost-effectiveness analysis
    - Scope, purpose, and desirability
Examples of areas of *non*-consensus:

- **Education & Communication issues:**
  - Feasibility of shared-decision making
  - Feasibility of providing comprehensive genetic counseling

- **Test issues:**
  - Reporting of secondary information
  - Evolving information
  - Duty to inform/re-contact
  - Potentially (or eventually) using whole genome sequencing as the screening method
Population-based carrier screening work group

- **Modified Policy Delphi**
  - Vast majority of issues had consensus
  - Some examples:
    - Informed consent should be required
    - Feasible to consider burden carrier screening puts on healthcare delivery system
    - Important to consider cost of follow up test/procedures/actions
    - Desirable to have shared decision making
    - Desirable to have kits/reagents widely available
    - Feasible to have comprehensive science and empirical evidence available to determine clinical utility
Anticipated SACHDNC actions:

- Review report
- Comment
- Make recommendations for eliciting input from relevant populations
- Determine final disposition of report
Questions?

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