Population-Based Carrier Screening Work Group
Interim Report

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- **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

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- **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

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- **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

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- (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

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- (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

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- (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

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- (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?)

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- **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

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- **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

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)

Questions?

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