



Update & Highlights

from the

Population-based Carrier Screening Work Group

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Population-based carrier screening work group

- ◆ **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
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Population-based carrier screening work group

◆ Work group composition:

- Ethicists
 - Legal experts
 - Providers
 - Advocates
 - Consumers
 - Representatives from commercial labs
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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



September 2010	Jan 2011	Feb 2011	Mar 2011
Charge given by SACHDNC			
	In-person meeting	Task force organized into work group	
			Delphi survey pilot tested



Population-based carrier screening work group



April 2011	May/June 2011	August 2011	Sept 2011	Nov 2011
First round of Delphi survey	Second round of Delphi survey			
		Work group divided into breakout groups to review primary data	Review of primary data	Conference call to discuss progress and next steps



Population-based carrier screening work group



Jan/Feb 2012	April 2012	May 2012	June-Aug 2012	Sept 2012
Breakout groups summarize findings	Breakout groups summarize members' recommendations	Summaries revised/collated into recommendations	Report revisions	
				Report submitted to SACHDNC for comments



Population-based carrier screening work group

Oct 2012	Nov/Dec 2012	Jan 2013
Report out for public comment		
	Report revised and finalized	
		Final Report presented to SACHDNC



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*
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◆ 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
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Population-based carrier screening work group

- **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
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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
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Population-based carrier screening work group

- ◆ **Anticipated SACHDNC actions:**
 - **Review report**
 - **Comment**
 - **Make recommendations for eliciting input from relevant populations**
 - **Determine final disposition of report**
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Population-based carrier screening work group

Questions?

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