



Population-Based Carrier Screening Work Group Report

Meredith Weaver, PhD, ScM, CGC
ACMG

January 31, 2013



Population-based carrier screening work group

◆ **Brief history of the project and timeline**

- In May, 2010 Dr. Sara Copeland addressed SACHDNC about population-based carrier screening and upon request from the Committee convened an expert panel (i.e., the work group) to examine these issues more fully
 - SACHDNC agreed that HRSA should gather, analyze and present additional data on the issue, with Dr. Copeland as the project leader
 - In August, 2010 HRSA partnered with ACMG staff to administer the data collection, analysis and report preparation under the guidance of Dr. Copeland
-



Population-based carrier screening work group

- January, 2011: in-person meeting of the work group
 - April, 2011: Delphi 1st round
 - May/June, 2011: Delphi 2nd round
 - May 18, 2012: reported on **Updates and highlights**
 - Reviewed the timeline and the progress
 - September 14, 2012: **Interim report** given
 - The 30,000 foot view of results
-



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

Sara Copeland (HRSA) & Meredith Weaver (ACMG)



Population-based carrier screening work group

◆ Today's goal:

- Have a discussion about the merits of sending this report to the Secretary either as is or with further revision or reframing



Population-based carrier screening work group

◆ Highlights of the final report and its recommendations

- Areas of consensus results summarized in tables on pages 10-14
 - Mirror the well-accepted general principles of carrier screening
 - Areas of non-consensus results on pages 14-23
 - Related to evolving technologies and moving carrier screening into the public health domain
 - Recommendations on pages 30-2
-



Population-based carrier screening work group

Recommendations

(30,000 foot view)

- ◆ (1) It is desirable to consider many ***social issues***, with variation in how feasible they may be to examine further
 - ◆ (2) It is desirable to consider many ***economic issues***, with variation in how feasible they may be
 - ◆ (3) There are ***psychological issues*** to consider prior to administering population-based carrier screening
 - ◆ (4) There are ***education and communication issues*** to consider prior to administering this screening
 - ◆ (5) It is desirable and important to consider many ***testing issues***, with variation in feasibility
-



Population-based carrier screening work group

- ◆ Points to consider when conducting population-based carrier screening programs
 - Both general to the screening process and condition specific
 - Not a list of which conditions to screen for and when to screen
-



Population-based carrier screening work group

NATURE OF SUPPORT	RECOMMENDATIONS AND/OR ACTIONS
Official SACHDNC Support	Products or Projects are FORWARDED TO SECRETARY FOR CONSIDERATION/ACTION - in purview of SACHDNC and under authority of HHS Secretary
SACHDNC Affirmation of Value to the Heritable Disorders and Newborn Screening Community	Products or Projects are FORWARDED TO SECRETARY FOR INFORMATION PURPOSES ONLY
SACHDNC Acknowledgment	Products or Projects are <u>NOT</u> FORWARDED TO THE SECRETARY. ACKNOWLEDGMENT CITED ON SACHDNC WEBSITE
No Support	N/A





Population-based carrier screening work group

◆ Discussion points

- Send to Secretary in present form as an FYI
 - OR
 - Re-craft based on today's discussion and send to Secretary as an FYI
 - Are there next steps related to defining a carrier screening panel or developing appropriate capacity?
-



Population-based carrier screening work group

Questions?

Meredith Weaver, PhD, ScM, CGC

mweaver@acmg.net
