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# **SACGHS Task Force on the Oversight of Genetic Testing: Status Report**

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SACGHS Oversight Task Force  
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# SACGHS Mandate

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- To explore, analyze and deliberate on the broad range of human health and societal issues raised by the development and use, as well as potential misuse, of genetic technologies
- To make recommendations to the Secretary of HHS and other Departments upon request



# SACGHS Scope

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- Integration of genetic technologies into health care and public health
- Clinical, ethical, legal, and societal implications of new medical applications
- Research and data collection
- Patent policy and licensing practices
- Broader social applications of genetics (forensics, education, etc.)
- Emerging applications and issues



# HHS Secretary's Charge

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Undertake the development of a comprehensive map of the steps needed for evidence development and oversight for genetic and genomic tests, with improvement of health quality as the primary goal.

- Evidence of harm attributable to analytic validity, clinical validity, or clinical utility
- Distinctions between genetic tests and other laboratory tests
- Existing pathways that examine the analytic validity, clinical validity, and clinical utility
- Roles and responsibilities of involved agencies and private sector organizations



# HHS Secretary's Charge

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- Information provided by and resources needed for proficiency testing
  - *Adequacy and transparency of proficiency testing processes*
- Potential communication pathways to guide test use
- New approaches or models for private and public-private sector engagement in demonstrating clinical validity and developing clinical utility (effectiveness measures)
- Added value of revisions/enhancements to government oversight



# Previous Reports on Oversight

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NIH-DOE Task Force issued a report in 1997 on assuring safe and effective genetic testing:

- Recommended consideration of a genetics testing specialty under CLIA
- Recommended that proficiency testing be mandated for all laboratories conducting genetic testing
- Led to the formation of SACGT



# Previous Reports on Oversight

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## **SACGT Report of 2000 recommended:**

- FDA should be responsible for the review, approval, and labeling of all new genetic tests that have moved beyond the basic research phase using a novel, streamlined process
- CLIA should be augmented with specific provisions to ensure the quality of laboratories conducting genetic tests
- Data collection efforts should continue after genetic tests reach the market and CDC should coordinate public-private sector collaborations



## HHS Response (January 2001)

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- Accepted recommendations and indicated that they would be implemented over time as resources allowed
  - FDA's oversight of genetic tests to include laboratory developed tests and genetic test kits
  - Post-market data collection to be performed by CDC and might be required of the test developer and other payers
  - CMS to develop new CLIA regulations for expanded oversight of genetic testing laboratories





# Developments in 2001-2007

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- Questions raised about FDA's authority to regulate LDTs
- FDA issues guidance clarifying
  - ASR regulation
  - review requirements for laboratory developed IVDMIAAs
- In 2006, CMS halts plans to establish a genetics specialty; other measures to be undertaken instead



# CMS-genetic testing

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- Provide CMS surveyors with expert guidance to assess genetic testing labs
- Develop alternative PT mechanisms (e.g., inter-laboratory comparisons)
- Develop educational materials
- Maximize expertise of accreditation organizations
- FDA and CDC to provide guidance for review of complex analytical test validations
- Collect data on genetic testing lab performance



# Oversight Task Force (n=33)

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**SACGHS Members** -- Andrea Ferreira-Gonzalez (Chair), Sylvia Au, Kevin FitzGerald, Steve Teutsch, Marc Williams

**Ad Hoc Members** -- Amy Brower, Barbara Evans, Mark Hoffman, Kathy Hudson, Paul Steven Miller, Richard Naples, Vicky Pratt, Sue Richards, Jim Robb, Gail Vance, Ann Willey

**Federal Experts** -- Michael Amos, Linda Bradley, Joe Boone, Phyllis Frosst, Steve Gutman, Muin Khoury, Tim O'Leary, Ira Lubin, Elizabeth Mansfield, Gurvaneet Randhawa, Judy Yost

**Consultants** -- Marie Earley, Scott Grosse, Lisa Kalman, Marie Mann, Joanne Mei, Glenn Palomaki



# SACGHS Oversight Task Force Activities

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- Beginning March 2007 – Created an expanded Task Force with *ad hoc* members/consultants
- Six meetings of the full Task Force – Developed an outline for a report, discussed the report’s scope, and debated the use of key terms
- Periodic meetings of the “Steering Committee” (which consists of the five SACGHS members)
- “Chapter” meetings – Teams assigned to each chapter received writing assignments and met as needed to refine drafts



# Focus of Activity

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- Identification of Gaps in knowledge
- Discussion of Harms
  - Real harms
  - Potential harms
- Develop policy options



# Report Outline

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- Chapter 1: Background, scope of the report, spectrum of harms, overview of each chapter
- Chapter 2: Technologies used to conduct genetic tests
- Chapter 3: Analytic validity, proficiency testing and clinical validity
- Chapter 4: Clinical utility and evidence development
- Chapter 5: Effective communication and Clinical Decision support
- Chapter 6: Summary of policy changes



# Chapter 1

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- What is oversight for the purposes of this report
  - Inclusive use of term rather than strict regulatory perspective
- Genetic exceptionalism will be acknowledged as a social and policy reality, but will not necessarily drive content
- Text to be written on broad ethical issues/spectrum of harms and benefits
  - Overestimation of ‘potential harm’ may interfere with realization of benefit
- Will address harm due to ‘reductionism’



# Chapter 1

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- Will explicitly tie this in with Secretary's Personalized Health Care initiative
- Roles of different entities (e.g. regulatory agencies, government, knowledge generation agencies, provider, payer, etc.)
- Will identify issues that are peripheral to focus explicitly that will not be addressed in the report
- **Status: Rough draft with content evolving based on content of other chapters**





## Chapter 2

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- Define genetic test for the purpose of the report
  - Incorporates definitions in use
  - Will include intended use of test (examples will be provided)
- Comprehensive list of methodologies being considered
- Identify future trends
- **Status: major portion completed; further refinements in progress**



# Chapter 3

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- Most extensive content area
- Analytic validity—Proficiency Testing—Clinical Validity
- **Status:**
  - **Exploring governmental, public/private and private oversight options**
  - **Second draft revised based on September 5 meeting and breakout**



## Chapter 4

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- At present no regulatory oversight for clinical utility (and this may not be appropriate)
- No existing infrastructure
- Largest gap in realization of benefit (value)
- Biggest opportunity to build processes for improvement



## Chapter 4

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- Group has chosen to take a broad approach for identification of actionable items
- Consistent with the direction of health care in the US
  - Quality improvement
  - Evidence based best practice
  - Pay for performance



# Chapter 4

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- **Status:**
  - Viewing utility from different perspectives (Patients, Providers, Payers, Public health, Quality improvement organizations, Guideline developers, etc.)
  - Exploring governmental, quasi-governmental, private methods for the generation, synthesis and management of new evidence
  - Second draft revised based on September 5 meeting and breakout



# Chapter 5

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- Focus on effective communication
  - Pre- and post-analytic
  - Roles of laboratory, provider and patient
  - Genetic specialty vs. non-genetic specialty (provider and laboratory)
  - Direct-to-consumer



## Chapter 5

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- Focus on clinical decision support
  - Pre- and post-analytic
  - Passive vs. active
  - Incorporation of evidence-based clinical guidelines
  - Opportunity to achieve greater impact based on experience in other sectors of health care
  - Clarify how CDS will be regulated



# Chapter 5

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- **Status**

- **Earliest and farthest along in development**
- **Second draft revised based on September 5 meeting and breakout**





# Development of Policy Options

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- Will follow 9/5 meeting
- Will synthesize based on gaps and harms
- Develop within each chapter.
- Steering committee members will review, consolidate and prioritize



# SACGHS Report Timeline

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- **May-June**
- **July 9**
  - Task Force met and developed first draft
  - In-person Task Force meeting to discuss first draft; work on gaps and recs.
- **July 10**
  - Progress report to SACGHS
- **July-Sept**
  - Second draft developed
- **Sep 5**
  - **In-person Task Force meeting to discuss draft; work on policy options**
- **Sept 6**
  - TF chair preset to CLIAC
- **Sept–Oct**
  - Report revised based on outcome of Sept meeting



# Report Timeline, Cont'd

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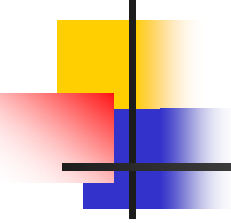
- **Sept 17**
  - TF chair presents to ACHDGDNC
- **Oct 15**
  - SACGHS reviews revised draft for release for public comment
- **Nov 5**
  - Draft released for public comment
- **Nov 19-20**
  - SACGHS devotes part of meeting to an extended comment period on oversight and a roundtable of professionals on the status of genetic education initiatives
- **Dec 21**
  - Close of public comment period
- **Dec 21-Jan 31**
  - Analysis of public comments



# Report Timeline, Cont'd

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- **~Feb 15**
  - SACGHS meets to discuss public comments and proposed revisions to draft report, reviews (approves) transmission of revised report informally to OS
- **February 18-Feb 28**
  - Final substantive edits to reflect SACGHS input
- **February 29**
  - Revised draft report submitted to OS informally
- **March 2008**
  - Final report developed
- **April 16**
  - Final review by SACGHS via email
- **April 30, 2008**
  - Final report formally submitted



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For more information about SACGHS,  
please visit:

<http://www4.od.nih.gov/oba/SACGHS.HTM>