



**HHS Priorities**  
*for America's Health Care*

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# **Update - Personalized Health Care Initiative Action Areas in Health Information Technology for Newborn Screening**

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Advisory Committee on Heritable Disorders and Genetic  
Diseases in Newborns and Children

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# PHC Initiative

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## Goal 1: Link Clinical and Genomic Information to Support Personalized Health Care

- Establish an interoperable public/private data partnership of networks to deliver information on individual medical outcomes and linking findings to genetic laboratory test.
- Establish Common Pathway for Data Integration through Electronic Personal Health Records

## Goal 2: Support the Appropriate Use of Genetic Information

- Protect individuals from genetic discrimination
- Encourage policies and practices that provide sufficient protections to consumers that genetic test information is used only for their medical benefit
- Provide oversight of genetic testing to assure analytical and clinical validity
- Standardize access policies to federally funded databases of genetic information



## PHC Initiative – Goal 1

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Link Clinical and Genomic Information to Support Personalized Health Care (PHC)-

### **Establish Common Pathway for Data Integration through Electronic Personal Health Records**

- PHC Workgroup formed through the American Health Information Community on October 31, 2006
- AHIC is the public-private collaborative that sets priorities and oversees and/or endorses HIT standards, certification, the National Health Information Network, and policies on a national level.
  - Supported through the Office of the National Coordinator for Health Information Technology
  - Seven work groups are now established involving over 100 experts and stakeholders – Population Health, Electronic Health Records, Chronic Care, Consumer Empowerment, Confidentiality, Privacy and Security, Quality, and Personalized Health Care
  - Workgroup evaluates needs and opportunities to utilize health information technology to advance personalized health care



# PHC Workgroup Overview

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Broad Charge:

**Make recommendations to the Community for a process to foster a broad, community-based approach to establish a common pathway based on common data standards to facilitate the incorporation of interoperable, clinically useful genetic/genomic information and analytical tools into electronic health records to support clinical decision-making for the clinician and consumer.**

Specific Charge:

**Make recommendations to the Community to consider means to establish standards for reporting and incorporation of common medical genetic/genomic tests and family health history data into electronic health records, and provide incentives for adoption across the country including federal government agencies.**



# PHC Vision and Priorities

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Personalized Health Care is a consumer-centric system in which clinicians customize diagnostic, treatment, and management plans

Four perspectives were identified as important to the vision

- Consumer
- Clinician
- Researcher
- Health Plan/Payer

Four priority areas across each perspective

- Genetic/Genomic Tests
- Family Health History
- Confidentiality, Privacy, and Security (CPS)
- Clinical Decision Support (CDS)

Recommendations accepted by the AHIC on July 31 to advance a PHC use case, addressing genetic/genomic tests and family health history

CPS and CDS will be discussed by the PHC workgroup this fall



## **Development of Workgroup Recommendations**

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**Background testimony to full workgroup**

**Workgroup determines if further work should be done**

**Subgroup of workgroup is formed to perform additional research and draft initial recommendations**

- Subgroup is constituted with workgroup members, senior advisors, and additional resources from communities of interest to the specific area of recommendation
- Subgroup does developmental work

**Subgroup co-chairs present recommendations to the full workgroup for comment and discussion**

**Once consensus is reached by the full workgroup, recommendations are presented to the AHIC for prioritization**



# Newborn Screening

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**During the spring visioning and priorities setting sessions, newborn screening was raised as an important category of genetic/genomic tests**

**Informational discussions have occurred throughout the summer with HRSA, ACMG, NICHD, NLM on this topic**

**First introduced for detailed discussion at the August 17 PHC workgroup meeting**



# Fostering Information Sharing for Newborn Screening (NBS)

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## Background

- Presentation by Michael Watson (ACMG) and Marie Mann (HRSA) at August 17, 2007 PHC Workgroup meeting
- Communities are well-developed (HRSA)
- Cross-cuts public health and primary care medical practice
- Provides incentives for consumer empowerment
- Clinical decision support information (ACT sheets) is available and widely accepted
- Recent RTI report to AHRQ and ONC: "Privacy and Security Solutions for Interoperable Health Information Exchange"
  - Need for secure information exchange between state/public health laboratories and public health registries
  - Complications of specially protected health information and consent requirements to provide a high level of privacy protection for sensitive health information



# Fostering Information Sharing for NBS

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## Overarching Goals for the PHC Workgroup

- Identify, develop, and encourage adoption of appropriate standards by instrument manufacturers, public health laboratories, and EHR vendors, to facilitate interoperable exchange of newborn screening test results (includes genetic, metabolic, and hearing tests)
- Ensure timely communication between state public health laboratories and newborn nurseries doing screening and immediate follow-up, and the primary care professionals and specialists who are involved in the diagnosis, treatment, and management of the infants identified
- Potential to have substantial public health benefit (coupled with national goal for standard test performance) and local interest



# Fostering Information Sharing for NBS

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## **The PHC Workgroup believes this is an area that deserves their attention**

- Newborn metabolic screens have different information needs than other genetic tests, including:
  - Ordering provider is often different than the PCP
  - Patient name can change
  - Positive screening results are often sent directly to the parents
  - Need for follow-up and confirmatory testing
  - Some positive screening results may require emergency intervention upon result reporting
  - Use of Point-of-Care devices to perform screening
  - Utility of this information for natural history and follow-up research
- Without Health IT, gathering enough information to evaluate natural history and evidence-based treatment protocols will not be possible



# Fostering Information Sharing for NBS

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## Topics of Workgroup Consideration:

- Examine the need for standards in areas of
  - Test information (LOINC)
  - Diagnostic codes (SNOMED, Medcin, etc)
  - Raw data from lab instruments
  - Cut-off values for positive/negative screens
- Tracking of qualitative vs. quantitative results to registries
- Confidentiality, Privacy, and Security concerns
- Research use of screening data
- Examine other registries (hearing screens, immunization)



# Fostering Information Sharing for NBS

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## Next Steps

- Newborn Screening subgroup formation: representation from CDC, HRSA, IHS, NICHD, NHGRI, NLM, Intermountain Healthcare, ACMG, Public Health Laboratory organizations, academia, advocacy communities
- Solicit widespread input on recommendation development from communities of interest
  - Common interests (e.g., need expertise in public health laboratories, health IT vendors, etc.) and broader community reach/input
- Foster advances in standards development and implementation for specialty laboratory health information exchange
- Examine linking test results with clinical decision support tools
- Leverage expertise and successes in AHIC recommendations and optimize use of resources
  - 2008 Personalized Health Care Use Case – reporting of genetic/genomic test results
  - 2008 Public Health Case Reporting Use Case – reporting and data sharing



## **NBS: PHC-ACHDGDNC Interactions**

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### **Input from your community should help shape the PHC workgroup's activities**

- Serve as a resource during recommendation development
- Help disseminate materials
- Outreach to stakeholders
- Inform the PHC workgroup of complimentary activities
- Discuss potential pilot projects to demonstrate the utility of HIT in this area