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

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

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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
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

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.
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

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
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.
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
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
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
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)
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
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