HEALTH HERITAGE

Leveraging the EMR for Research

Sustaining the Digital Research Enterprise
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Wendy F. Cohn PhD
Department of Public Health Sciences
The University of Virginia School of Medicine
Health Heritage V.1.0

• Originally developed in 2000 to help patients gather and assess their family health history (FHx)

• Assessed risk for 89 conditions in: oncology, endocrinology, neurology, cardiovascular and cerebrovascular disease

• Developed in response to the anticipated increase in need for FHx assessment; the intense & lengthy process of genetic counseling

• Free standing, patient-focused, with all information input by patient

• Primary care physicians were able to access the patient’s HH record (pedigree & risk recommendations) with permission
<table>
<thead>
<tr>
<th>Genome Enabled Electronic Medical Record (GenE EMR)</th>
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<tr>
<td>• Enable the efficient <strong>collection, integration, and transfer</strong> of personal medical, family and social history and new genomic and other biomedical data among EMRs and Personal Health Records at the level of the individual</td>
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<td>• Provide for delivery of <strong>decision support</strong> to providers and individuals at various points of care</td>
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<td>• Enhance continuity of care for patients and promote efficiency for clinicians through common <strong>protocol portability and automated communication</strong></td>
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<td>• <strong>Empower patients</strong> to better understand, manage, improve, and control their health, health information, and healthcare.</td>
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<td>• Facilitate both individual and population based improvements in outcomes through <strong>comparative effectiveness studies</strong> and other novel discoveries based on large scale data integration</td>
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Health Heritage Goals V.2.0

• Incorporate health-related data from multiple sources / Integrate HH with electronic medical records / Share family data among relatives
  
  – Make it easier for people to collect and maintain family health history
  – Increase completeness & accuracy

• Enhance Decision Support
  
  – Incorporate personal and genomic risk factors into family health history risk assessment and risk management recommendations
THE DOMAINS AND TEAMS OF GenE EMR

INTEGRATION/INFORMATICS TEAM
Electronic and Personal Medical Records

CONTEXTUAL TEAM
Personal and Public Privacy, Security and Ethics

COMPARATIVE EFFECTIVENESS STUDY TEAM
Comparative Effectiveness Research

EVIDENCE REVIEW/RECOMMENDATION TEAM
Genomic and Personalized Medicine
PATIENT SIGNS ON TO HH
Please wait while we check for medical record updates.

This should only take a moment.
We need some information about your family's medical history.
Click "Add" if you or any relative have had a condition listed below. Click "Remind me to come back to this page" if you are not finished yet. If you don't see the condition you're looking for, click "Show More".

Have you or any of your blood relatives ever had any of the following types of cancer?

- Breast Cancer
- Colon or Rectal Cancer
- Kidney Cancer
- Leukemia
- Lymphoma
- Pancreatic Cancer
- Prostate Cancer
- Skin Cancer
- Stomach Cancer
- Thyroid Cancer
- Uterine (or Endometrial) Cancer

Remind me to come back to this page  |  Go Back  |  Next Question
We need some information about your family’s medical history. Click "Add" if you or any relative have had a condition listed below. Click "Remind me to come back to this page" if you need assistance.

Add Relative

Choose the relative(s) who had this condition.

- Joe Patient (self) [ ]
- (father) [ ]
- (mother) [X]
- (paternal grandfather) [ ]
- (maternal grandfather) [ ]
- (paternal grandmother) [ ]
- (maternal grandmother) [ ]

Is your relative not in this list? [Add them.]

[Add Relative] [Cancel]
• HH compares data available to data needed and generates follow up questions
HH provides patients with an opportunity to ask relatives if they want to share data.
• HH decision support determines risk and develops personalized risk reports
Patient views risk report; has access to provider-facing report
What is my risk for Colorectal Cancer?

Population: High

You are at moderate risk for colorectal cancer.

Why am I at risk?

The health information you provided shows that:

- you already have been diagnosed with inflammatory bowel disease (IBD)

Inflammatory bowel disease can cause chronic inflammation of the colon and rectum. It is sometimes called IBD for short. People with inflammatory bowel disease are at higher risk of getting colorectal cancer. Two main types of IBD are ulcerative colitis and Crohn's disease. IBD is more common in people of Jewish heritage. It also tends to run in families: 10 to 25 percent of people with IBD have a first-degree relative (parent, sibling, child) with either Crohn's disease or ulcerative colitis.

What can I do?
Patient risk report is sent to her doctor’s Inbox with a pedigree
# SCIENTIFIC & TECHNICAL DETAILS

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| Information gathering & sharing              | • Generating follow-up questions  
• Ethical, legal issues in sharing among relatives                                                                                     | • Question logic  
• Technical sharing details                                                   |
| Decision support & risk algorithms           | • Evidence necessary to determine risk and recommendations  
• Decision trees                                                                                                                            | • Programming (Reverse polish notation)                                   |
| Risk Report & Behavior Change                | • “Patient friendly language”  
• Conceptual model development                                                                                                                 | • Programming to populate template  
• Markdown                                                                       |
INFORMATION FLOW BETWEEN AN ELECTRONIC MEDICAL RECORD, PERSONAL HEALTH RECORD AND HEALTH HERITAGE

Medical, Surgical, Family History, Labs

EMR
EpicCare

PHR
MyChart

Health Heritage

Risk Results, Recommendations & Pedigree

Relevant personal health history data flows to HH; Tailored risk report and pedigree flows back to provider & patient
Terminology

• HH attributes have been mapped to UMLS / SNOMED concepts
• We extracted a mapping between Epic’s diagnosis ID and SNOMED for all diagnosis records (approximately 260k), representing 36,700 unique SNOMED codes
• We used the NCBO Bioportal to identify all SNOMED “parents” for the 36k SNOMED codes in the Epic diagnosis master file
• Identified 4,700 SNOMED codes that have at least one “parent” concept that represents a HH attribute
• If a web service call for a patient’s problem list includes one of those 4,700 SNOMED codes, than it “sets” an HH attribute to “present”.
• Diagnosis mapping files and hierarchy paths from Bioportal are stored locally and need to be periodically re-created to reflect terminology updates.
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<tr>
<td>Question</td>
<td>Answer Choices</td>
<td>Question Subject(s)</td>
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<td>Have you or any of your blood relatives ever had any of the following types of cancer?</td>
<td>Y/blank</td>
<td>Pt, FDR, SDR, TDR</td>
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<td>At what age did [person] have adrenocortical cancer? If you don’t know, estimate a range.</td>
<td>(direct) AND Select Single: &lt;18, 19-35, 36-44, 45, 46-59, 60+, DK1, DK2</td>
<td>Pt, FDR, SDR, TDR</td>
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<td>Brain cancer</td>
<td>Y/blank</td>
<td>Pt, FDR, SDR, TDR</td>
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<td>Astrocytoma (What type of brain cancer did [person] have?)</td>
<td>Y/blank</td>
<td>Pt, Sibling, FDR, SDR, TDR</td>
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<td>[PREREQ: astrocytoma]] At what age did [person] have astrocytoma? If you don’t know, estimate a range.</td>
<td>(direct) AND Select Single: &lt;18, 19-24, 25-35, 36-44, 45, 46-59, 60+, DK1, DK2</td>
<td>Pt, Sibling, FDR, SDR, TDR</td>
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Information sharing among family members

• Technically feasible; socially “tricky”

• ELSI team providing guidance on how to develop sharing protocols
  – Conducted series of focus groups
  – Issue Brief underway to provide guidance to GenE EMR development team & community
  – Focus on controls - who gets information, when, how often, what information?
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Developing Decision Support

- Tree structure
  - Risk of carrying a deleterious mutation
  - Other familial risk
  - Clinical diagnoses
  - Cancer-specific risk factors
  - Risk calculators
  - Other factors associated with risk (eg. Lifestyle)
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Clinical Benefits

• Primary Care
  – Efficient way to practice standard of care; implement guidelines; saves time
  – Allows PCP to begin to practice in genomic environment
  – Acquire knowledge

• Genetic Services
  – Increase appropriate referrals; decrease inappropriate
  – Streamline process; able to increase volume; practice in genomics (DTC; whole genome sequencing)

• Patients
  – Improved care; communication
  – Potential to be basis for complete personal health record
Research & System Benefits

- Health System
  - Harness power of data to improve care; reduce costs; improve outcomes

- Research
  - “Free the data” from EMRs
  - Connects patient-oriented data, clinical data, family network data
  - Facilitates genomic (genome/phenome) research
  - Supports large scale comparative research studies
FUTURE DEVELOPMENT & RESEARCH

- **DEVELOPMENT**
  - Health Heritage is the first of many applications to begin to support data needs in clinical care and patient oriented research
  - Continued tracking of genomics risk evidence & inclusion as ready
  - Increased interoperability with multiple types of data sources (e.g. home monitoring)
  - Knowledge management

- **RESEARCH**
  - Clinical Validity
    - How well does Health Heritage predict risk? How well does Health Heritage predict development of disease?
  - Clinical Utility
    - Do patients and providers change behavior?
    - Are there improved health outcomes for patients?
    - Is the availability of HH useful for personal or clinical decision-making