Genetic data and electronic health records: a discussion of ethical, logistical and technological considerations

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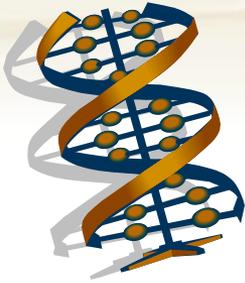
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Agenda

- Genetic Data and Electronic Health Records: A Discussion of Ethical, Logistical and Technological Considerations

  - Competing Interest Statement
  - Co-authors: N. Fost, U. Tachinardi, E.A. Mendonca
  - Example Case
  - Motivation
  - Methods
  - Insights Gained
  - Future Steps

Co-Authors and Mentors

- Norman Fost, MD, MPH
  - Professor of Pediatrics and Bioethics at the University of Wisconsin School of Medicine and Public Health, Madison, WI
  - Founder of the Bioethics Program and the Child Protection Team at the University of Wisconsin in 1973
  - Current chair of the University of Wisconsin Hospital Ethics Committee since 1984
  - Former chair of the Health Sciences Institutional Review Board for 31 years
  - Recipient of lifetime achievement awards from the American Academy of Pediatrics and the Office of Human Research Protections,
  - First recipient of the Patricia Price Browne Prize in Bioethics
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  - Fellow of the American College of Medical Informatics (FACMI)

  - Fellow of American Academy of Pediatrics (FAAP)

  - Former member of the Columbia University Medical Center Institutional Review Board

Example Case

- Asymptomatic, 57 year old male of European descent, presents to family physician requesting genetic testing for esophageal squamous cell carcinoma. Patient's brother tested positive for a genetic variant associated with an increased risk of this cancer during personal genomic testing through 23andMe (an online genomic testing agency marketed to consumers).

  - The genetic variant of concern has only been studied in Asian populations (no known increase risks in other ethnic groups)

  - At the patient's insistence, genetic consultation is performed. No further testing or endoscopy is recommended.

  - Patient calls family practice physician and geneticists two more times after final meeting requesting upper endoscopy.

Based on a case in: Sturm AC, Manickam K. Direct-to-consumer personal genomic testing: a case study and practical recommendations for "genomic counseling". *J Genet Couns 2012;21(3):402-12.*

Questions from this Case

- What is the validity and utility of the genomic test?

  - Who decides what is valid and useful?

    - Is this the same at every institution, in every setting?

  - Where can a provider quickly locate this information?

- What follow-up should be recommended?

  - Who pays for visit to the primary care provider, the genetic consultation and/or any follow-up?

  - How is the follow-up plan documented and updated if necessary (if genetic understanding changes)?

  - How is the patient updated if necessary?

More Questions from this Case

- Should results go in the patient's EHR?

  - If genomic studies are obtained, what results are put in the EHR?

    - **Where** are these results stored in the patient's record?

    - **How** are these results stored?

    - **Are results updated** if understanding (genome interpretations) change?

      - Is the **patient updated**? By whom? How often? How long?

- Should patient's family be notified of a familial increased risk of cancer if a genomic study is positive?

  - Where is a **patient's preference** documented?

  - Where would information to family members be documented in their records?
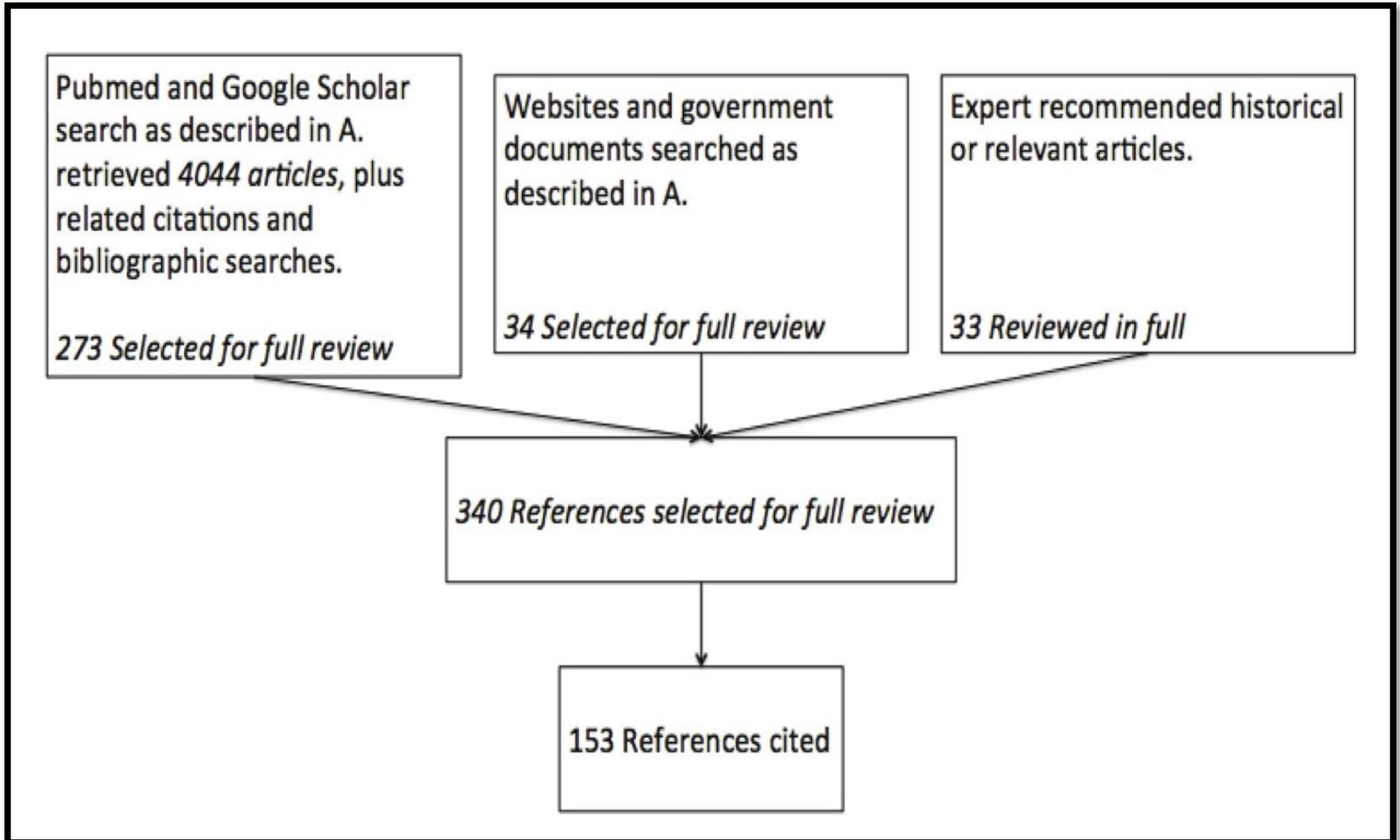
Motivation for this Project

- **Increasing genetic data**

- **Provider information overload**

  - Desire to develop comprehensive computerized genetic clinical decision support (**CDS**) tool for providers

  - Preliminary research showed

    - **Provider knowledge gaps** (CDS tool needed)

    - **Scientific knowledge gaps** (comprehensive genetic CDS tool not yet feasible)

  - **Research question: what is needed** to facilitate incorporation of genetic data into electronic health records (EHRs)?

Genetic Data and EHRs

- Methods

  - **Literature review** that included journal articles, government documents and websites in ethics, genetics and informatics as related to EHRs

  - **Interviews** with experts and vendors in EHRs, ethics, informatics, data warehousing, and members of the Electronic Medical Records and Genomics (eMERGE) Network research project

  - AMIA workshops and lectures on genotyping, EHRs, data warehousing, facilitation of research, and informatics-related ethics

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Reference Search

Pubmed and Google Scholar search as described in A. retrieved *4044 articles*, plus related citations and bibliographic searches.

273 Selected for full review

Websites and government documents searched as described in A.

34 Selected for full review

Expert recommended historical or relevant articles.

33 Reviewed in full

340 References selected for full review

153 References cited

Ethical Issues Related to Genetic Data Incorporation into EHRs

- Ethics discussions must include information on test characteristics:

  - **Analytical validity** – likelihood that reported results are correct

  - **Clinical validity** – how well the test assesses risk of health or disease (e.g. sensitivity, specificity, positive and negative predictive value)

  - **Clinical utility** – how well the test guides medical management (availability of safe and effective treatments)

Ethics discussion must include classification of genetic tests based on type of test

	Research Genetic Test	Traditional Clinical Genetic Test	Genetic or Genomic Test of Unproven Validity/Utility Ordered by Clinician	Personal Genomic Test Ordered by Consumer
Analytical Validity	Uncertain*	Good	Good	Uncertain*
Clinical Validity	Uncertain	Good	Uncertain	Uncertain
Clinical Utility	Uncertain	Good	Uncertain	Uncertain

*May or may not be CLIA certified laboratory

CLIA = Clinical Laboratory Improvement Amendments
federal oversight of quality of laboratory tests

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Ethical Issues Related to Genetic Data Incorporation into EHRs

- Potential harms of genetic testing
  - Personal
  - Social
  - Professional
  - Financial
  - Insurance problems

- Legislative acts to minimize harms related to genetic testing
  - Health Information Portability and Accountability Act of 1996 (HIPAA)
  - Genetic Information Nondiscrimination Act of 2008 (GINA)
  - Patient Protection and Affordable Care Act of 2010 (ACA)

Potential Harms of Genetic Testing

Personal	
anxiety, depression, **confusion**, changes in life plans, changes in reproductive plans, parental guilt about passing on a deleterious mutation, survivor guilt about not having a deleterious genetic mutation when other family members do, refusal of recommended medical care because of false reassurance from an invalid genetic test	
Social	
stigmatization, breach of confidentiality, misattributed parentage, privacy concerns or desire not to know a genetic result family members do desire to know, misuse of genetic data (surreptitious DNA testing or transfer of genetic data to third parties after sale of direct to consumer testing company)	
Professional	
employment and training concerns (see legislative discussion below)	
Financial	
increased use of medical consultation and follow-up tests, employment and insurance concerns (see legislative discussion below)	
Insurance	
discrimination in obtaining health, disability, life and long term care insurance (see legislative discussion below)	

Legislative Acts Relevant to Genetic Testing

Health Information Portability and Accountability Act of 1996 (HIPAA)

Sets standards on how protected health information should be controlled

Does not apply to many companies or labs that perform direct to consumer genetic testing and analysis

Protects against genetic discrimination in employer-sponsored group health plans

Genetic Information Nondiscrimination Act of 2008 (GINA)

Extends HIPAA protections by making it **illegal to use genetic information to underwrite group and individual health insurance**

Prohibits employers from making employment decisions based on genetic information

Does not address life insurance, disability insurance or long-term care insurance discrimination

Does not apply to health benefits for federal employees, members of the military, veterans seeking health care through the Department of Veterans Affairs, or the Indian Health Service

Does not apply to athletic programs

Patient Protection and Affordable Care Act of 2010 (ACA)

Prohibits health insurers from determining eligibility for coverage based on signs and symptoms of genetic disease

Changes in 2014: prohibits differences in premiums according to health status and genetic information

Proposed Health Information Technology (HIT) Actions to Minimize Harms

- **Security appropriate for the sensitive nature of genetic data**
  - Minimizing risk of breach of confidence minimizes several other harms

- **Ready access and use of validated tests**
  - Documentation of when, how, and where data is obtained
  - Easily interpretable result forms

- **Storage of genetic data not ready for EHR inclusion**

- **Informed consent**
  - Online models to assist with counseling on risks and benefits
  - Documentation of consent, patient wishes

Logistical Issues Related to Genetic Data Incorporation into EHRs

- Lack of central validation of genetic/genomic tests

- Scarcity of genetic specialists

  - Too few geneticists and genetic counselors

  - Inadequate training in medical school for most physicians

  - Inadequate support tools for providers

    - CDS tools to be used at the point of care

    - Online training to be used at provider convenience

- Inadequate understanding of genetics and statistics by laypersons and providers

Proposed HIT Actions to Assist with Logistical Problems

- Infrastructure development (knowledge bases) for validated genetic tests

- CDS tool development

- Provider online training tool development

- Online genetic and statistics courses for the public (adults and children)

Technological Issues Related to Genetic Data Incorporation into EHRs

- Multiple standards used in genetic data communication

  - Biomolecular data analysis and interpretation

  - Clinical interpretation

  - Reporting and sharing of clinical data

- Need for agreement on and use of **standardized genetic terminology in EHRs**

  - Specific standards reviewed in paper

Workflow Schematic with Example Standards
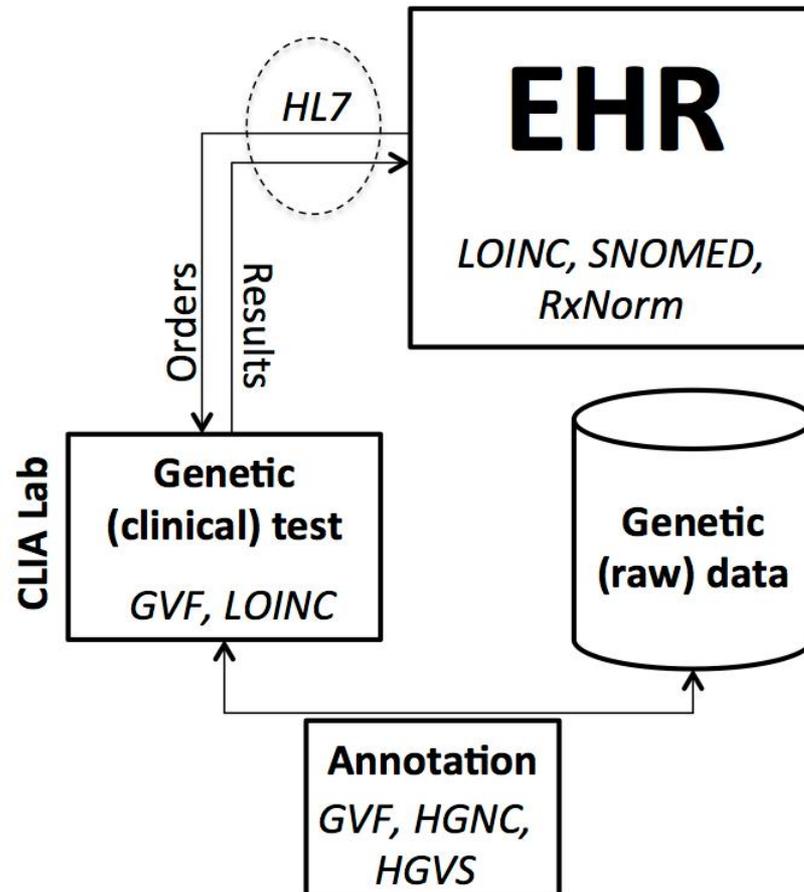
Technological Challenges Related to Genetic Data Incorporation into EHRs

- Need for methods to **store** permanent (biomolecular) genetic data and impermanent (interpreted) genetic data

  - Ability to **update** interpretations as understanding of biomolecular findings advances

  - Ability to **distinguish** validated findings from unvalidated findings

Technological Challenges Related to Genetic Data Incorporation into EHRs

- Need to **compress** genetic data and enable storage without losing information

  - No standard method for lossless compression yet

    - Options discussed in the paper

      - Masys et al: "Clinical Standard Reference Genome"[1]

      - Qiao et al: "SpeedGene"[2]

      - Vey: "Differential Direct Coding algorithm"[3]

[1]Masys DR, Jarvik GP, Abernethy NF, et al. Technical desiderata for the integration of genomic data into Electronic Health Records. *J Biomed Inform 2012;45:419-22.*

[2]Qiao D, Yip WK, Lange C. Handling the data management needs of high-throughput sequencing data: SpeedGene, a compression algorithm for the efficient storage of genetic data. *BMC Bioinformatics 2012;13:100.*

[3]Vey G. Differential direct coding: a compression algorithm for nucleotide sequence data. *Database (Oxford) 2009;2009:bap013.*

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Technological Challenges Related to Genetic Data Incorporation into EHRs

- Need to improve **computational** speed for genetic data analysis

  - Big Data challenges

  - No standard method

    - Options reviewed in paper

  - Concerns about patient privacy, security and HIPAA compliance

Technological Challenges Related to Genetic Data Incorporation into EHRs

- Need to **adapt workflows**
- EHRs currently handle genetic data as lab data
  - Genetic data is both permanent and impermanent
  - Must re-evaluate methods for:
    - Ordering
    - Storage (biomolecular data and clinical interpretation)
    - Retrieval
    - Interpretation and reporting
    - Re-Interpretation and reporting
    - Patient Notification

Future Steps
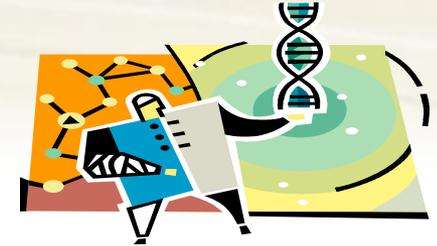
- Genetic data incorporation into EHRs will be a multi-step process requiring an international, collaborative effort to accomplish ethically guided:

  - Standards development

  - Genetic test validation

  - Infrastructure development

    - Human factors – education, training

    - Machine factors – knowledge base, CDS tools, computation, storage, retrieval, research and clinical access, security/privacy procedures

  - Workflow adaptation

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- Questions?

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