Making use of our genome for life: strategic opportunities and challenges

Marilyn D. Ritchie, PhD
Department of Genetics
Institute for Biomedical Informatics
University of Pennsylvania
http://ritchielab.org
@MarylynRitchie
@theCALMpodcast
http://marylynritchie.com/podcast
Past paradigm for using genetics in clinic*

* Different from prenatal/newborn screening
Future paradigm for using genetics in clinic

- Identify patients “at risk” for follow-up, deeper evaluation, and phenotyping
- Identify patients for genotype-guided clinical trials
- Return information to patients about their genetic risks

Physician develops prevention strategy for patient based on genetic testing results

If patient develops illness, treatment based on genetic testing
Future paradigm for using genetics in clinic

- For this to become reality, we need to have access to sequence information across a patient’s lifetime.

Mendelian Disease Risk Genes

Pharmacogenomics

Polygenic Risk Scores
Opportunities for Clinical Informatics

- EHR systems are starting to build capacity for storing genetic information as structured data
  - Example: Epic Genomics Module
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<td>Final result</td>
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**Patient Information**

- **Patient Name**: Zzzz, DAO (9330009978)
- **Sex**: Male
- **DOB**: 10/5/1959

**Result Information**

Status: Final result (Resulted: 7/16/2019)

**Order-Level Results - 07/10/2019:**
Scan on 7/17/2019 10:38 AM by Thai, Mark: Genetic Testing - Final Report

**Result Read / Acknowledged**

- Acknowledge result

No acknowledgement history exists for this order.

7/17/2019 10:38 AM - Thai, Mark

**Additional Information**

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**Lab and Collection**

CPD ORDER (Order: 397921) - 7/16/2019

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Opportunities for Clinical Informatics

- EHR systems are starting to build capacity for storing genetic information as structured data
  - Example: Epic Genomics Module

- With Clinical Decision Support (CDS) built to disseminate the relevant genetic sequence information, healthcare providers across the system can find and use the information
  - Enormous potential to use genetic information outside of Medical Genetics Clinics
  - More on this topic in “Challenges”

- Through Health Information Exchanges (HIE), this relevant genetic information can be shared across health systems → following the patients wherever they go
  - More on this topic in “Challenges”

- As more knowledge is gained in Genomic Medicine, informatics systems can push annotation updates to patient records based on the genetic sequence data stored
  - More on this topic in “Challenges”
Challenges to overcome

**Clinical Decision Support**
- Need to build individual genomic indicators with CDS
- Need to update regularly
- Need to educate providers across the system (current medical education does not cover genetics extensively)

**Health Information Exchange**
- This *could* provide the infrastructure to have important genetic information follow patients throughout their healthcare journey
- Need genetic data to be shared—currently not always part of HIE
- Need to educate providers across systems

**Genome Annotations**
- Knowledge about genome function changes regularly
- Need to update annotations and interpretations
- Need infrastructure to alert providers
Challenges to overcome

- **Compute limitations**
  - Cloud storage costs and egress costs are high
  - These costs may exacerbate health disparities

- **Phenotype ontology/standards**
  - HPO (or any ontology) is not well supported in EHR
  - Broad ontology adoption is lacking

- **Genomic annotation standards**
  - There are great standards for genomics (e.g., GA4GH)… but they are poorly adapted to broad clinical standards yet
  - Similarly true for functional annotations
Future Needs: A strategy to address gaps and barriers