Novel and Disruptive Opportunities in Genomic Medicine

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Interpreting genetic information in EHR
How can we keep clinicians and patients up-to-date with genomics?

**Genetic Data Sources**
- eMERGE Core Labs
- External Reference Labs
- Internal Hospital Labs
- DTC Sources

**ClinVar**

Genetic test results come from many sources

Knowledge is evolving

**eMERGE IV areas of opportunity:**
- Build on standards for structured reporting of genetic test report content, working with ALL sources of genetic data.
- Develop approaches for determining when and how to update genetic knowledge and alert physicians and patients.

**App for Genetic Knowledge**
(Ancillary System integrated with the EHR)

- Alerts to physicians
- Alerts to patients if physicians are non-responsive

**Opportunity for ClinGen-eMERGE Partnership**
A future model for real-time use of genomic data to improve clinical care?

- Expert Interpreted Pathogenic variant
- Immediate clinical test order
- Suspicious or novel variant
- Consider ordering a test depending on clinical picture
- Care based on symptoms
- No variants
- Order genetic analysis only if likelihood of genetic etiology high

**Symptoms**

**Family History**

EHR with Genome data (VCF File)
Limitation: Only reportable (Pathogenic and Likely Pathogenic) variants from a limited set of genes are integrated into the EHR.
Current eMERGE Workflow – Variant Interpretation

Site 1
Site 2
Site 3
Site 4
Site 5
Site 6
Site 7
Site 8
Site 9

ClinVar
PubMed

Public knowledge sources only

Variant Interpretation by eMERGE CSGs

Limited patient data shared (e.g. indication for testing)
Can we support real-time access to populations of individual level phenotype data to inform genetic variant interpretation?

**Needed:**
1. Better common and rare disease phenotyping data in EHR
2. Ability for patient to contribute phenotype data
3. Infrastructure for labs and clinicians to access aggregate (and individual level) patient data from many sources
Opportunity for eMERGE IV – Integration of Genomic Data

Can we develop approaches to make a patient’s entire genome accessible for real-time decision making in the clinical care setting?

Needs:
1. Improve standardized data models for genomic data (vcf file)
2. Quality standards for which variants are brought in and how we qualify limitations (coverage/variant type detection)
3. Decision logic for using uninterpreted data
Novel Sources of Data for Genomic Medicine
Novel data sources for genomic medicine

- DTC genomic test results
- Social media and networks crowdsourcing
- Environmental variables
  - Patient reported (family history, surveys, medication adherence)

API
Big (‘omic’) data linked to the EHR phenotypes

- Microbiome
- Genome
- Epigenome
- Transcriptome
- Metabolome
- Lipidome
- Proteome

Chain of processes:
- Data warehouse
- Big data analytics
- Discovery & Implementation

Integration of Big (‘omic’) data with EHR phenotypes.
Impact on Stakeholders

Patients/Participants
Public
Providers
Payors
Patients

- Perception of genetic results, -ve, risk scores vs pathogenic variants
- ROR new methods, GC vs non GC
- Match attitudes, beliefs to actions
- Improve genomic literacy
- Health disparities, underserved communities
- Attitudes to sharing with family members, family discussion
- Use EHR to facilitate family sharing HIPAA?
- Novel methods to facilitate family sharing
Patient-centered data governance

Health care institution centered

Input from patient, community or advocacy groups

Patient centered data governance

Portability/Storage/Security of data

Genome in an APP

patientslikeme

myresults.org
Tier 1 genomic disorders
- Familial Hypercholesterolemia
- Colorectal cancer
- Breast cancer

Health Information Exchanges

State Public Health Programs
- CDC
- FQHCs
Providers and payors

Providers

- Burden of interpretation
- Complexity, Education needs
- Views on CDS, Apps (Q & Q)
- ‘Versioning’
- Medical uncertainty

- CDS for genomic medicine
- Knowledge resources
- Shared decision making

Payors

- Cost of genetic testing
- Coverage and reimbursement
- Variable test quality
- Cascade screening

- RCTs to demonstrate clinical utility and cost effectiveness
- Economic modeling and forecasting
Thank you

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