

Enhancing Functionality of EHRs for Genomic Research, Including E- Phenotyping, Integrating Genomic Data, Transportable CDS, Privacy Threats

Genomic Medicine 8 meeting

Alexa McCray

Christopher G Chute

Rex L Chisholm

Questions to be Addressed

- 1) importance of the topic and potential impact for genomic medicine implementation;
- 2) focus and related programs currently addressing the topic;
- 3) gap areas or opportunities for addressing it more fully;
- 4) potential synergies across programs; and
- 5) training opportunities or needs.

Use Case I : eMERGE

- Show value for discovery
 - Accelerate gene to phenotype association
 - Introduce high-throughput phenotyping
 - Demonstrate phenotyping portability
- Show value for implementation
 - Integration of actionable genomic data into EHR
 - Deploy reusable decision algorithms (CPIC)
 - Demonstrate scalable knowledge integration

Use Case II:

Undiagnosed Diseases Network (UDN)

- Show value for discovery
 - N-of-1 -> N-of-many
 - Prospective deep phenotyping correlated with genotype
 - Patient voice
- Show value for implementation
 - Broad sharing of de-identified data
 - Standard data collection across sites supported by research tools
 - Central IRB; Formal agreements for sharing identifiable data within Network; Strong information security framework

Focus Groups Currently Pursuing

- Integrate sequence data into patient care:
 - eMERGE, IGNITE, CSER
- Incorporate actionable variants into EMR, develop CDS
 - eMERGE, IGNITE, CSER
- Develop electronic phenotypes
 - eMERGE
- Define and share processes of implementation, sustainability
 - UDN, eMERGE, IGNITE, CSER,

Gap Areas / Opportunities for improving

- Standards (clinical and genomic)
- Interoperability
 - APIs (FHIR)
- Sustainability
 - Long-term storage of and access to the data
- Consents for using and sharing EHR data for research purposes
 - Encapsulated with data

Synergies Across Programs

Objectives Related to Genomic Medicine Implementation	Focus Programs										Related Programs							
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GA4GH	GAPH	IOM	ISCC	LSAC	PAGE	PhenX	
Improve genomic diagnostic methods	X	X	X						X									
Facilitate research in undiagnosed and/or Mendelian diseases	X		X						X									
Expand scale of genomic data available in newborns		X																
Advance understanding of disorders of newborns		X																
Research ethical/legal/social issues in genome sequencing		X	X										X					
Interpret sequence data in variety of clinical contexts			X	X														
Integrate sequence data into patient care			X	X	X								X					
Incorporate actionable variants into EMR, develop CDS			X	X	X								X					
Educate clinicians and patients on genomics in clinical care			X	X	X			X		X			X	X			X	
Develop electronic phenotypes				X													X	
Identify variants related to complex traits	X		X	X								X			X	X		
Characterize pharmacogenetic variants and use in care				X	X					X								
Assess outcomes of using genomic information in clinical care				X	X							X						
Assess penetrance of potentially actionable variants				X														
Translate implementation outside highly specialized centers					X	X												
Define and share processes of implementation, sustainability	X		X	X	X		X	X					X					
Share genotype/phenotype info through open databases						X					X							
Standardize clinical annotation and interpretation						X											X	
Improve understanding of variation in diverse populations				X		X										X		
Assess action ability of genes and variants for clinical use						X				X								
Identify, address barriers to genomic medicine implementation				X			X	X										
Promote interaction and collaboration, reduce duplication	X					X	X	X	X		X		X	X				
Serve as clearinghouse, knowledge base for genomic medicine						X		X		X								
Use genomics to enable new drug development												X	X					
Create genomics-enabled learning health care systems				X	X								X					
Develop evidence base for clinical use of novel diagnostics												X						
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GA4GH	GAPH	IOM	ISCC	LSAC	PAGE	PhenX	

Training Opportunities

Existing:

- NLM Informatics Fellowship and Training Program
- Training for Clinical Informatics sub-specialty
- BD2K training efforts

Needs:

- Improve pipeline of math and coding training in graduate school and even high schools

Discussion Questions

- While individual projects might agree on data standards specific to their needs, how do we plan for and *promote* large-scale data sharing across projects and beyond?
- How can we leverage data in existing institution-specific EHR's to ease the task of data collection and data comparability for research purposes?
- Is it sufficient to incorporate summary level genetic test result data in the EHR, or are raw data needed?
- How do we design clinical decision support systems to ease the clinician's task in interpreting genetic test results?
- How do we represent novel genomic findings consistently to fit within decision-support algorithms?