Novel and Disruptive Opportunities in Genomic Medicine

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Reaction: Carol Horowitz, Matt Might

Summary: Dan Masys

Interpreting genetic information in EHR

How can we keep clinicians and patients up-to-date with genomics?

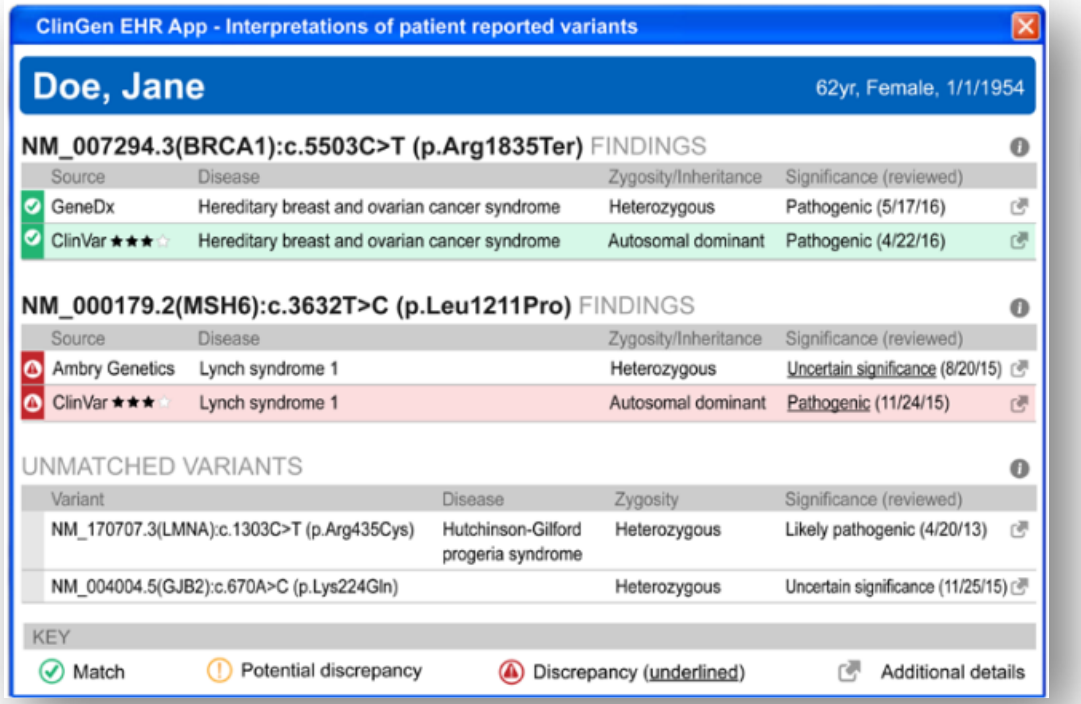
Genetic Data Sources
eMERGE Core Labs
External Reference Labs
Internal Hospital Labs
DTC Sources

Genetic test results come from many sources

ClinVar

Knowledge is evolving

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



Alerts to physicians

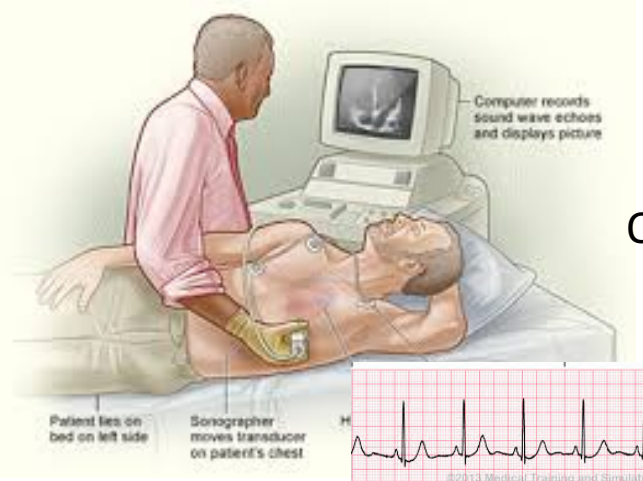
Alerts to patients if physicians are non-responsive

eMERGE IV areas of opportunity:

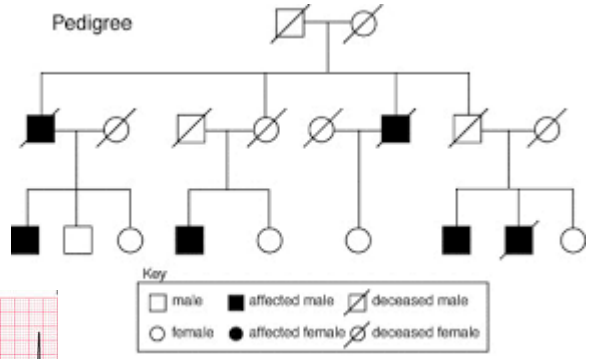
- Build on standards for structured reporting of genetic test report content, working with ALL sources of genetic data
- Develop approaches for determining when and how to update genetic knowledge and alert physicians and patients

Opportunity for ClinGen-eMERGE Partnership

A future model for real-time use of genomic data to improve clinical care?



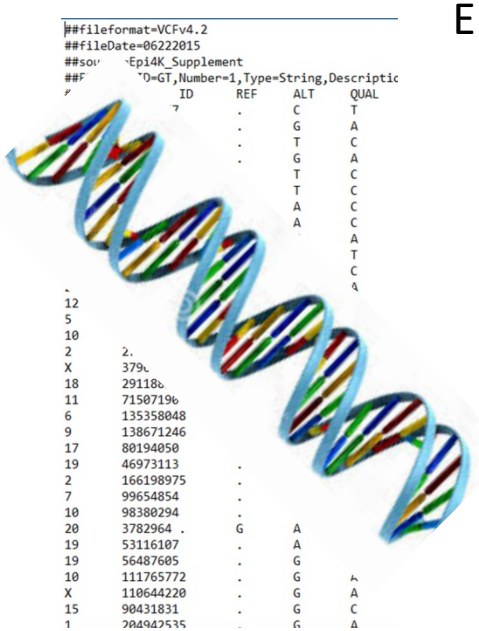
OR

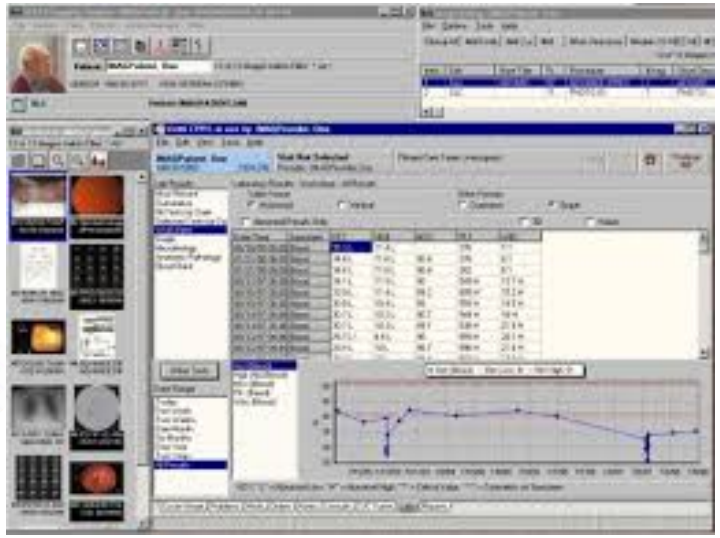


Family History

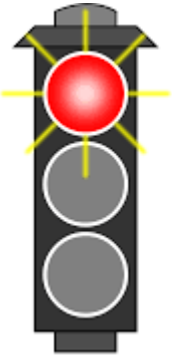
Symptoms

EHR with Genome data (VCF File



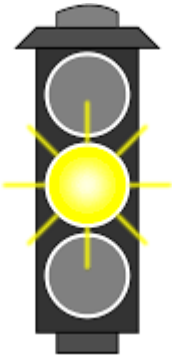


- Expert Interpreted Pathogenic variant



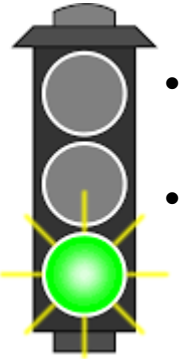
- Immediate clinical test order

- Suspicious or novel variant



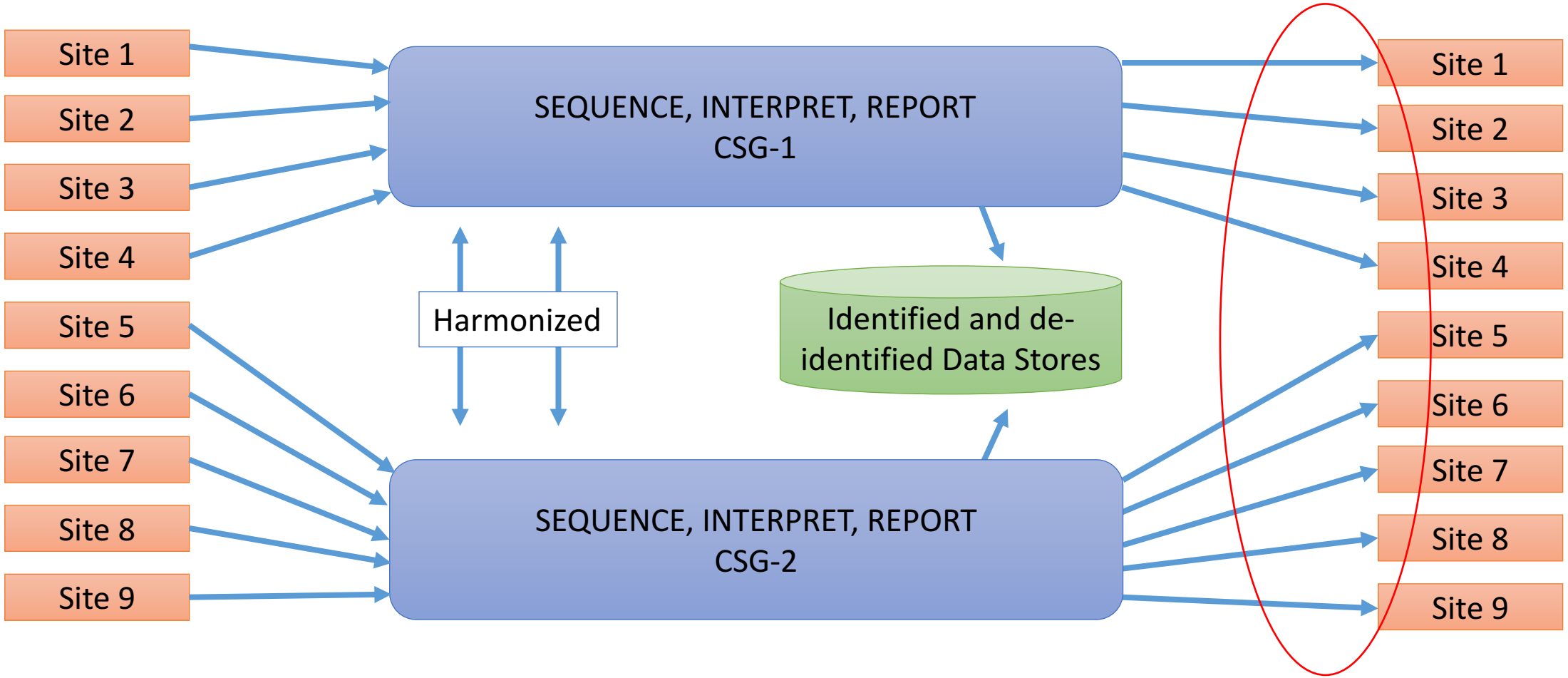
- Consider ordering a test depending on clinical picture

- No variants



- Care based on symptoms
- Order genetic analysis only if likelihood of genetic etiology high

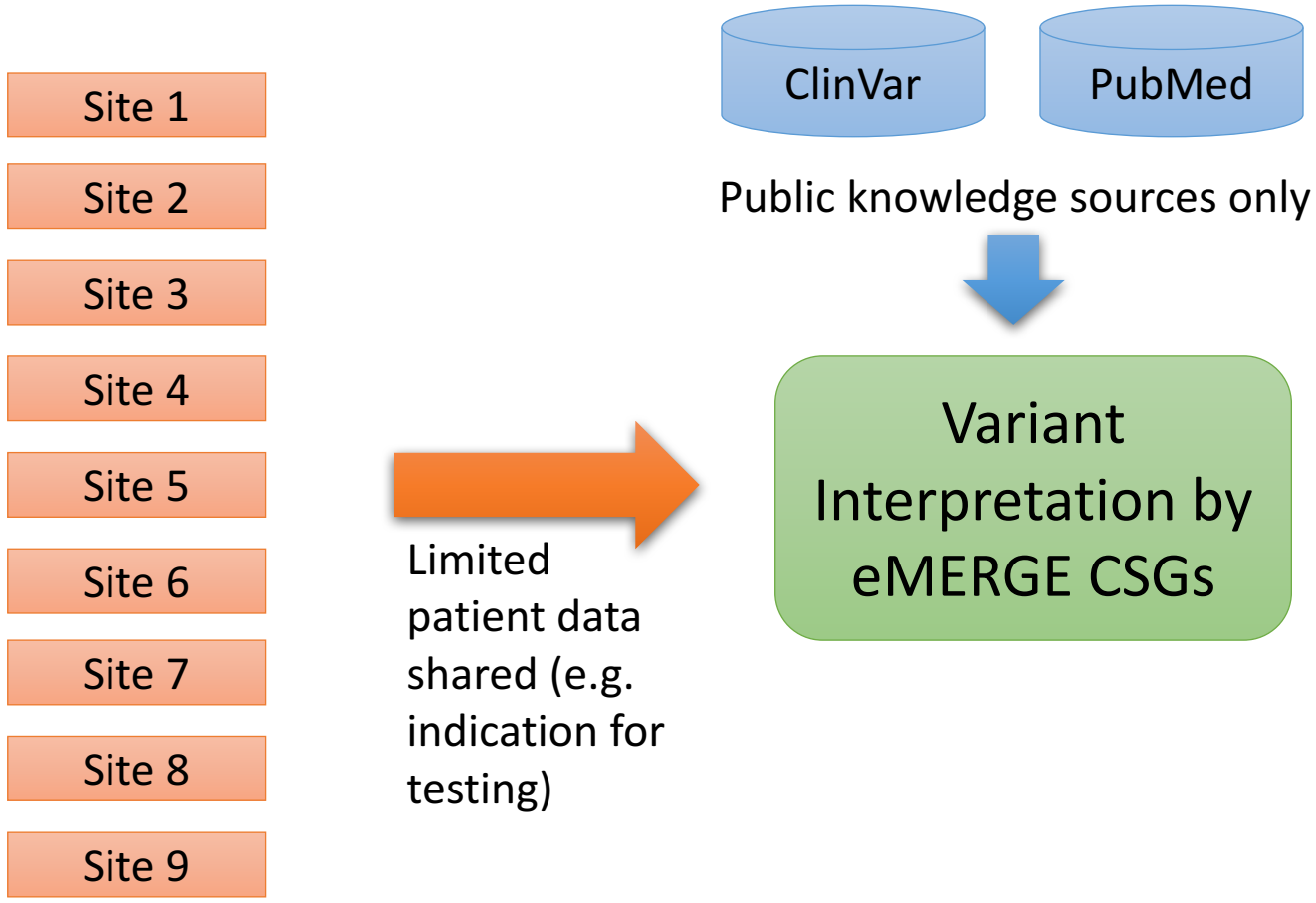
Current eMERGE Workflow – Data Delivery



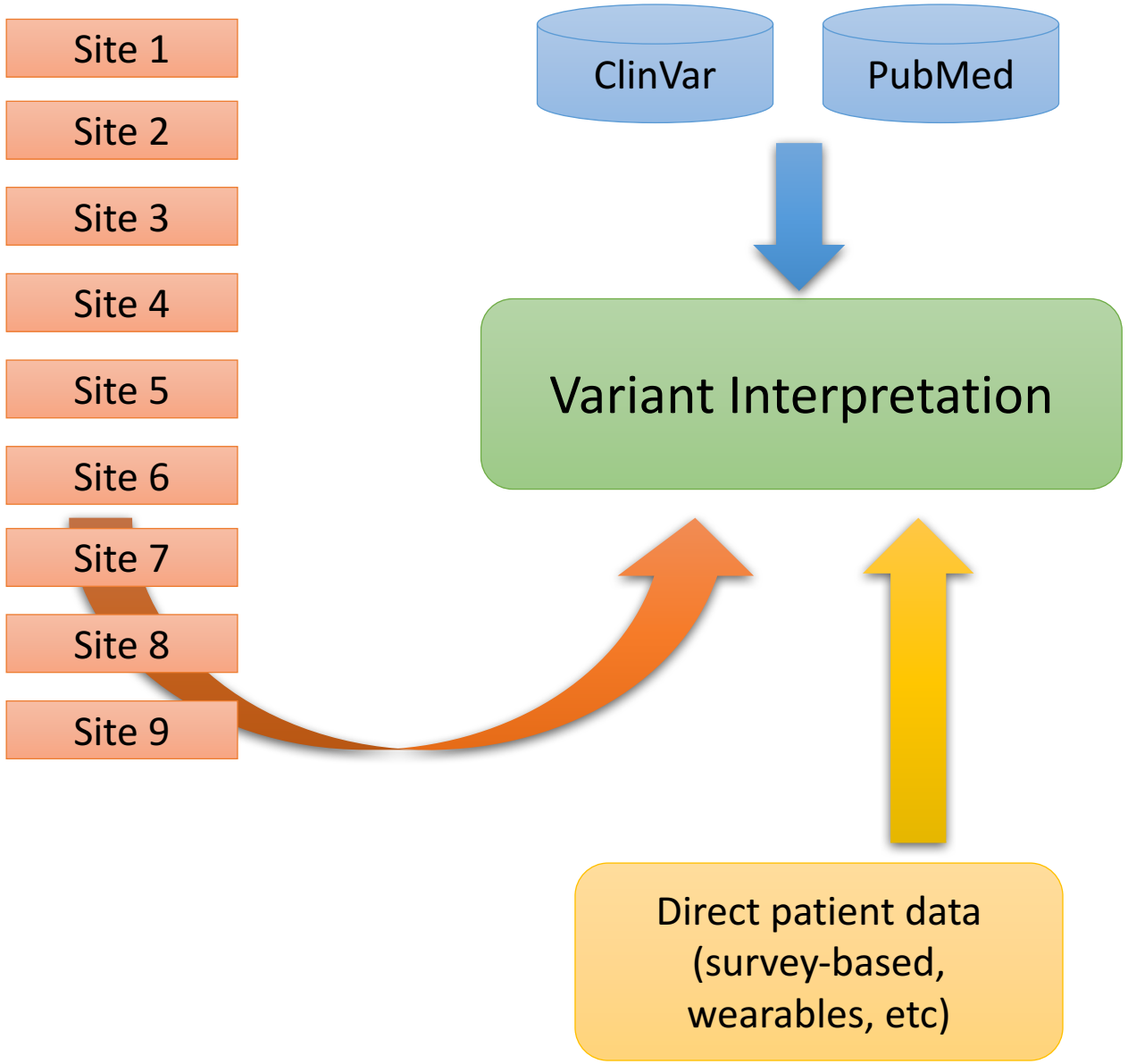
Limitation: Only reportable (Pathogenic and Likely Pathogenic) variants from a limited set of genes are integrated into the EHR

Many custom processes for data integration

Current eMERGE Workflow – Variant Interpretation



Opportunity for eMERGE IV – Variant Interpretation

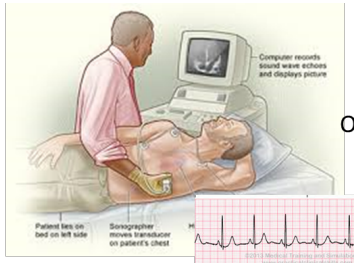


Can we support real-time access to populations of individual level phenotype data to inform genetic variant interpretation?

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



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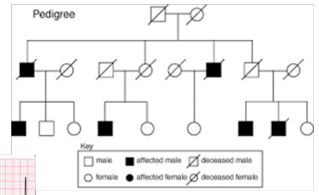
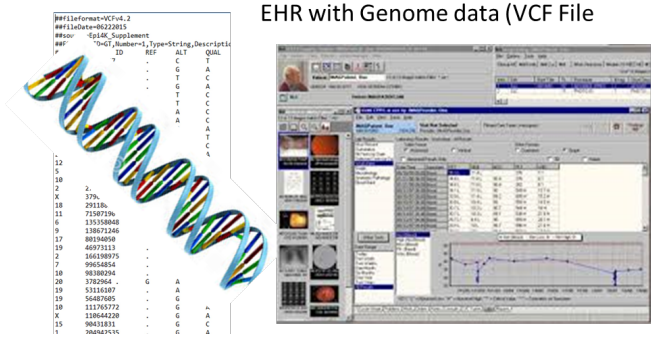
OR



Family History

EHR with Genome data (VCF File



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

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

Needs:

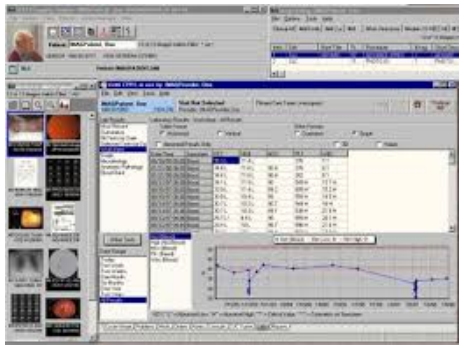
1. Improve standardized data models for genomic data (vcf file)
2. Quality standards for which variants are brought in and how we qualify limitations (coverage/variant type detection)
3. Decision logic for using uninterpreted data

Novel Sources of Data for Genomic Medicine

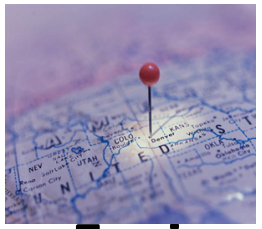
Novel data sources for genomic medicine

DTC genomic test results

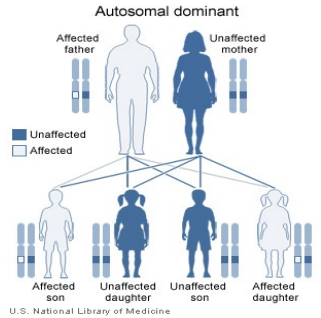
API



Social media and networks crowdsourcing

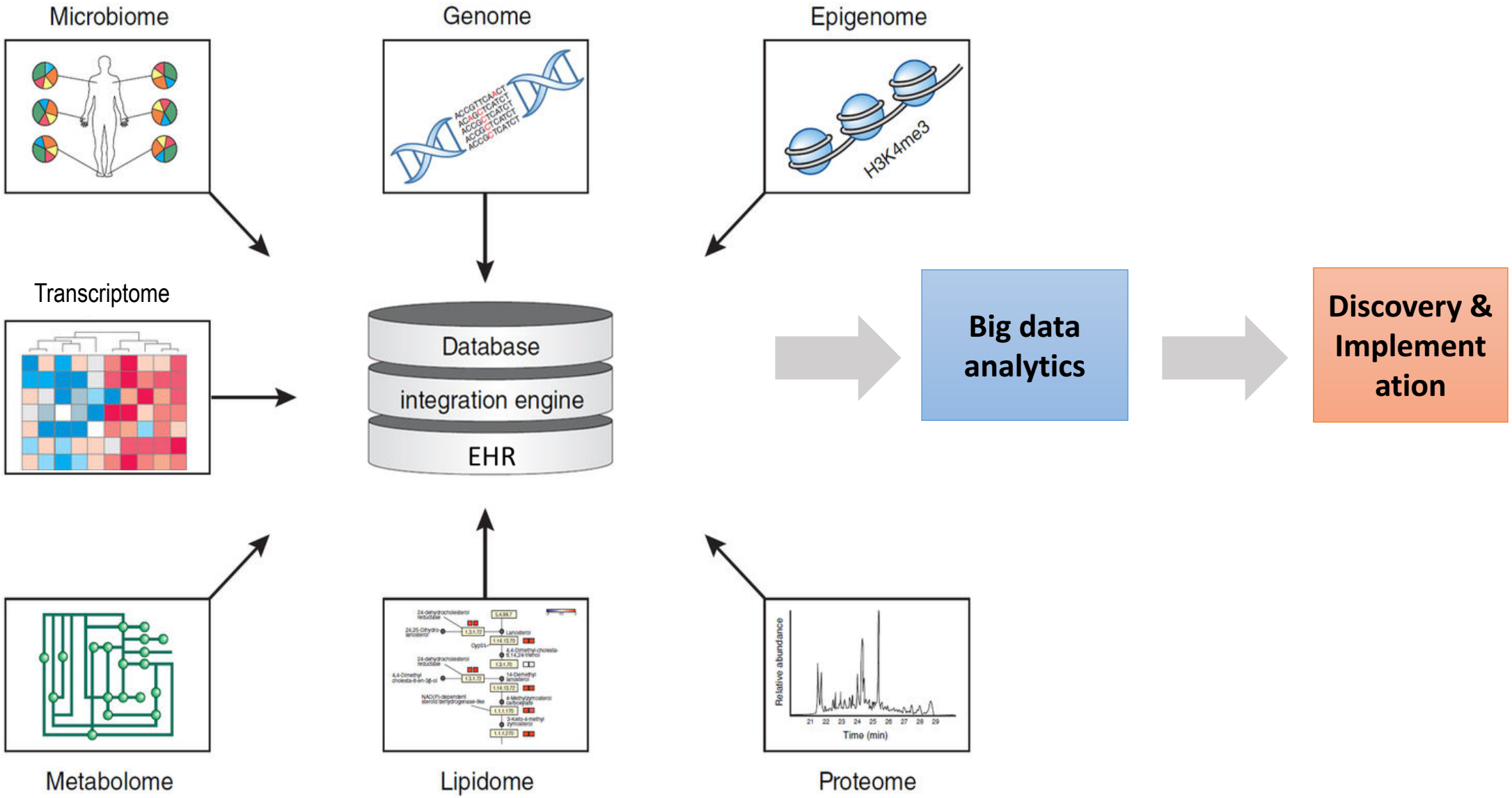


Environmental variables



Patient reported (family history, surveys, medication adherence)

Big ('omic') data linked to the EHR phenotypes



Impact on Stakeholders

Patients/Participants

Public

Providers

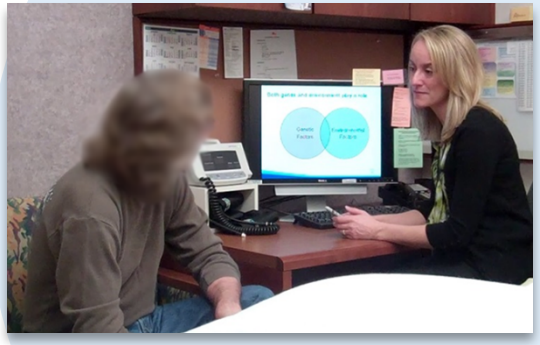
Payors

Patients

Perception of genetic results, -ve, risk scores vs pathogenic variants

Improve genomic literacy

Attitudes to sharing with family members, family discussion



ROR new methods, GC vs non GC

Use EHR to facilitate family sharing HIPAA?

Match attitudes, beliefs to actions

Novel methods to facilitate family sharing

Health disparities, underserved communities

Patient-centered data governance

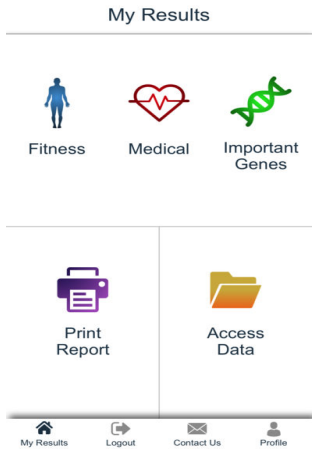
Health care institution centered

VS

Input from patient, community or advocacy groups

Patient centered data governance

Portability/Storage/Security of data



Genome in an APP

patientslikeme

FH FOUNDATION

myresults.org

Public Health Genomics

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

Chunhua Weng
Josh Peterson
Dan Masys
Rex Chisholm
Marc Lichtenfeld
Matt Might