Genomics and the Electronic Health Record

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GENOMICS

Using genetic data to inform medical decisions across the entire continuum of care
PRECISION MEDICINE

- Family History
- Genetics
- Microbiome
- Social Factors
- Environment

TREATMENT A

TREATMENT B

TREATMENT C
Learning Health System

Care

Evidence

Science
Determine Testing

- Gather info
- Plan
- Educate
- Consent

Testing Methods

- Cytogenetic
- Molecular
- Next Gen. Sequencing
  - Alignment
  - Variant Calling
  - Variant Annotation
  - Determine Clinical Utility

- FastQ
- BAM
- VCF

Analysis and Application

- Reconcile
- Plan
- Engage

Family Hx
Phenotype
Environment

Genotype
Knowledge
Learning Health System
Unprecedented GWAS research

The RPGEH then combined the genetic data with information derived from Kaiser Permanente’s comprehensive longitudinal electronic medical records, as well as extensive survey data on participants’ health habits and backgrounds, providing researchers with an unparalleled research resource. These data form the basis of genome-wide association studies (GWAS) that can look at hundreds of thousands to millions of SNPs at the same time in relation to many different health conditions.

“The GERA cohort represents the largest number of people — of any age — with genetic, health and environmental data to be deposited in dbGaP,” said Hodes in his announcement. “New approaches to genomics were developed for this project and I’m pleased that it’s ready for researchers’ use in the dbGaP database. I look forward to new insights that such a unique resource might offer for better health with age.”
Models

Where are the DATA?

Where is the TESTING?

Treatment recommendation

Web Services

Epic

...with the patient at the heart
Epic’s Research Community
Clinical Pharmacogenetics Implementation: Approaches, Successes, and Challenges

KIRSTIN W. WEITZEL, AMANDA R. ELSLEY, TAIMOUR Y. LANGAEE, BENJAMIN BURGESS, DAVID R. NESSEL, ANIWKA OWUSU OBEN, BENJAMIN J. STALEY, HUI-JIA DONG, ROBERT W. ALLAN, J. FELIX LIU, RHONDA M. COOPER-DEHOF, R. DAVID ANDERSON, MICHAEL CONLON, MICHAEL J. CLARE-SALZLER, DAVID R. NELSON, AND JULIE A. JAFFE
“…testing enabled [U of Florida] to virtually eliminate the major cardiovascular (CVD) events that can occur when patients with the genotype are given clopidogrel.”
# Use Case Types

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<tr>
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<th>Use Case Types</th>
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<tbody>
<tr>
<td>1</td>
<td>Incorporating Genetic Results into EHR User Interfaces</td>
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<tr>
<td>2</td>
<td>Adding genetic tests in order sets</td>
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<tr>
<td>3</td>
<td>Clinical Decision Support (CDS) identifies when a test should be ordered (pre-test alert*)</td>
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<tr>
<td>4</td>
<td>CDS identifies when a drug order is inconsistent with a test result (post-order alert*)</td>
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* Note pre and post order status refers to the status of the test order as opposed to the drug order
Establishing Connectivity and Pharmacogenomic Clinical Decision Support Rules to Protect Patients Carrying HLA-B:5701 and TPMT Variants

An Implementation Guide
Challenges

Data exchange standards
Validated knowledge
Patient preferences and consent
Patient and provider education
Reimbursement
International considerations

These are international!