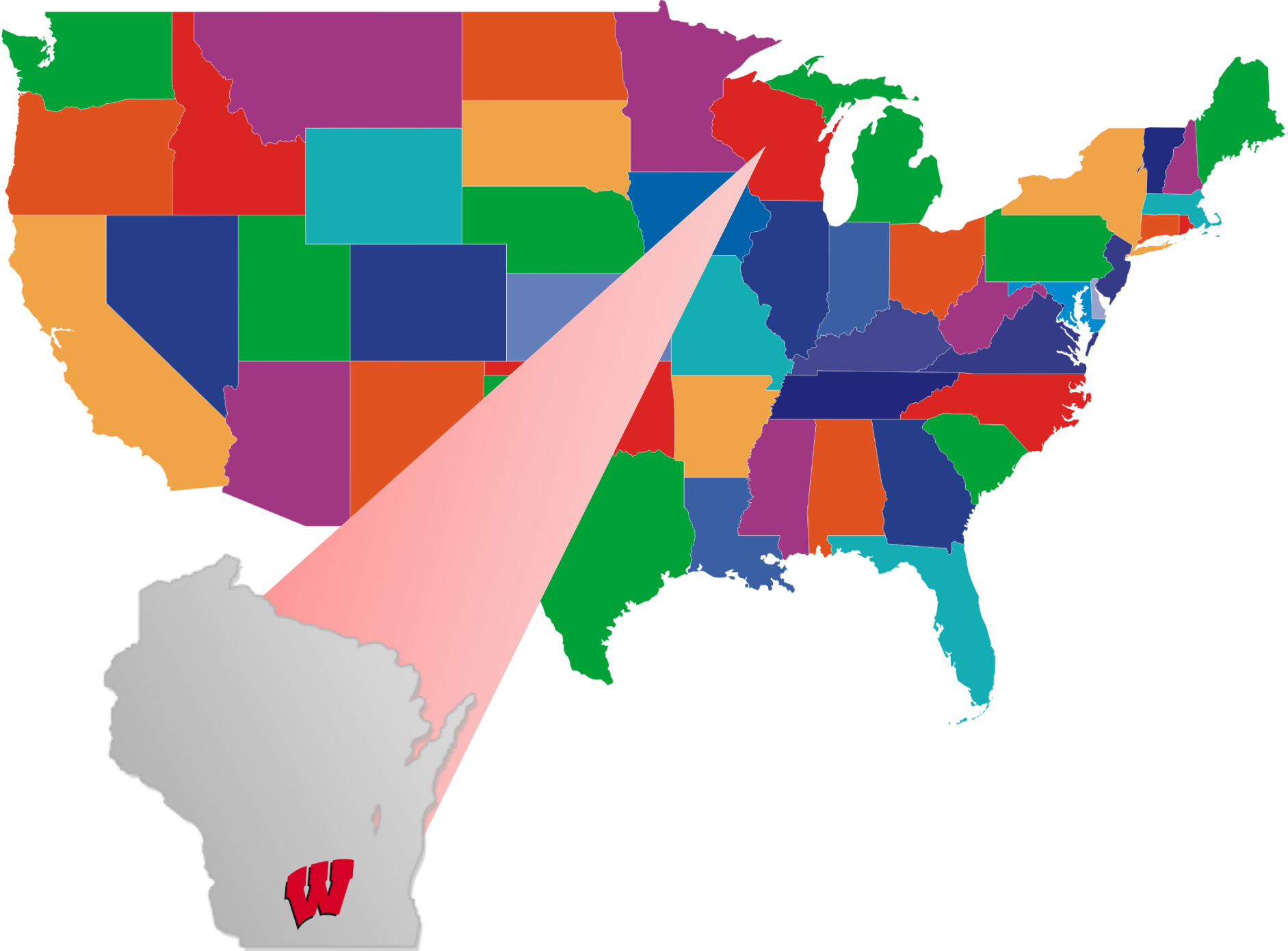




Genomics and the Electronic Health Record

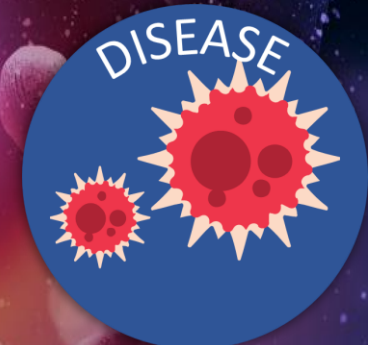
Scott Moss
Software Developer
Research Informatics, Epic
G2MC: November 7, 2015



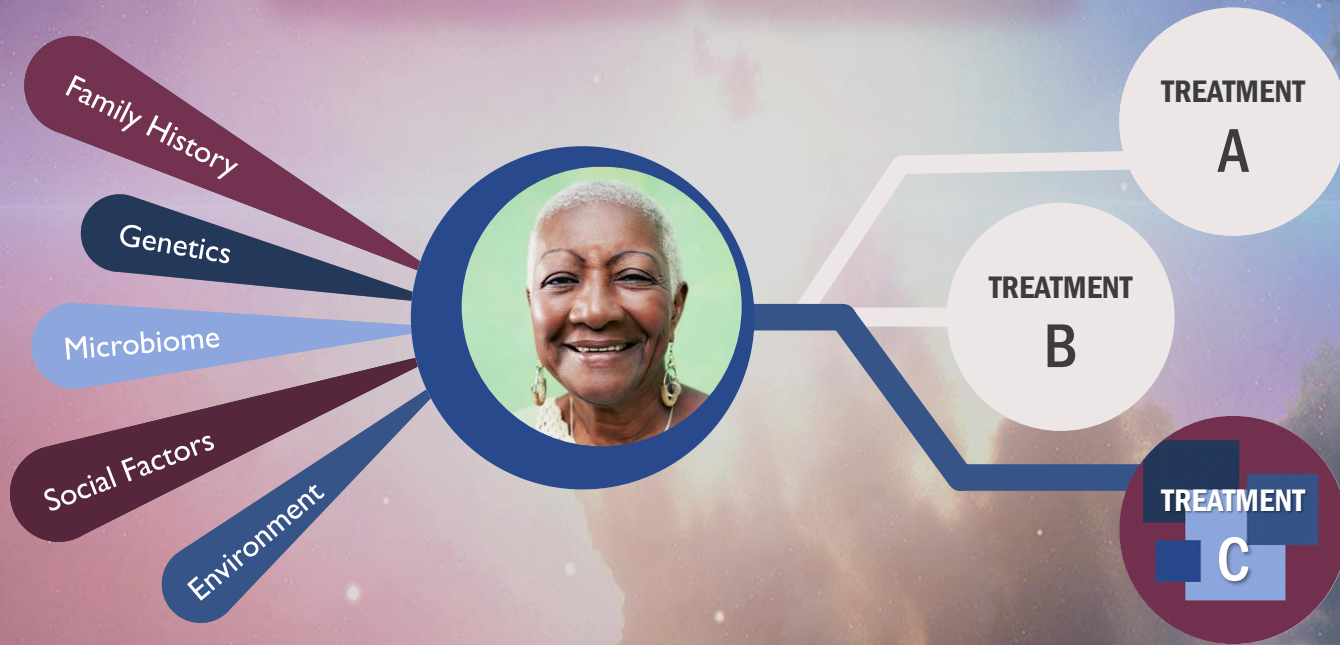


GENOMICS

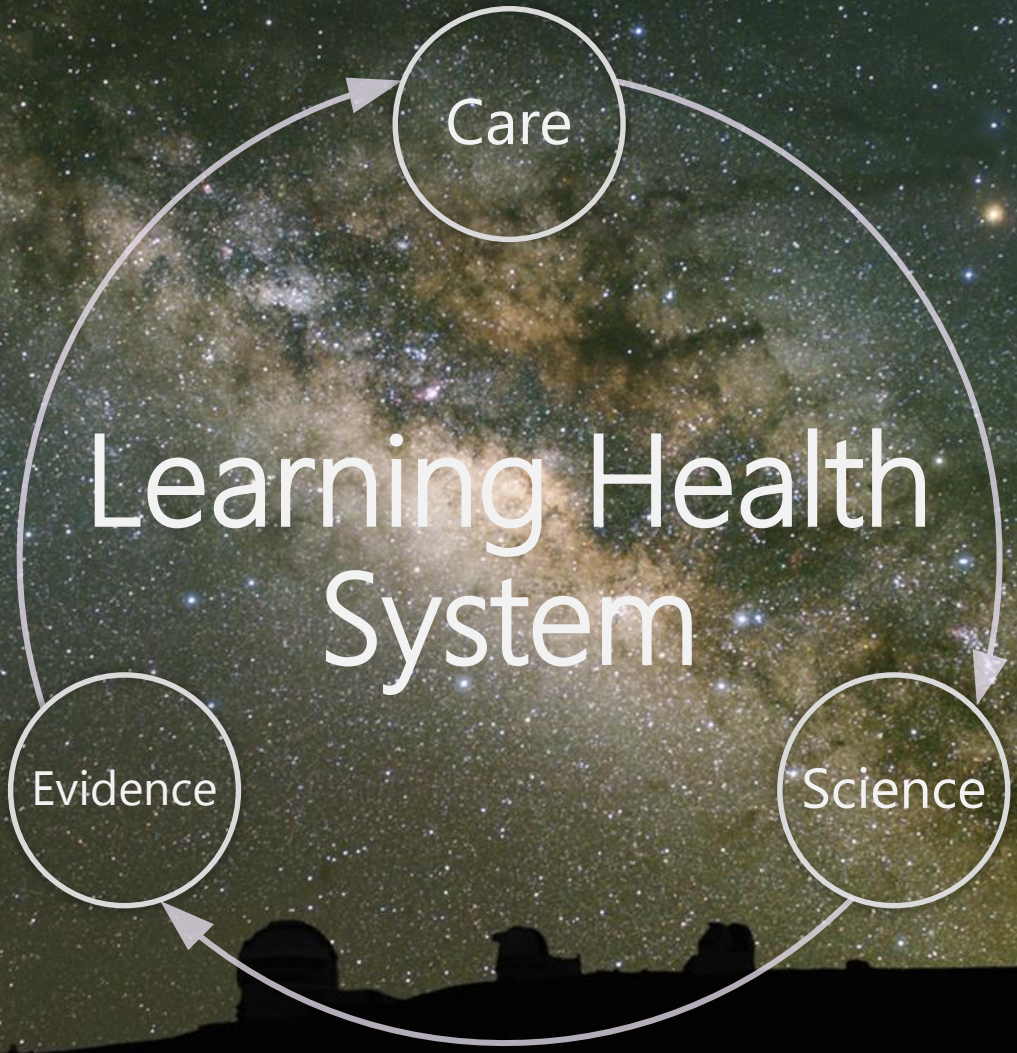
Using genetic data to inform medical decisions across the entire continuum of care



PRECISION MEDICINE



Learning Health System



Care

Evidence

Science

DETERMINE TESTING

Gather info

Plan

Educate

Consent



Family Hx



Phenotype



Environment



TESTING METHODS

Cytogenetic

Molecular

Next Gen. Sequencing

Alignment

↳ Variant Calling

↳ Variant Annotation

↳ Determine Clinical Utility



FastQ



BAM



VCF



Genotype



Knowledge

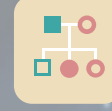


ANALYSIS AND APPLICATION

Reconcile

Plan

Engage



Family Hx



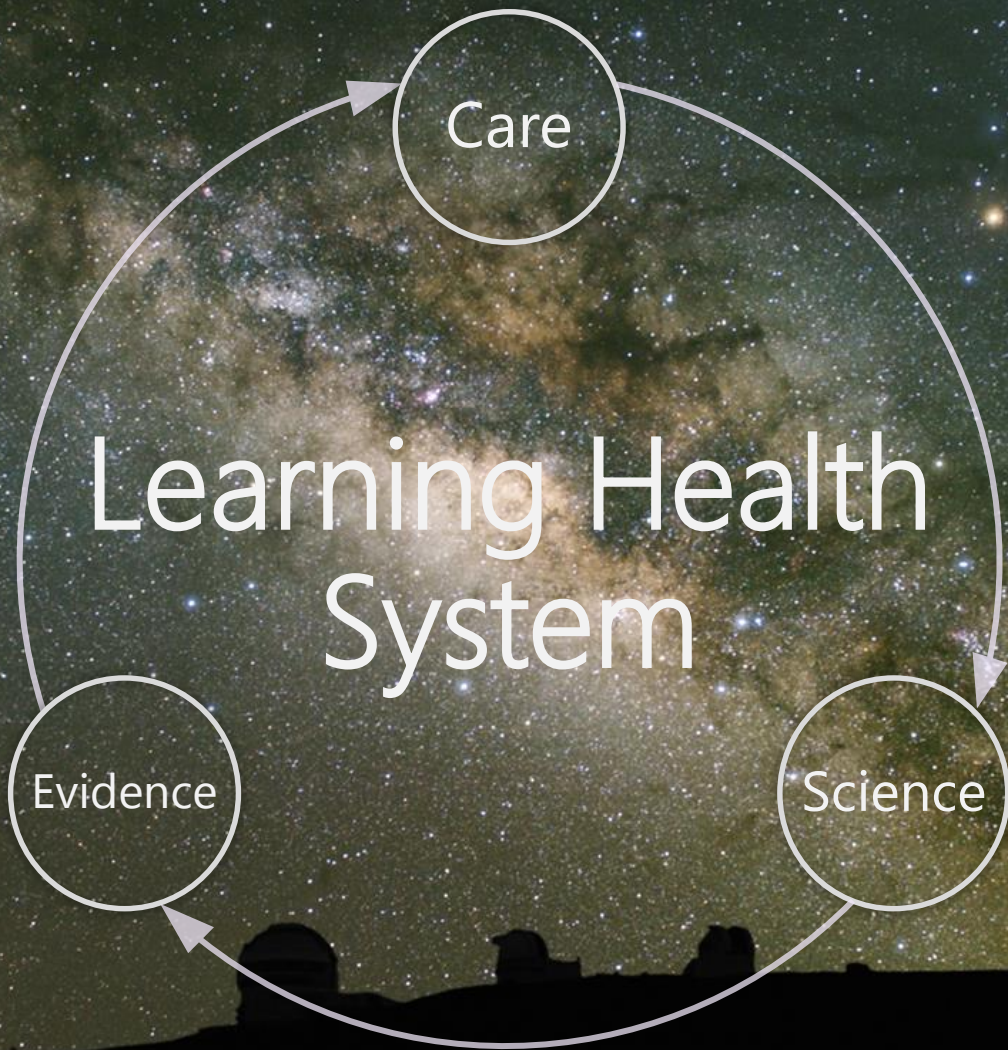
Phenotype



Environment



Learning Health System



Unprecedented GWAS research

Kaiser Permanente and UCSF Add Substantial Genetic, Health Information to NIH Online Database

Information is Largest-Ever Genetic Resource for Researchers

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By **Kristen Bole** on February 26, 2014

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Researchers worldwide will now have access to genetic data linked to information on a diverse group of more than 78,000 people, enabling it into many diseases and conditions.

The data have just been made available to qualified researchers through the data Genotypes and Phenotypes (dbGaP), the online database of the National Institute of Health (NIH). The announcement was made February 26 at the National Advisory Council on Aging, directed by the National Institute on Aging (NIA).

The data come from one of the nation's largest and most diverse genomic projects: the Genetic Epidemiology Research on Adult Health and Aging (GERA) cohort — developed collaboratively by the Kaiser Permanente Research Program on Genes and Health (RPGEH) and UC San Francisco (UCSF). The addition of the data to dbGaP was made possible with \$24.9 million in support from the NIA and the National Institute of Health at NIH, as well as from the Office of the NIH Director.

"Data from this immense and ethnically diverse population will be a tremendous science," said NIH Director Francis Collins. "It offers the opportunity to identify genetic risks and influences on a broad range of health conditions, particularly those related to aging."

The GERA cohort is part of the RPGEH, which includes more than 430,000 adults from the Kaiser Permanente Northern California health plan who volunteered to participate in the research program. Data on this larger cohort include electronic medical records, behavioral and demographic information from surveys, and saliva or blood samples from 200,000 participants obtained with informed consent.

This work was made possible with support from the Johnson Foundation, which supports research. "This massive influx of data will speed breakthroughs faster," said Nan Johnson, CEO of the Johnson Foundation. "Researchers use this data to study genomic samples for improving health by studying individuals in dbGaP."

Additional support for development of the database was provided by the Gladys Valley Foundation, the E

"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."

The genetic information on more than 78,000 individuals translates into over 55 billion bits of genetic data for the cohort. The researchers conducted genome-wide genotyping using the newly developed Affymetrix Axiom arrays.

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.

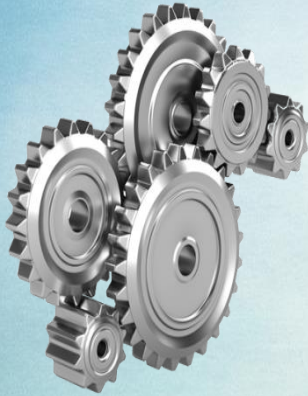
phenotypic
& genotypic
information

<http://www.ucsf.edu/news/2014/02/112161/kaiser-permanente-and-ucsf-add-substantial-genetic-health-information-nih-online>

Epic ...with the patient at the heart

Models

Epic



Treatment
recommendation

Where are the

DATA ?



Where is the

TESTING ?



Epic
Web Services



Treatment
recommendation



Epic's Research Community



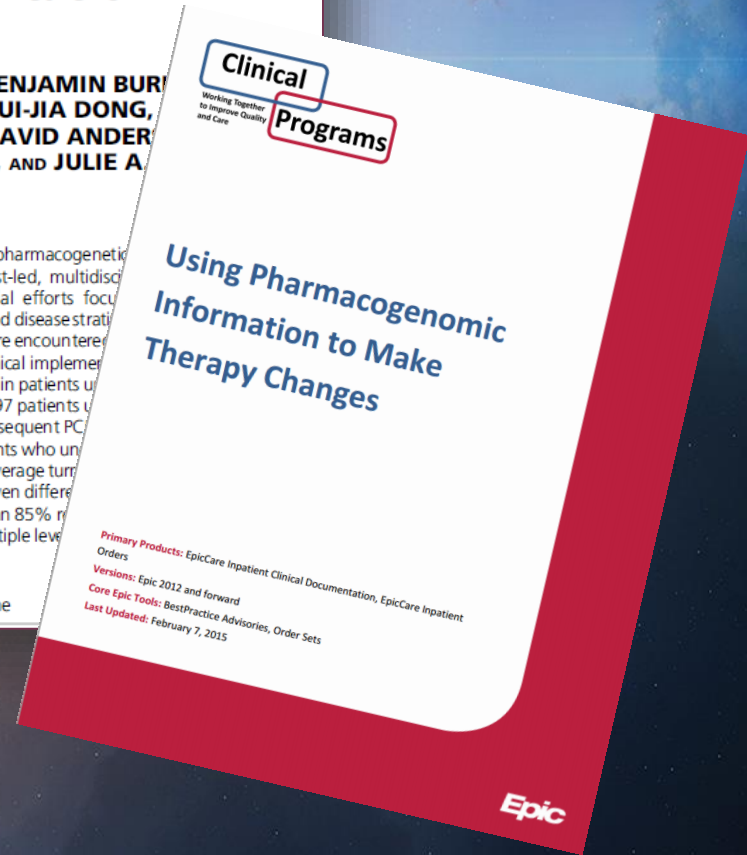
ARTICLE

Clinical Pharmacogenetics Implementation: Approaches, Successes, and Challenges

KRISTIN W. WEITZEL, AMANDA R. ELSEY, TAIMOUR Y. LANGAEE, BENJAMIN BURK, DAVID R. NESSL, ANIWAA OWUSU OBENG, BENJAMIN J. STALEY, HUI-JIA DONG, ROBERT W. ALLAN, J. FELIX LIU, RHONDA M. COOPER-DEHOFF, R. DAVID ANDERSON, MICHAEL CONLON, MICHAEL J. CLARE-SALZLER, DAVID R. NELSON, AND JULIE A. ...

Current challenges exist to widespread clinical implementation of genomic medicine and pharmacogenetics. The University of Florida (UF) Health Personalized Medicine Program (PMP) is a pharmacist-led, multidisciplinary program established in 2011 within the UF Clinical Translational Science Institute. Initial efforts focused on clinical pharmacogenetics, with long-term goals to include expansion to disease-risk prediction and disease stratification. This article describes the processes for development of the program, the challenges that were encountered, and the success stories by clinicians of the genomic medicine implementation. The initial clinical implementation began in June 2012 and targeted clopidogrel use and the *CYP2C19* genotype in patients undergoing percutaneous-coronary intervention (PCI). After 1 year, 1,097 patients undergoing PCI were genotyped preemptively, and 291 of those underwent subsequent PCI. Data were reported to the medical record for 100% of genotyped patients. Eighty patients who underwent PCI with a high-risk genotype, with drug therapy changes implemented in 56 individuals. Average turnaround time from genotype result entry in the medical record was 3.5 business days. Seven different insurance carriers, including Medicare, reimbursed for the test during the first month of billing, with an 85% reimbursement rate for claims that were submitted in the first month. These data highlight multiple levels of success in the implementation of genomic medicine. © 2014 Wiley Periodicals, Inc.

Keywords: genomic medicine; implementation; *CYP2C19*; personalized medicine

Clinical Programs
Working Together to Improve Quality and Care

Using Pharmacogenomic Information to Make Therapy Changes

Primary Products: EpicCare Inpatient Clinical Documentation, EpicCare Inpatient Orders
 Versions: Epic 2012 and forward
 Core Epic Tools: BestPractice Advisories, Order Sets
 Last Updated: February 7, 2015

Epic

PHARMACY PRACTICE NEWS

Clinical

ISSUE: OCTOBER 2015 | VOLUME: 42:10

CYP2C19 Testing Post-PCI Slashes Heart Risk

by David Wild

New Orleans



Percutaneous coronary intervention (PCI) guidelines do not recommend basing post-procedure clopidogrel therapy on cytochrome P450 2C19 (CYP2C19) genotype, but a study from the University of Florida suggests that may be a dangerous oversight.

“...testing enabled [U of Florida] to **virtually eliminate** the major cardiovascular (CVD) events that can occur when patients with the genotype are given clopidogrel.”



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DIGITizE Initial PGx Use Cases

#	Use Case Types
1	Incorporating Genetic Results into EHR User Interfaces
2	Adding genetic tests in order sets
3	Clinical Decision Support (CDS) identifies when a test should be ordered (pre-test alert*)
4	CDS identifies when a drug order is inconsistent with a test result (post-order alert*)

* 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!