

Genomic Evidence to Improve Quality

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Fall NENIC Webinar



Conflict of Interest Disclosure

Kathleen A. McCormick, PhD, RN

Conflicts of Interest Reported –

McGraw-Hill - Royalties,

SAIC- Stock,

LEIDOS- Stock,

Varied Mutual Funds



Session Objectives

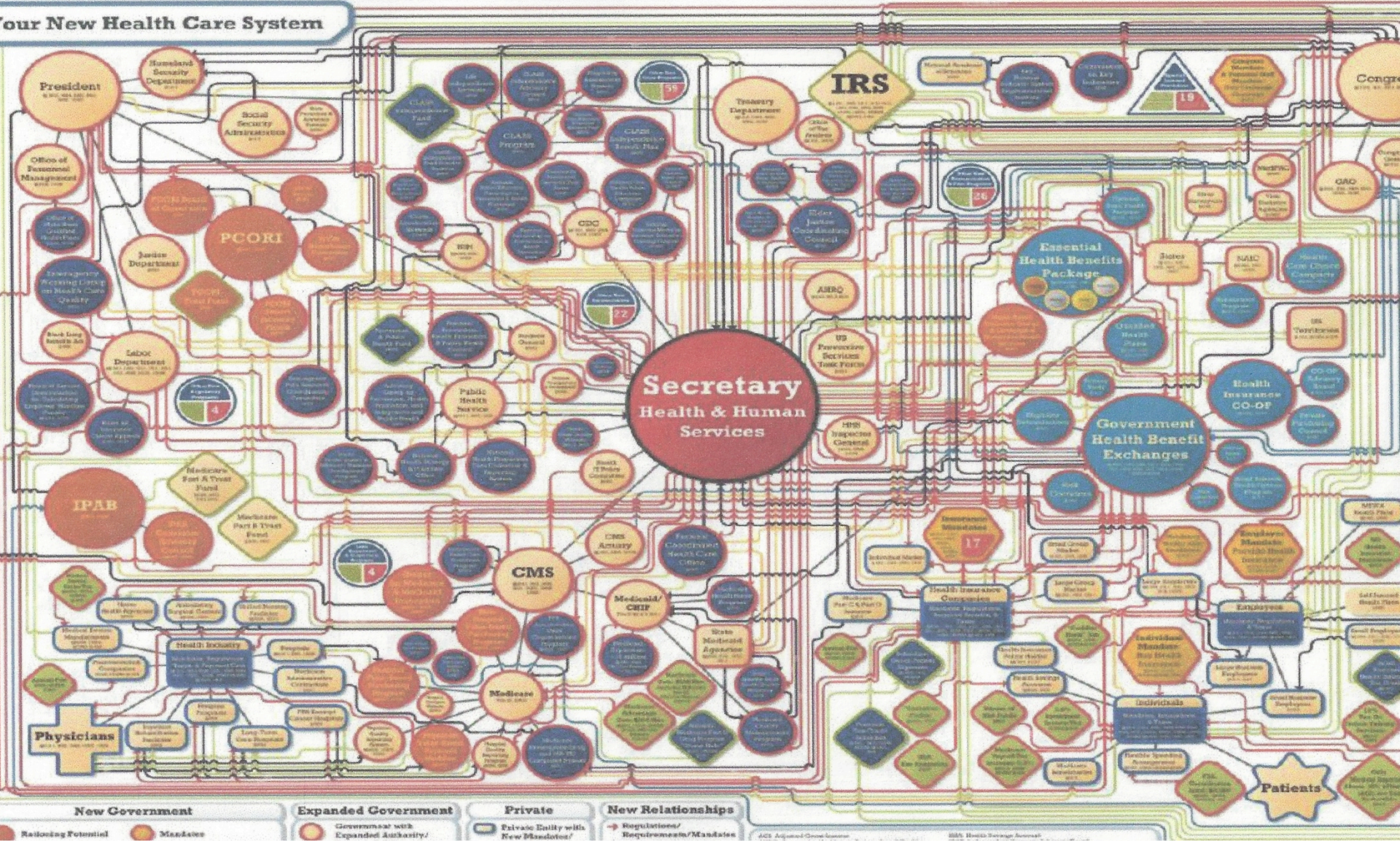
1. To describe what evidence exists in genomics.
2. To define roadmaps to including genomic evidence into EHRs.
3. To identify resources that continually update information.
4. To understand the impact on nursing policies, education, documentation, and innovation.

- Acknowledge Dr. Kathleen Calzone-Co-author Essentials of Nursing Informatics, V6, 2015. Big Data Initiatives: Genomics and Information Technology for Personalized Health



Focus Regulation

Our New Health Care System



•These are the Regulatory Changes of ACA- Fritsma SINI, July, 2015

- So how do you make time for yet another implementation?
- Use the products that are already developed.

This is a Historical Moment

- October 1, 2015 marks **the 25th Anniversary** of the Launch of the Human Genome Project
- Time to reflect on the progress since that time



Headline February 26, 2015: “...in many instances there is sufficient evidence to justify the use of genetic testing to inform choice or dosage of medications.” IOM

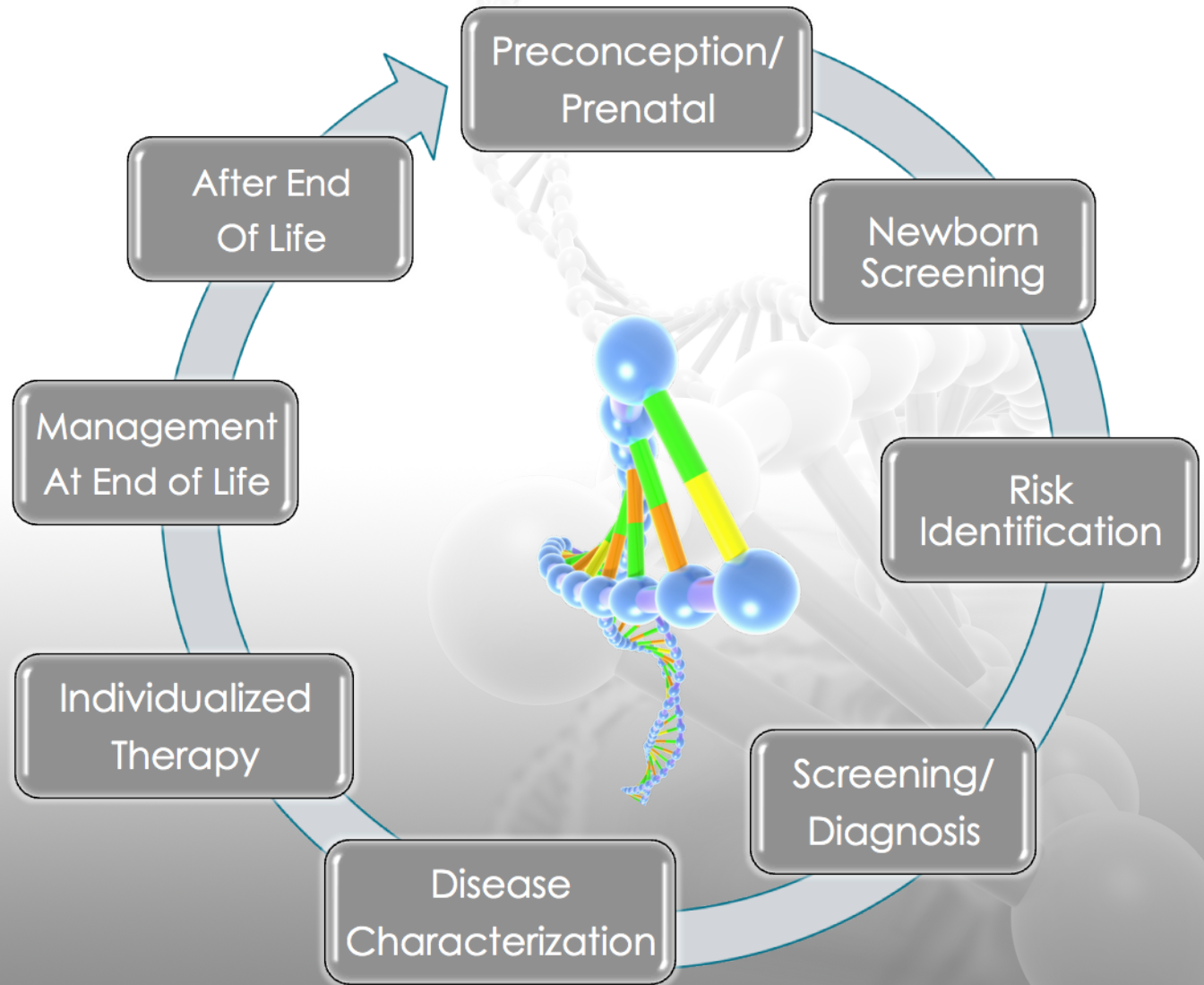
- These topics will be described in this session, as well as exemplars where the incorporation of evidence can improve quality medication outcomes.
- Outcomes can be achieved when guidelines and CDS is incorporated into the EHR.
- Algorithms are being developed to test the incorporation of genetic/genomic evidence into EHRs.



**Where can the
evidence be found?**

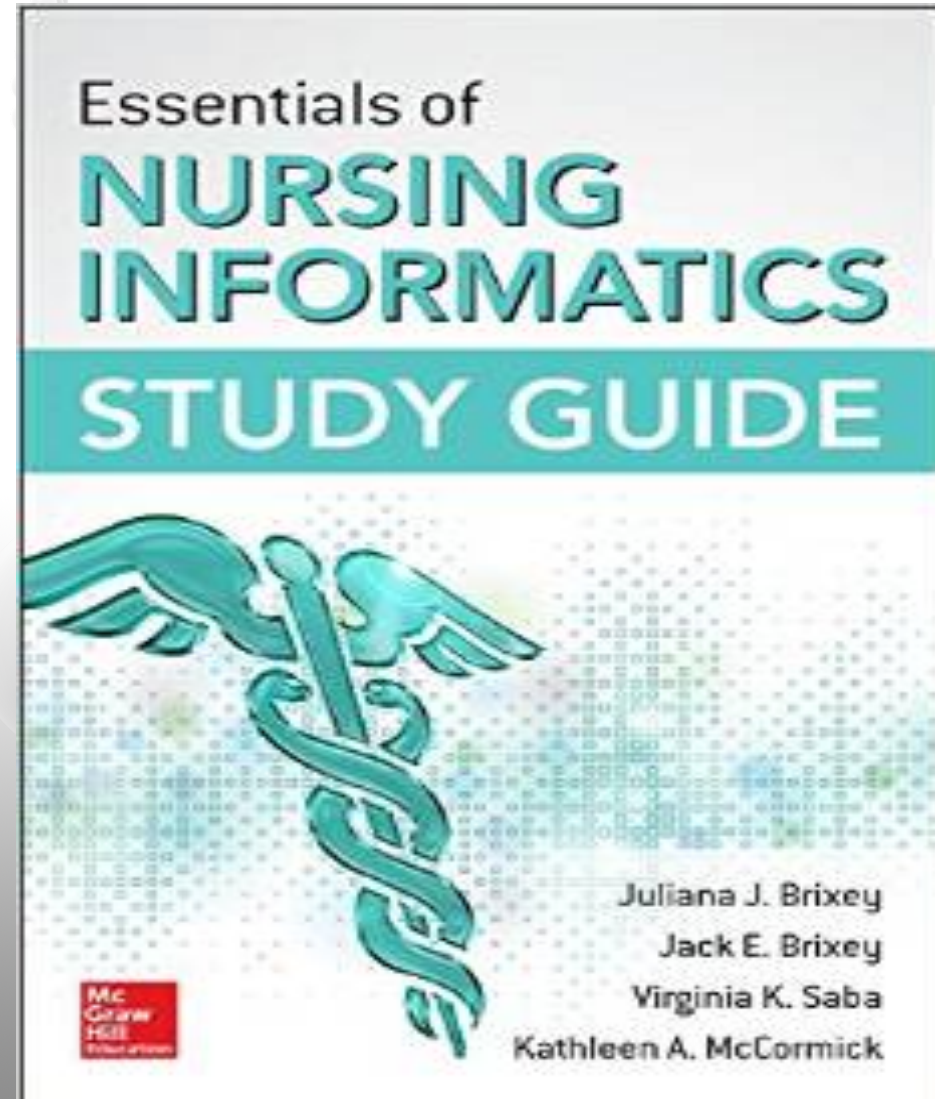
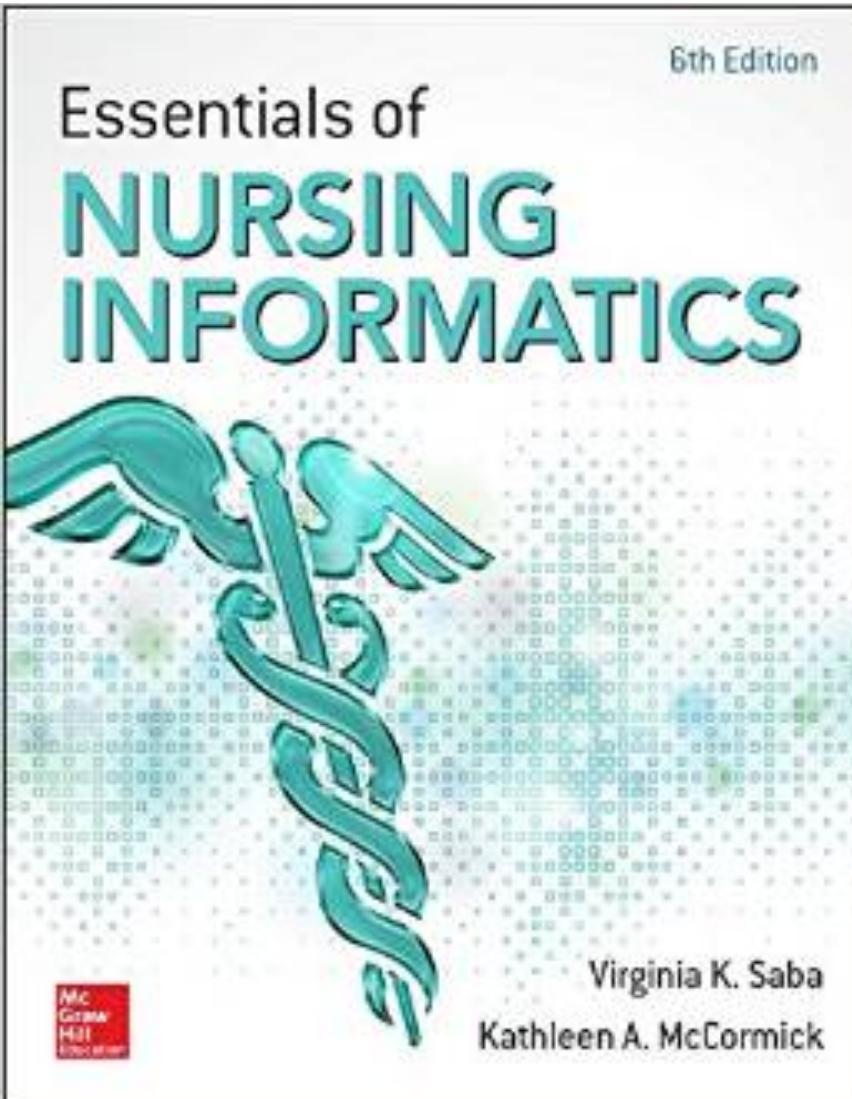
Genetic and Genomic Influences Across the Healthcare Continuum

Interdisciplinary Collaboration to Improve Patient Outcomes

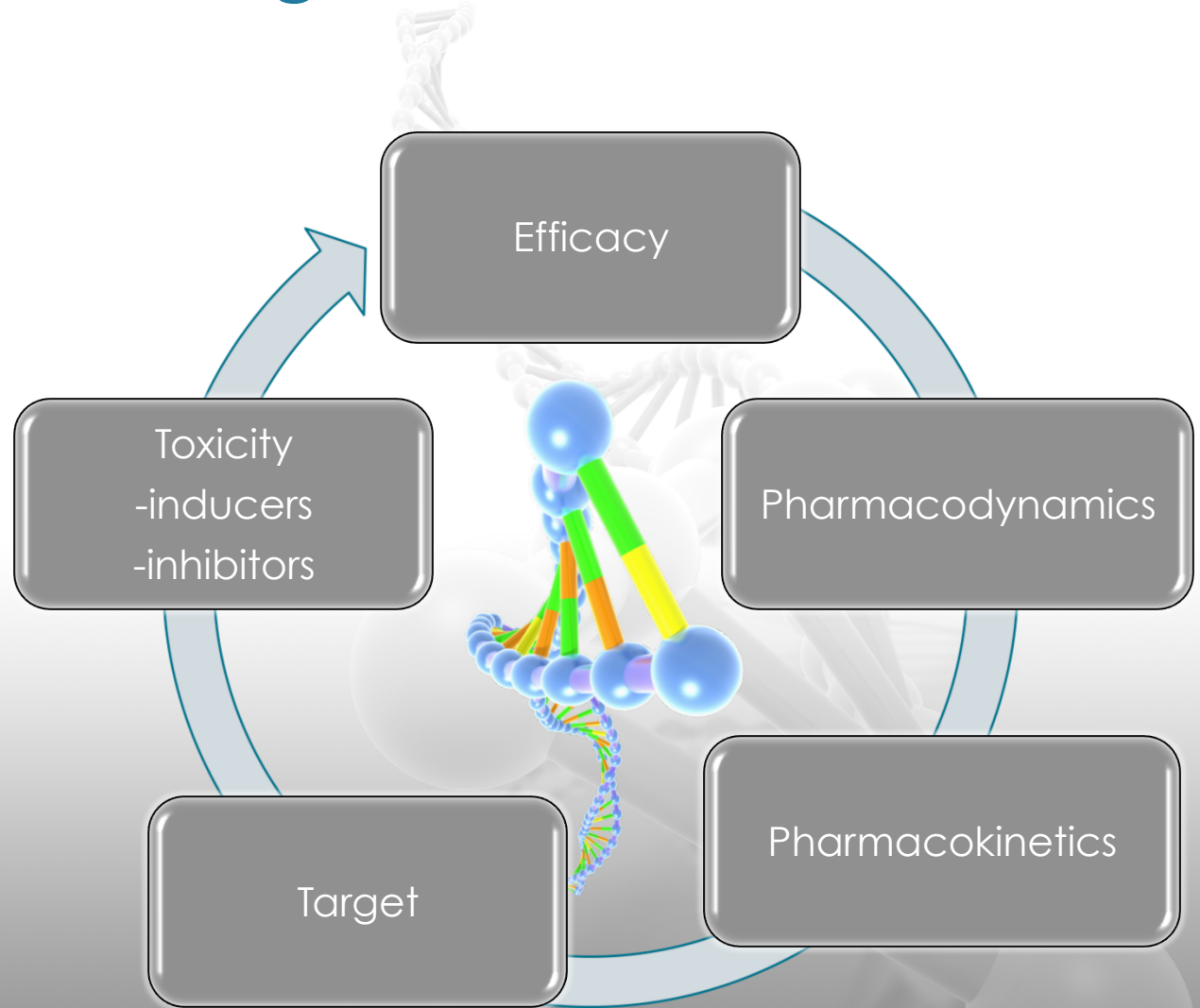


Calzone, et al. (2013). Relevance of genomics to healthcare and nursing practice. *Journal of Nursing Scholarship*, 45, 1-2.

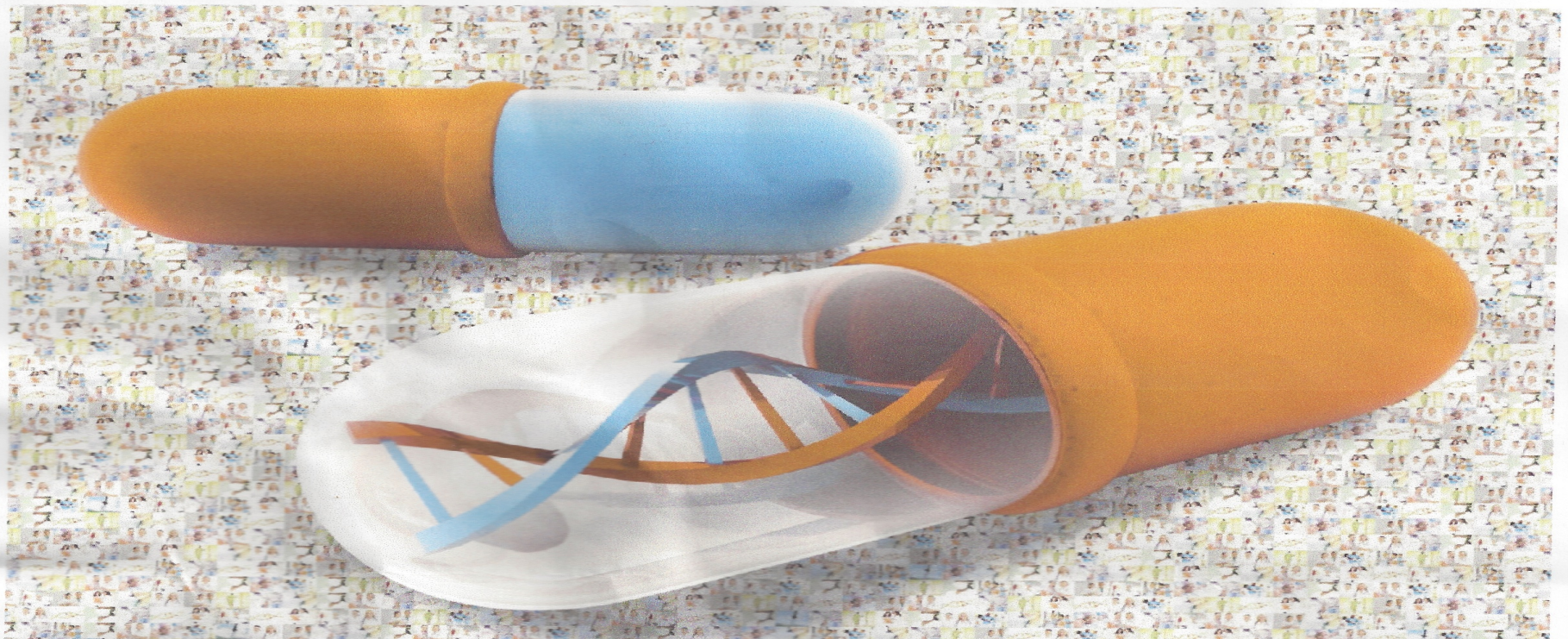
Content from: McCormick and Calzone, Chapter 49. Genomics and Information Technology for Personalized Health



Pharmacogenomic Influences



PK = absorption, distribution, metabolism and excretion
PD = mechanism of action, drug concentration and effect



MASTERING

PHARMACOGENOMICS

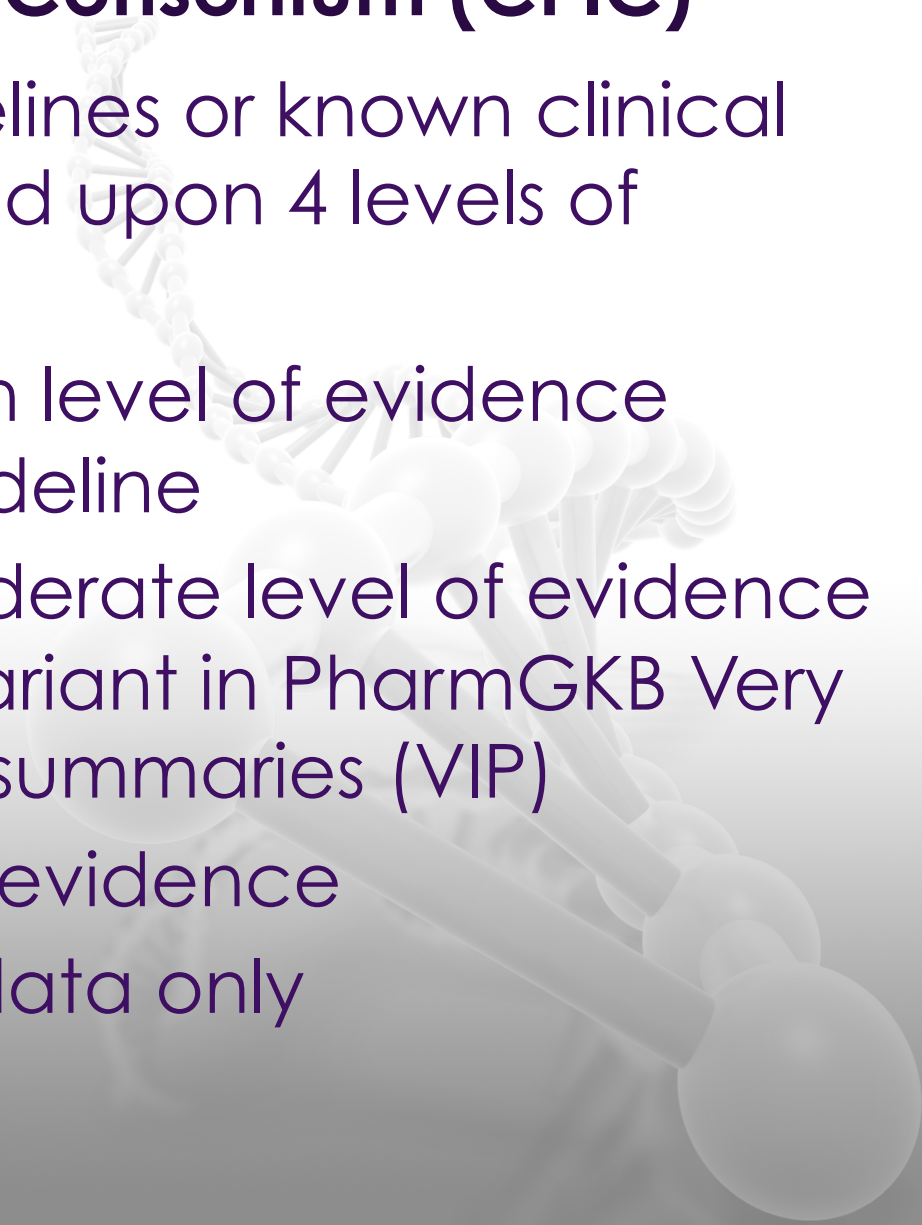
A Nurse's Handbook for Success

**DALE HALSEY LEA | DENNIS CHEEK
DANIEL BRAZEAU | GAYLE BRAZEAU**

•What is PharmGKB?

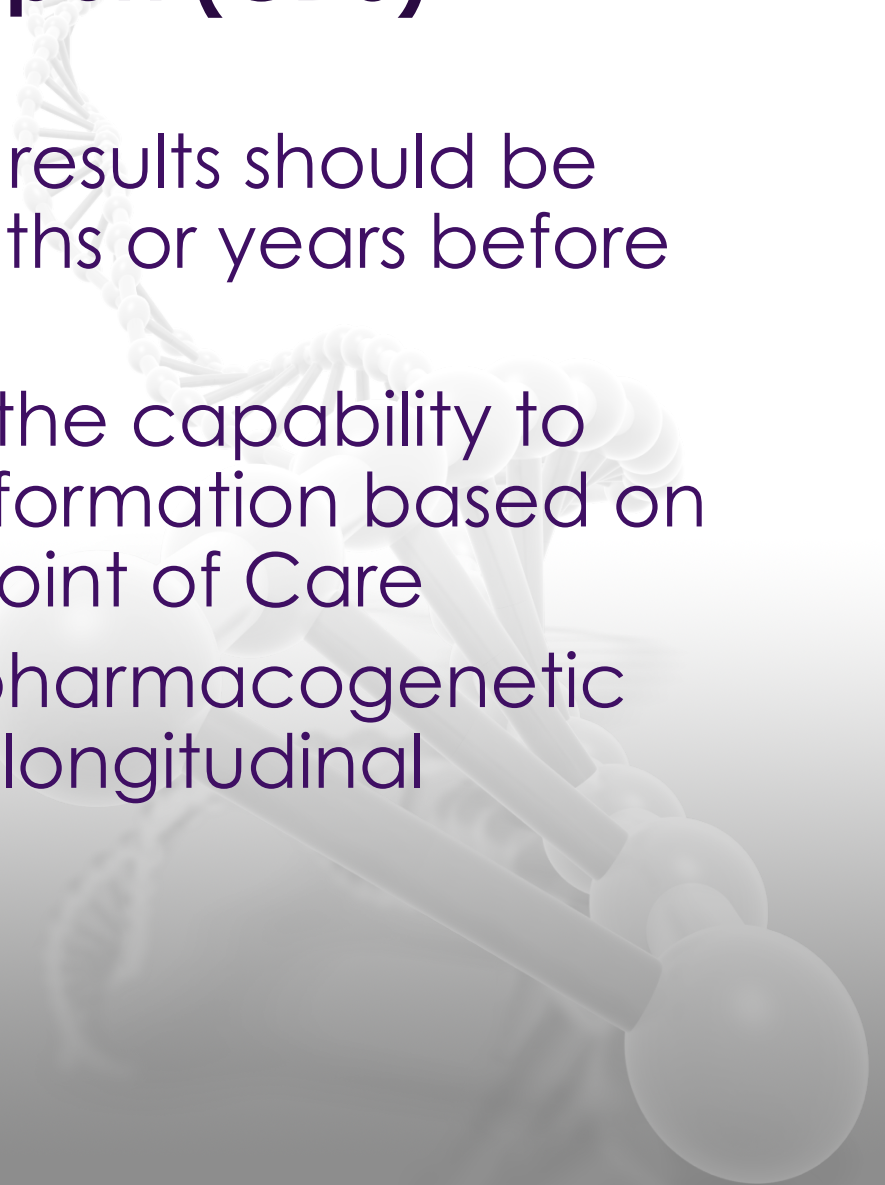
- A knowledge base that collects, curates, and disseminates knowledge about the impact of human genetic variation on drug response
- This knowledge base represents Primary pharmacogenomic literature, Knowledge extraction, Knowledge annotation, aggregation and integration, Clinical interpretation, and Clinical Implementation (where available)
- PharmGKB contains clinical information including dosing guidelines and drug labels, potentially clinically actionable gene-drug associations and genotype-phenotype relationships- links to CPIC
- It is NIH Funded and include consortium for Warfarin, Tamoxifen, SSRI, and Clopidogrel

What is the Clinical Pharmacogenetics Implementation Consortium (CPIC)

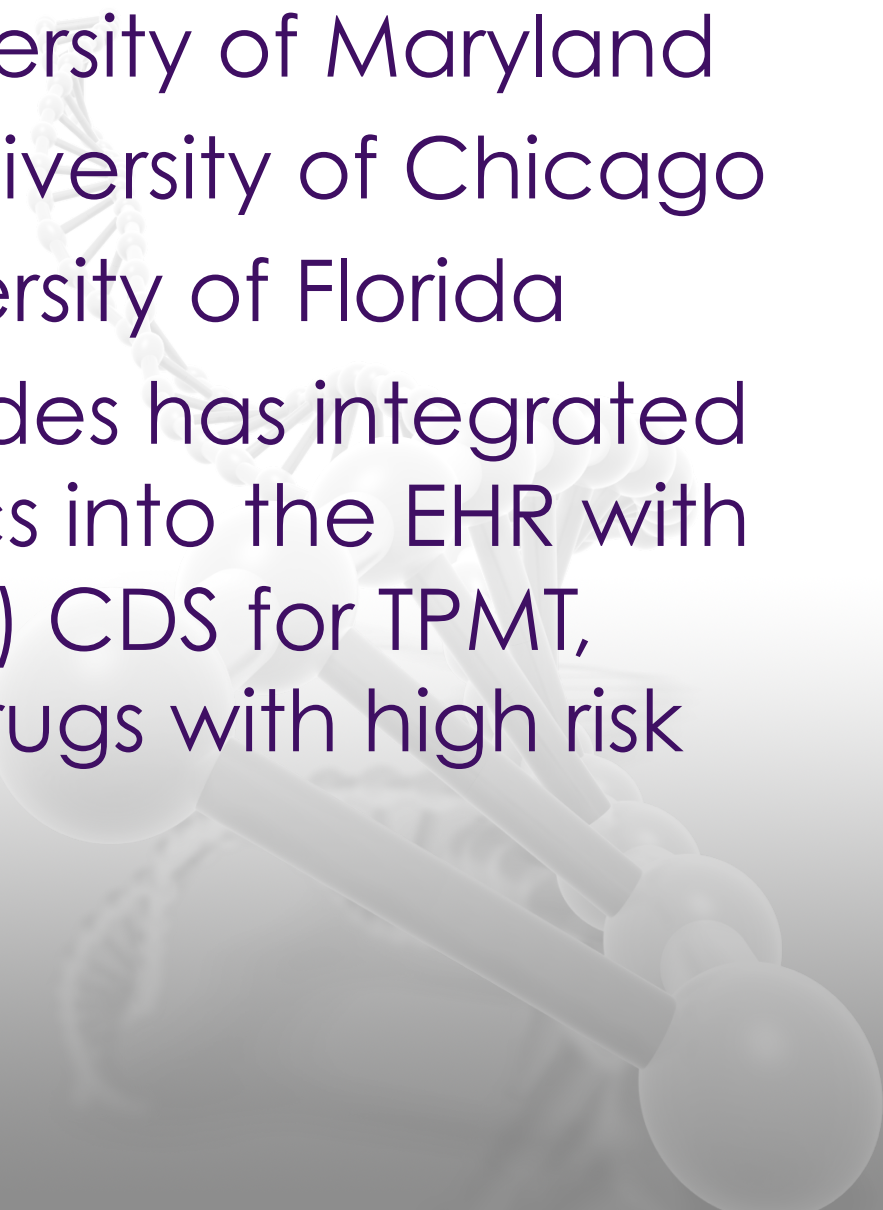
- CPIC produces guidelines or known clinical implementation based upon 4 levels of evidence:
 - Level 1 a and b - High level of evidence becomes a CPIC guideline
 - Level 2 a and b - Moderate level of evidence is represented as a variant in PharmGKB Very Important PGx gene summaries (VIP)
 - Level 3 - Low level of evidence
 - Level 4 - Preliminary data only
- 

•CPIC guidelines in EHR and using Clinical Decision Support (CDS)

- Today preemptive test results should be placed in the EHR months or years before relevant drug is used
- It is necessary to have the capability to deliver drug specific information based on genetic results at the Point of Care
- CDS facilitates use of pharmacogenetic results over a patient's longitudinal healthcare



•Key Players in CPIC

- Alan Shuldiner, University of Maryland
 - Peter O'Donnell, University of Chicago
 - Julie Johnson, University of Florida
 - Hoffman, JM. St. Jude's has integrated pharmacogenomics into the EHR with CDS (Cerner based) CDS for TPMT, matches high risk drugs with high risk genotype patients
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•Leveraging Existing Government Supported EHR Requirements

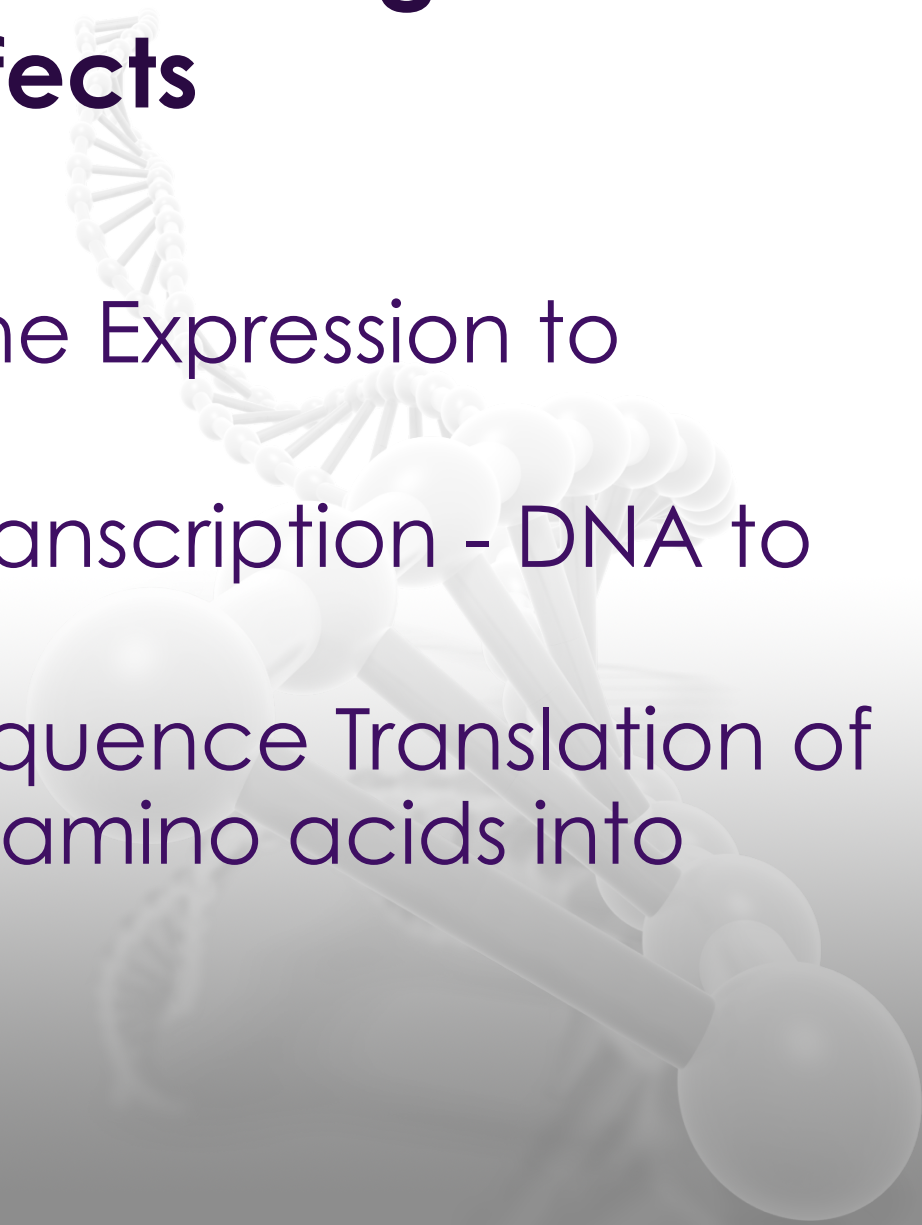
- ONC Meaningful Use
- I2B2
- PopMedNet
- CommonDocumentArchitecture (HL7 CDA)
- CommonDataModel (CDM)
- Health Insurance Portability Accountability Act (HIPAA)
- Physician quality reporting initiatives
- Value based purchasing
- eMERGE and PGRN Networks
- Clinical Pharmacogenetics Implementation Consortium (CPIC)- Guidelines
- Pharmacogenomics Knowledge Resource (PharmGKB -

—

•Sample Warning

- “Based on the genotype result, this patient is predicted to have intermediate TPMT activity. The patient is at risk for myelo-suppression with normal doses of 6-mercaptopurine. Consider starting 6-mercaptopurine doses at 30-70% of normal dose. Please consult a clinical pharmacist or review the pharmacogenetics tab for more information.”
- Action statement:
- Cancel Entry
- Dose Altered Accordingly
- Modify
- Source : James Hoffman, St. Jude

•Basic types of Pharmacogenomic genetic code defects

- Alternations in Gene Expression to assemble proteins
 - Abnormal Gene Transcription - DNA to RNA
 - Abnormal Base Sequence Translation of mRNA to organize amino acids into proteins
- 

•UGT1A1 Deactivates Itinotecan (Camptpsar)

- Patients on this drug with this UGT1A1 gene cannot deactivate the medication, leading to severe toxicities including neutropenia and diarrhea. Since 2005 the FDA has recommended patient with colorectal cancer be tested for UGT1A1 before administering this drug

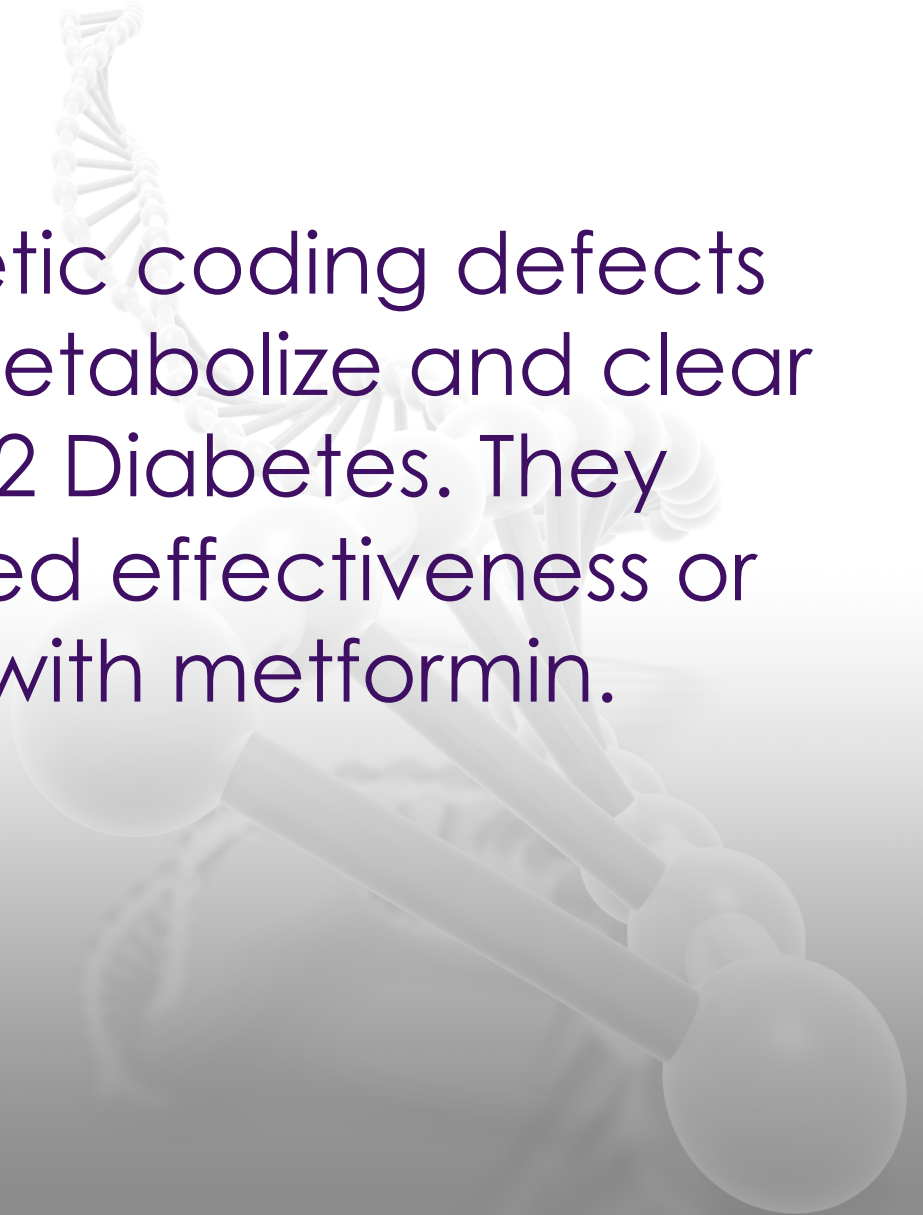
• Genes that Regulate Transport of P-Glycoprotein_

- Interfering with transport using anti-epileptic drugs, leads to reduced transfer of drugs into cells and prevents accumulation of anti-epileptic drugs in brain cells.



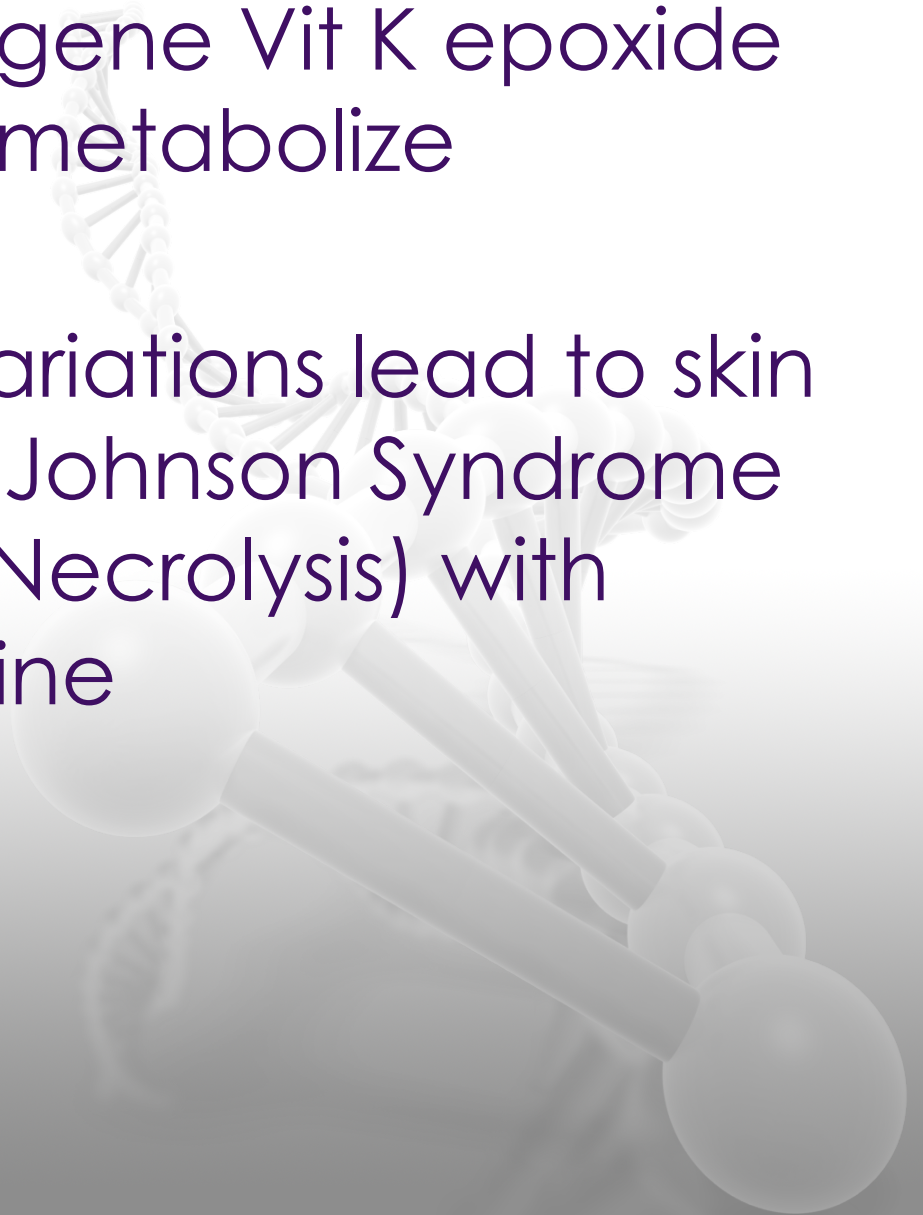
• Genes that affect drugs Transport in Renal Tubules

- Patients with Genetic coding defects cannot properly metabolize and clear metformin in Type 2 Diabetes. They either have reduced effectiveness or increased toxicity with metformin.



•Genes in Inheritance

- Huntington's - one gene Vit K epoxide cannot effectively metabolize coumadin
- HIV - HLA-B gene variations lead to skin condition (Stevens-Johnson Syndrome or Toxic Epidermal Necrolysis) with abacivir or nevirapine



• Genes targeting Protein coding on the DNA

- Classic case of IVACAFTOR alters folding of defective proteins coded by DNA in G551D mutation resulting in Cystic Fibrosis. The drug improves the chloride ion channel flow of salt and fluids across lung cells that helps thick sticky mucus. More than 400 mutations of this gene exist.



•Pharmacogenetic Testing and Drug Response (Part I)

9/23/2014


| | |
|---|---|
| Cytochrome P450 2C19 | This test detects variants in genes which may affect an individual's response to approximately 5% - 10% of all medications. |
| Cytochrome P450 2C9 & VKORC1 | This test detects variants in genes which may affect an individual's response to approximately 15% of all medications (including Warfarin Sensitivity via 2C9 and VKORC1). |
| Cytochrome P450 2D6 | This test detects variants in genes which may affect an individual's response to approximately 25% of all medications |
| Cytochrome P450 3A4 & 3A5 | This test detects variants in genes which may affect an individual's response to approximately 40% - 45% of all medications. |
| Factor II Prothrombin Genetic Profiling | This test detects a genetic change in the Factor II gene called Factor II Prothrombin. Patients with this Prothrombin variant are at an increased risk of blood clot formation (thrombosis) when exposed to other risk factors such as smoking, pregnancy, obesity, oral contraceptive use, and immobility. The risk is approximately 3-10 times higher in individuals who have one copy of the genetic variant. The risk in people who carry two copies of the genetic variant is unknown. Individuals who do not have a Factor II Prothrombin mutation may still be at increased risk. Other changes in the Factor II gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis. |

•Pharmacogenetic Testing and Drug Response (Part II)

9/23/2014

| | |
|--------------------------------------|---|
| <p>Factor V Leiden Mutation Test</p> | <p>This test detects a genetic change in the Factor V gene called Factor V Leiden. Individuals who have this variant are at an increased risk of blood clot formation. This risk is approximately 2-10 times higher in individuals who have one copy of the genetic variant, and greater than 10 times higher for individuals who carry two copies of the genetic variant. Individuals who do not have the Factor V Leiden mutation may still be at increased risk. Other changes in the Factor V gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis.</p> |
| <p>MTHFR Mutation Testing</p> | <p>This test detects two genetic changes in the MTHFR gene. Individuals who are found to have two mutations are at an increased risk for serious blood clot formation. Individuals who have only one or no copies of either genetic change in the MTHFR gene may still be at increased risk. Other changes in the MTHFR gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis.</p> |

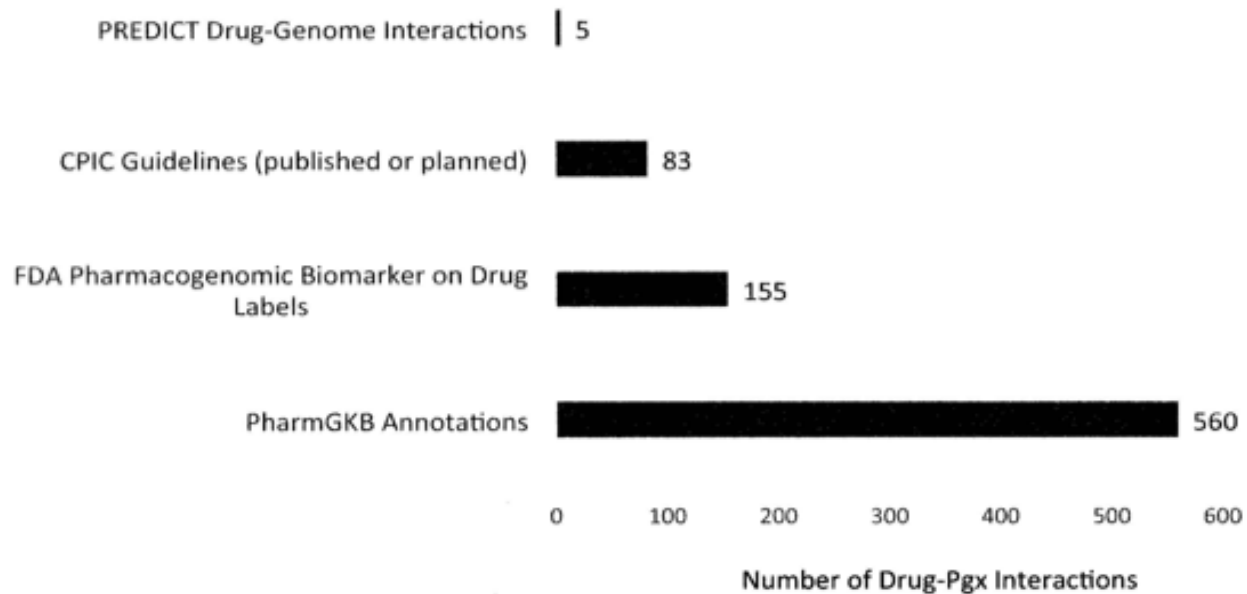
•Examples of CPIC Guidelines

- Azathioprine, TPMT
 - -CPY2C19- Clopidogrel
 - CPY2C9, VKORC1 - Warfarin
 - CYP2D - Codeine
 - HLA-B - Abacabir
 - SLOC1B1 - Simvastatin
 - HLA-B - Allopurinal
 - CYP2D6, CYP2C10 - TCAs
 - HLA-B - Carbamazepine
 - DPYD - 5FU / Capecitabine
 - IL28B- PEG Interferon alpha
 - CFTR - Ivacaftor
 - C6PD-Rasburicase
 - CYP2C9 and HLA-B- - Phentoin
 - CYP2D6 - SSRIs
 - CPP3A5 - Tacrolimus
 - CPY2X19 - Variconazole
- 

•From Bob Freimuth



Implemented vs. Potential Scope of Content



Lawsuits Have Begun



- [Lawsuit in Hawaii against Plavix Sponsors Alleges Burden is on Pharma to Market PGx Information - March 19, 2014](#)
 - Plavix has "diminished or no effect" on people of East Asian or Pacific Islander descent because they metabolize it poorly.
 - Suits also filed against Plavix Sponsors in Louisiana, Mississippi, West Virginia and California
- [California Clinical Laboratory Association sues HHS over local coverage determinations-May 2, 2014](#)
 - The [complaint](#) goes on to discuss (starting on page 11) LCDs developed by two influential MACs, [Noridian](#) and [Palmetto GBA](#), which together control 20 states, for molecular genetic testing, and how these LCDs restricted access genetic testing for Medicare beneficiaries.

•Questions still raised

- Structure of the data
- Standardization of the data
- National and international data sharing
- Structure of guidelines
- Structure of the decision supports



• **A Case for Integrated Data in EHR**

- Resources are available on current pharmacogenetics and pharmacogenomics from:
- Guidelines produced by the Clinical Pharmacogenomics Implementation Consortium (CPIC) - free, peer-reviewed, updated and detailed gene-drug clinical practice guidelines

• Building Personalized (Precision) Healthcare into your EHR

- Bioinformatics
 - Includes Genetic/Genomic/Molecular processes, pathways, workflows, biorepositories, statistical analysis, and decision support
- Imaging Informatics
 - Includes X-Ray, MRI, CT Scans, Tissues and Organs
- Healthcare Informatics
 - Individual
 - Home
 - Outpatient/Retail Clinics
 - Personal Health Data
 - Hospital
 - Nursing Home/Rehab
 - Hospice
 - Population
 - Public Health
 - State Surveillance
 - Emergency Preparedness and Response
 - Global Surveillance
- Includes Genetic/Genomic/Molecular processes, pathways, workflows, biorepositories, statistical analysis, and decision support
- Includes X-Ray, MRI, CT Scans, Tissues and Organs
- Includes EHR, PHR, Telehealth, Insurance data, Population health, Statewide repositories and surveillance, Global Surveillance



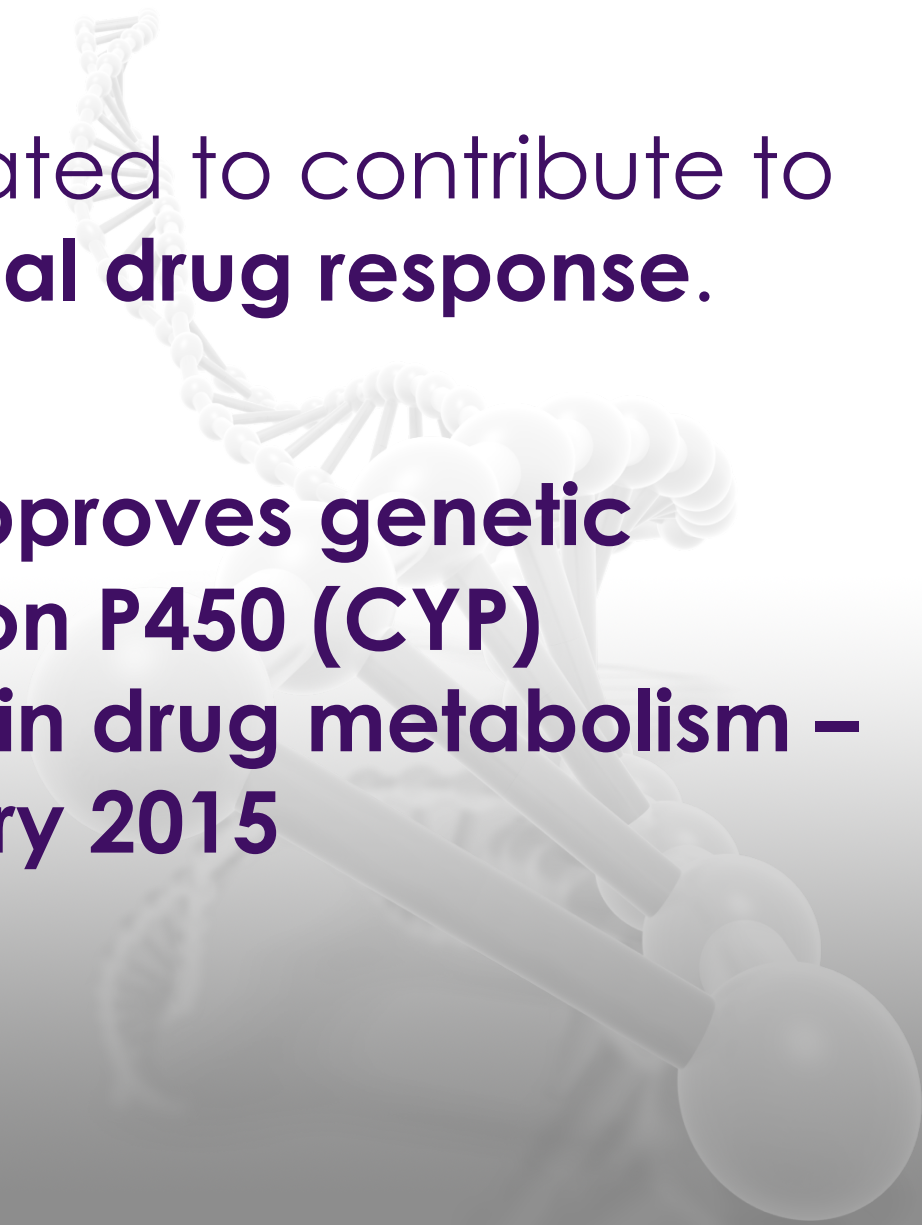
• **New FDA October 8, 2015**

- Soon to be launched – PrecisionFDA
- A cloud based open source platform for evaluating bioinformatics workflows it has been building with its partner DNAnexus.
- FDA White Paper – Optimizing FDA's Regulatory Oversight of Next Generation Sequencing Diagnostic Tests – Preliminary Discussion Paper

• **Genomics Contributions to Individual Drug Response**

- Genomics is estimated to contribute to **20-50 % of individual drug response.**

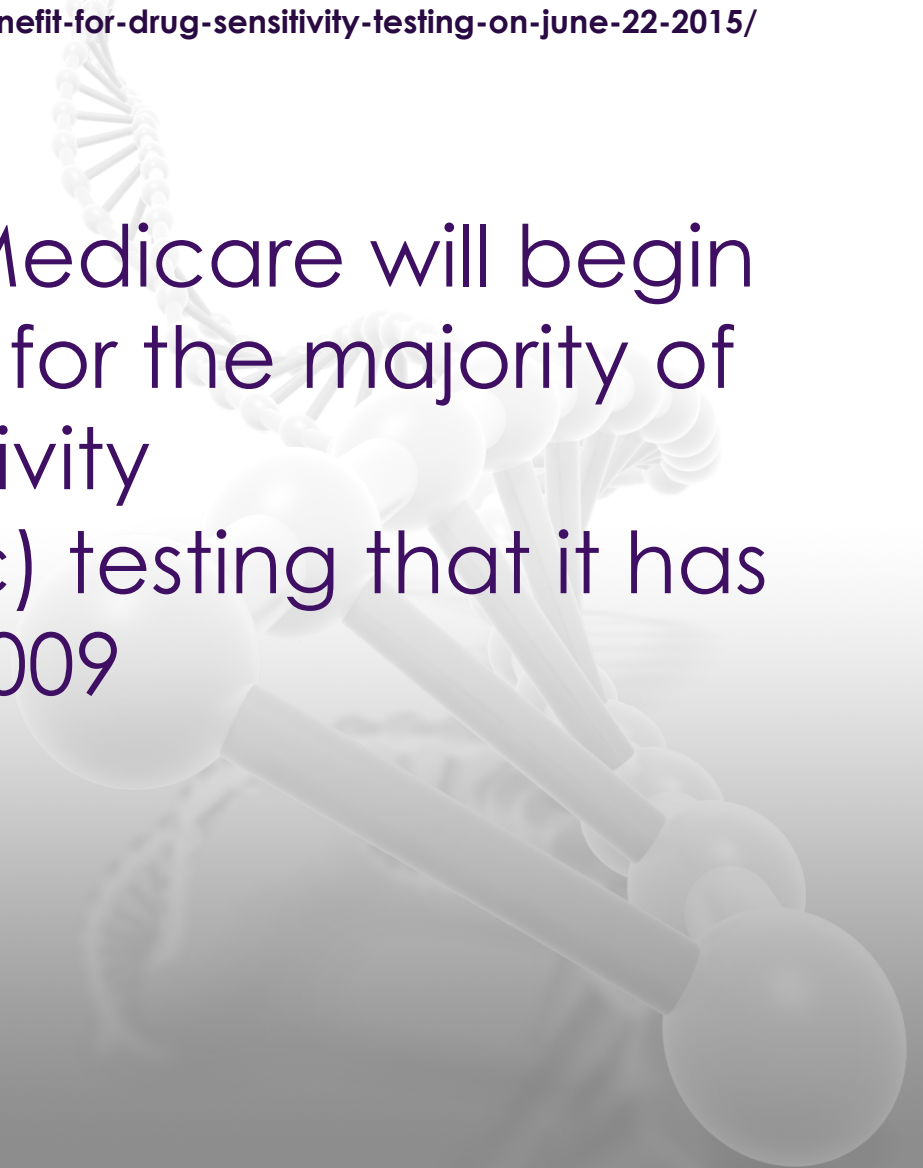
- **Even MEDICARE approves genetic testing of 9 common P450 (CYP) enzymes involved in drug metabolism – That was in February 2015**



• Medicare Limiting Coverage Benefit for Drug Sensitivity Testing on June 22, 2015

<http://genelex.com/medicare-limiting-coverage-benefit-for-drug-sensitivity-testing-on-june-22-2015/>

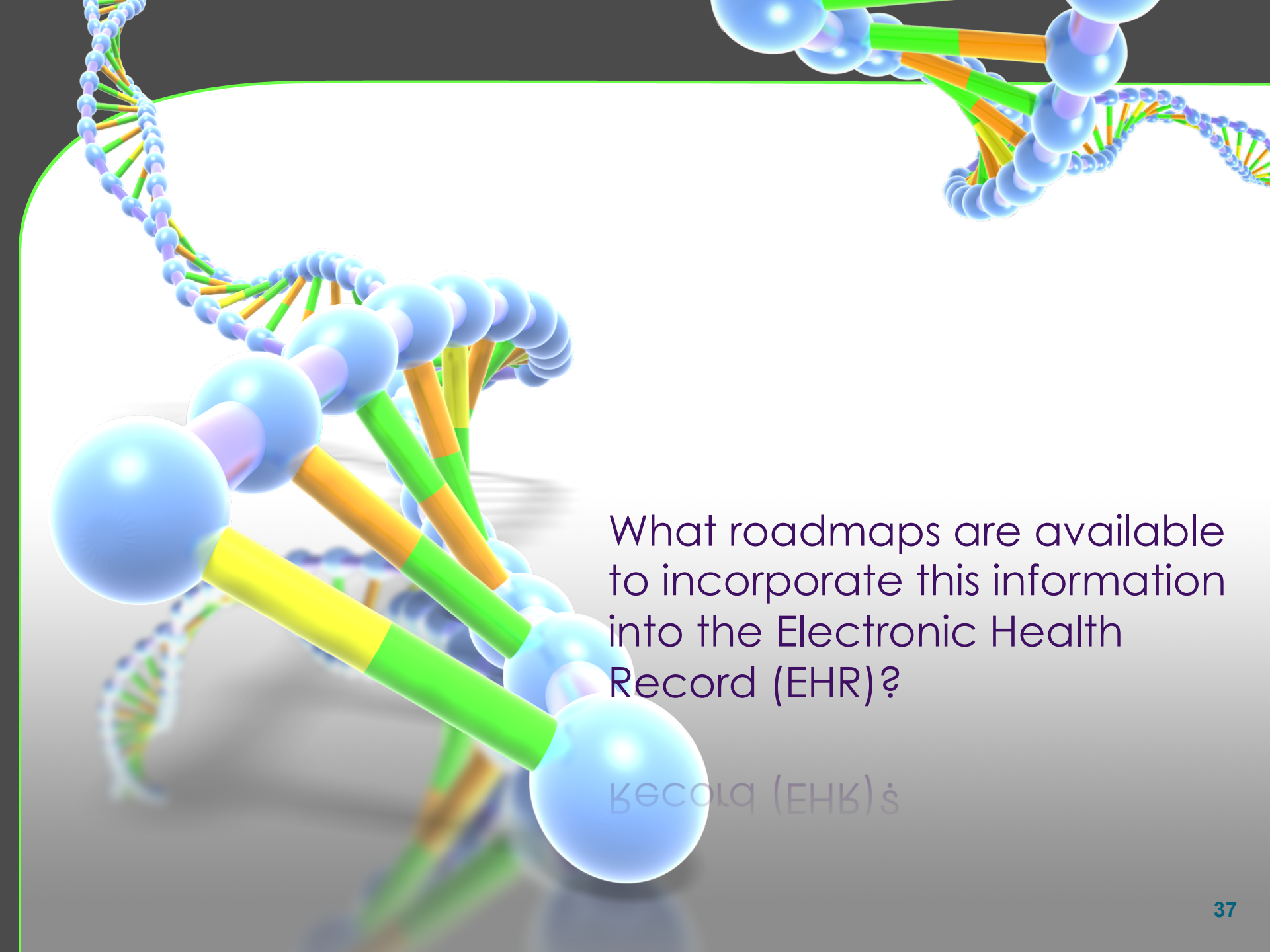
- On June 22, 2015 Medicare will begin denying coverage for the majority of genetic drug sensitivity (pharmacogenetic) testing that it has reimbursed since 2009



• Example CMS Position on Warfarin

<https://www.cms.gov/Medicare/Coverage/Coverage-with-Evidence-Development/Pharmacogenomic-Testing-for-Warfarin-Response.html> NEW AUGUST 18, 2015

- CMS believes that the available evidence does not demonstrate that pharmacogenomic testing of CYP2C9 or VKORC1 alleles to predict warfarin responsiveness improves health outcomes in Medicare beneficiaries. There we have determine that pharmacogenomic testing of CYP2C9 or VKORC1 alleles to predict warfarin responsiveness is not reasonable and necessary under Rule 1862 (a)(1)(A) of the Social Security Act. However, we do believe the available evidence supports that Coverage with Evidence Development (CED) under rule 1862 (a) (1)(E) of the Social Security Act is appropriate. Thus, we are making the following decision.
- Pharmacogenomic testing of CYP2C9 or VKORC1 alleles to predict warfarin responsiveness is covered only when provided to Medicare beneficiaries who are candidates for anticoagulation therapy with warfarin who:
 - 1. have not been previously tested for CYP2C9 or VKORC1 alleles; and
 - 2. have received fewer than five days of warfarin in the anticoagulation regimen for which the testing is ordered; and
 - 3. are enrolled in a prospective, randomized, controlled clinical study when that study meets the standards specified in the decision memorandum (DM)



What roadmaps are available to incorporate this information into the Electronic Health Record (EHR)?

What roadmaps are available to incorporate this information into the Electronic Health Record (EHR)?

Patients

Clinicians

Laboratories

Researchers

Sharing Genetic and Health Data

ClinGen's Critical Questions

Is this gene associated with a disease?
Clinical Validity

Is this variant causative?
Pathogenicity

Is this information actionable?
Clinical Utility

Building a Genomic Knowledge Base
ClinVar & Other Resources

Improved Patient Care
Through Genomic Medicine

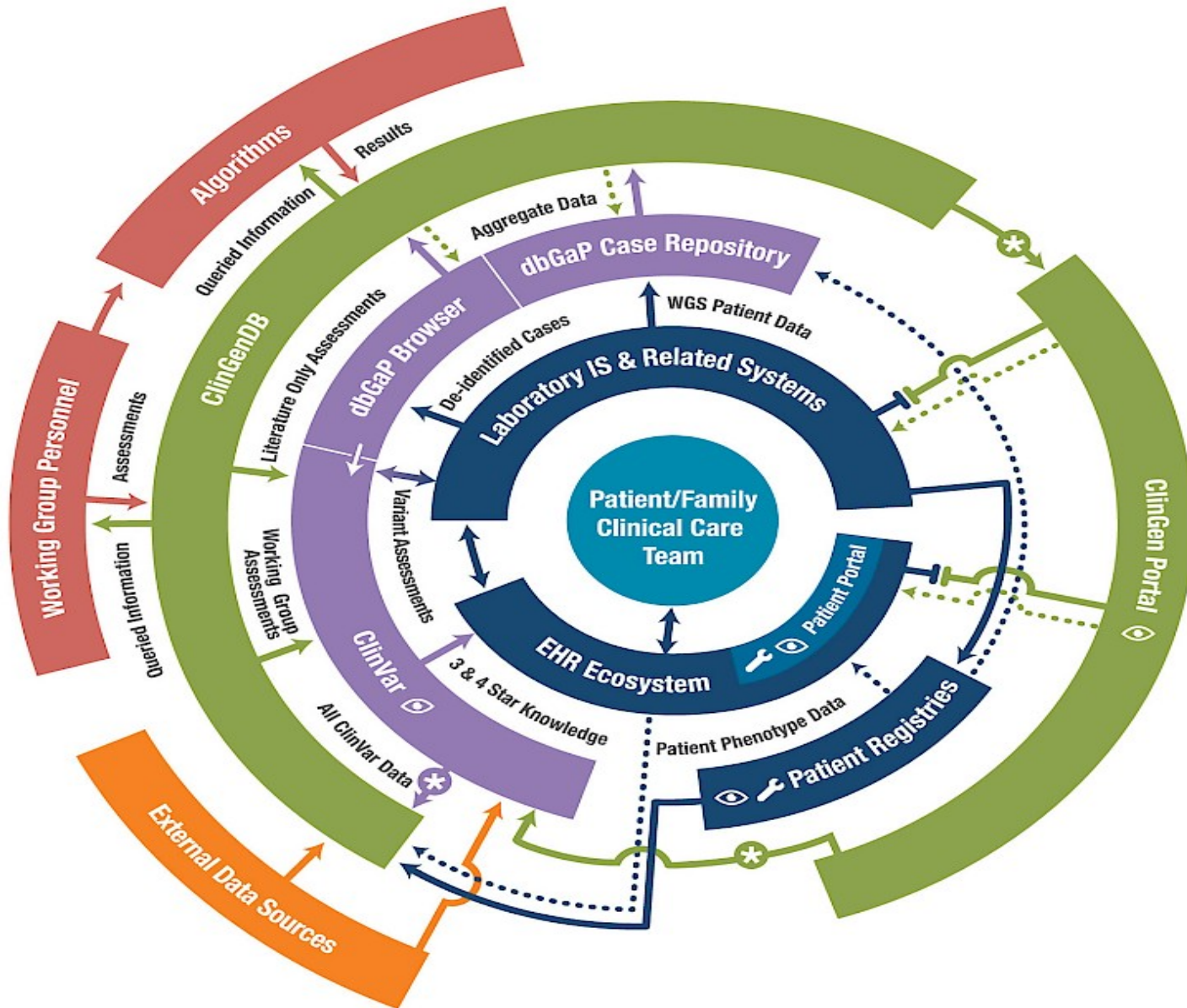


•ClinicalGenome.org

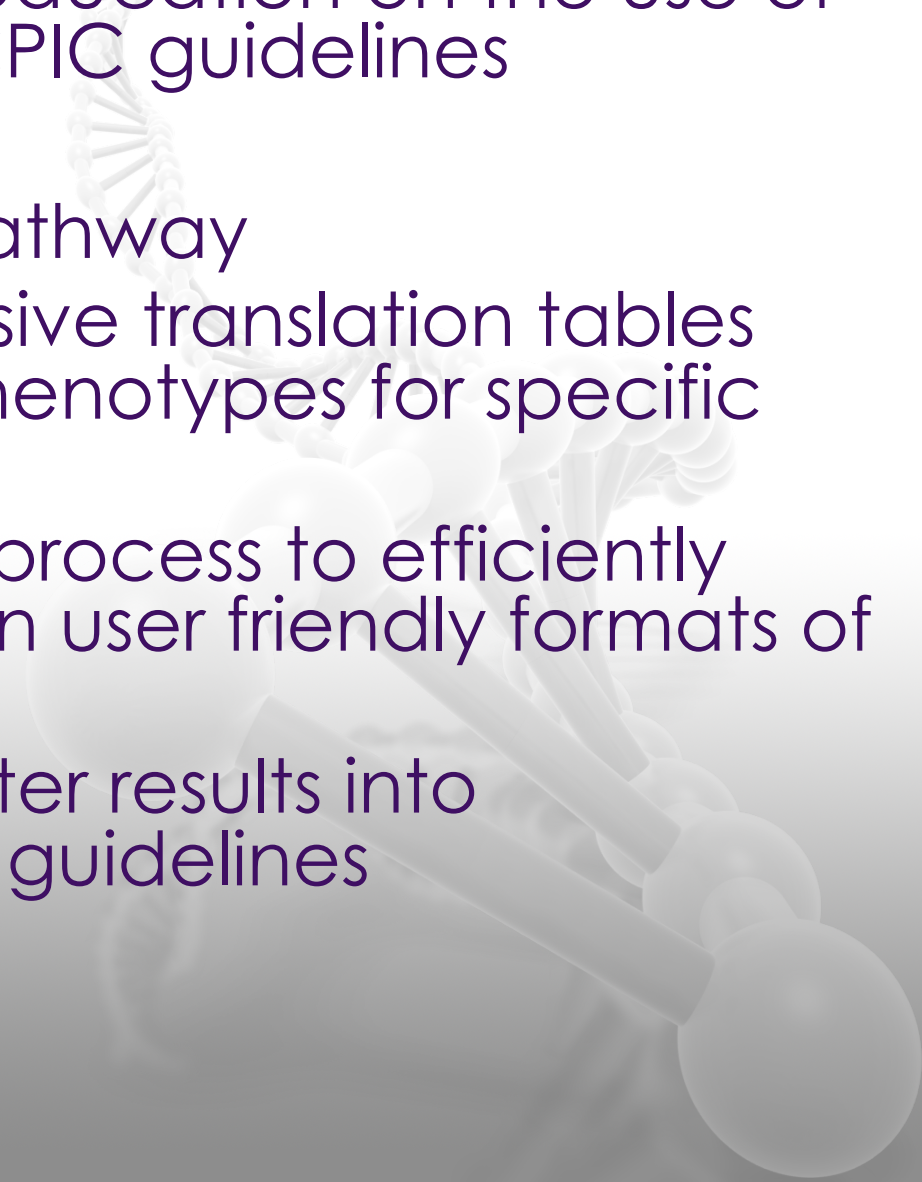
ClinGen is a National Institutes of Health (NIH)-funded resource dedicated to building an authoritative central resource that defines the clinical relevance of genomic variants for use in precision medicine and research.

Working Groups: Actionability, Clinical Domain, **Standards and Data Submission**, Data Model, Education, Engagement and Counseling, EHR, Consent and Disclosure, External Scientific Panel, Gene Curation, Informatics and Analysis, Phenotyping, Sequence Variant, Steering Committee, Structural Variant

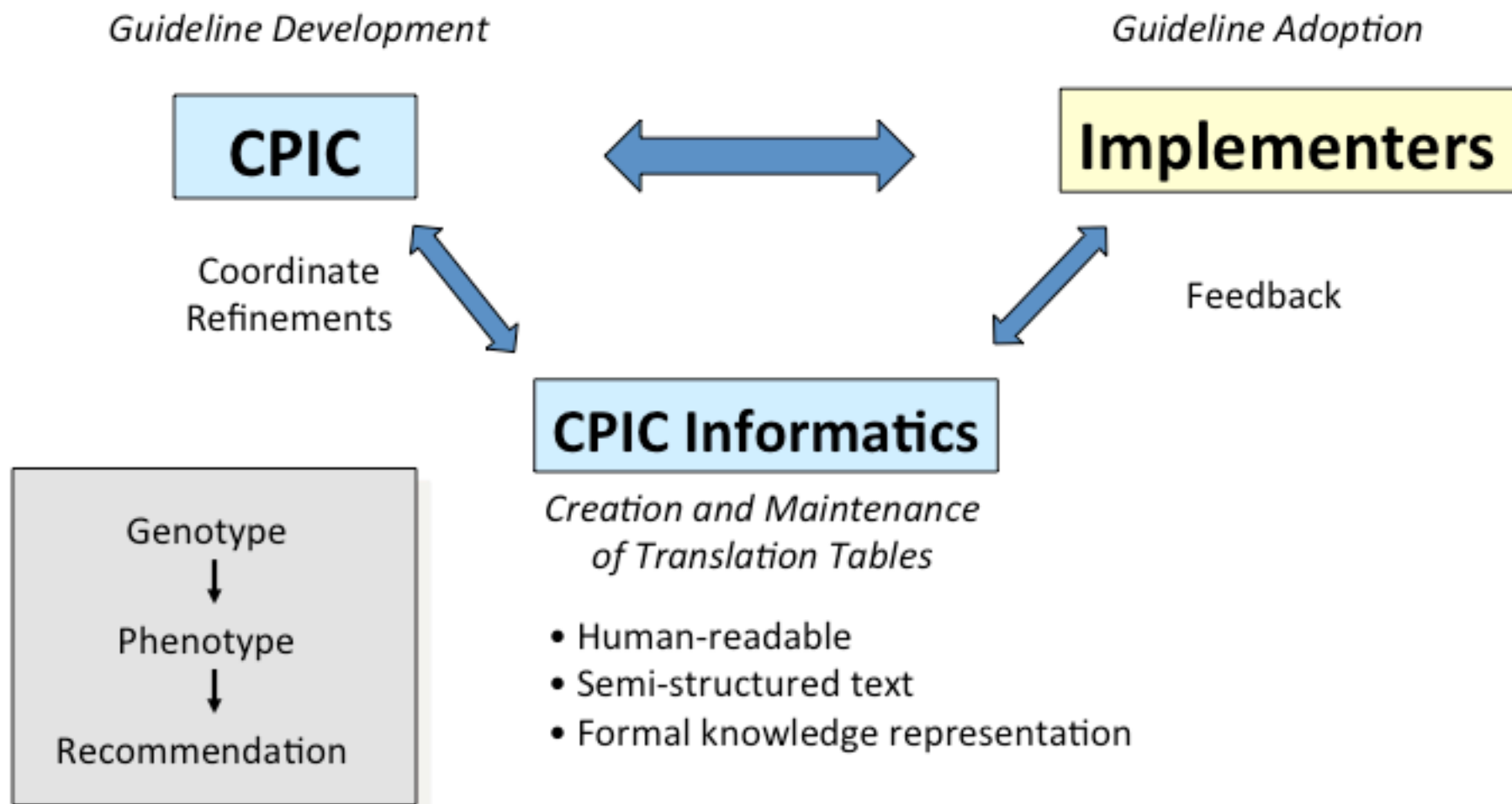
•ClinGen Resource Map



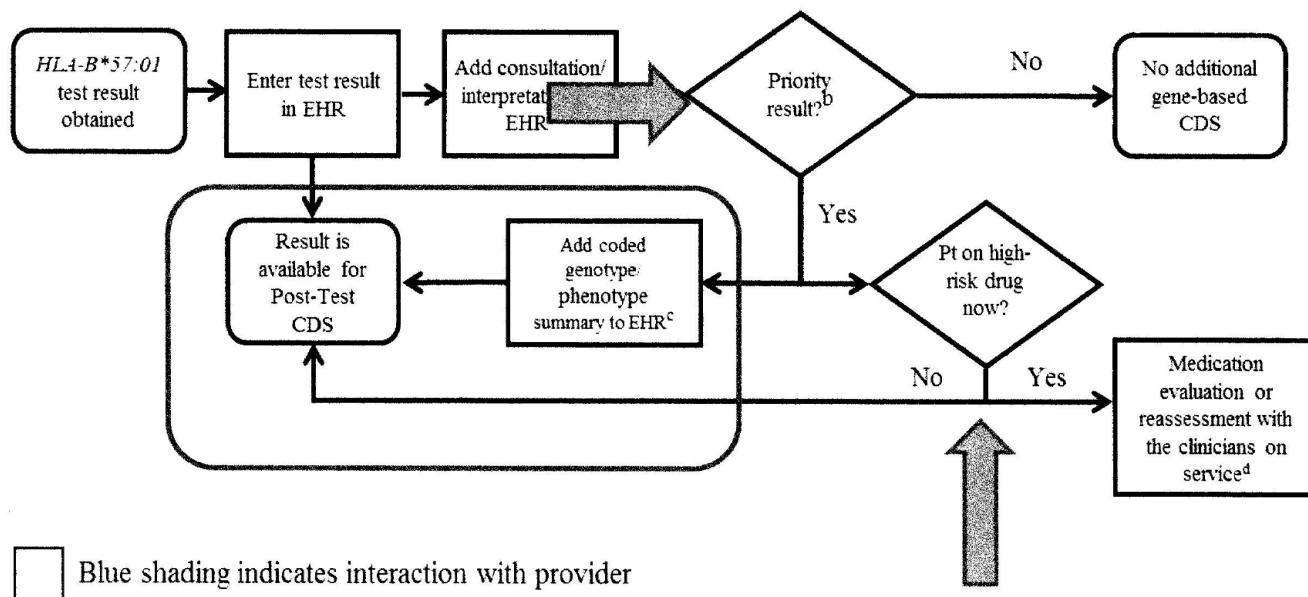
• **Roadmap to CDS from CPIC into EHR**

- Provide access and education on the use of the PharmGKB and CPIC guidelines
 - Describe workflow
 - Develop algorithm pathway
 - Develop comprehensive translation tables from genotypes to phenotypes for specific drugs
 - Define structure and process to efficiently develop and maintain user friendly formats of CDS text
 - Publish results and enter results into PharmGKB and CPIC guidelines
- 

CPIC Informatics: Supporting Guideline Implementation



HLA-B*57:01 Pharmacogenetic Test Result: Clinical Implementation Workflow for EHR



•DIGITize – July 20, 2015 at HL7 Genomics Policy Conference + IOM Roudtable for Translating Genomic-Based Research For Health

- Discussed Cerner, Epic and Allscripts vision for integrating genomic information into the EHR
- Consensus that APOS need to plug into the EHR – like SMART and FHIR (Fast Healthcare Interoperability Resources)
- FHIR can be used to mediate decision support and data dumps
- SMART on FHIR can allow consumer apps to connect their data through a patient portal
- IOM will launch pilot studies that concentrate on pharmacogenomics examples with vendors.

•DIGITize from IOM Use Cases (Sandy Aronson Partners Chair)

- Building on the eMERGE, IGNITE, and ClinGen projects
- Will start with HLA-B - Abacabir
- because 6% of patients have hypersensitivities that are life threatening
- Second use case will be Azathioprine, TPMT
 - Risk for patient for myelosuppression
- Also developing an Implementation Guide, LOINC Transfer Codes, and Allele Registry with ClinGen

•Allscripts bought NantHealth for \$200M

- June 30, 2015 – Allscripts bought NantHealth which will integrate NantHealth's genomic proteomic sequencing (GPS Cancer) diagnostic test and Eviti treatment protocol planning tool into Allscripts' EHR and Practice Management Software.



**What are some
Resources to Follow?**



Goals of Precision Medicine Initiative



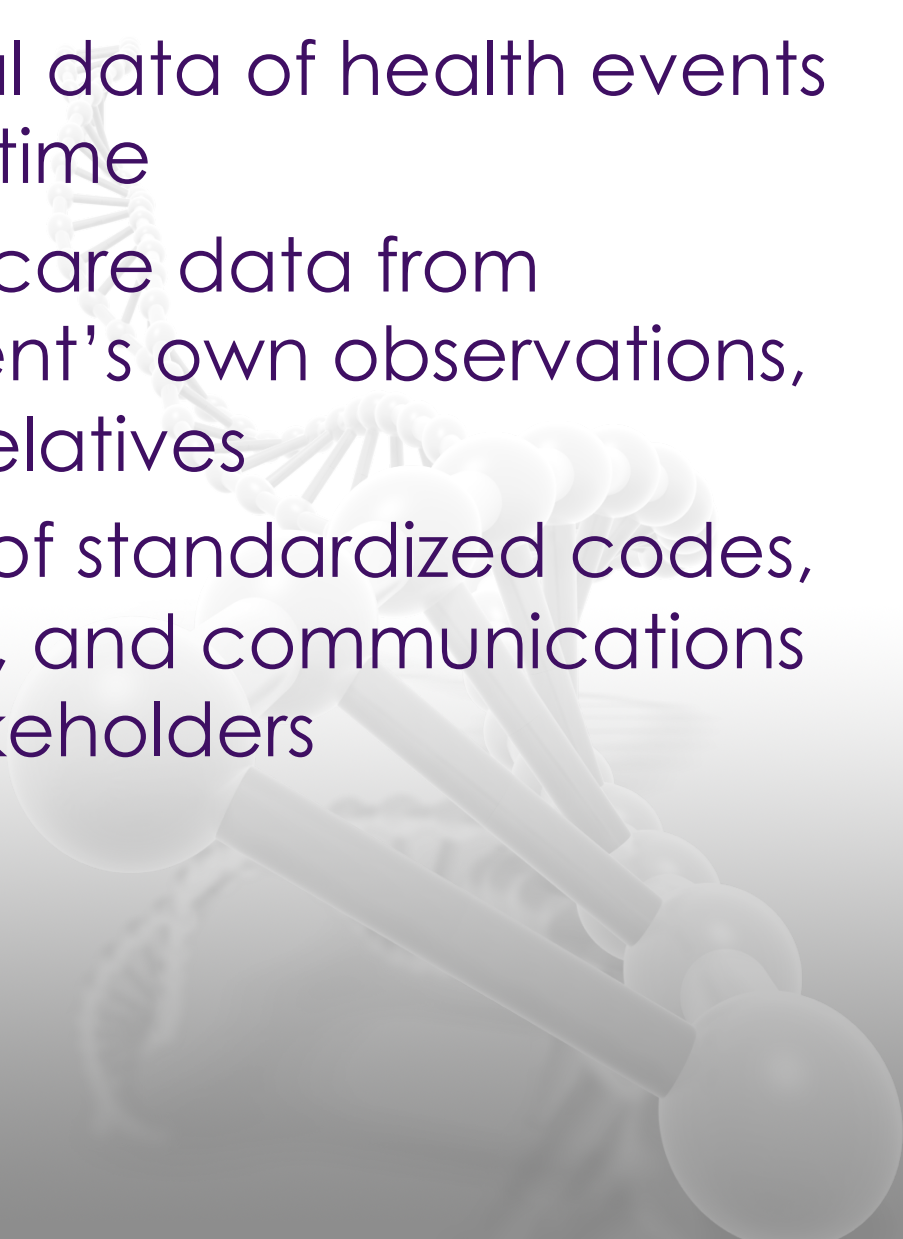
- Near Term Goals

- Innovative clinical trials of targeted drugs for adult and pediatric cancers
- Use of combination therapies
- Knowledge to overcome drug resistance

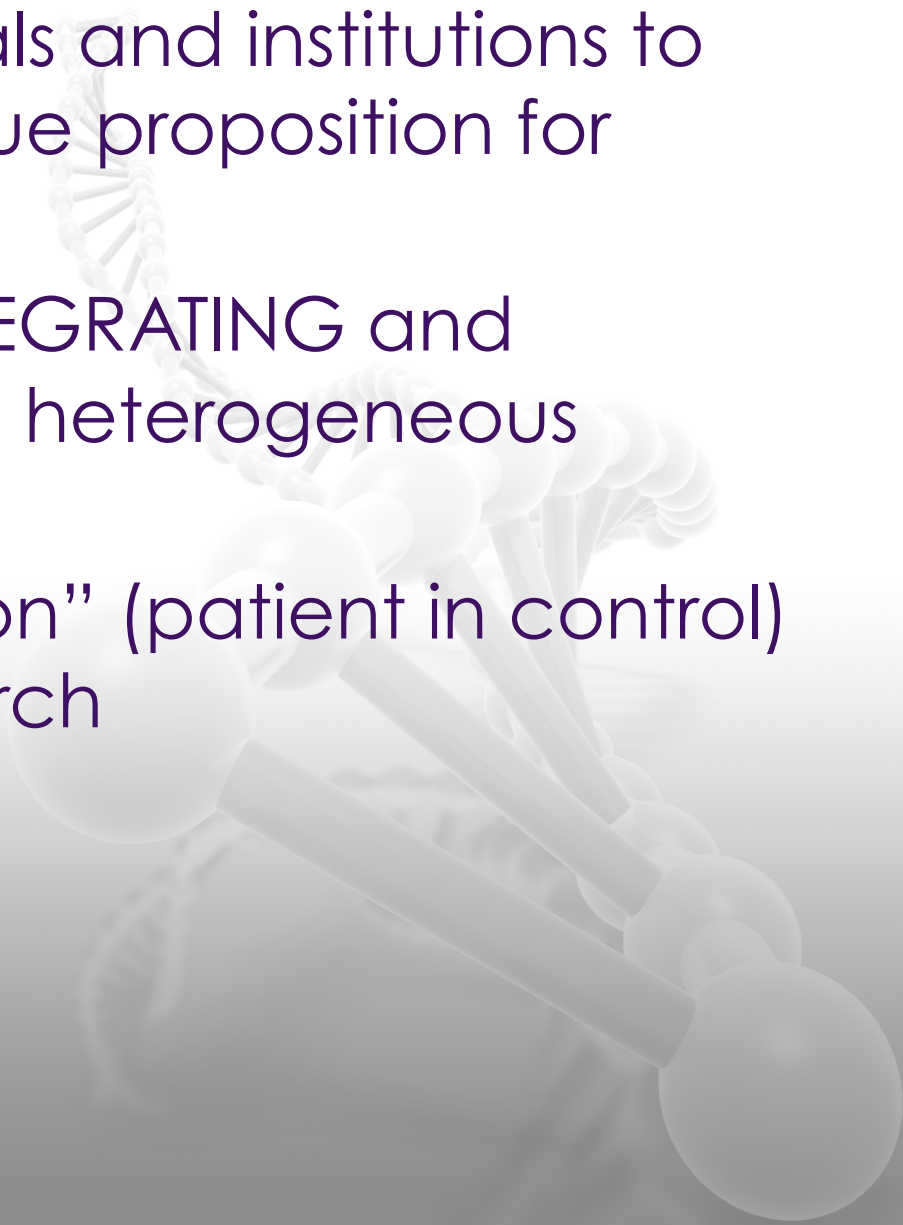
- Longer Term Goals

- Pioneer a new model for doing science that emphasizes engaged participants, responsible data sharing, and privacy protection.
- Research based upon cohort data:
 - Advance pharmacogenomics, the right drug for the right patient at the right dose
 - Identify new targets for treatment and prevention

•Why the EHR?

- It contains longitudinal data of health events over a long period of time
 - It may contain healthcare data from professional, the patient's own observations, and family history of relatives
 - The data can consist of standardized codes, text, video and audio, and communications between multiple stakeholders
- 

•High Priority Issues

- The ability of individuals and institutions to participate and a value proposition for project participation
 - Technical issues in INTEGRATING and ANALYZING data from heterogeneous systems
 - Enhancing “Blue Button” (patient in control) functionality for research
 - Industry engagement
 - Cyber security
- 

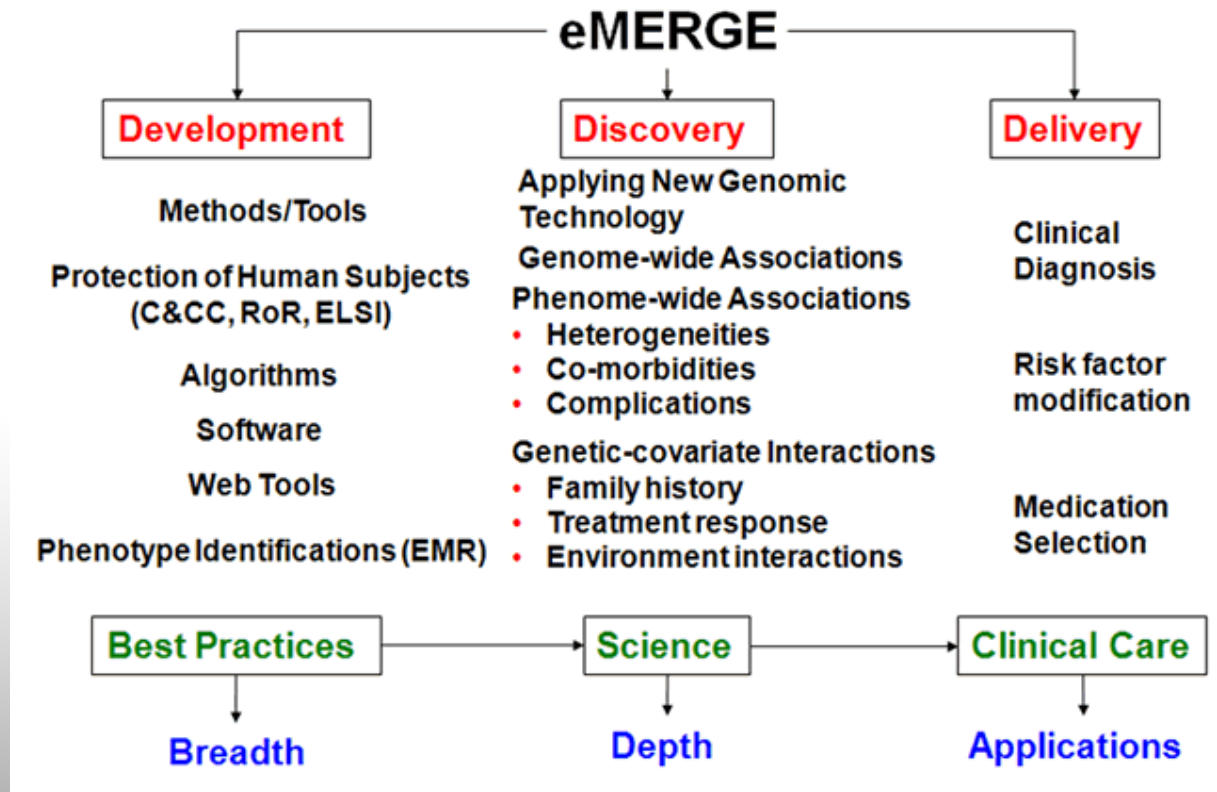


4 White Papers for Precision Medicine February 11-12, 2015

- Building a Consortium of Cohorts – Cohort Identification and Participant Recruitment
- Participant Engagement, Data Privacy, and Novel Ways of Returning Information to Participants
- Data Collection and Mobile Technologies
- Opportunities and Challenges related to the use of Electronic Health Records data for research

[http://www.nih.gov/precisionmedicine/
whitepapers.htm](http://www.nih.gov/precisionmedicine/whitepapers.htm)

•Goals of eMERGE consortium



Resources to Assure Security and Privacy of Clouds

- Cloud Threats, Technical Risks and Safeguards NIST Grance and Jansen, Guidelines on Security and Privacy in Public Cloud Computing 2011 http://www.nist.gov/manuscript-publication-search.cfm?pub_id=909494
- NIST Cloud Guidelines for Managing Security and Privacy 2012 <http://www.nist.gov/itl/csd/cloud-012412.cfm>

•NEW NIH POSITION STATEMENT ON STORAGE OF GENETIC DATA ON CLOUDS

NEW- NIH Position Statement on Use of Cloud Computing Services for Storage and Analysis of Controlled-Access Data Subject to the NIH Genomic Data Sharing Policy (March 23, 2015) :

http://gds.nih.gov/pdf/NIH_Position_Statement_on_Cloud_Computing.pdf

•Some Resources to Follow to Keep Up to Date

From McCormick, K. A., Calzone, KA (2015). *Genomics and Information Technology for Personalized Health*.
 In V. K. Saba & K. A. McCormick (Eds.), *Essentials of Nursing Informatics*, version 6. New York, NY: McGraw-Hill.

| Title | Website |
|---|---|
| Electronic Medical Records and Genomics (eMERGE) Network | http://www.genome.gov/27540473 |
| Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees | http://www.nursingworld.org/MainMenuCategories/EthicsStandards/Genetics-1/Essential-Genetic-and-Genomic-Competencies-for-Nurses-With-Graduate-Degrees.pdf |
| Genetics/Genomics Competency Center for Education (G2C2) | http://www.g-2-c-2.org |
| Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators, 2nd Edition | http://www.genome.gov/27527634 |
| Genetics and Genomics for Health Professionals | http://www.genome.gov/27527599 |
| Genetics and Your Practice | http://www.marchofdimes.com/gyponline/index.bm2 |
| Genetics Home Reference | http://ghr.nlm.nih.gov |
| GeneTests/Gene Reviews | http://www.genetests.org |
| Genetics is Relevant Now: Nurses' Views and Patient Stories | http://www.cincinnatichildrens.org/ed/clinical/gpnf/resources/curriculum/relevant-genetics.htm |
| Genomics and Disease Prevention | http://www.cdc.gov/genomics/ |
| Genetics, Health, and Society | http://oba.od.nih.gov/SACGHS/sacghs_home.html |
| Global Genetics and Genomics Community (G3C) | http://www.g-3-c.org |
| International Society of Nurses in Genetics | http://www.isong.org |
| National Institute of Nursing Research Summer Genetics Institute | http://www.ninr.nih.gov/Training/TrainingOpportunitiesIntramural/SummerGeneticsInstitute/ |
| Pharmacogenomics Education Program | http://pharmacogenomics.ucsd.edu/ |
| Teaching Genetics Genetic Science Learning Center | http://teach.genetics.utah.edu/ |
| U.S. Surgeon General's Family History Initiative | http://www.hhs.gov/familyhistory/ |


•Resources to Follow for Education

- Genetic and Genomic Competencies for all nurses
<http://www.nursingworld.org/MainMenuCategories/EthicsStandards/Genetic-1/Essential-Genetic-and-Genomic-Competencies-for-Nurses-With-Graduate-Degrees.pdf>
- ANA Learning the Basics of Genomics -
<http://ananursece.healthstream.com>
- PDQ Cancer Information Summaries-Genetics Continuing Education=
<http://www.cancer.gov/cancertopics/genetics>
- Six weeks to Genomic Awareness offered by the Michigan Public Health Training Center =
http://www.practice.sph.mich.edu/mphtc/site.php?module=courses_one_online_course&id=108
- Cincinnati Children's Hospital Independent self-paced modules: Interpreting Family History, Ethical and Social Issues Related to Genetic Testing, Nurses' Role in Pharmacogenetics/pharmacogenomics, and promoting informed decision making about genetic testing=
<http://www.cincinnatichildrens.org/education/clinical/nursing/genetics/cont/self/default/>
- Coursera courses, Useful Genetics, Parts 1 and 2 =
<https://www.coursera.org/course/usefulgenetics>
- <https://www.coursera.org/course/usefulgenetics2>
-

•Four Additional Resources

1. Lee, D.H., Cheek, D.J., Brazeau,D., Brazeau, G. (2015) Mastering Pharmacogenomics. Sigma Theta Tau International, IN: Indianapolis.
2. McCormick, K. A., Calzone, KA (2015). Genomics and Information Technology for Personalized Health.
 - In V. K. Saba & K. A. McCormick (Eds.), Essentials of Nursing Informatics, version 6. New York, NY: McGraw-Hill.
3. Journal of Nursing Scholarship, Special Issue on Genomics, 45(1), 1–103. Also available at <http://www.genome.gov/27552093>.
4. Journal of Nursing Scholarship- May 2015. Jenkins, J. Et al. Methods of Genomic Competency Integration in Practice. 47:3,200-210.

•Further Resources

- CDC Listserv
 - Nursing Genomic Listserv International Society of Nurses in Genetics
 - Annual Meeting October
 - Genetic Testing Registry
 - OMIM
 - National Human Genome Research Institute
 - National Cancer Institute
 - PDQ
 - NCI NCIP -Cloud Initiative
 - PharmGKB
- 



**What have some
Magnet Hospitals
Done to Include
Genetics in Practice?**

•NI Examples

- Assure the family history section in an EHR elicits a minimum of three generation family history and the physical assessment section includes information regarding genetic and environmental information and risk factors.
- Assist in identification of current genetic and genomic information resources that should be included in clinical practice guidelines orchestrated in the EHR.
- Works on policies regarding access to genomic information stored within the EHR.
- Understands the issues around genomic privacy and identifies appropriate state legislation, legal and social issues related to use and potential misuse of genomic information.

Family History Data Feero, Bigley and Brinner, 2008 JAMIA

Data Category (Individual)

Identification

Age

Date of death

Cause of death

Ethnicity/race (self-identified)

Biological sex

Multiple-birth status

Biological parents identified

Consanguinity

Adoptive status

Disorders

Research identifier placeholder

Relevant genetic/genomic test results

Data field for "unknown" Denotes that question was asked but answer was not known

Approximate dates/ages for data fields

Sensitive fields

"Certainty of data"

Integration with other EHR elements

Systems should not force

duplicate entry of family history data that is already stored in a legacy system (e.g., age, past medical history relevant to family history, and self-described ethnicity data).

Ability to define data sharing status

Global means to provide patient maximum control over sharing of his/her data with individuals including family members.

Text box for annotations

• Example of documenting toxicities - NIH Advisory Council BD2K-

Medidata RAVE® Toxicity (Adverse Event) Page

DEV iMedidata Messages My Profile Help Home Logout
User: Whitnev Smith Project Manager
John Smith

9177 MDSOL New Subject Ongoing Adverse Events

Patient Initials (LFM):

Subject [New Subject](#)
Page [Adverse Events - Ongoing](#)

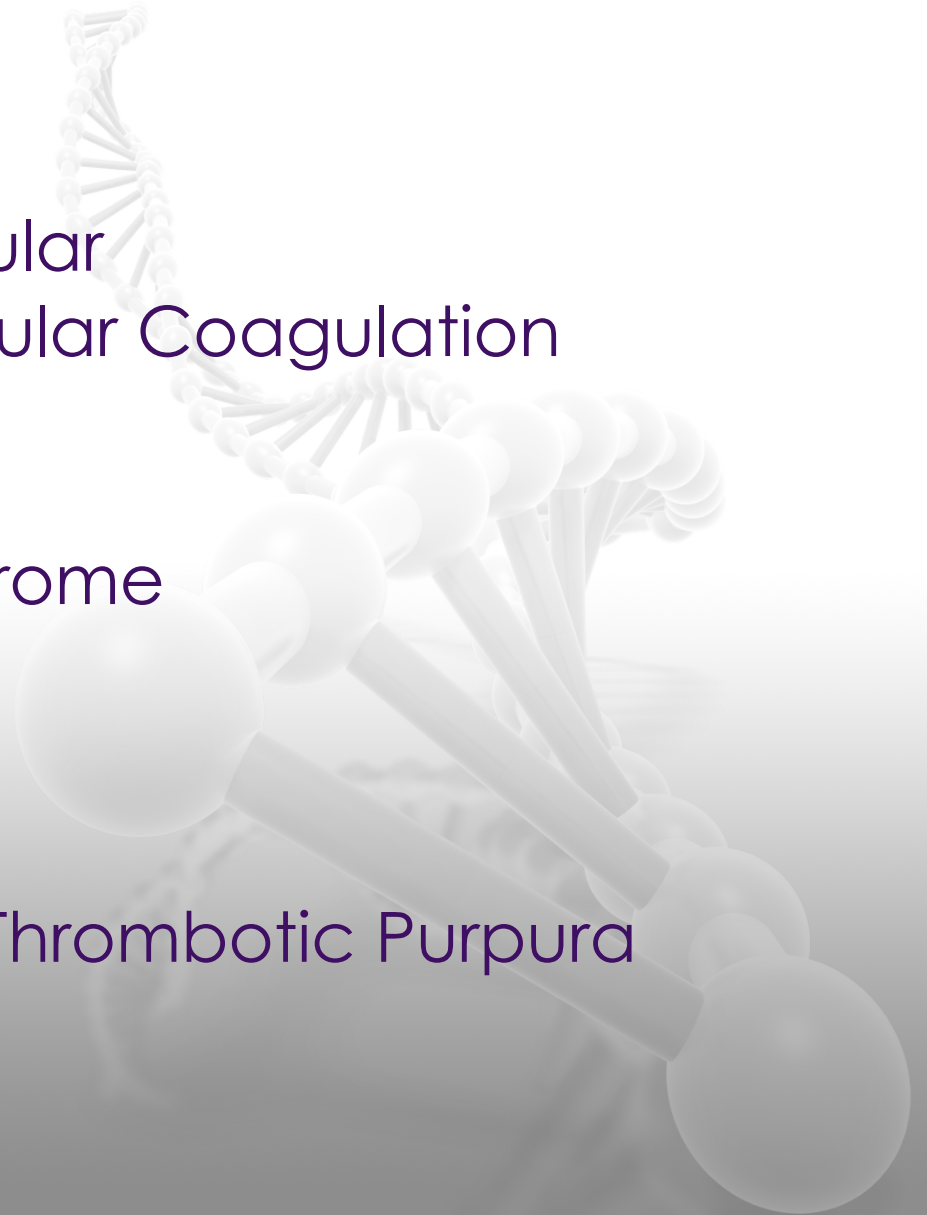
Visit

Record all Grade 3 or higher AEs. Record only Unexpected Grade 2 AEs. Record Grades 1 and higher for all events listed in protocol section 8.1.1. Record each event only one time per cycle of treatment, identifying the highest grade of the event.

| # | Adverse Event Text Name (CTCAE v4.0) | MedDRA Adverse Event Code (v12.0) | Adverse Event Grade | Adverse Event Grade Description | CTC Adverse Event Attribution Scale | Has an event reported? |
|---|--|-----------------------------------|---------------------|---------------------------------|-------------------------------------|---|
| 1 | <div style="border: 1px solid black; padding: 5px;"> <ul style="list-style-type: none"> Anemia Bone marrow hypocellular Disseminated intravascular coagulation Febrile neutropenia Hemolysis Hemolytic uremic syndrome Leukocytosis Lymph node pain Spleen disorder Thrombotic thrombocytopenic purpura </div> | | | | | <input type="radio"/> No <input type="radio"/> Yes |

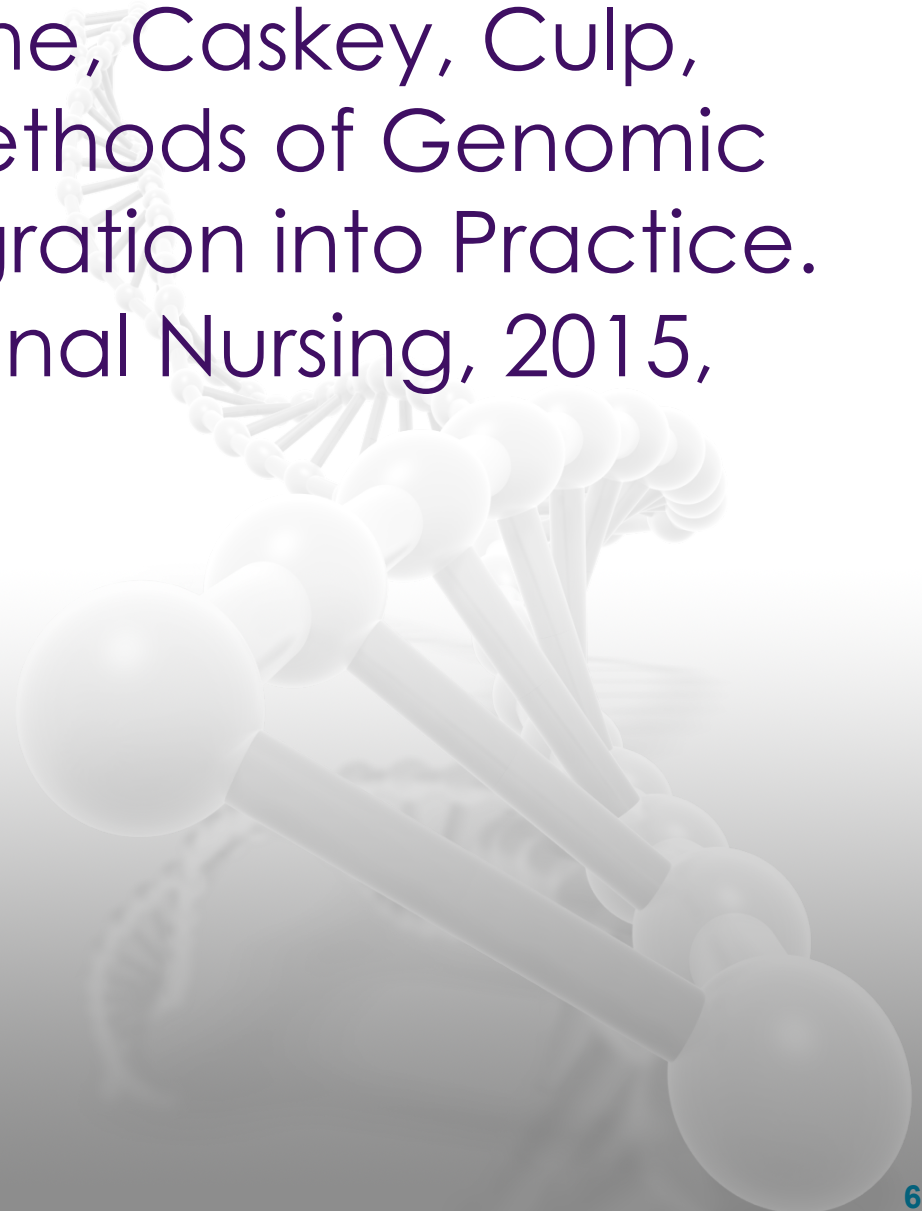
• Adverse toxic events

- Anemia
- Bone Marrow Hypocellular
- Disseminated Intravascular Coagulation
- Febrile Neutropenia
- Hemolysis
- Hemolytic Uremic Syndrome
- Leukocytosis
- Lymph Node Pain
- Spleen Disorder
- Thrombocytopenic Thrombotic Purpura

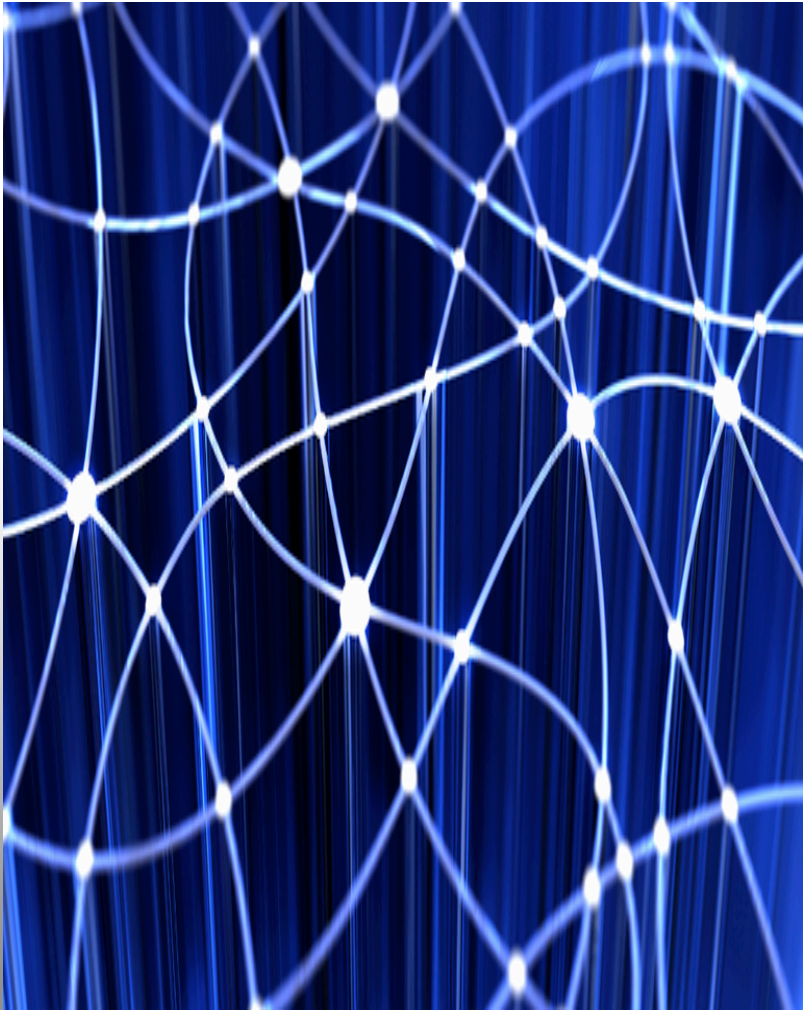


•Integration into Practice

- See Jenkins, Calzone, Caskey, Culp, Weiner, Badzek. Methods of Genomic Competency Integration into Practice. Journal of Professional Nursing, 2015, 47-3, 200-210)



•A Peek Into the Future



- Complete a mutational atlas for all cancer tumors, all types of heart, immunological, neurological diseases
- Expand beyond the atlas to: metastases, recurrence to full recovery
- Systematic functional annotation
- Systematic clinical utilization
- Data sharing nationally and internationally
- Attempts to genotype 1 million Americans and share data



Questions/Discussion

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