

Clinical Informatics to Support Genomic Medicine for Diverse EHR systems

*IGNITE Clinical Informatics Interest Group
(CIIG)*

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Clinical Informatics and Genomic Medicine

- Implementation of genomic medicine requires:
 1. Clear, actionable representations of genomic risks and *clinical guidance* within the EHR
 2. Communication with the patient and exchange of structured information between sites of care for long-term management of genomic risks
- The Clinical Informatics Interest Group (CIIG) within IGNITE is positioned to make contributions toward both of these objectives with a **mission of supporting an open forum for clinical informatics and summarizing and disseminating best practices.**

Adoption of EHRs 2004- 2014

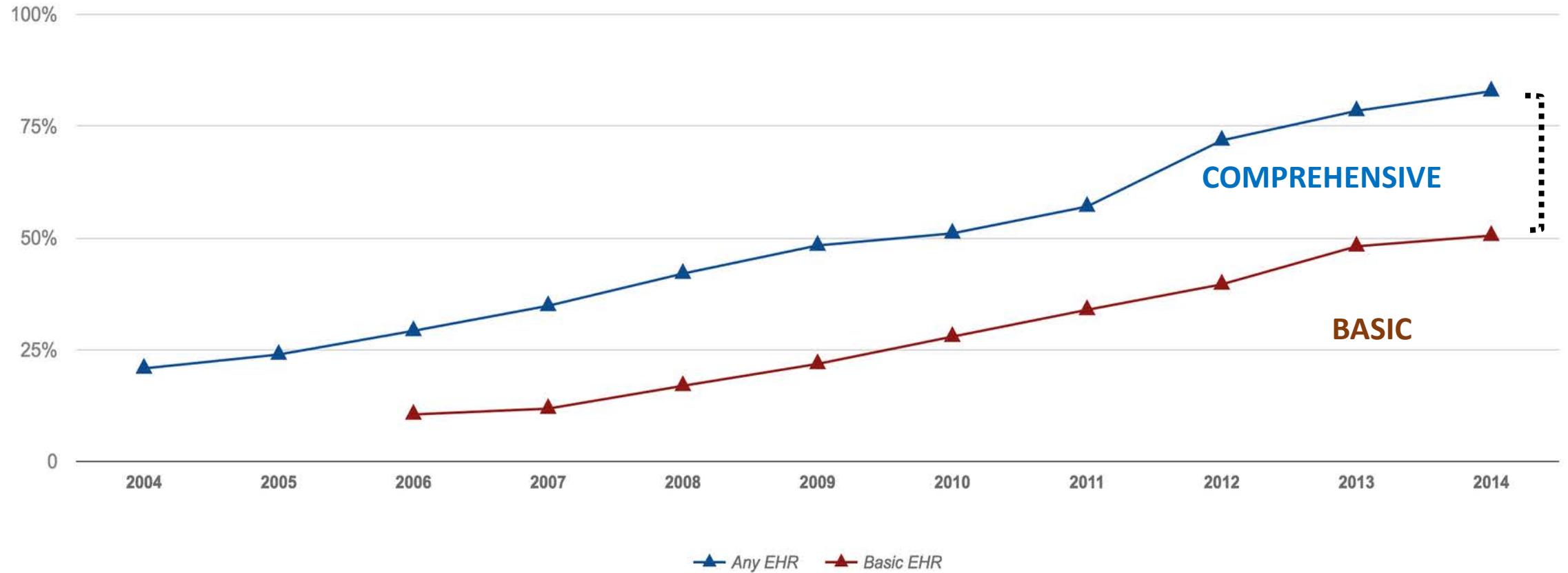


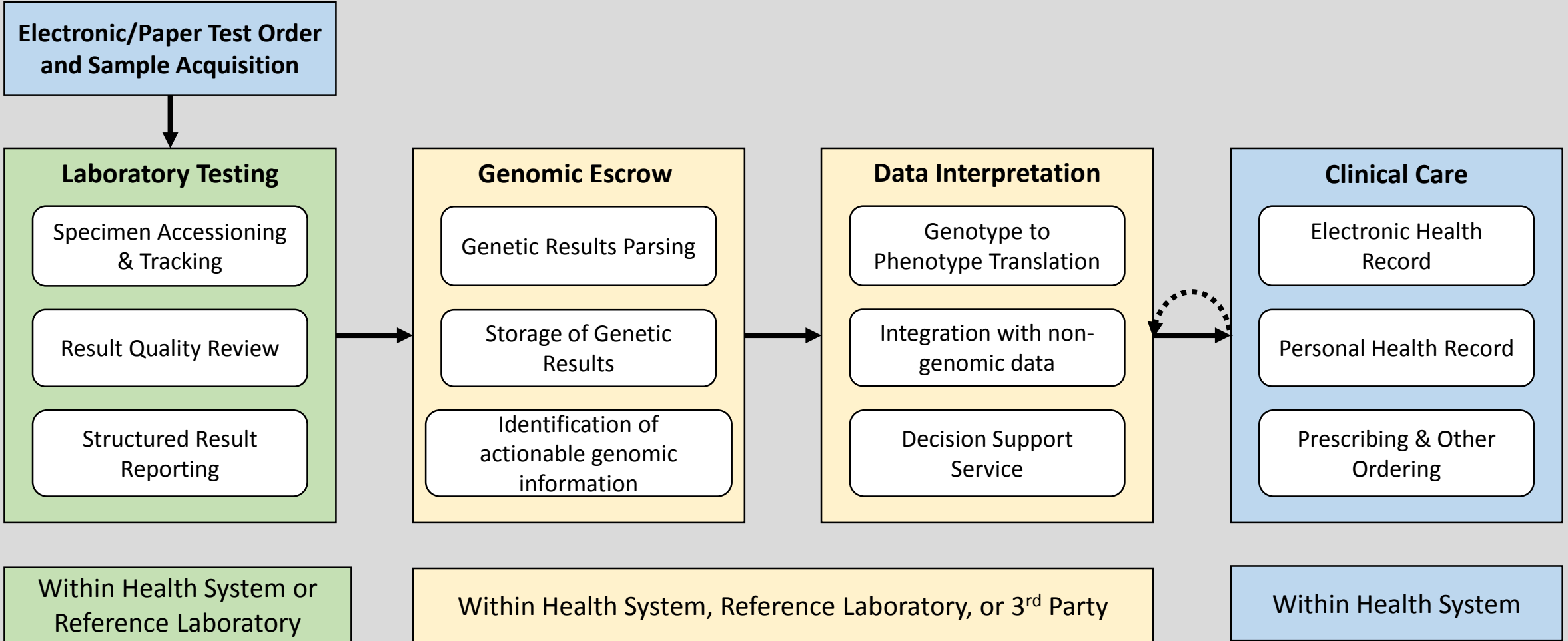
Table 2: Electronic Functions Required for Hospital Adoption of Basic or Comprehensive EHR Systems

EHR Functions Required	Basic EHR without Clinician Notes	Basic EHR with Clinician Notes	Comprehensive EHR
Electronic Clinical Information			
Patient demographics	★	★	★
Physician notes		★	★
Nursing assessments		★	★
Problem lists	★	★	★
Medication lists	★	★	★
Discharge summaries	★	★	★
Advance directives			★
Computerized Provider Order Entry			
Lab reports			★
Radiology tests			
Medications	★		
Consultation requests			
Nursing orders			
Results Management			
View lab reports	★		
View radiology reports	★		
View radiology images			
View diagnostic test results	★		
View diagnostic test images			
View consultant report			
Decision Support			
Clinical guidelines			
Clinical reminders			
Drug allergy results			
Drug-drug interactions			
Drug-lab interactions			
Drug dosing support			

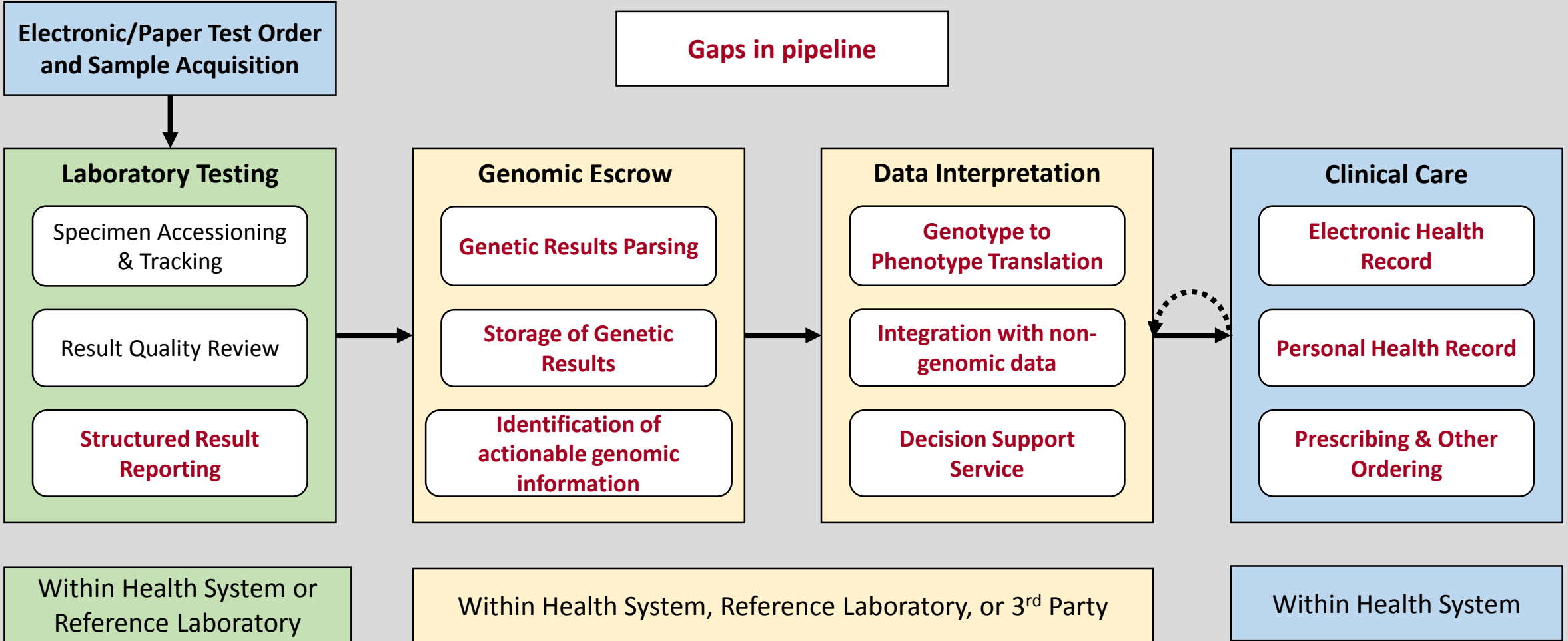
Decision Support
Clinical guidelines
Clinical reminders
Drug allergy results
Drug-drug interactions
Drug-lab interactions
Drug dosing support

NOTES: Basic EHR adoption requires each
 Comprehensive EHR adoption requires each

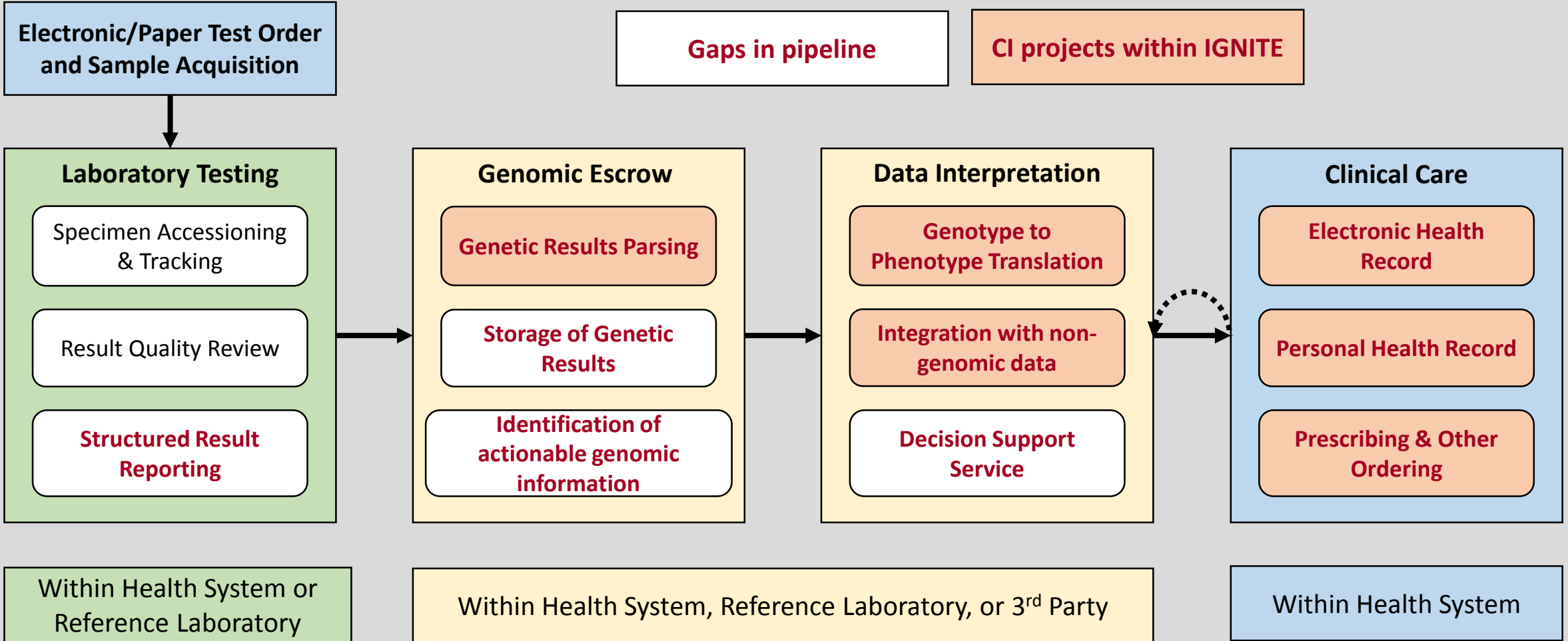
CLIA-compliant Genomic Data Pipeline



CLIA-compliant Genomic Data Pipeline



CLIA-compliant Genomic Data Pipeline

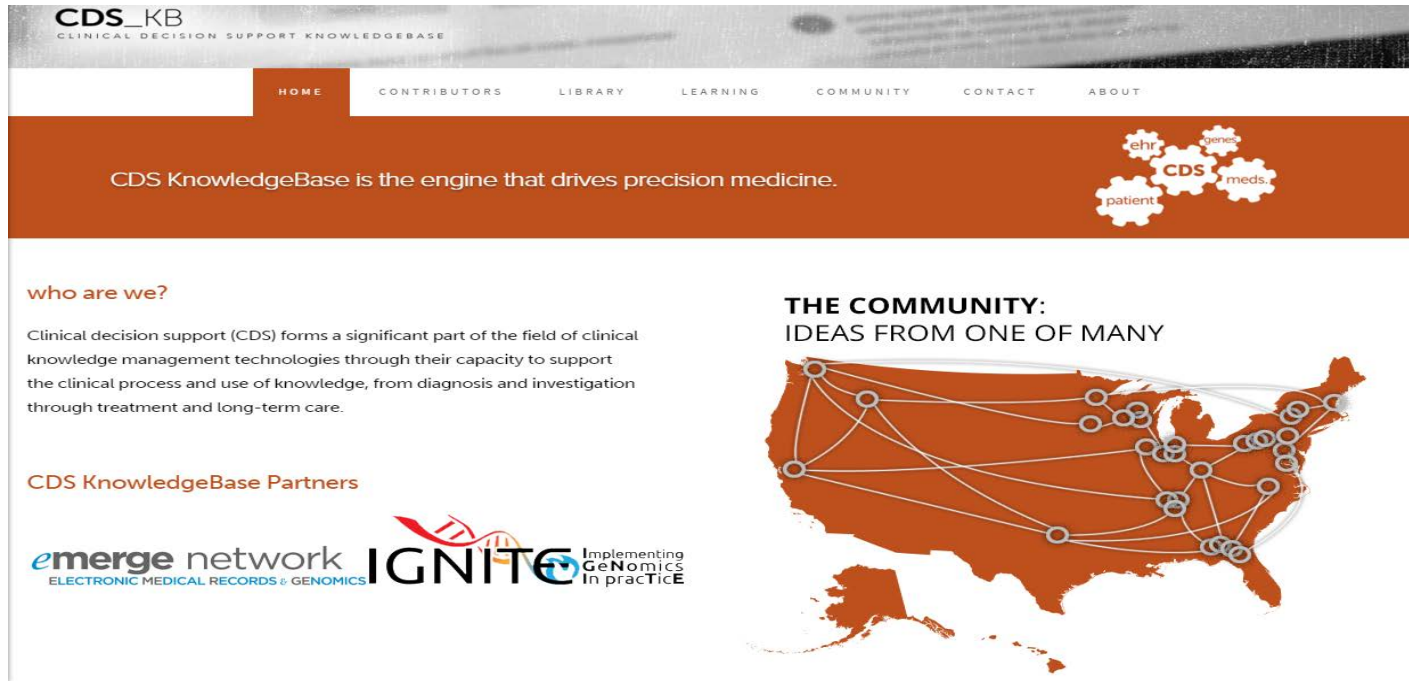


Major Informatics Challenges

- Standard representation of genetic results
- Standard representation of metadata (sequencing or genotyping scope)
- Preserving original genetic data for future reanalysis
- Detecting need for re-analysis based on new findings
- Delivering clinical guidance based on genetic risks to the right person and place within the workflow
- Securely communicating findings to patients and their families and facilitating long term access to results
- Coordination between institutions, reference labs, and vendors
- Transfer of discrete genetic data between sites

CIIG Highlights: Establishing CDSKB.ORG

- Develop a clinical decision support document resource: Clinical Decision Support Knowledge Base (CDS-KB) – Access at <http://cdskb.org>




The screenshot shows the homepage of the CDS KnowledgeBase. At the top, the logo reads "CDS_KB CLINICAL DECISION SUPPORT KNOWLEDGBASE". A navigation menu includes "HOME", "CONTRIBUTORS", "LIBRARY", "LEARNING", "COMMUNITY", "CONTACT", and "ABOUT". Below the menu is a dark blue banner with the text "CDS KnowledgeBase is the engine that drives precision medicine." and a gear icon containing the words "ehr", "genes", "CDS", "meds", and "patient".

who are we?
Clinical decision support (CDS) forms a significant part of the field of clinical knowledge management technologies through their capacity to support the clinical process and use of knowledge, from diagnosis and investigation through treatment and long-term care.

CDS KnowledgeBase Partners

emerge network **IGNITE** Implementing GeNomics In pracTICE

THE COMMUNITY: IDEAS FROM ONE OF MANY



- 59 artifacts from 13 institutions and organizations
- Registered users: 211

LOGIN TO CDS_KB

Want to add to this library? [Click here.](#)

SEARCH BY CONTRIBUTOR:

Icahn School of Medicine at

FILTER RESULTS BY REQUIRED
ELEMENTS:

- CDS Architecture diagrams
- Usage scenarios
- CDS Presentation
- Workflow
- Mapping and translation tables
- Algorithms and pseudocode
- Patient materials
- Uncategorized

CLOPIDOGREL

Icahn School of Medicine at Mount Sinai ^

WORKFLOW DIAGRAM AND TEXT DISPLAY

WORKFLOW : MOUNT SINAI WORKFLOW DIAGRAM AND DISPLAY TEXT --
CLOPIDOGREL, CYP2C19.PDF

DOWNLOAD:

[pdf version](#)

STEVE ELLIS | AVAILABLE FROM: [HTTPS://CDSKB.ORG/ARTIFACT/?ID=30](https://CDSKB.ORG/ARTIFACT/?ID=30)

CDS ARCHITECTURE

Icahn School of Medicine at Mount Sinai ^

ARCHITECTURE FOR GENOMIC-INFORMED CLINICAL DECISION SUPPORT.

CDS ARCHITECTURE DIAGRAMS : MOUNT SINAI CDS ARCHITECTURE.PDF

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NO CONTACTS | AVAILABLE FROM: [HTTPS://CDSKB.ORG/ARTIFACT/?ID=39](https://CDSKB.ORG/ARTIFACT/?ID=39)

CYP2C19 / Clopidogrel Alert

Vanderbilt University Medical Center

Drug-Genome Advisor

Intermediate Metabolizer - clopidogrel (Plavix) - Rare Risk Allele
Substitution recommended due to increased cardiovascular risks

If not otherwise contraindicated:

- Prescribe prasugrel (Effient) 10 mg daily

Prasugrel should not be given to patients:

- history of stroke or transient ischemic attack
- ≥ 75 years of age [Current patient age: 51]
- with body weight < 60 kg [Current patient weight: 59.0 kg as of 10/12/2012]

- Prescribe ticagrelor (Brilinta) 90 mg twice daily

Ticagrelor should not be given to patients:

- history of severe hepatic impairment
- intracranial bleed

- Continue with clopidogrel (Plavix) prescription

Primary override reason:



- Contraindicated for prasugrel or ticagrelor
- Potential side effects
- Provider/Patient opts for clopidogrel
- Cost

[Evidence Link](#)

Screenshot from Vendor System – CYP2C19 / Clopidogrel Alert

Northwestern University

▼ Genetic test results indicate patient may be a poor metabolizer of clopidogrel Consider alternative- medication may be ineffective if prescribed

Acknowledge reason:  

[Discussed result with patient](#)

- ↗ Click here to add "Resistance to clopidogrel" to Prob List
- ↗ Click here to view Medications
- ↗ Click here to change therapy or order a consultation
- ↗ Clopidogrel Genetic Results Fact Sheet

[Accept & Stay](#) [Accept](#) [Cancel](#)

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APOL1 Alert for Risk of Renal Failure

Mount Sinai

Genomic Medicine -- GUARDD Study

POSITIVE RESULT:

This patient has ***INCREASED RISK*** for END STAGE KIDNEY FAILURE, according to APOL1 genetic testing (result: APOL1 G2/G2)

Evidence suggests that good blood pressure control and renal function testing may forestall kidney failure.

Recent blood pressure readings for this patient were:

2014-09-28	2014-09-28	2014-09-18
138/100	120/80	120/80

[Click here for provider information.](#)

[Click here for patient materials.](#)

Note: These results will be filed under Labs / Genetics.

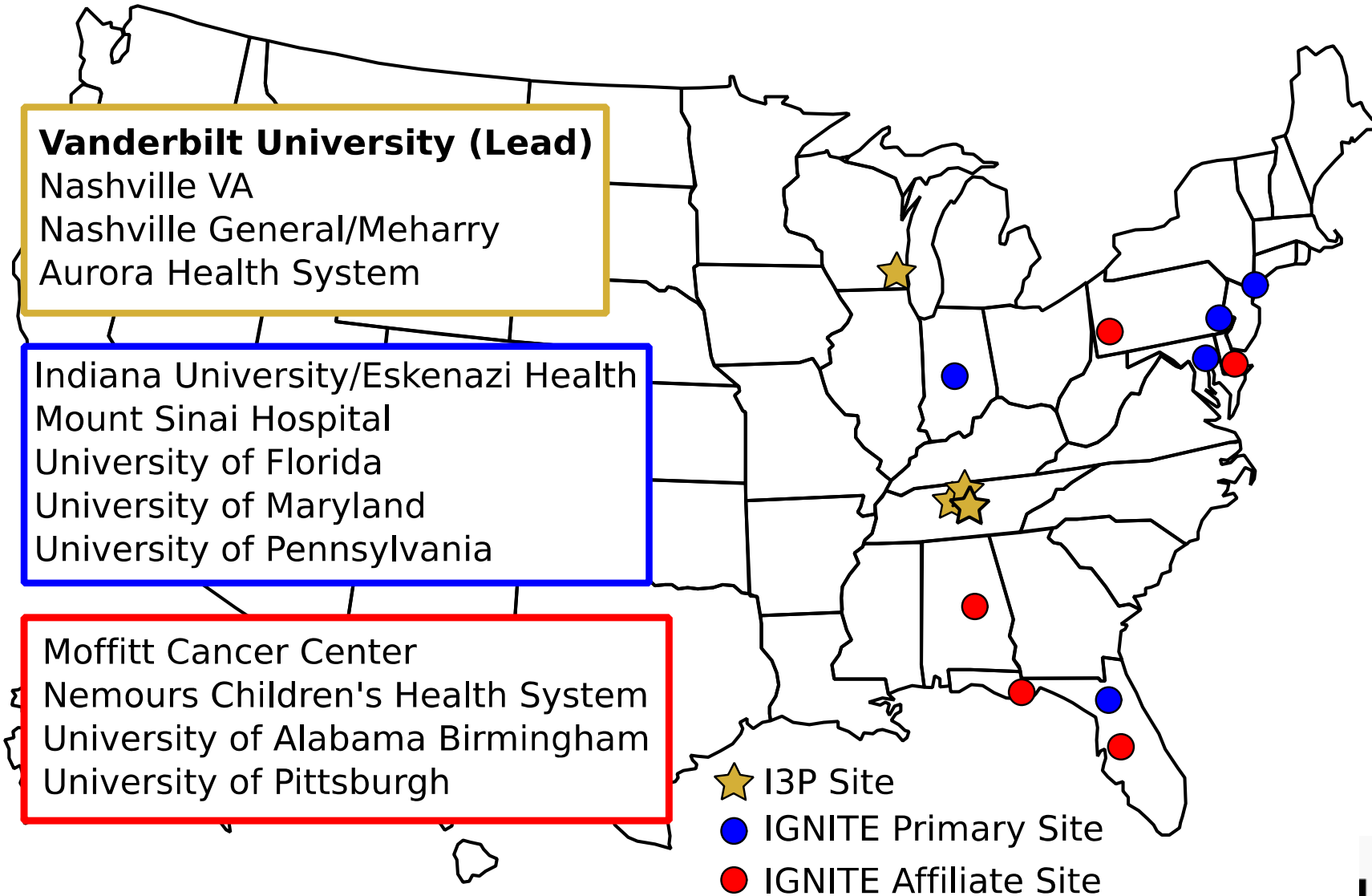
CIIG Highlights: Monthly Webinars

Date	Topic	Presenter(S)	Attendance
10.01.2015	Toward effective knowledge delivery: a proposed framework	Casey Lynnette Overby	18
11.05.2015	SMART on FHIR Genomics	Gil Alterovitz	48
12.03.2015	Genomics and Electronic Health Record	Brad Strock/ Scott Moss (Epic)	38
01.06.2015	DIGITiZE AC	Sandy Aronson (Harvard)	31
02.04.2016	Moffitt Cancer Center	Gillian Bell (Mission Health)	37
03.03.2016	Clinical Decision Support for Precision Oncology	Mia Levy (Vanderbilt)	42
04.21.2016	PGRN PGx guidelines repository	Bob Freimuth (Mayo)	15
05.05.2016	Innovation Around EHR	Ricky Bloomfield (Duke)	19
06.02.2016	The HSPC Open Services Platform	Scott Narus (University of Utah)	13
07.07.2017	Laboratory and precision medicine	John David Larkin Nolen (Cerner)	22
08.04.2016	Deciphering the Genome: Community Driven Efforts	Heidi L. Rehm (Partners)	28
09.01.2016	OpenInfoButton and EMR integration with EPIC and Cerner	Guilherme Del Fiol (University of Utah)	n/a

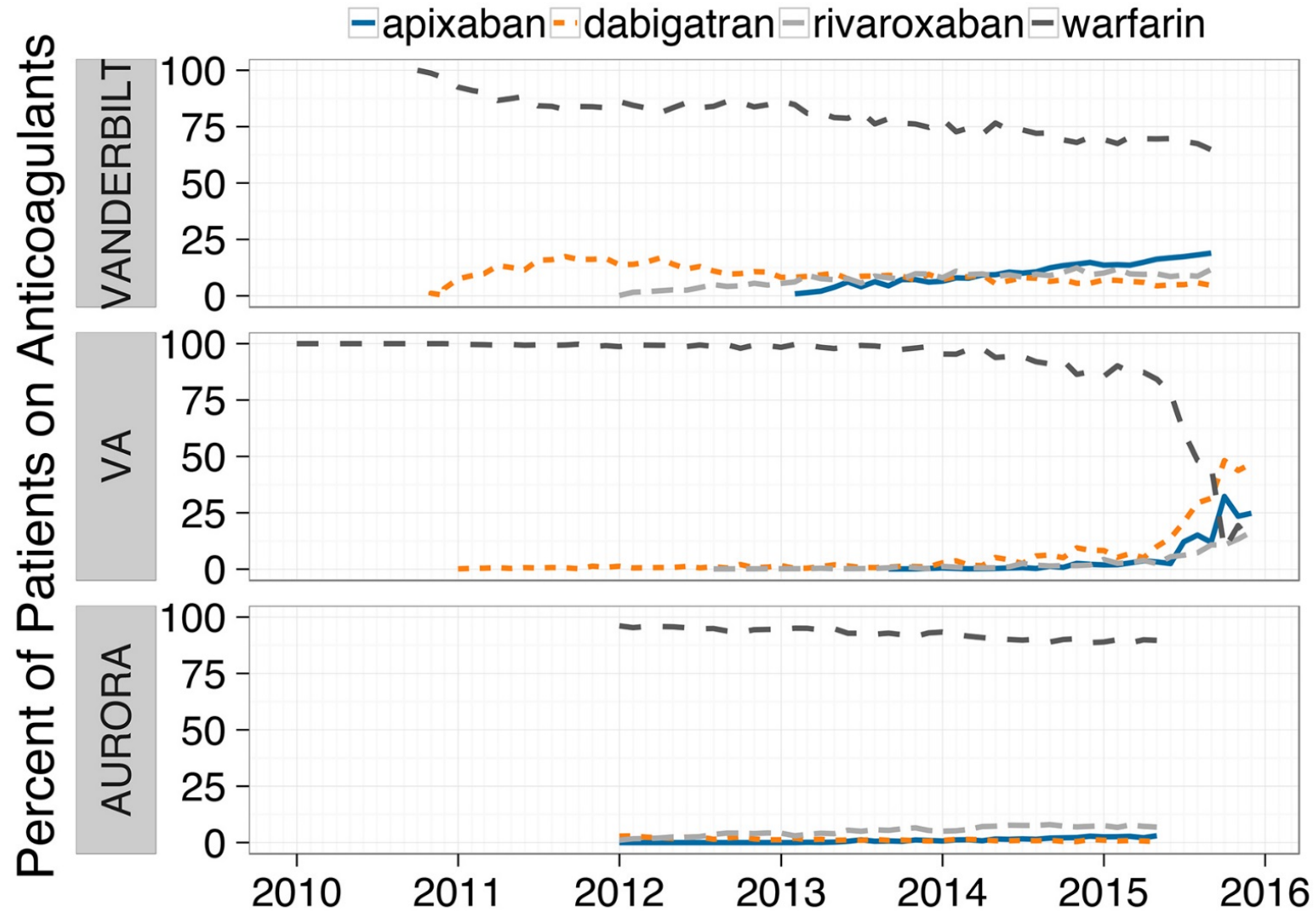
New CIIG Project: Site Survey of Data Pipeline

- Framework describing key components of an ideal informatics pipeline
- Comparison of implemented systems to ideal pipeline across academic and non-academic settings
- Structured data collection:
 - Laboratory: Where are discrete genomic results stored and transformed to non-discrete results?
 - Institution: Where are gaps in internal or reference lab interface?
 - How is variant -> phenotype performed and maintained?
 - CDS Rules: process of creating, storing, and modifying rules
 - Presentation: UI issues, “right time, person, and context”
 - Clinical Effectiveness: how is data related to EHR interactions stored and accessed

IGNITE CPIC Prescribing Study



Comparison of Anticoagulant Utilization



Summary: Future Opportunities for CIIG

- Study and address gaps in data pipeline
- Support comparative effectiveness activities of the network
 - Phenotyping of drug response or disease response outcomes
 - Linking data from broader sources; e.g. health information exchange or state registry
- Focus on the user experience of accessing genomic data and interpretations within EHRs