

PennChart Genomics Initiative: from Integration to implementation

LESSONS LEARNED

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PennChart Genomics Initiative

Large team of individuals from Clinical Genetics, Molecular Genetics, Legal/Privacy and Penn Medicine IS

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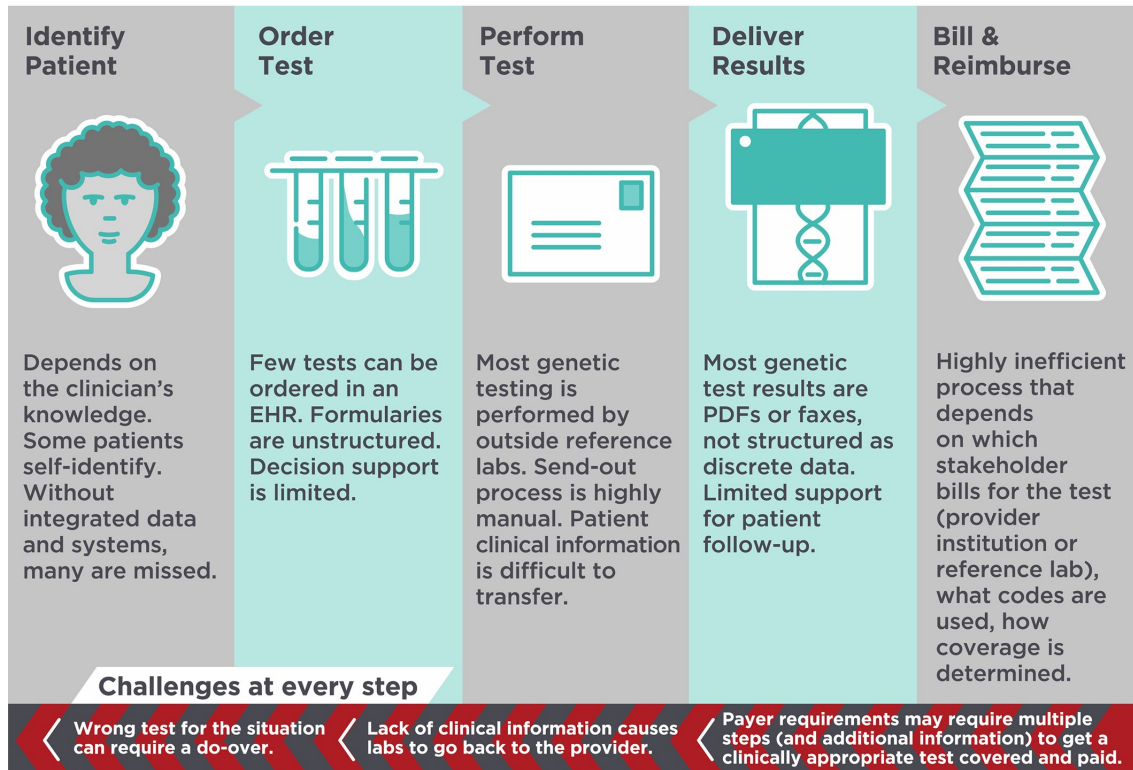
It takes a (well-supported) village
(with buy-in at all levels)



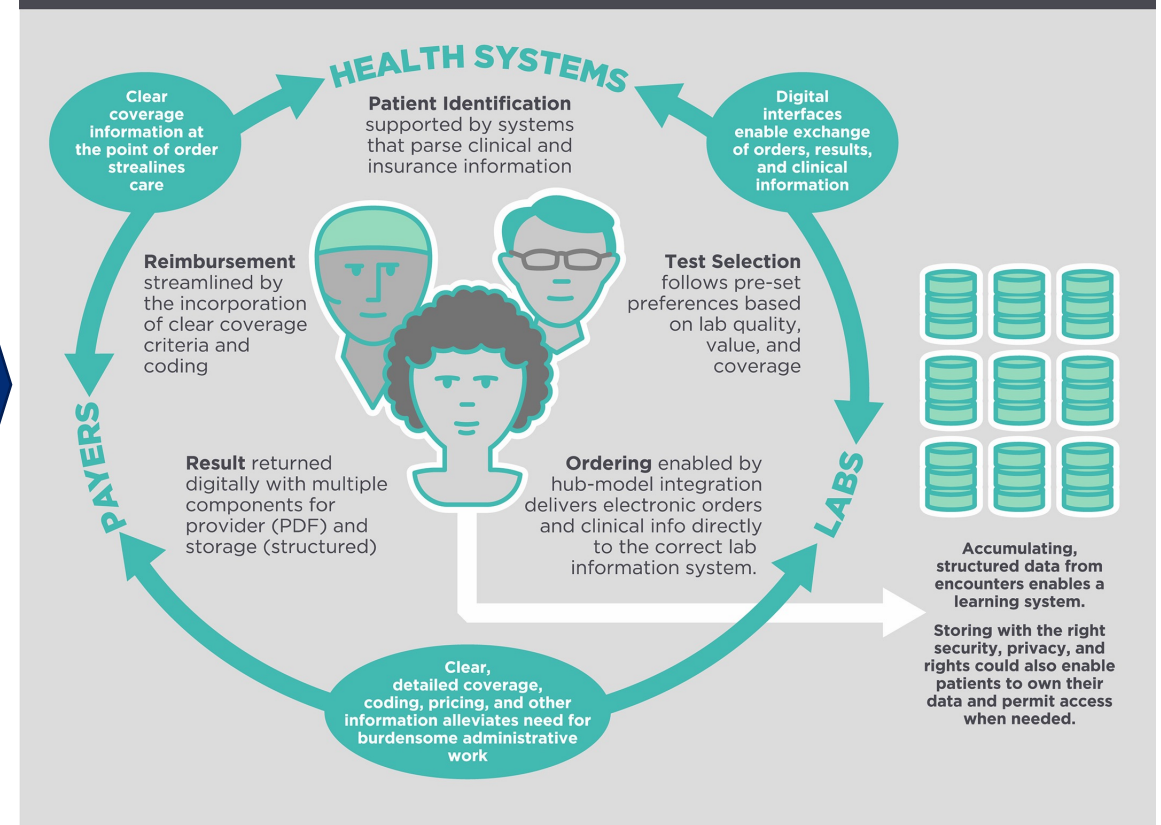
Barriers to implementation

Genetic Testing Today: An Inefficient Value Chain

The Fragmented Genetic Testing Value Chain (current)

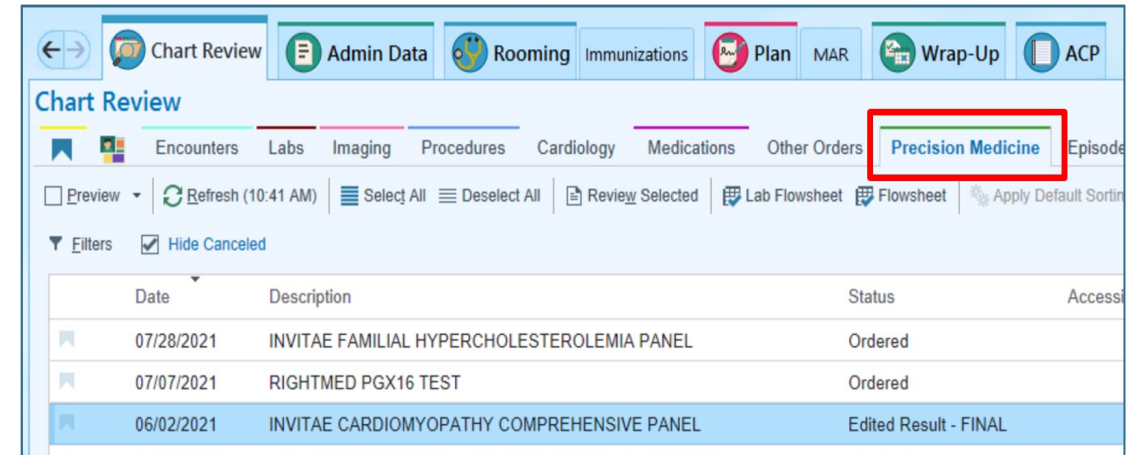


Vision: An integrated value chain that enables learning



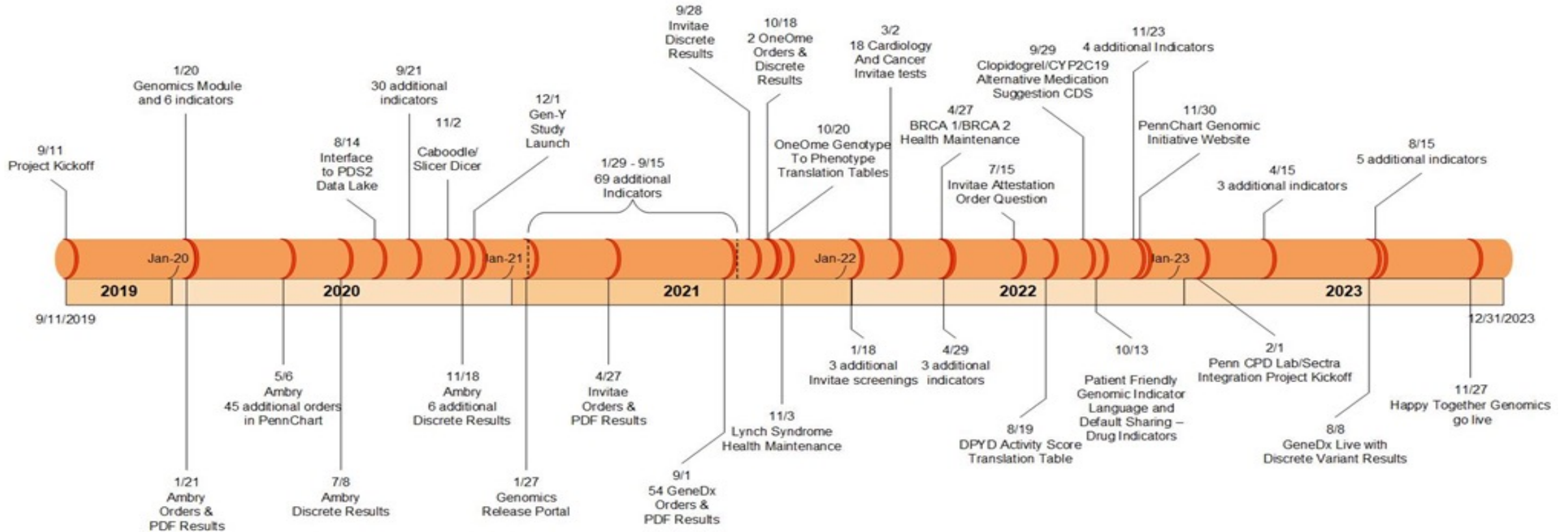
First steps to a genetics friendly EHR

- ▶ Standardized naming conventions in the EHR mid-late 2010s
 - All genetics programs within the institution agreed upon standards for naming and labeling of genetic testing results data
- ▶ Created a 'Precision Medicine' tab within PennChart
 - 'Genetics' specific document type
 - All scanned results in that document type go into the tab
 - Allows isolation of genetic data (to prevent upload into health exchanges)
- ▶ Legacy data moved into the tab (over 17.5K results)
- ▶ Precision Medicine Tab – 35,570 views in last three months by 6,748 providers in 1,058 department



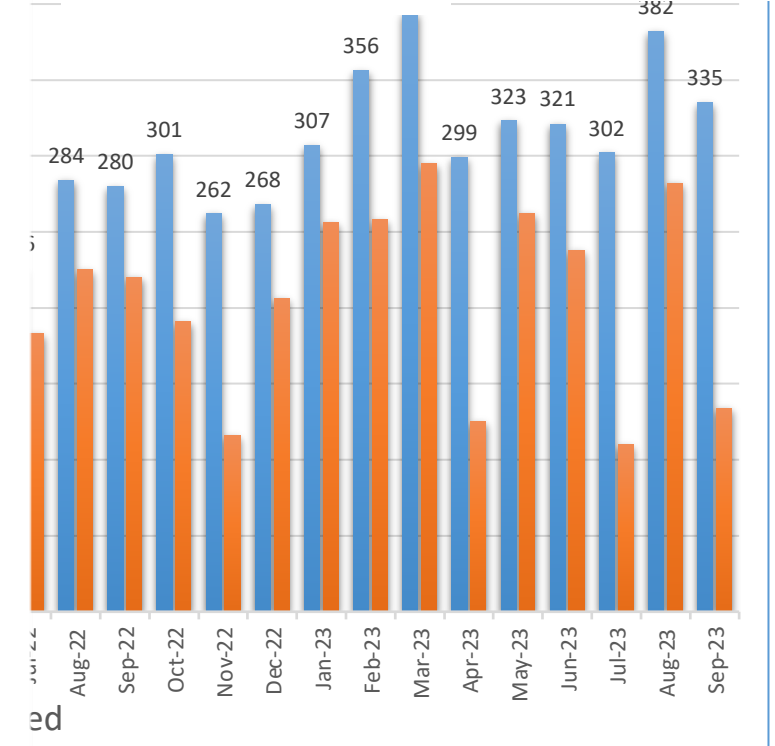
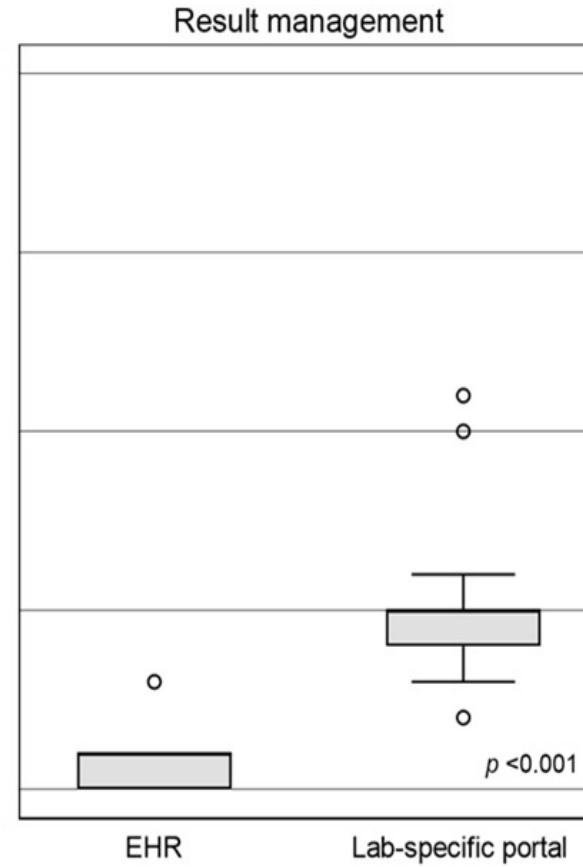
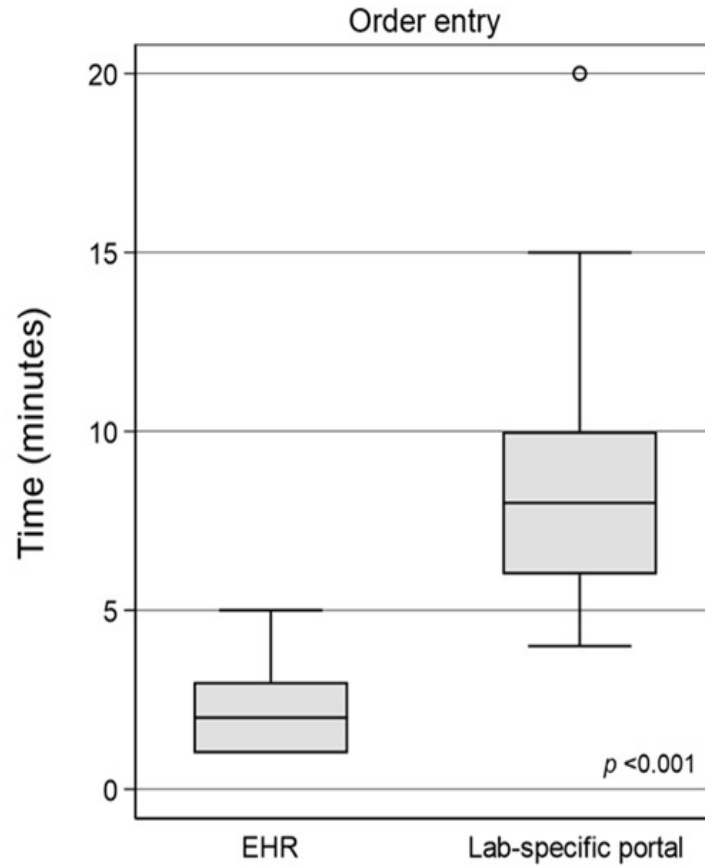
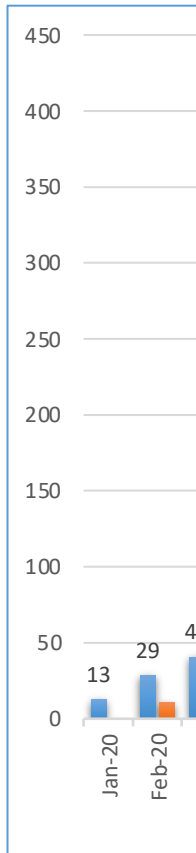
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PennChart Genomics Initiative timeline



The overnight success story that took five years.

Uptake of genetic test ordering/resulting

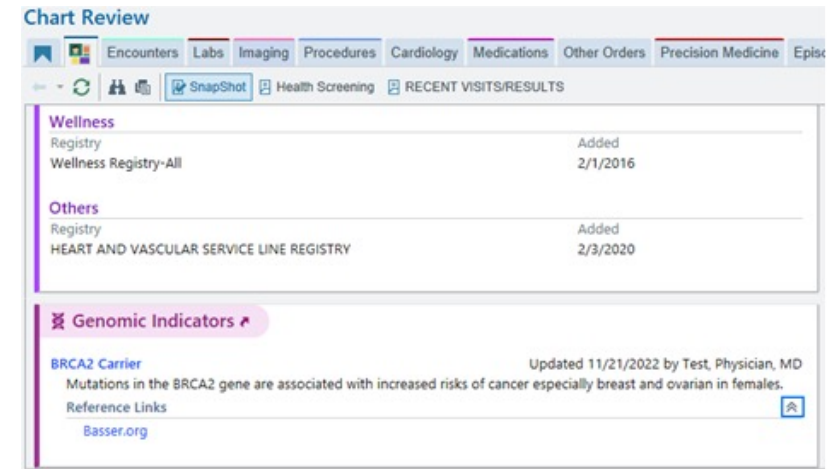
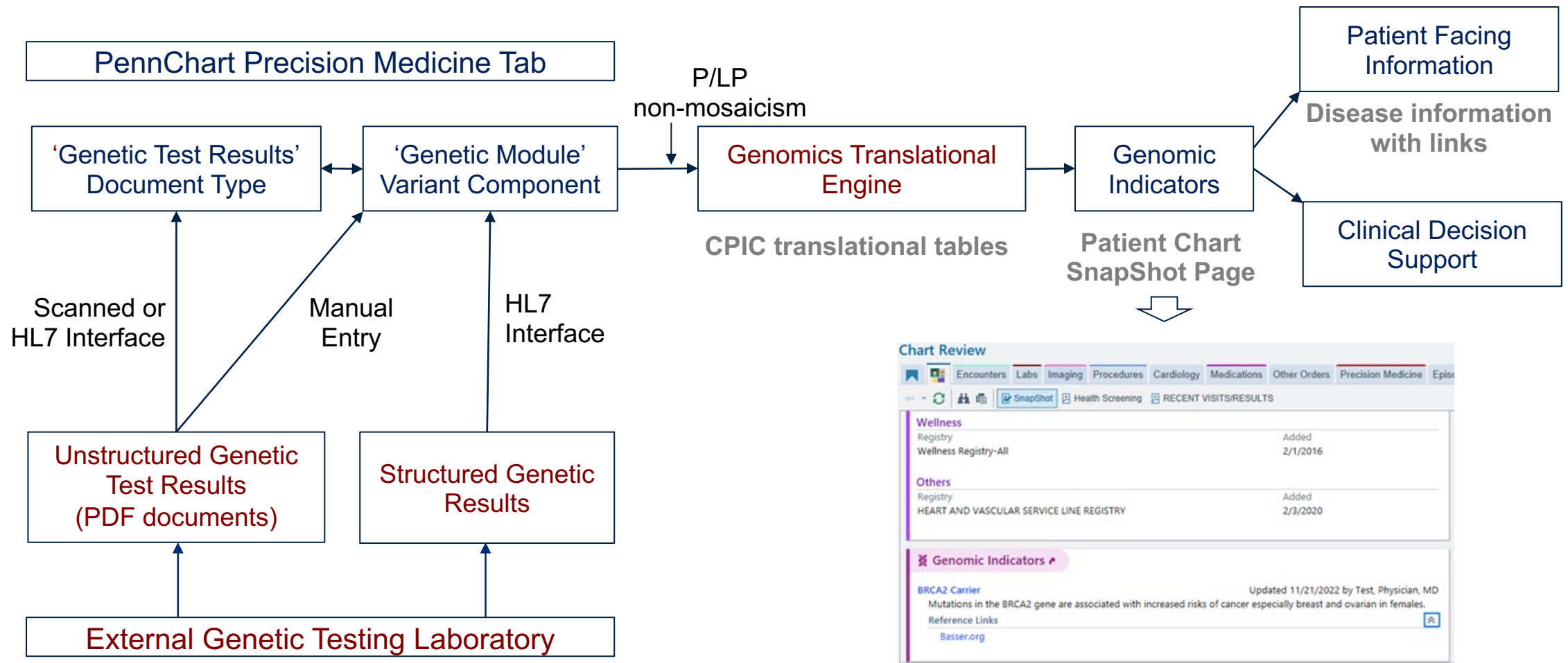


► 928 genetic testing orders placed as a part of clinical care

- 278 different ordering providers

If you build it, they will come.

Overview of flow of genetic test results – Mendelian and PGx



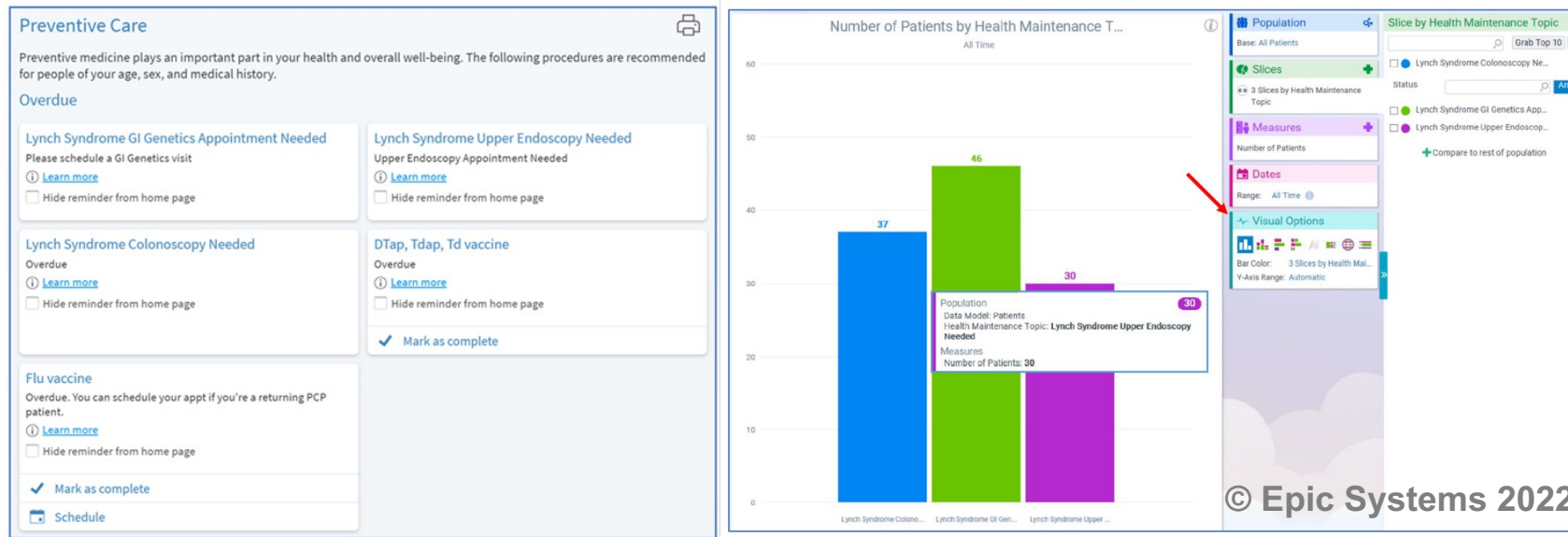
If you build it, they want improvements.

Impact of genomic indicators

BRCA2 Carrier	963	CYP2C19 Intermediate Metabolizer	89	MUTYH-associated polyposis syndro...	21	GREM1	5
BRCA1 Carrier	788	CYP2C19 Rapid Metabolizer	83	Hereditary Leiomyomatosis and Renal...	20	SMAD4	5
Lynch Syndrome	531	Von Hippel-Lindau Syndrome	79	MYBPC3-Related Cardiomyopathy	20	Vascular Ehlers-Danlos syndrome	5
DPYD Normal Metabolizer	343	IFNL4 Unfavorable response genotype	78	MYH7-Related Cardiomyopathy	17	DPYD Intermediate Metabolizer Activit...	4
DPYD Normal Metabolizer Activity Sc...	276	CYP2B6 Normal Metabolizer	74	CYP2B6 Poor Metabolizer	15	Multiple endocrine neoplasia, type 1	4
ATM (heterozygous)	230	CDH1	73	DPYD Intermediate Metabolizer	12	Multiple endocrine neoplasia, type 2	4
Li-Fraumeni Syndrome	168	SDHB Related Hereditary Pheochromo...	69	HOXB13	12	SDHD Related Hereditary Pheochromo...	4
UGT1A1 Normal Metabolizer	164	CYP2D6 Normal Metabolizer	62	CYP2D6 Poor Metabolizer	11	CYP2C19 Ultrarapid Metabolizer	3
UGT1A1 Intermediate Metabolizer	162	UGT1A1 Poor Metabolizer	59	PTEN Hamartoma Tumor Syndrome	10	CYP2C9 Poor Metabolizer	3
CYP2C19 Normal Metabolizer	142	CYP2D6 Intermediate Metabolizer	58	CYP2C19 Poor Metabolizer	9	LMNA-Related Cardiac Disease	3
HLA-B*15:02 Negative	131	BRIP1	55	DPYD Intermediate Metabolizer Activit...	9	Malignant hyperthermia susceptibility	3
NUDT15 Normal Metabolizer	129	IFNL4 Favorable response genotype	47	CYP2B6 Rapid Metabolizer	8	TTN-Related Dilated Cardiomyopathy	3
TPMT Normal Metabolizer	125	CYP2B6 Intermediate Metabolizer	46	CYP3A5 Normal Metabolizer	8	DPYD Poor Metabolizer	2
HLA-A*31:01 Negative	124	CYP2C9 Intermediate Metabolizer	46	HLA-B*57:01 Positive	8	DSP-Related Arrhythmogenic Cardiom...	2
HLA-B*57:01 Negative	120	CYP3A5 Intermediate Metabolizer	39	CYP2D6 Needs Review	7	Familial hypercholesterolemia (MIM 1...	2
HLA-B*58:01 Negative	118	CDKN2A	38	HLA-A*31:01 Positive	7	Long QT Syndrome Type 1 (KCNQ1-Re...	2
PALB2	115	RAD51C	37	HLA-B*58:01 Positive	7	TTR	2
Familial Adenomatous Polyposis	105	Birt-Hogg-Dubé Syndrome	30	TPMT Intermediate Metabolizer	7	ACTA2 Thoracic aortic aneurysm susc...	1
SLCO1B1 Normal Function	99	BARD1	27	CYP2D6 Ultrarapid Metabolizer	6	DPYD Poor Metabolizer Activity Score 0	1
CYP2C9 Normal Metabolizer	93	SLCO1B1 Decreased Function	27	Marfan Syndrome and related disorders	6	DPYD Poor Metabolizer Activity Score...	1
CYP3A5 Poor Metabolizer	90	RAD51D	24	SDHC Related Hereditary Pheochrom...	6	MUTYH-associated polyposis (MAP) c...	1

- ▶ 138 unique genomic indications available
 - 61 disease indicators
 - 76 drug indicators
- ▶ 4037 total patients with an indicator
- ▶ 6590 total indicators active on patient charts
 - 2877 automatic
 - 3090 added manually
 - 623 added via import
- ▶ 2538 indicators shared to myPennMedicine (MyChart)

Clinical decision support



Active for 450 Lynch syndrome patients and 1607 patients with BRCA1/2 mutations across Penn Medicine

- ▶ Clinical decision support has required **review and iteration** to make operational
- ▶ Surgeries and procedures are not coded correctly so are not automatically picked up by the algorithms (e.g. reminding a woman with a BRCA1 mutation to have mammograms post-mastectomy)

Challenges of EHR integration

Anticipated

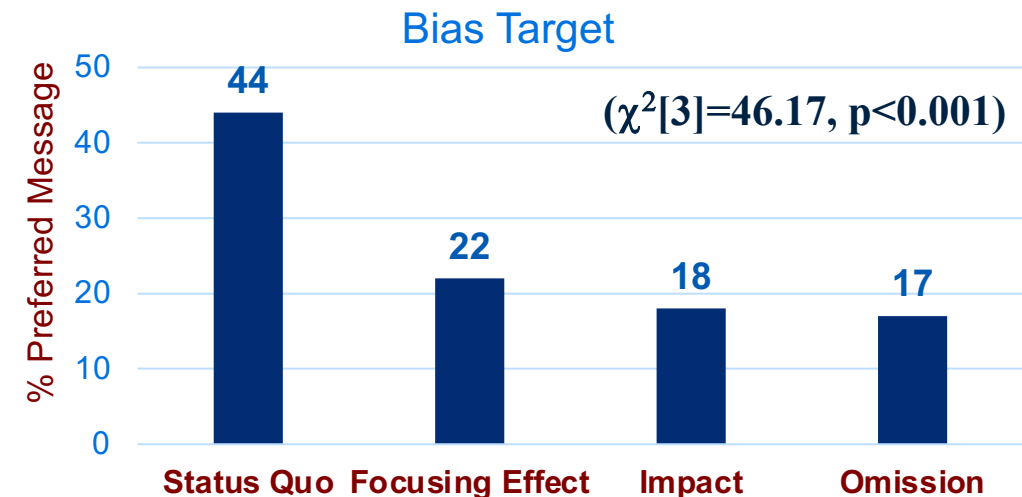
- ▶ Project Scope – started small with very specific use cases
- ▶ Technical Build – established standards early for required data elements and entry
- ▶ Language Barriers – both IS and genetics have their own jargon
- ▶ Vendor Relationship – vary substantially
- ▶ Privacy Concerns – release of results to all, involved privacy and legal

Unanticipated

- ▶ Project Cadence – what is the right timing to move integration forward
- ▶ Different Stakeholder Needs – different genetic provider groups were engaged at varying points in the process
- ▶ Impact on Clinical Workflows – change is hard
- ▶ Knowledge Dissemination – demand for dissemination was much higher than anticipated

Prompting non-genetic providers to order genetic testing

- ▶ Behavioral economics aims to target situations where patients or care teams don't make choices that can achieve optimal health or operational outcomes
- ▶ Behavioral economics relies on **nudges**, which are changes in the way choices are presented or information is framed that are meant to **guide or motivate decision-making** - **low-cost, scalable** interventions, **implemented in EHR**
- ▶ Performed discrete choice experiment to see which nudge providers preferred
 - 43/79 (54%) providers completed RedCap Survey
 - 44% women
 - 16% cardiology, 47% neurology, 2% endocrine
 - 49% physician
 - M=37 patients/month (SD=38)
 - Compared each message and varied order



Supporting non-genetic providers to order genetic testing

- ▶ Developed fully supported genetic testing for non-genetic providers, including language addressing *status quo bias*

ⓘ Patient is eligible for genetic testing.

Research Participant

Rhodes, Corinne M, MD, MPH
PCP - General
Coverage: Aetna/Aetna Choice P...

ⓘ Patient is eligible for genetic testing.

Your patient has been identified as a candidate for Pheochromocytomas (PCCS) and Paragangliomas (PGLS) genetic testing. In the past, genetic testing may not have informed clinical management. But now, genetic testing is strongly recommended to help guide management of patients with this condition. Click the highlighted option below to order the appropriate genetic testing.

ⓘ Patient is eligible for genetic testing.

Your patient has been identified as a candidate for Pheochromocytomas (PCCS) and Paragangliomas (PGLS) genetic testing. In the past, genetic testing may not have informed clinical management. But now, genetic testing is strongly recommended to help guide management of patients with this condition. Click the highlighted option below to order the appropriate genetic testing.

Open Order Set Do Not Open **Order Genetic Testing for Pheochromocytomas (PCCs) and Paragangliomas (PGLs)** Preview

For more information about genetic testing in general, ordering genetic testing and how the results of genetic testing would influence your patient's medical management, please click here.

Override Reason

Not clinically indicated Defer to a later date Patient not interested Already had genetic testing Other

Referred patient to genetics

Accept

Passive BPA with nudge language and linked genetic test ordering

Order Genetic Testing for Pheochromocytomas (PCCs) and Paragangliomas (PGLs)

▼ **Ambry Order - Default Based on Insurance**

▼ **UPHS RSH GENOMICS PCCS AND PGLS PANEL - DEFAULT**

PGLNext: 14 Genes
Ambry Lab, Routine
For Multi-Gene Orders: Select an Indication for Testing: Other
Resulting Lab Billing Method: Insurance
Indication for Testing: Diagnostic
Responsible Party Account Type: Personal/Family
Ship saliva kit to patient? Yes
Shipping method: Ground 3-5 days
Resulting Agency - AMBRY GENETICS

Patient Instructions - Ambry Panel

Progress Note Para/Pheo - Ambry Lab

My Note

Note Details

Cosign Request?

Summary:

Arial 11 B P U L

Genetic testing was recommended for Ms. Zzpcp today based on her history of pheochromocytoma or paraganglioma.

Information about genetic testing was reviewed with the patient, and they were provided with a copy of this information in their After Visit Summary

Genetic testing was ordered and a saliva kit will be sent to the patient from Ambry Genetics. If the patient has any billing questions they can reach out to the lab at Ambry's Patient Financial Service team at 949-900-5795 or billing@ambrygen.com.

The results will be sent to our office approximately 3-4 weeks after the patient submits their sample to the lab, and we will contact the patient with the results when they are available.

Genetic test ordering smart set with insurance sensitive testing, autopopulates provider note and aftervisit summary

Document Link

For instructions about how to place a genetics referral, more information about interpretation of genetic testing results and how the results of genetic testing would influence your patient's medical management, click here

Results

BREAST/GYN CANCER PANEL [PROC10005] (Accession 999914288) (Order 556114572)

Results include static link to provider website with information about referral and genetic testing

- Genetic testing is done on a blood or saliva sample and works by looking for these variants in the DNA.
- Why have genetic testing? Genetic testing may:**
 - Help your doctors better understand your diagnosis or provide targeted treatment options or better ways to monitor your health.
 - Give family members information about their chances to develop a medical condition.
- What is being tested?**
 - A panel including genes associated with your health condition.
 - This includes testing of genes that are related to your medical condition. Sometimes, the lab can find a change in a gene that causes your known health condition, but also other health conditions.
- The results of genetic testing could be:**
 - Positive: means that a genetic variant was found. This can increase your risk for a medical condition or help to determine a course of treatment or method to monitor your health.
 - Negative: means that no genetic variant that affects your health was found. This can also help determine treatment options and how you are monitored.
 - Uncertain: means that a genetic variant was found that the lab is not sure causes an increased chance to have a medical condition.
- Risks and limits of genetic testing:**
 - There is a small risk for errors, like errors in processing a sample and technical problems.
 - If a genetic diagnosis is not found, we cannot completely rule it out. You might have a condition that was not tested for, not discovered yet, or that cannot be found with current technology.
- Privacy and Protections for genetic test results:**
 - Genetic test results are protected health information. Penn Medicine privacy practices apply to genetic test results.
 - The Affordable Care Act (ACA) does not allow the use of pre-existing conditions (like cancer or heart disease), to deny health insurance coverage or raise premiums. If you would like to learn more, visit [About the ACA | HHS.gov](http://www.hhs.gov).
 - The Genetic Information Non-discrimination Act (GINA) is another law with more protections.
 - GINA does not allow the use of genetic information by employers and health insurance companies.
 - GINA does NOT apply to employers smaller than 15 employees. GINA does NOT apply for Life, Long-term Care, or Disability insurance.
 - If you would like to learn more, visit <http://www.ginahelp.org>
- Cost of testing:**
 - Genetic testing is usually billed to your insurance, just like any other medical lab or procedure.
 - In many cases insurance fully covers the test. If there is a cost, it is usually less than \$100.
 - You will be contacted by the lab if you owe more than \$100. The lab's billing department will discuss payment options with you.
- Testing process:**
 - The lab will ship a saliva kit to your home address with instructions and supplies.
 - The office that ordered your testing will contact you to review the results.
 - You may have the option to review these results in the patient portal before your healthcare provider has a chance to talk about them with you.
 - You should still talk with your healthcare provider about the results, even if you see the results first. They will let you know if there are any next steps for your medical care, or for your family.
- Billing Information**
Ambry Genetics will send you a text asking you to call into their billing line if your out-of-pocket cost is determined to be over \$100. If you receive that text, please call them to discuss patient assistance (including \$249 option) or payment plans. If you do not respond to their text message within 2 attempts, insurance will be billed automatically. You will be responsible for the estimated out-of-pocket cost. For billing questions, contact Ambry's Patient Financial Service team at 949-900-5795 or billing@ambrygen.com.

PennChart Genomics Initiative Website

<https://www.med.upenn.edu/pgi/>



PennChart Genomics Initiative
Optimizing the EHR for Use in Genomic Medicine

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Home



Welcome to the PennChart Genomics Initiative (PGI), a collaborative that aims to optimize the electronic health record (EHR) infrastructure supporting genomic medicine. In response to repeated requests from other institutional platforms to easily order genetic testing directly through the EHR, and to ensure results in a standardized way, we have developed tools making it accessible to other institutions globally.

On this website, you will find:

[Home](#) [Videos](#) [Resources](#) [Personnel](#) [Publications](#) [Feedback](#)

[Home](#) > [Resources](#) > [Penn Overview Tutorials](#)

Penn Overview Tutorials

Please click the links below to go to the Epic UserWeb to view the videos of tutorials. You will be required to use your Epic login to access resources.

[Orders](#)

A brief walkthrough of searching for genomic tests based on external lab, examples of varying order questions within the tests, and how to ensure colleagues are informed when results come back from a lab.

[Results](#)

See what genomic results can look like inpatient charts when filed to Epic from an external lab, containing discrete variant data.

[Genomic Indicators](#)

Explore the different ways Genomic Indicators can be documented on a patient chart, and examples of the content you can display within each drug/disease indicator.

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[Home](#) > [Resources](#) > [Epic Genomics Galaxy Guides](#)

Epic Genomics Galaxy Guides

Please click the links below to go to the Epic UserWeb to view the documents. You will be required to use your Epic login to access resources.

BestPractice Advisories

- [Show Clinicians BestPractice Advisories Based on a Patient's Genomic Indicators](#)
- [Show BestPractice Advisories Based on Genomic Variants](#)
- [Configure Interaction Settings for BestPractice Advisories That Appear as Medication Warnings](#)

Genomic Indicators

- [Genomic Indicators Setup: Essentials](#)
- [Show Clinicians a Genomic Indicators Navigator Section](#)
- [Summarize a Patient's Genomic Indicators in a Report](#)
- [Add and Update Patient Genomic Indicators Using the Translation Engine](#)
- [Automatically Add Genomic Indicators Based on Results \(LRR\)](#)

on results, Genomic Indicators, and clinician before deciding to order.



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Christine VanZandbergen
Colin Wollack
Dan Biros
Danielle McKenna
Jacob Sims
Jaquelyn Powers
Jeff Landgraf

**Clinical Genetics, Molecular Genetics,
Legal/Privacy and Penn Medicine IS**

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Susan McGarvey



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