Penn Medicine

PennChart Genomics Initiative: from Integration to implementation

LESSONS LEARNED

Katherine L. Nathanson, MD
Pearl Basser Professor of BRCA-Related Research
Medicine, Translational Medicine and Human Genetics
Deputy Director, Abramson Cancer Center
Director, Penn Center for Genomic Medicine

PennChart Genomics Initiative

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

Anjali Owens
Anna Schoenbaum
Christine VanZandbergen
Colin Wollack
Dan Biros
Danielle McKenna
Jacob Sims
Jaquelyn Powers
Jeff Landgraf
Jessica Chen
Jessica Chowns
Joe Bleznuck
Katherine Nathanson
Kelsey Lau-Min
Kim McClellan
Lauren Steinfeld
Mark Thai
Marylyn Ritchie
Michael Feldman
Peter Gabriel
Rose Giardine
Shawn Thompson
Stephanie Asher
Sony Tuteja
Susan Domchek
Susan McGarvey

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

CANCER MOONSHOT

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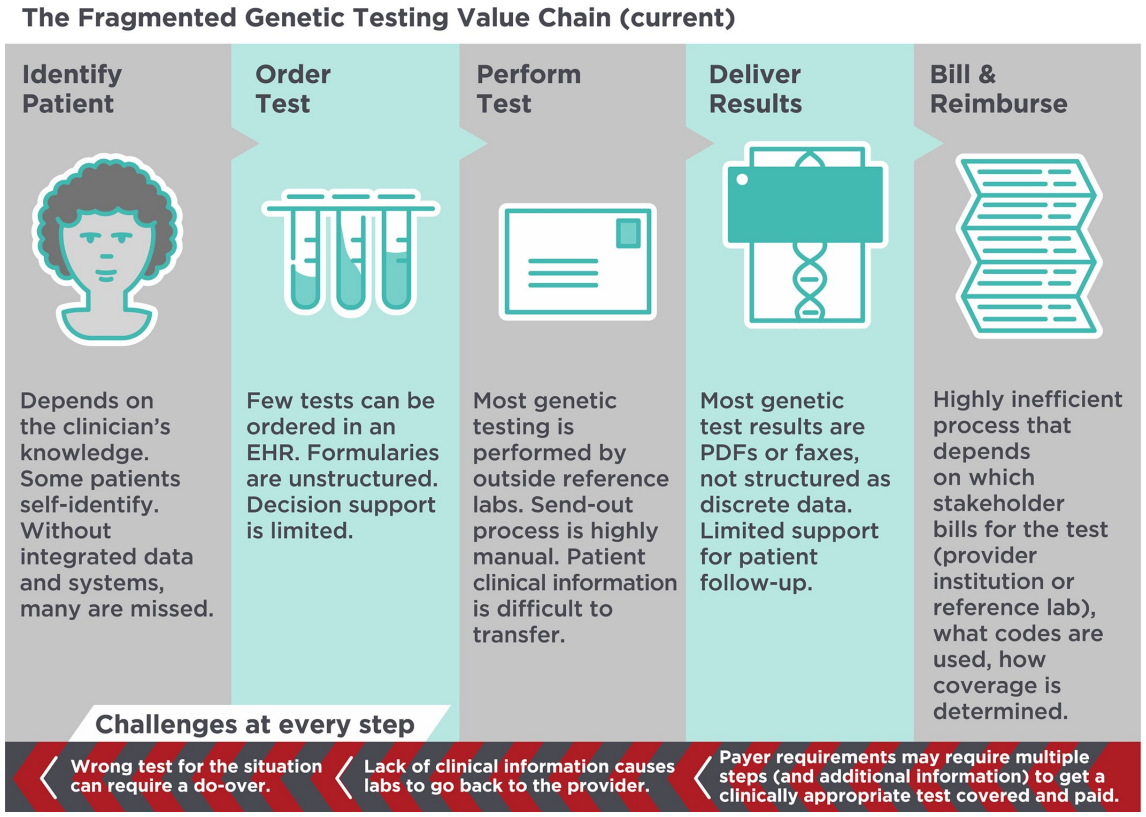
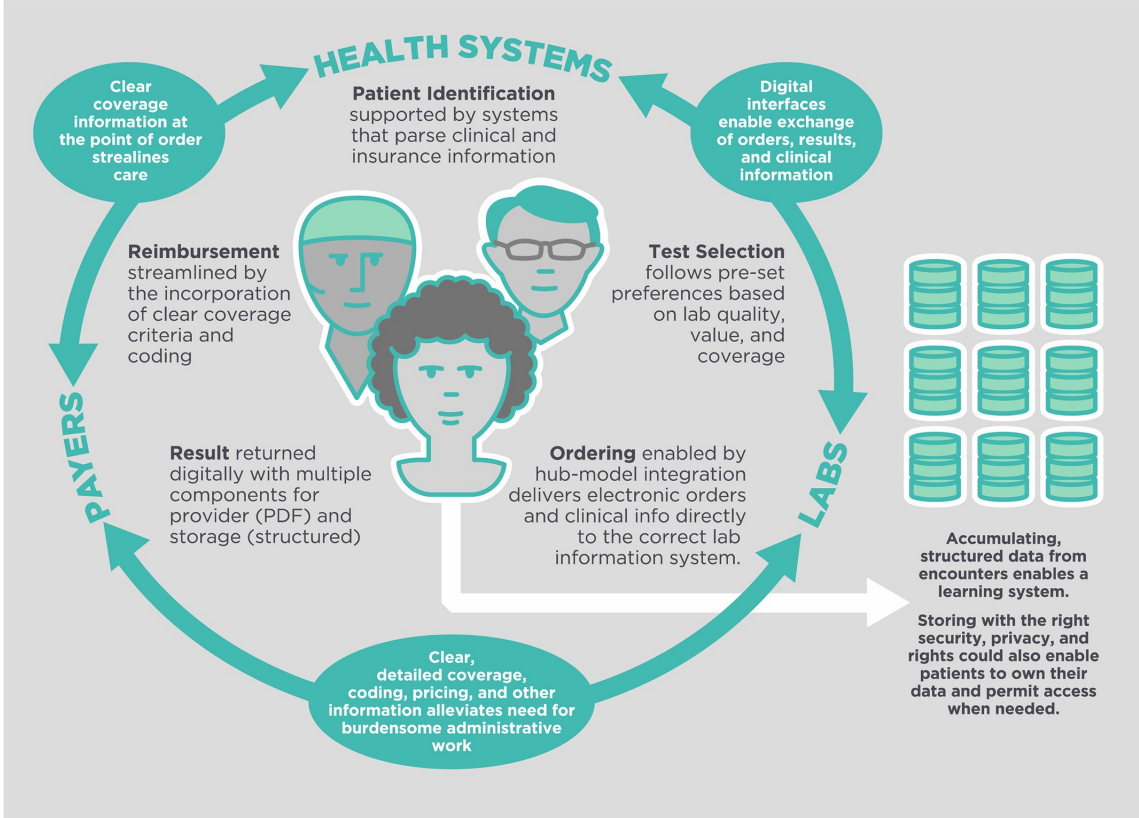
Barriers to implementation

Genetic Testing Today: An Inefficient Value Chain



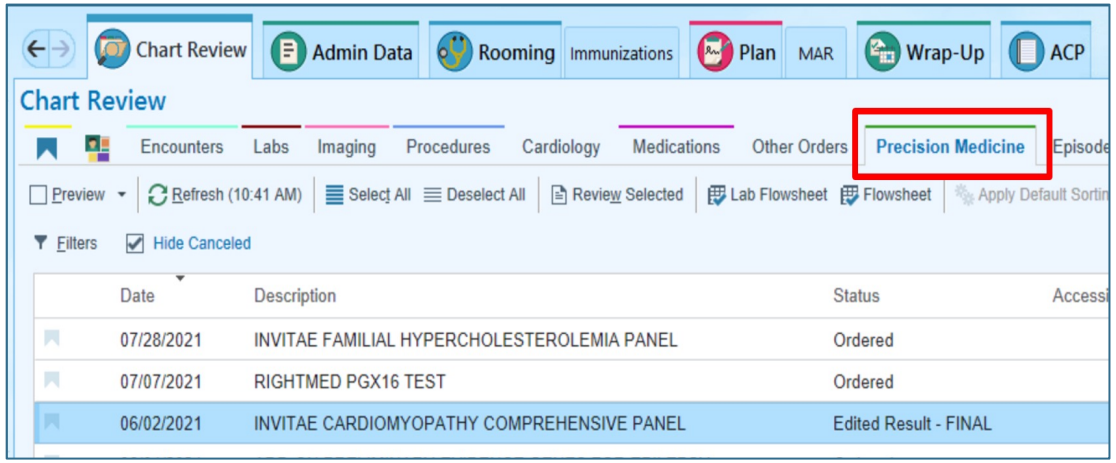
Vision: An integrated value chain that enables learning



Hooker, *Am J Med Genet*, 2021

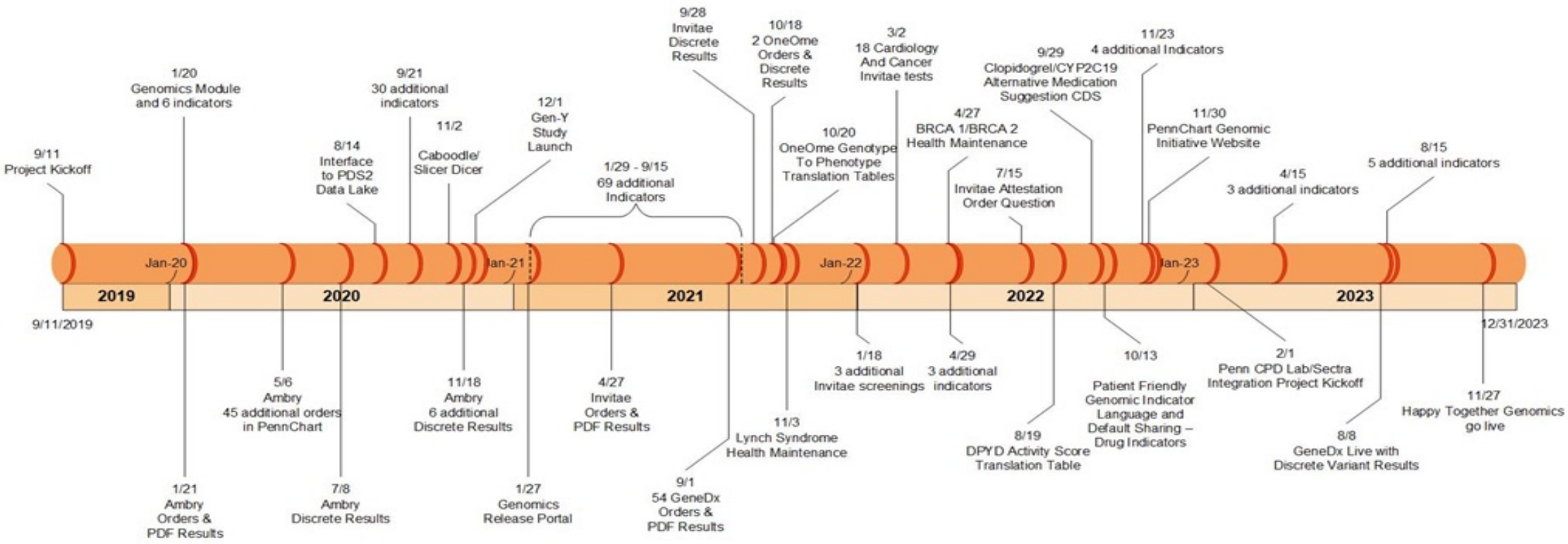
First steps to a genetics friendly EHR

- 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 up-load into health exchanges)
- Legacy data moved into the tab (over 17.5K results)



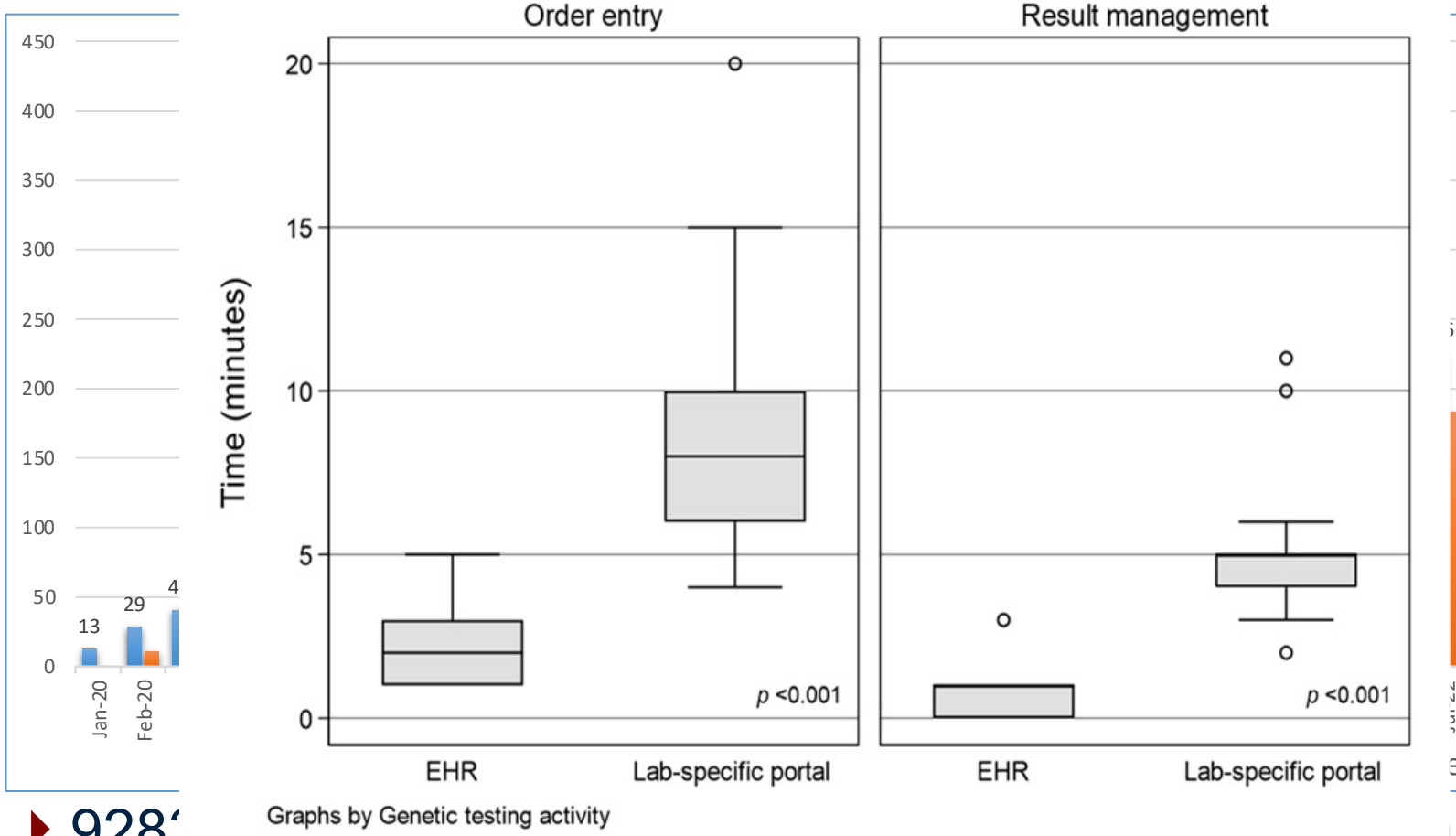
- Precision Medicine Tab – 35,570 views in last three months by 6,748 providers in 1,058 department

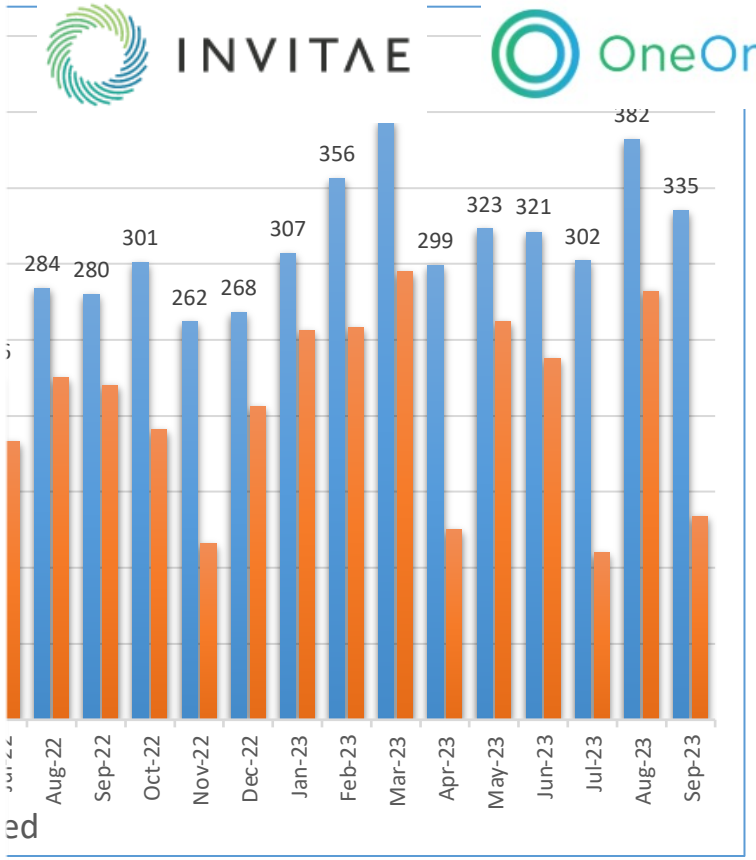
PennChart Genomics Initiative timeline



The overnight success story that took five years.

Uptake of genetic test ordering/resulting



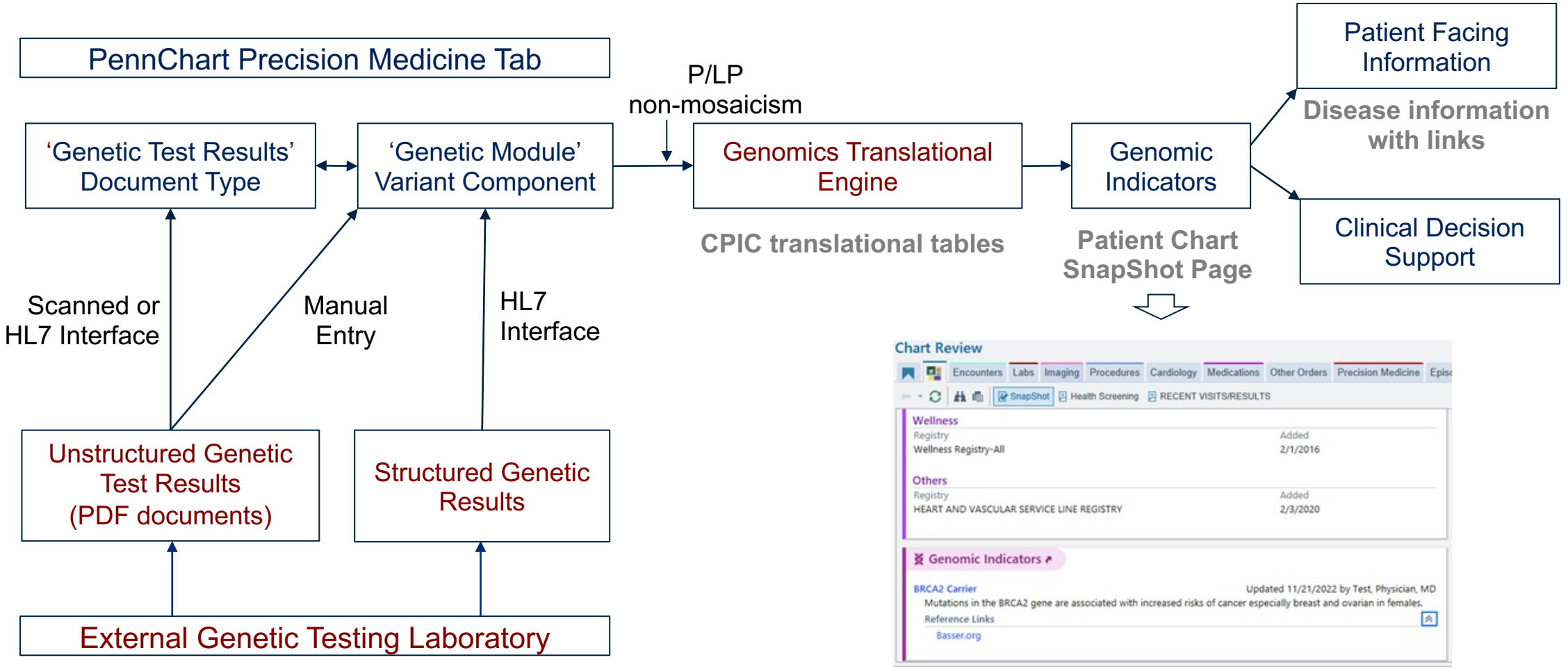


- 928 genetic testing orders placed as a part of clinical care
  - 278 different ordering providers

If you build it, they will come.

Lau-Min et al, *Genet Med*, 2022

Overview of flow of genetic test results – Mendelian and PGx



If you build it, they want improvements.

Impact of genomic indicators

BRCA2 Carrier	963
BRCA1 Carrier	788
Lynch Syndrome	531
DPYD Normal Metabolizer	343
DPYD Normal Metabolizer Activity Sc...	276
ATM (heterozygous)	230
Li-Fraumeni Syndrome	168
UGT1A1 Normal Metabolizer	164
UGT1A1 Intermediate Metabolizer	162
CYP2C19 Normal Metabolizer	142
HLA-B*15:02 Negative	131
NUDT15 Normal Metabolizer	129
TPMT Normal Metabolizer	125
HLA-A*31:01 Negative	124
HLA-B*57:01 Negative	120
HLA-B*58:01 Negative	118
PALB2	115
Familial Adenomatous Polyposis	105
SLCO1B1 Normal Function	99
CYP2C9 Normal Metabolizer	93
CYP3A5 Poor Metabolizer	90

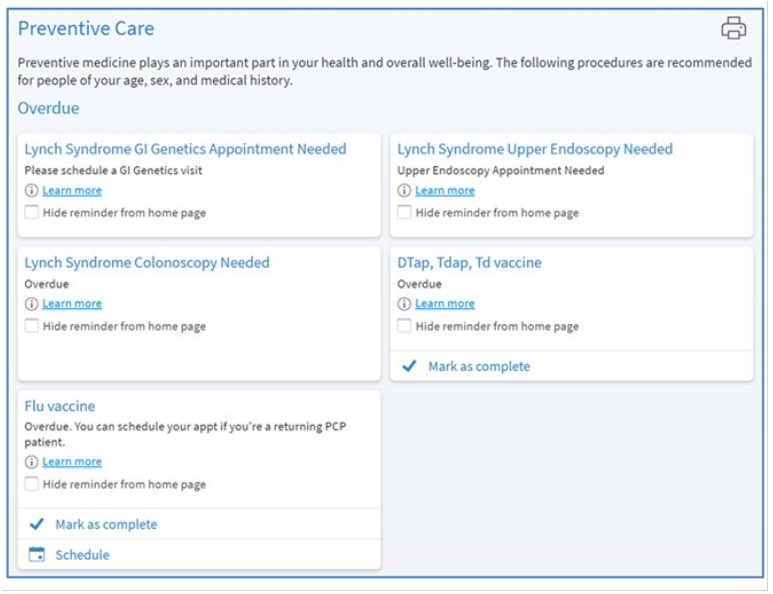
CYP2C19 Intermediate Metabolizer	89
CYP2C19 Rapid Metabolizer	83
Von Hippel-Lindau Syndrome	79
IFNL4 Unfavorable response genotype	78
CYP2B6 Normal Metabolizer	74
CDH1	73
SDHB Related Hereditary Pheochromo...	69
CYP2D6 Normal Metabolizer	62
UGT1A1 Poor Metabolizer	59
CYP2D6 Intermediate Metabolizer	58
BRIP1	55
IFNL4 Favorable response genotype	47
CYP2B6 Intermediate Metabolizer	46
CYP2C9 Intermediate Metabolizer	46
CYP3A5 Intermediate Metabolizer	39
CDKN2A	38
RAD51C	37
Birt-Hogg-Dubé Syndrome	30
BARD1	27
SLCO1B1 Decreased Function	27
RAD51D	24

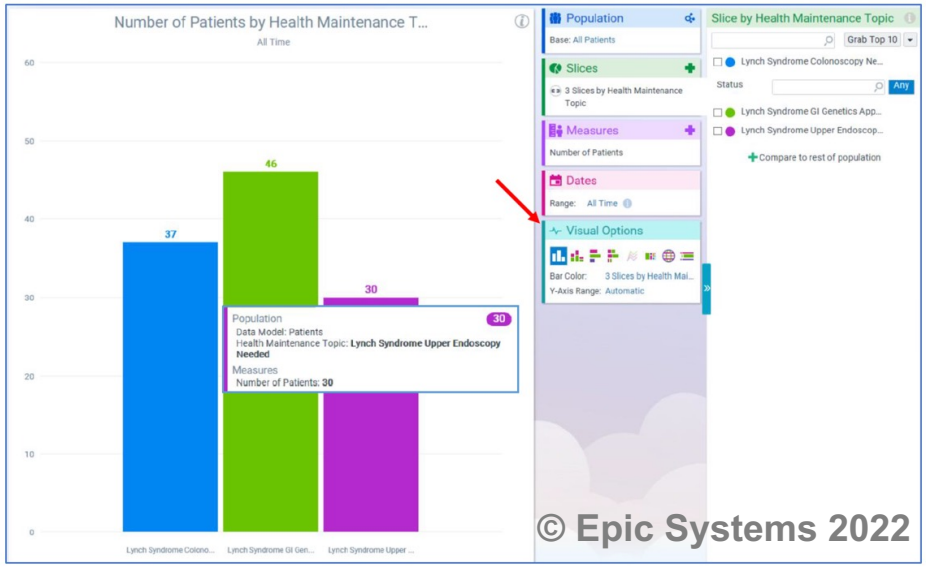
MUTYH-associated polyposis syndro...	21
Hereditary Leiomyomatosis and Renal...	20
MYBPC3-Related Cardiomyopathy	20
MYH7-Related Cardiomyopathy	17
CYP2B6 Poor Metabolizer	15
DPYD Intermediate Metabolizer	12
HOXB13	12
CYP2D6 Poor Metabolizer	11
PTEN Hamartoma Tumor Syndrome	10
CYP2C19 Poor Metabolizer	9
DPYD Intermediate Metabolizer Activit...	9
CYP2B6 Rapid Metabolizer	8
CYP3A5 Normal Metabolizer	8
HLA-B*57:01 Positive	8
CYP2D6 Needs Review	7
HLA-A*31:01 Positive	7
HLA-B*58:01 Positive	7
TPMT Intermediate Metabolizer	7
CYP2D6 Ultrarapid Metabolizer	6
Marfan Syndrome and related disorders	6
SDHC Related Hereditary Pheochrom...	6

GREM1	5
SMAD4	5
Vascular Ehlers-Danlos syndrome	5
DPYD Intermediate Metabolizer Activit...	4
Multiple endocrine neoplasia, type 1	4
Multiple endocrine neoplasia, type 2	4
SDHD Related Hereditary Pheochromo...	4
CYP2C19 Ultrarapid Metabolizer	3
CYP2C9 Poor Metabolizer	3
LMNA-Related Cardiac Disease	3
Malignant hyperthermia susceptibility	3
TTN-Related Dilated Cardiomyopathy	3
DPYD Poor Metabolizer	2
DSP-Related Arrhythmogenic Cardiom...	2
Familial hypercholesterolemia (MIM 1...	2
Long QT Syndrome Type 1 (KCNQ1-Re...	2
TTR	2
ACTA2 Thoracic aortic aneurysm susc...	1
DPYD Poor Metabolizer Activity Score 0	1
DPYD Poor Metabolizer Activity Score...	1
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)

Lau-Min et al., *JCO Clin Informat*, 2023

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

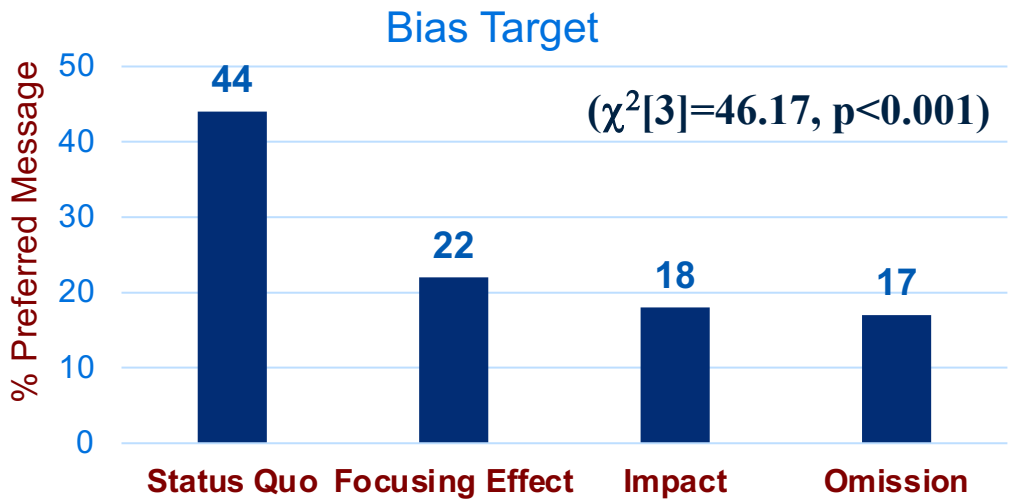
Unanticipated

- 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

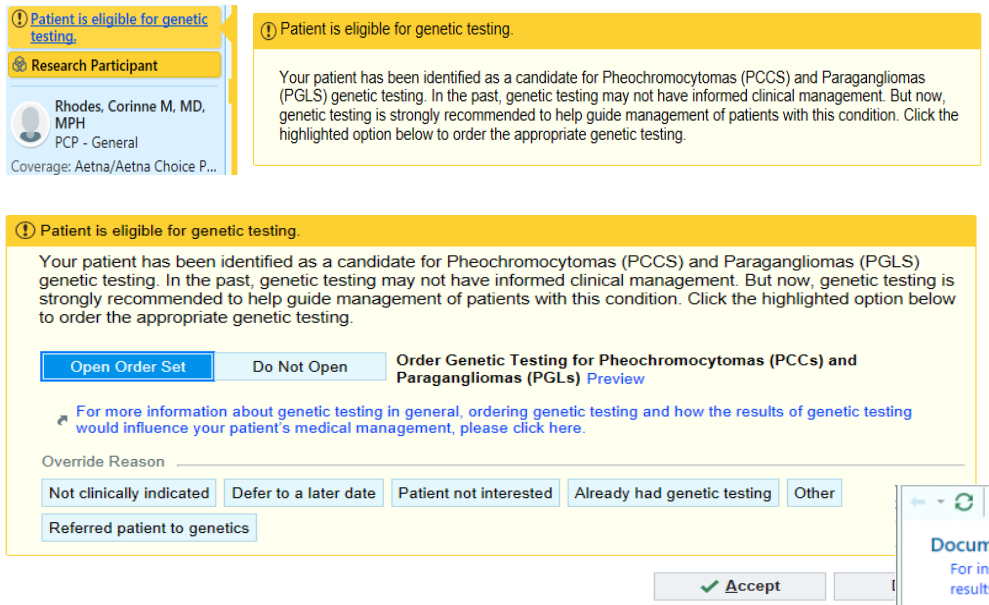
- Behavioral economics aims to target situations where patients or care teams don't make choices that can achieve optimal health or operational outcomes
- Behavioral economics relies on **nudges**, which are changes in the way choices are presented or information is framed that are meant to **guide or 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

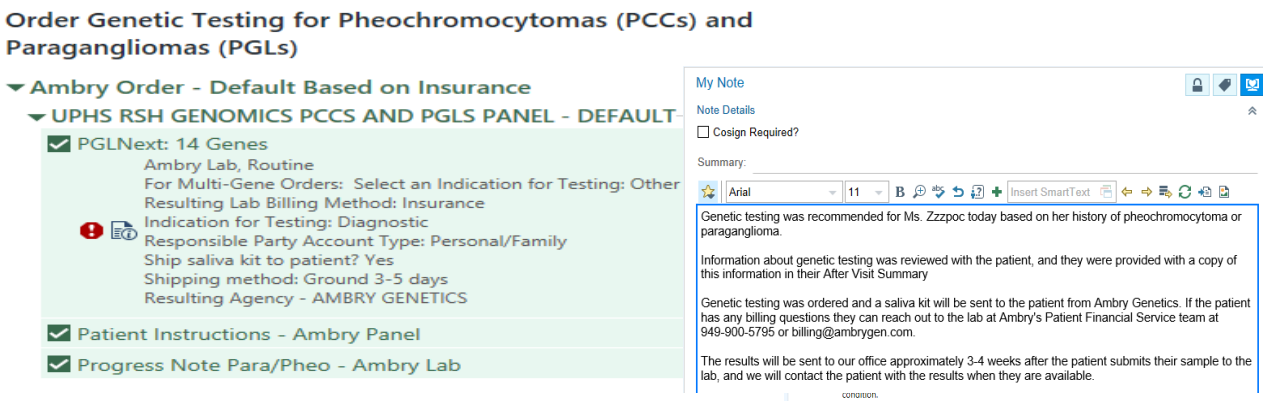


Supporting non-genetic providers to order genetic testing

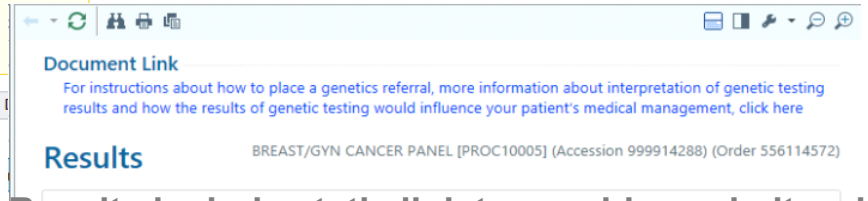
- Developed fully supported genetic testing for non-genetic providers, including language addressing *status quo bias*



Passive BPA with nudge language and linked genetic test ordering



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



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

PennChart Genomics Initiative Website

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

Perelman School of Medicine
University of Pennsylvania

PennChart Genomics Initi
Optimizing the EHR for Use in Genomic

Home | Videos | Resources | Personnel | Publications | Feedback

Home

Welcome to the PennChart Genomics Initiative (PGI)
collaborative that aims to optimize the electronic h
the EHR infrastructure supporting genomic medicin
response to repeated requests from other institutic
platforms to easily order genetic testing directly thr
results in a standardized way, we have developed tl
making it accessible to other institutions globally.

On this website, you will find:

Home | Videos | Resources | Personnel | Publications | Feedback

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.

Home | Videos | Resources | Personnel | Publications | Feedback

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

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)

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

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