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Implementing a Precision Health Program Using the Principles of a Learning Healthcare System

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Overview

- Define precision health and learning healthcare system
- Introduce the Geisinger MyCode Community Health Initiative and the Genomic Screening and Counseling Program
- Discuss the key processes required to achieve the objectives of a large-scale population sequencing program

Precision Health

- Emphasizes prevention while encompassing the interventions inherent in precision medicine (The provision of care for diseases that can be precisely diagnosed, whose causes are understood, and which consequently can be treated with rules-based therapies that are predictably effective.*)
- We view our project as a population precision health effort, and have renamed it the MyCode Community Health Initiative to distinguish it from the biorepository
- Inherent in this are educational efforts directed at participants, providers, payers, administrators and other stakeholders
- This is endorsed at the highest level of the organization as a strategic initiative

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*Adapted from The Innovator's Prescription A Disruptive Solution for Healthcare. Christensen , Grossman and Hwang, 2009

MyCode Community Health Initiative

- 250,000+ Geisinger Patients Will Have Their Exomes Sequenced.
- We will Look For Medically Actionable Results In That Data And Then Return Results To Patients And Providers.
- We will support the patients and providers in the follow-up to the results and long term management planning.
- We will be Operationalizing A Scalable Genomic Return Of Results Infrastructure In A Large Integrated Healthcare System

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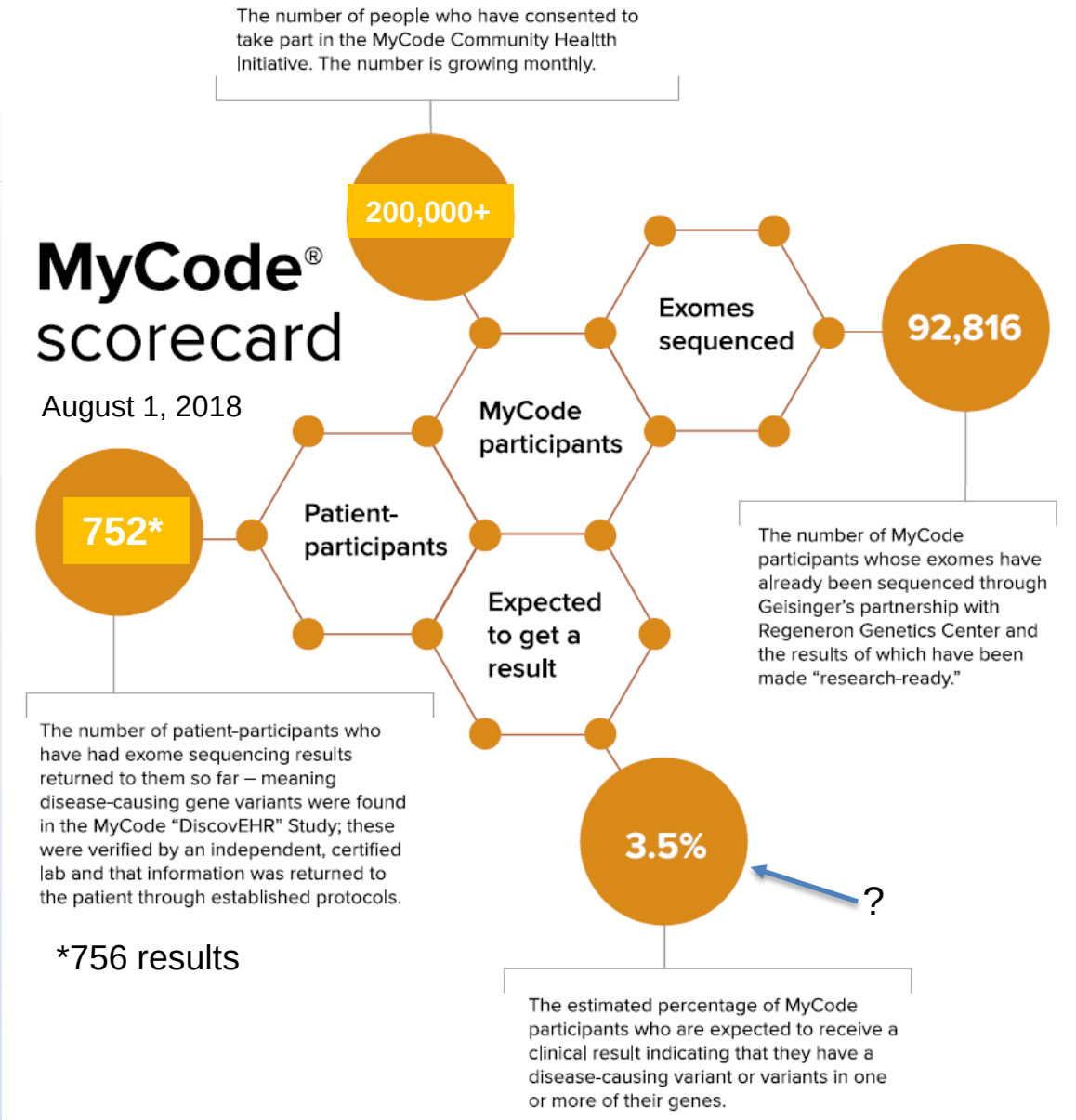
- GHS Biorepository started in 2007
  - Followed extensive consultation with GHS patients and other stakeholders that informed design of project
  - Defined as Community Health Initiative as opposed to biorepository
- Participants sign broad consent to combine EHR data (prospective, de-identified) and biospecimens
- Consent includes the ability to re-contact participants for future projects and communicate medically actionable results

Progress to date



*756 results

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Reporting Results

The Geisinger 80

- Focus on 30 conditions (80 genes)
- Builds on the ACMG Incidental Findings List (published 2013, updated 2016)
- Cancer predisposition (e.g. *BRCA1* and *BRCA2*)
- Cardiovascular disease (e.g. FH)

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Dewey FE, et al. Science. 2016 Dec 23;354(6319). pii: aaf6814

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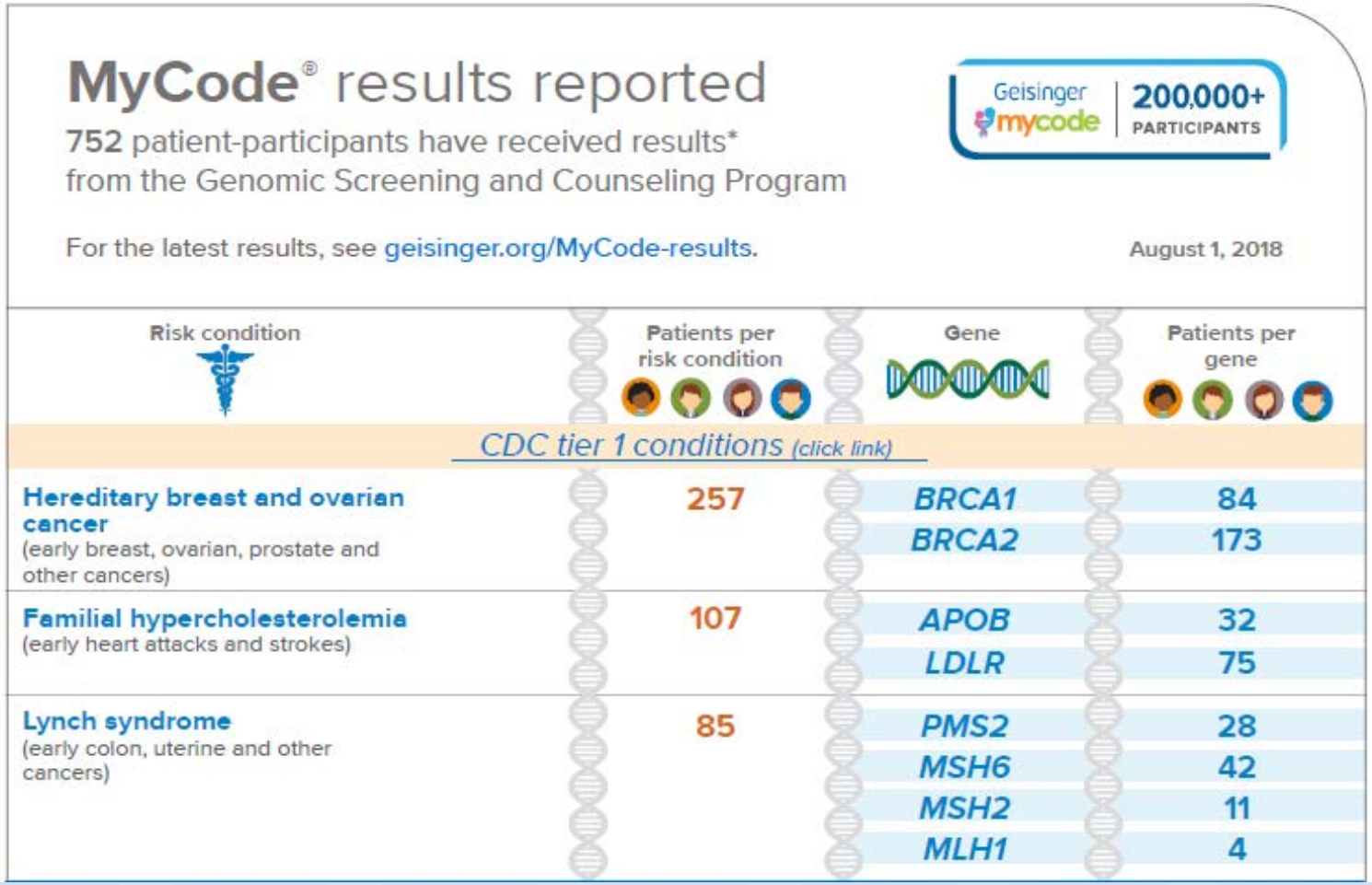
Progress to date



MyCode® results reported

752 patient-participants have received results*
from the Genomic Screening and Counseling Program

Geisinger mycode | 200,000+ PARTICIPANTS

For the latest results, see geisinger.org/MyCode-results.

August 1, 2018

Risk condition	Patients per risk condition	Gene	Patients per gene
CDC tier 1 conditions (click link)			
Hereditary breast and ovarian cancer (early breast, ovarian, prostate and other cancers)	257	*BRCA1*	84
		BRCA2	173
Familial hypercholesterolemia (early heart attacks and strokes)	107	*APOB*	32
		LDLR	75
Lynch syndrome (early colon, uterine and other cancers)	85	*PMS2*	28
		MSH6	42
		MSH2	11
		MLH1	4

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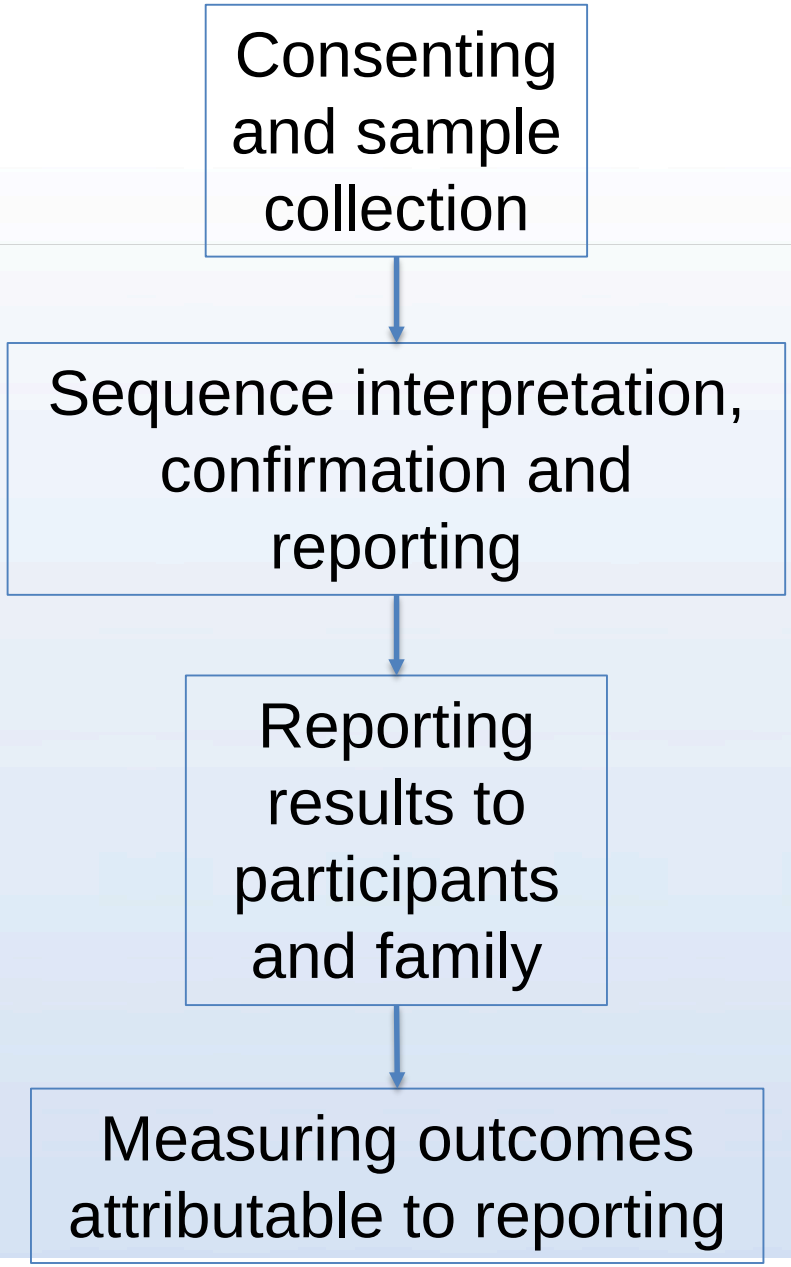
High Level Process



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Consent and Sample Collection

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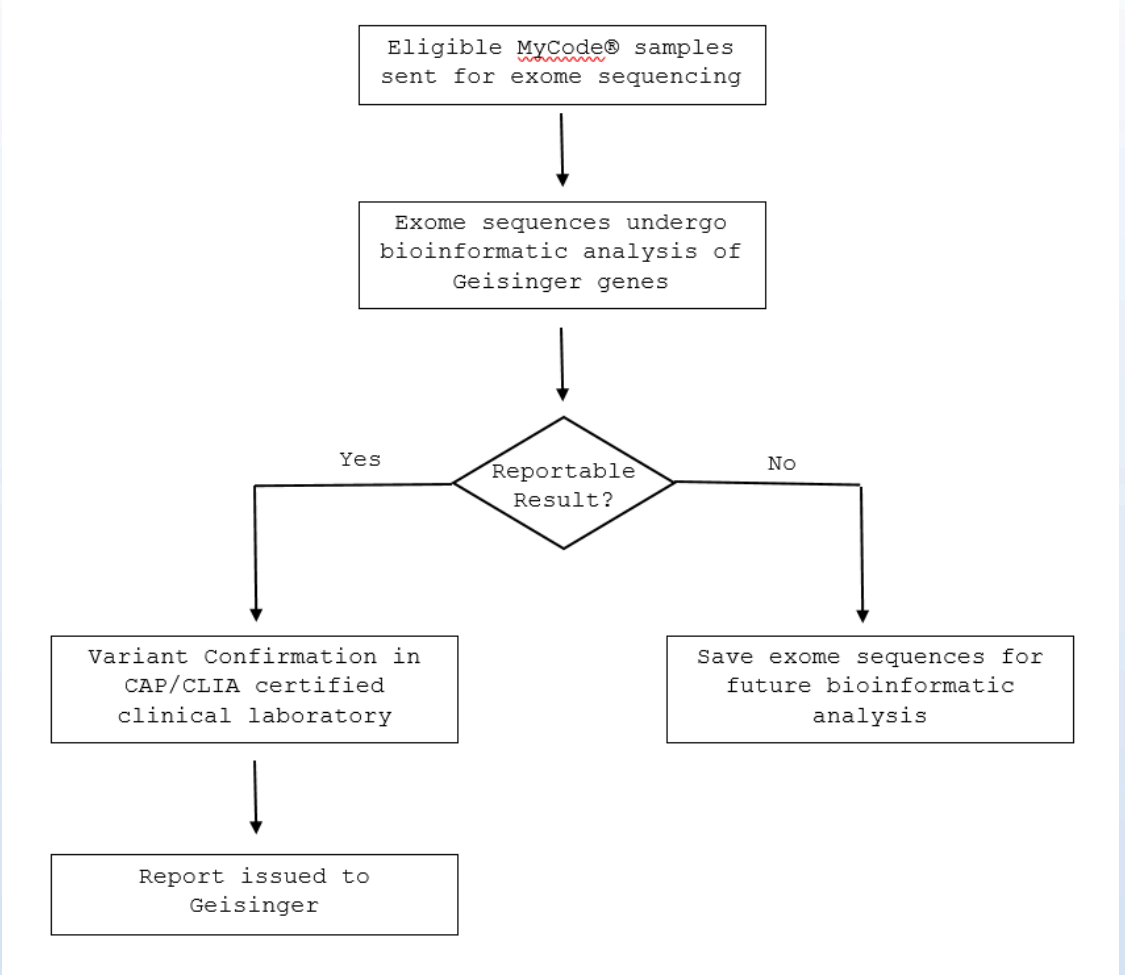
Consenting and sample collection

- Participants can consent in person, online or smart device (through patient portal)
- Consenting 800-1200 participants per week
- Heavy reliance on in person consenting
- Sample collection done as part of a routine blood draw
- Additional samples can be collected over time to replenish biospecimens

Sequencing, confirmation, and reporting- In theory



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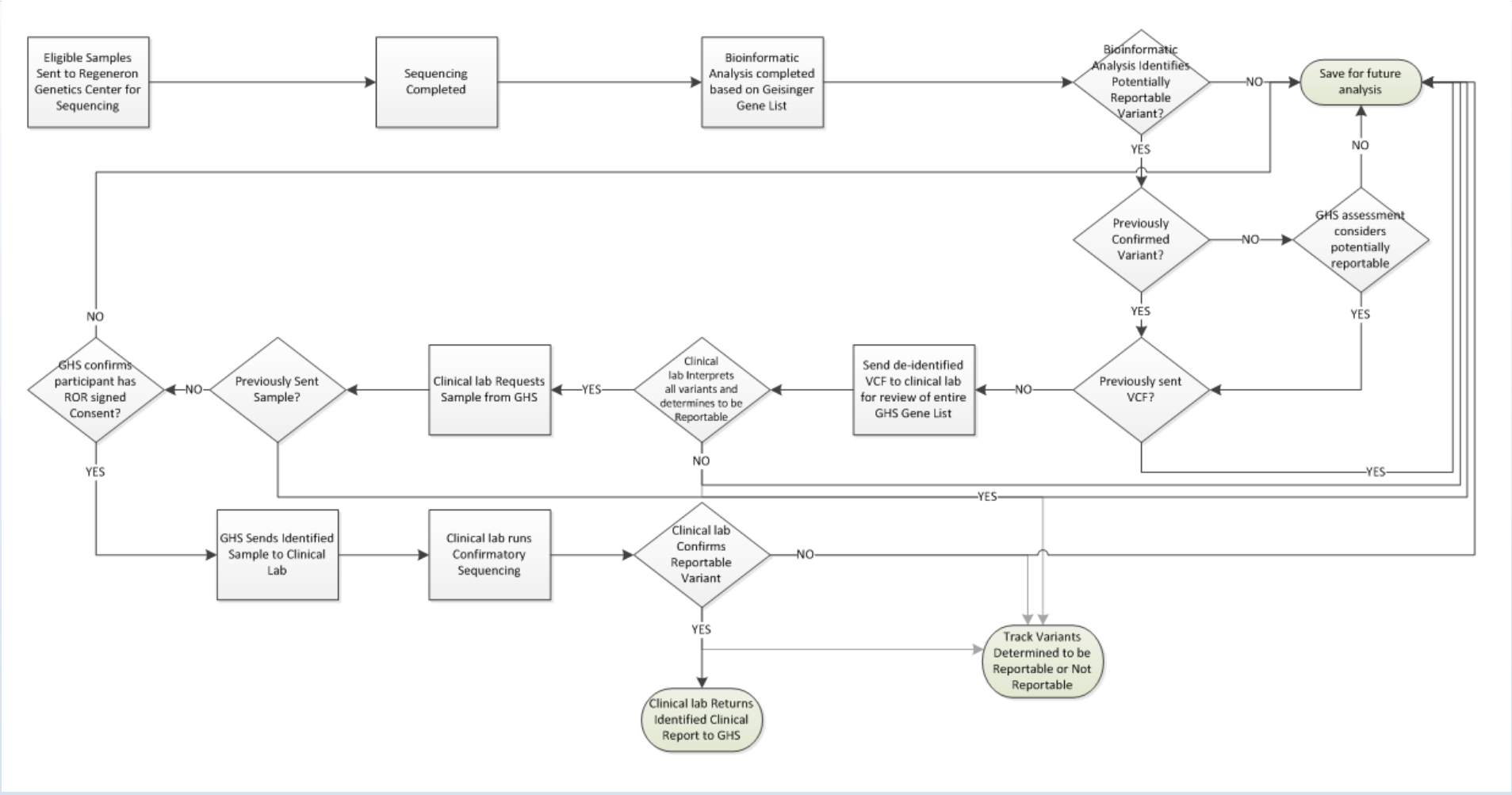
Sequencing, confirmation, and reporting- In practice



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Reporting Results to Participants and Families

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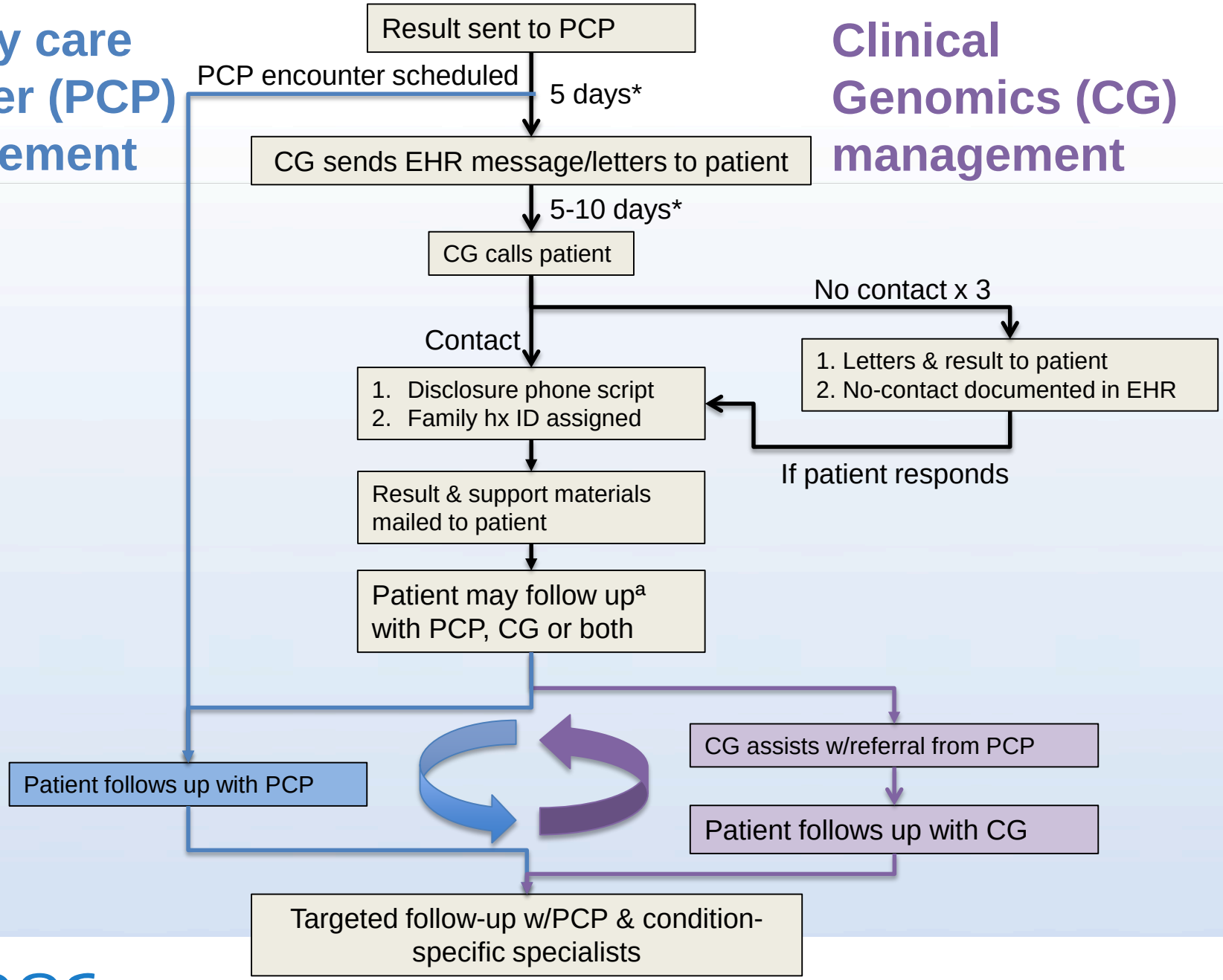
Primary care provider (PCP) management

Clinical Genomics (CG) management



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*Business; [a]Follow-up includes genetic counseling & medical evaluation

Measuring Outcomes Attributable to Reporting

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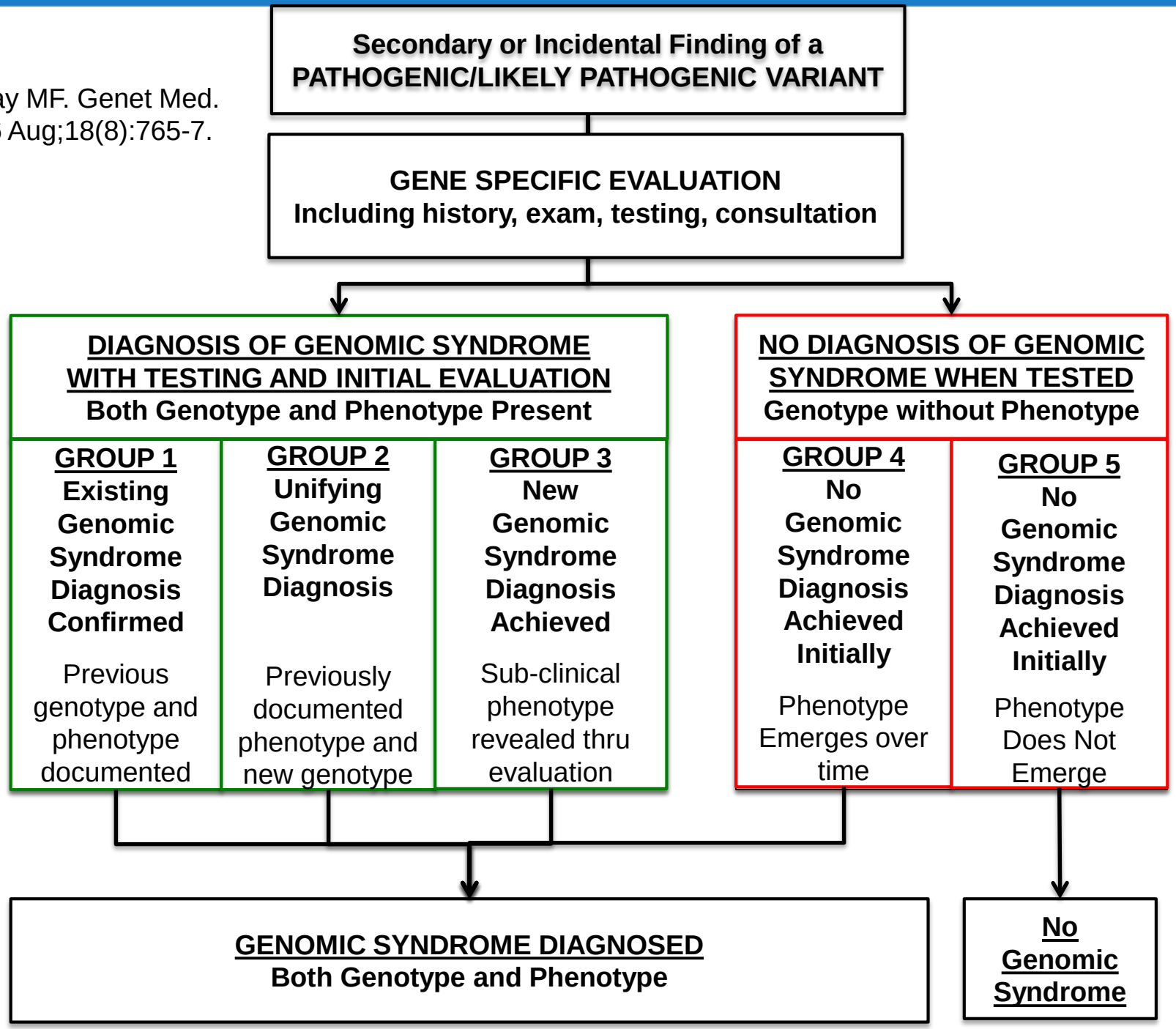
Murray MF. Genet Med. 2016 Aug;18(8):765-7.



Outcomes

Outcome Type	Description	Examples
Process	These measures are the specific steps in a process that lead – either positively or negatively – to a particular health outcome	Lipid profile performed after return of a pathogenic variant in *LDLR* a gene associated with familial hypercholesterolemia
Intermediate	A biomarker associated – either positively or negatively – to a particular health outcome	An LDL cholesterol level at or below the target level of 100 mg/dl in response to interventions recommended based on presences of a pathogenic variant in *LDLR*
Health	Change in the health of an individual, group of people or population which is attributable to an intervention or series of interventions	Decrease in myocardial infarction, or cardiac revascularization procedures in response to interventions recommended based on presences of a pathogenic variant in *LDLR*
Cost	Standard costs associated with the interventions and health states experienced by the patient. Can also include costs associated with patient report outcomes from self-reported health state and life disruption.	Cost of sequencing Cost of genomics results delivery infrastructure Direct costs of care related to return of genomic information Utilization
Behavioral	Change in patient or provider behavior attributable to genomic information	Improved adherence to medication Modification of care based on condition-specific recommendations
Patient-reported	Report of the status of a patient's health condition, knowledge, or service outcomes that comes directly from the patient, without interpretation of the patient's response	Satisfaction with service Engagement with self-care Knowledge about gene and disease Access to recommended care Self-assessed well being Family communication of genomic risk result and uptake of cascade testing

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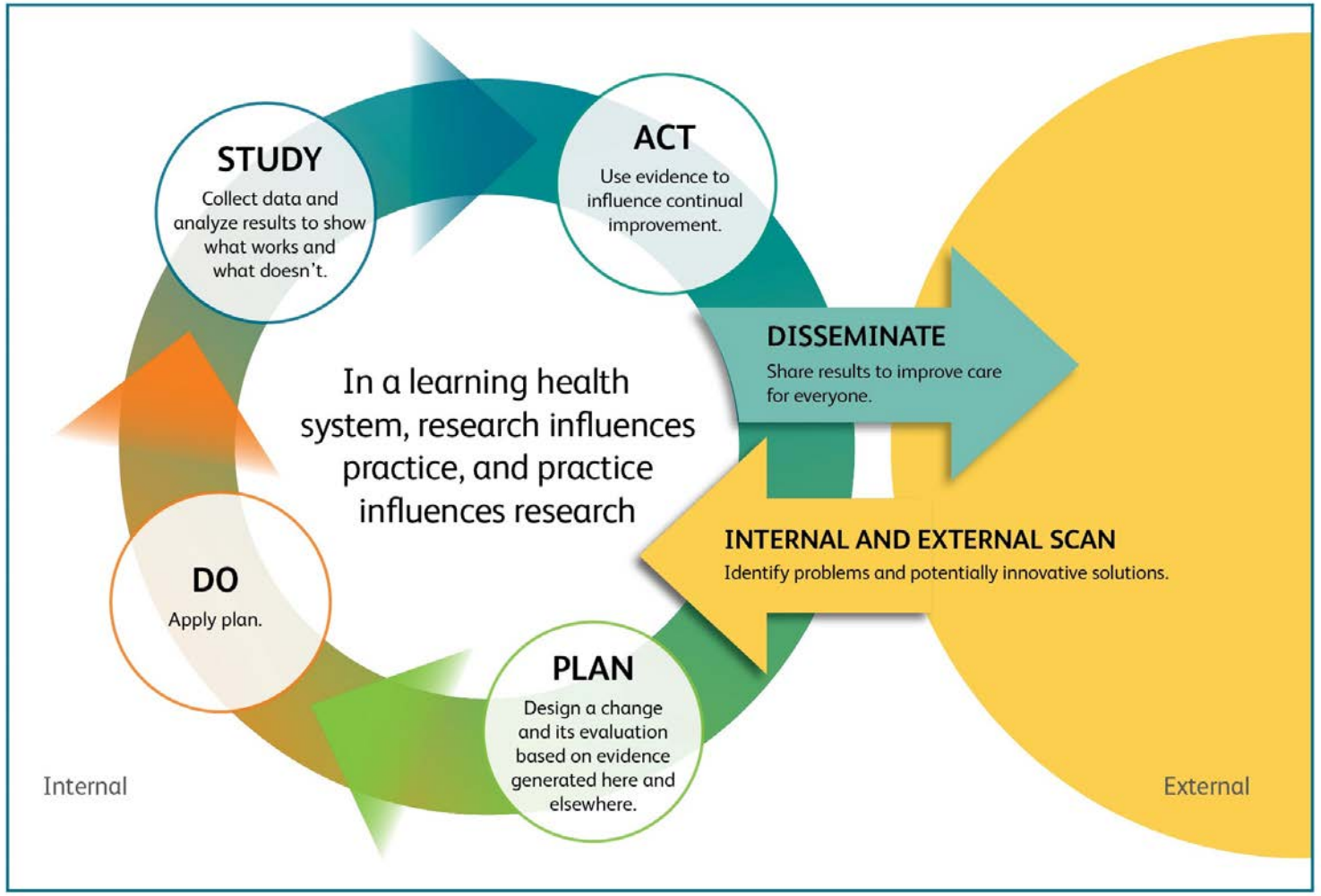
System Outcomes

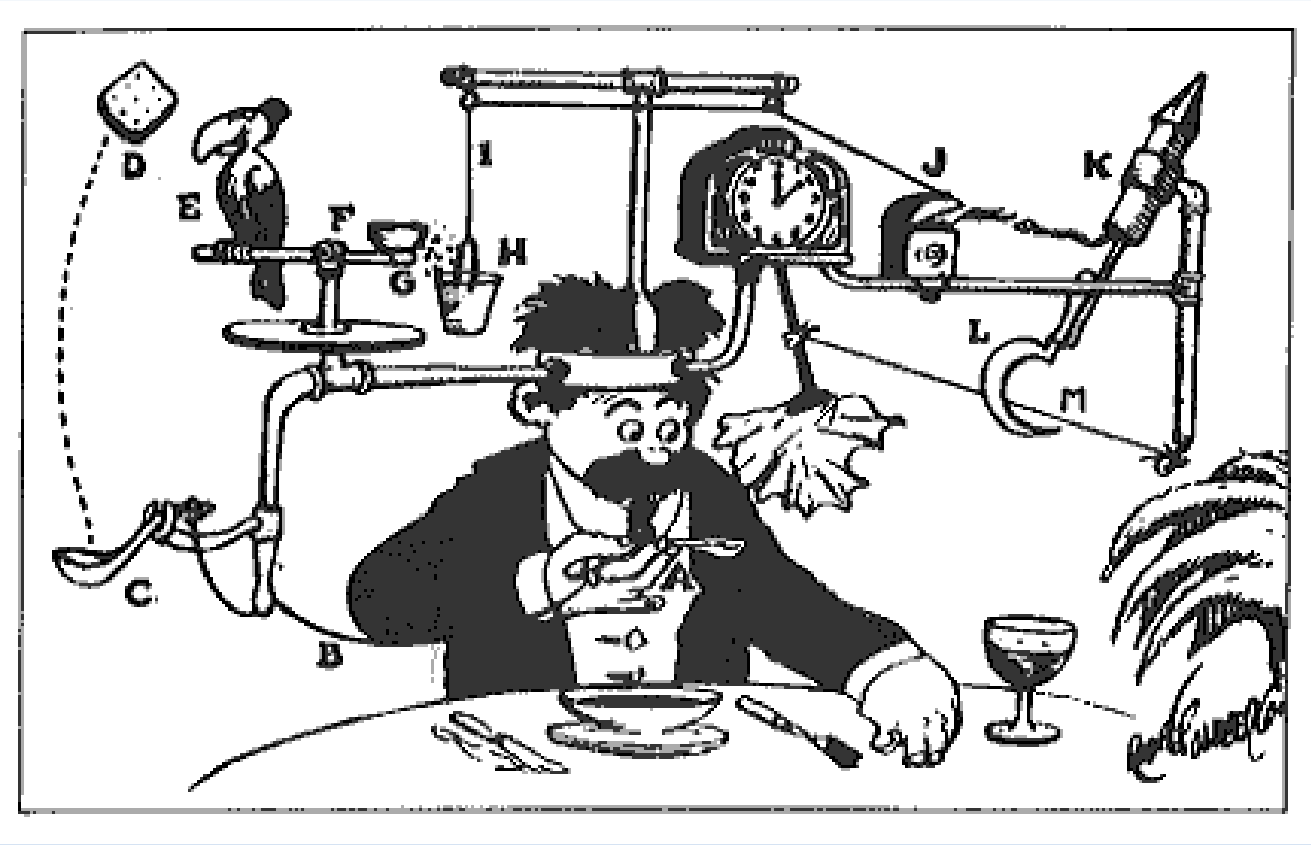
- Costs incurred/avoided
- Utilization
- Visibility/reputation
- Patient experience

Precision Health in a Learning Healthcare System



Source: KP Washington Health Research Institute

Does it work?



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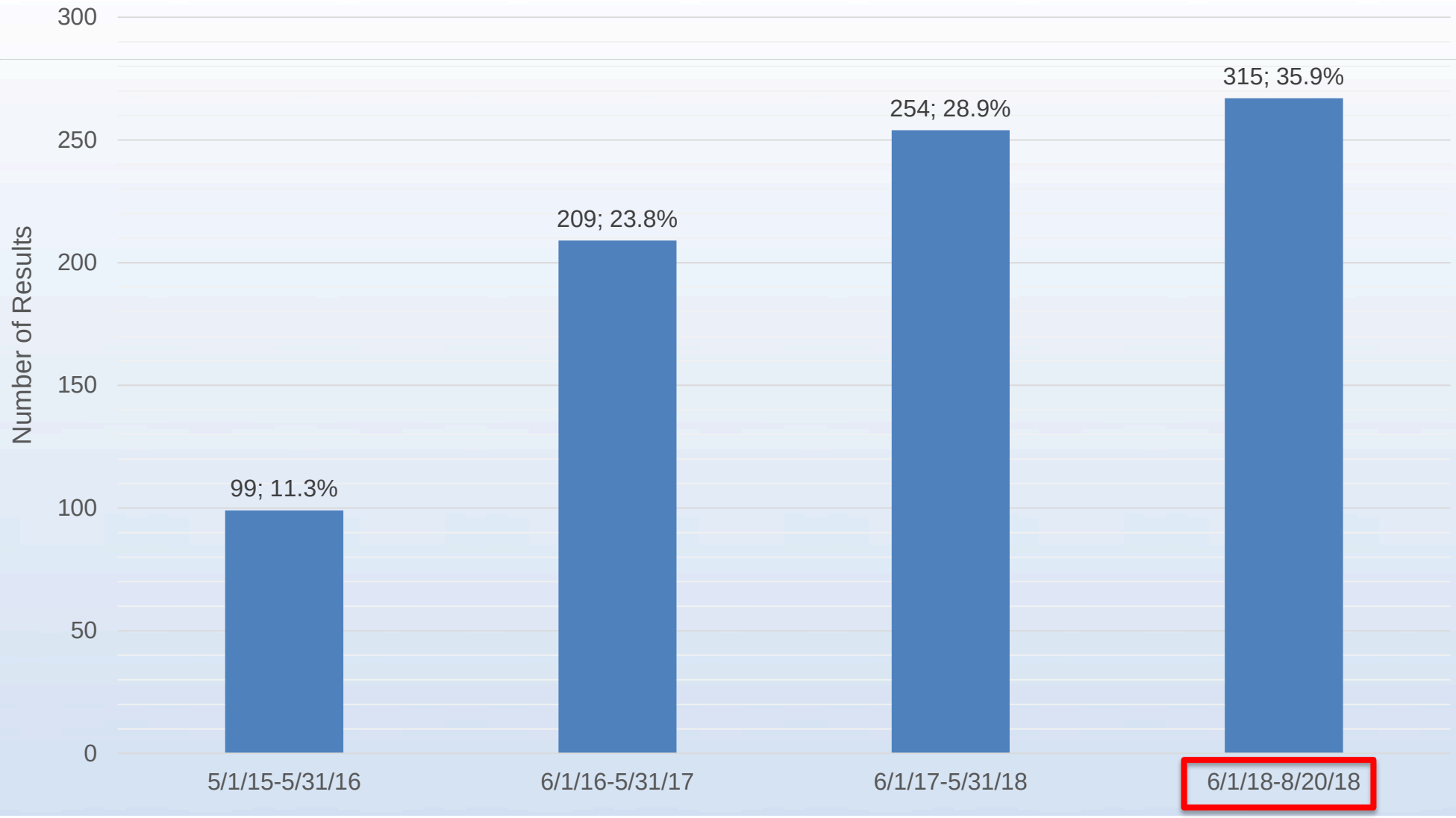
Results Returned to Date



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Re-engineered Sequence interpretation, confirmation and reporting, and Reporting results to participants and family

Conclusions

- Precision health is an emerging technology that must be able to demonstrate improved value in the health care delivery setting before it will be adopted
- Outcomes must defined and systems built to support measurement to determine which services add value
- Implementation is complex and requires a systematic approach. The learning health care system framework may represent a robust implementation model*

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Williams MS, et. al. Patient-Centered Precision Health In A Learning Health Care System: Geisinger's Genomic Medicine Experience. Health Aff (Millwood) 2018 37:757-764.

Acknowledgments

- MyCode Participants: over 200,000 Geisinger Patients
- Genomic Screening and Counseling Program
- DiscovEHR Collaborators: At Regeneron and Geisinger