Future Directions in Genomic Medicine

Inter-Society Coordinating Committee for Practitioner Education in Genomics

Teri Manolio, M.D., Ph.D.
Division of Genomic Medicine, NHGRI

April 5, 2024

National Human Genome Research Institute

The Forefront of Genomics®

Future Directions in Genomic Medicine

- NHGRI's approaches to identifying future directions
- Implementation research (UDN/NICU/PGx)
- Genomic Learning Health Systems
- eConsults
- Population screening
- Hidden Mendelians – Phenotype Risk Scores (PheRS)
- Training opportunities and priorities
- Not covered: Data sharing, genomic influences on lab values, AI methods, multi-omic technologies
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NACHGR Genomic Medicine Working Group Members

Carol Bult	Jackson Labs
Rex Chisholm	Northwestern
Pat Deverka	Deverka Consulting
Geoff Ginsburg	All of Us Research Program
Gillian Hooker	Concert Genetics
Gail Jarvik	U Washington
George Mensah	NHLBI
Casey Overby Taylor	Johns Hopkins
Dan Roden	Vanderbilt
Marc Williams	Geisinger

<u>NHGRI</u>

Eric Green	Erin Ramos
Teri Manolio	Robb Rowley

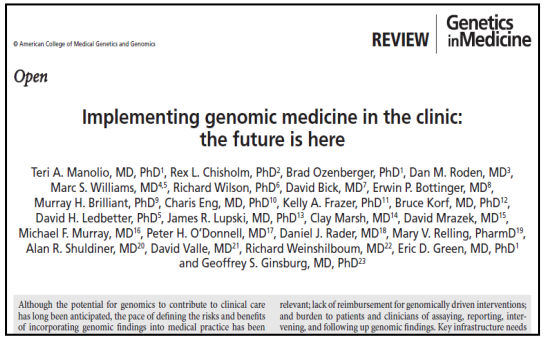
Jahnavi Narula

Genomic Medicine Working Group Charge

Assist in advising NHGRI on research needed to evaluate and move genomics into routine medical practice

- Review current progress, identify research, implementation, and education gaps and approaches for filling them
- Identify and publicize key advances
- Plan genomic medicine meetings on timely themes
- Facilitate collaborations, coordination, long-term availability of genomic resources

Genomic Medicine Colloquium, June 2011

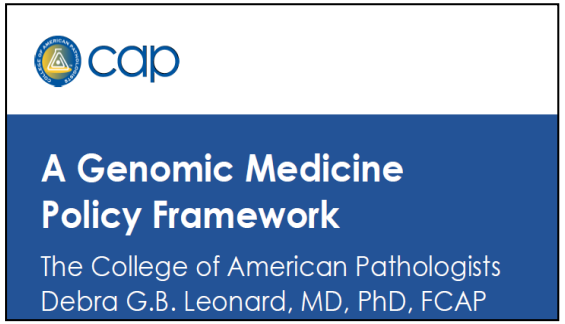


GM II: Forming Collaborations, Dec 2011

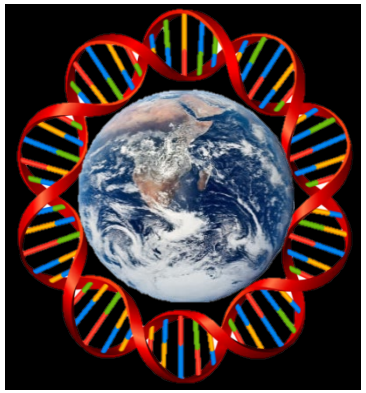
GM III: Stakeholders, May 2012

GM IV: Physician Education, Jan 2013

GM V: Federal Strategies, May 2013



GM VI: Global Leaders, Jan 2014



GM VII: Genomic CDS, Oct 2014

GM VIII: NHGRI's Genomic Medicine Programs, June 2015

GM IX: Bedside Back to Bench, April 2016

GM X: PGx Implementation, May 2017

GM XI: Clinical Implementation, Sept 2018

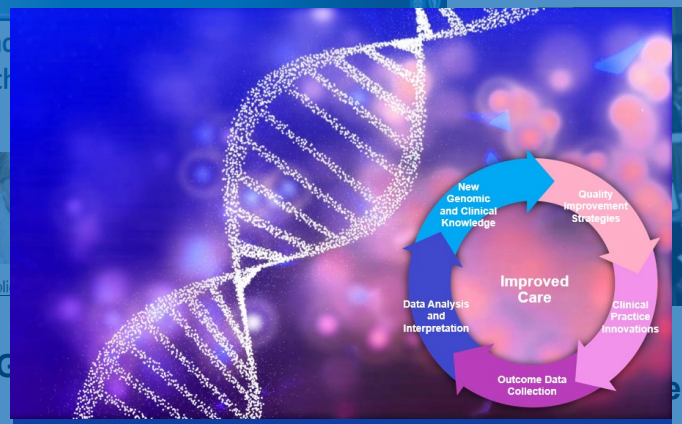
GM XII: Genomics and Risk Prediction, May 2019

Genomic Medicine Colloquium, June 2011

GM II: Forming Collaborations, Dec 2011

GM XIII: Clinical Informatics Research Agenda, Feb 2021

GM III: Stakeholders, May 2012



GM XIV: Genomic Learning Healthcare Systems, Aug 2022

GM IV: Physician Education, Jan 2013

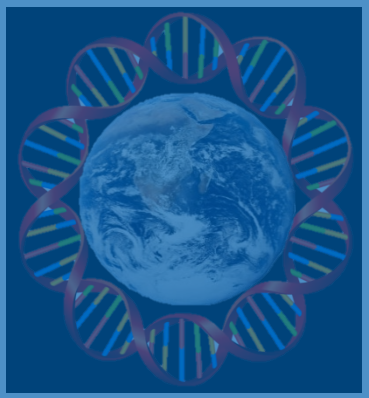
GM X: PGx Implement… May 2017

…ederal Strategies, May 2013

A Genomic Medicine Policy Framework

The College of American Pathologists
Debra G.B. Leonard, MD, PhD, FCAP

GM VI: Global Leaders, Jan 2014



GM IX: Bedside Back to Bench, April 2016

GM VIII: NH… Medicine Pro…

GM XV: Population Screening and Genomics, Nov 2023

…CDS, Oct 2014

Key Products of Genomic Me eetir

GenCC

A global effort to harmonize gene-level resources.

Home | GenCC Database | About | Members | News & Articles

PA-18-167
Variant, Function, Disease

NOT-HG-20-020
Genomic Medicine Modules

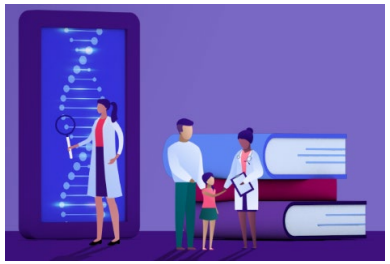
Employers Health and Wellness

RFA-HG-24-001
PAR-24-106/7
eConsults

NOT-HG-22-011
Patient-Centered Informatics

Population Screening Concept

ClinGen
Clinical Genome Resource



ISCC-PEG

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15

Clin Action

ADOPT-PGx

emerge network

Genomic Risk Assessment

G2MC

eMERGE PGx

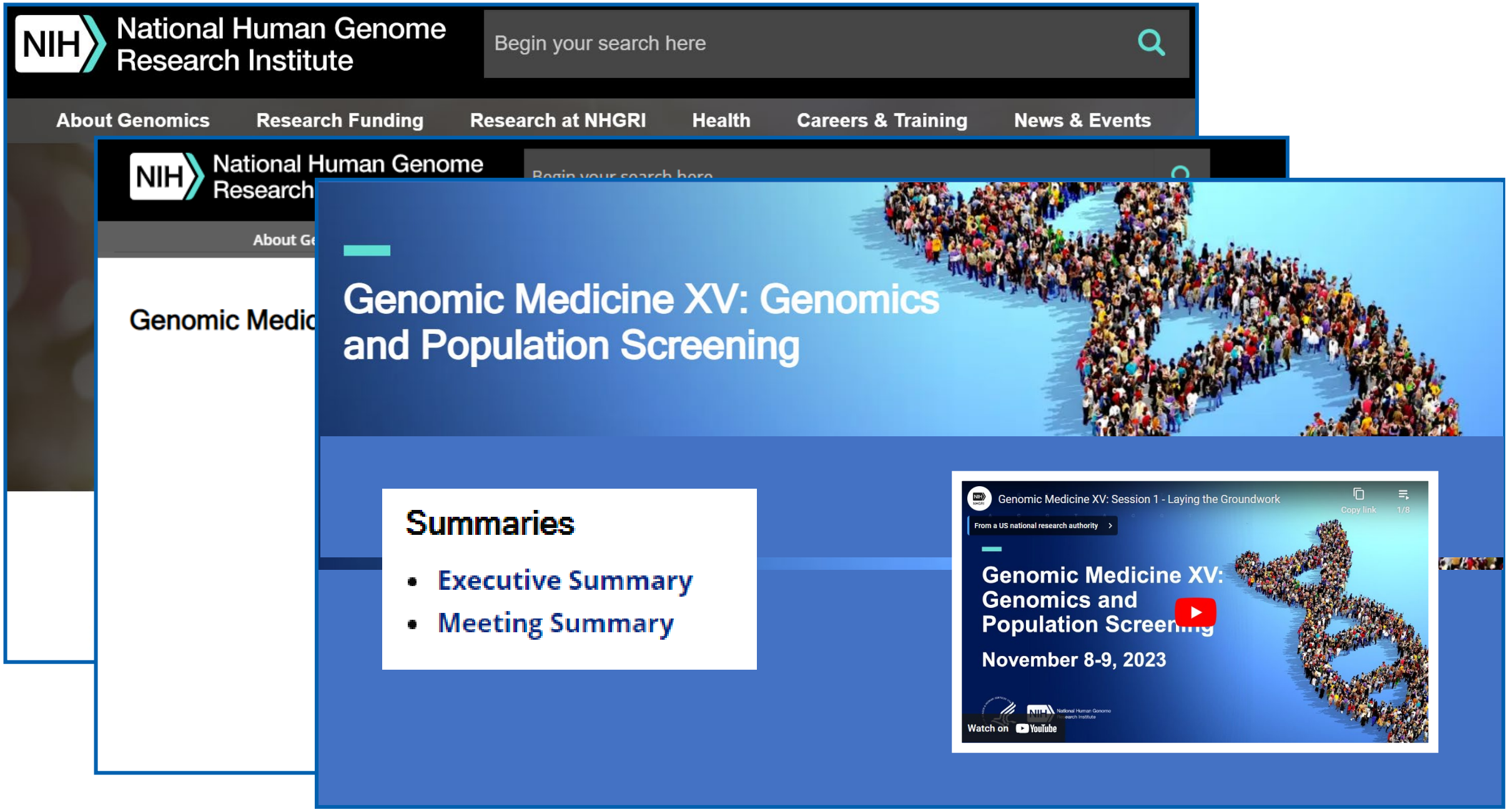
IGNITE
Implementing GeNomics In pracTicE

IHCC

PRIMED consortium

RFA-HG-23-041/2
Genomic LHS

Genomic Medicine Meetings



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Research-Practice Gap

17-20 years to get clinical innovations into practice

Fewer than 50% of clinical innovations make it to practice

80% of dollars spent on research and implementing innovations do not make public health impact

Only 14% of research and innovations reach target recipients

Courtesy A Kulchak Rahm, NHGRI

Bauer MS, Kirchner J. Implementation science: what is it and why should I care? Psychiatry Research 283 (2020)
Balas EA, Boren, SA. Managing clinical knowledge for healthcare improvement. In Yearbook of Medical Informatics. 2000.
Chagnon F, et al. Comparison of determinants of research knowledge utilization by practitioners and administrators in the field of child and family social services. Implementation Science. 2010:5:41.

If you implement a new practice, guideline, process and it doesn't work……

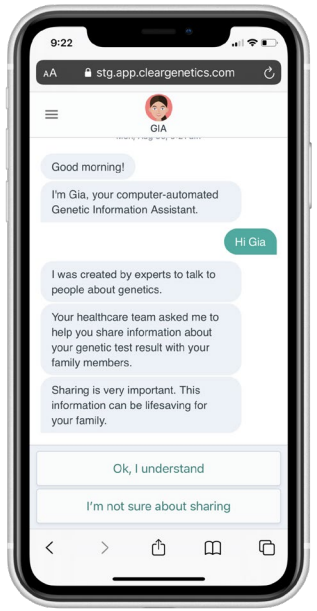
Was it because…

- it isn't *effective* in your setting/population? (an efficacy or effectiveness issue)

- the *operationalization* of it in your setting/population affected the utilization? (an implementation issue)

Courtesy A Kulchak Rahm, NHGRI

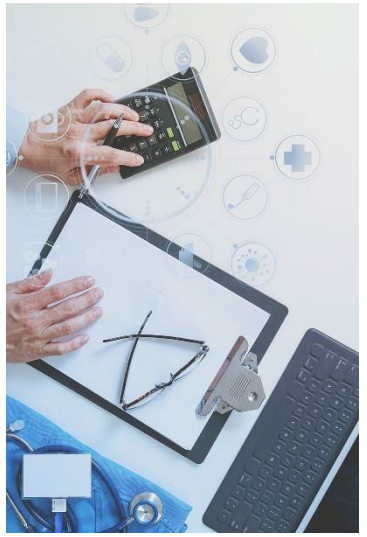
Implementation Science in Plain Language



The intervention / Program/ Innovation Is **THE THING**

Effectiveness research asks **DOES THE THING WORK**

Implementation Strategies are the stuff we do to try and help people/ places **DO THE THING**

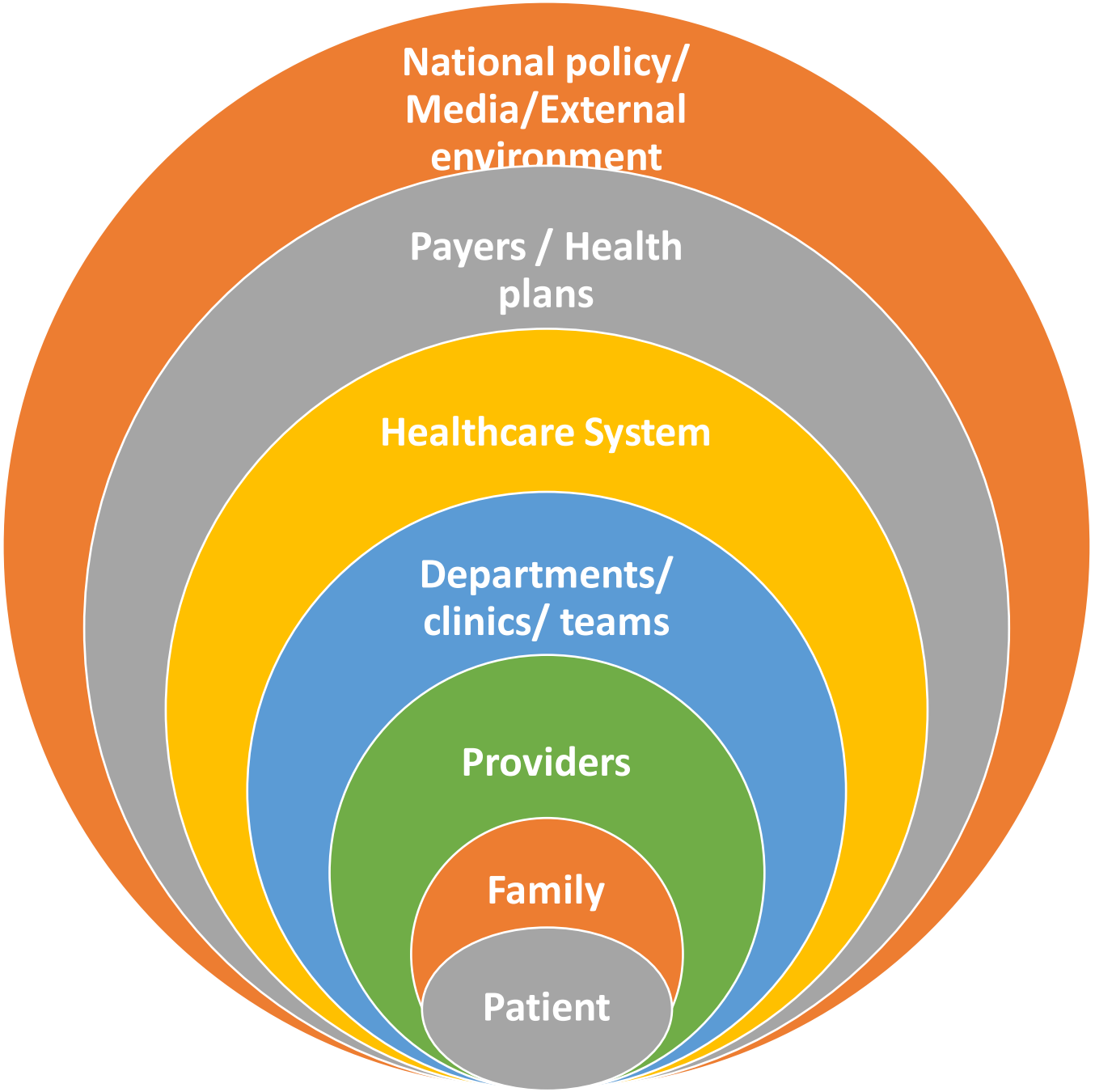


Implementation Outcomes are **HOW MUCH** and **HOW WELL** they **DO THE THING**

Courtesy A Kulchak Rahm, NHGRI

Curran GM. Implement Sci Commun. 2020

Implementing genomic information into care is a multi-level complex issue



Courtesy A Kulchak Rahm, NHGRI

Why You Should Care about Implementation Science

- The gap between what we know works and what we do is still large
- Most evidence-based practices never get implemented in healthcare systems or public health programs
- Personalizing programs to the populations for whom they will work and designing them in the way they will work for the system implementing the program promotes adoption
- Precision health is changing rapidly – we need to learn as we go and adapt over time, but also help others learn from what we discover

Courtesy A Kulchak Rahm, NHGRI

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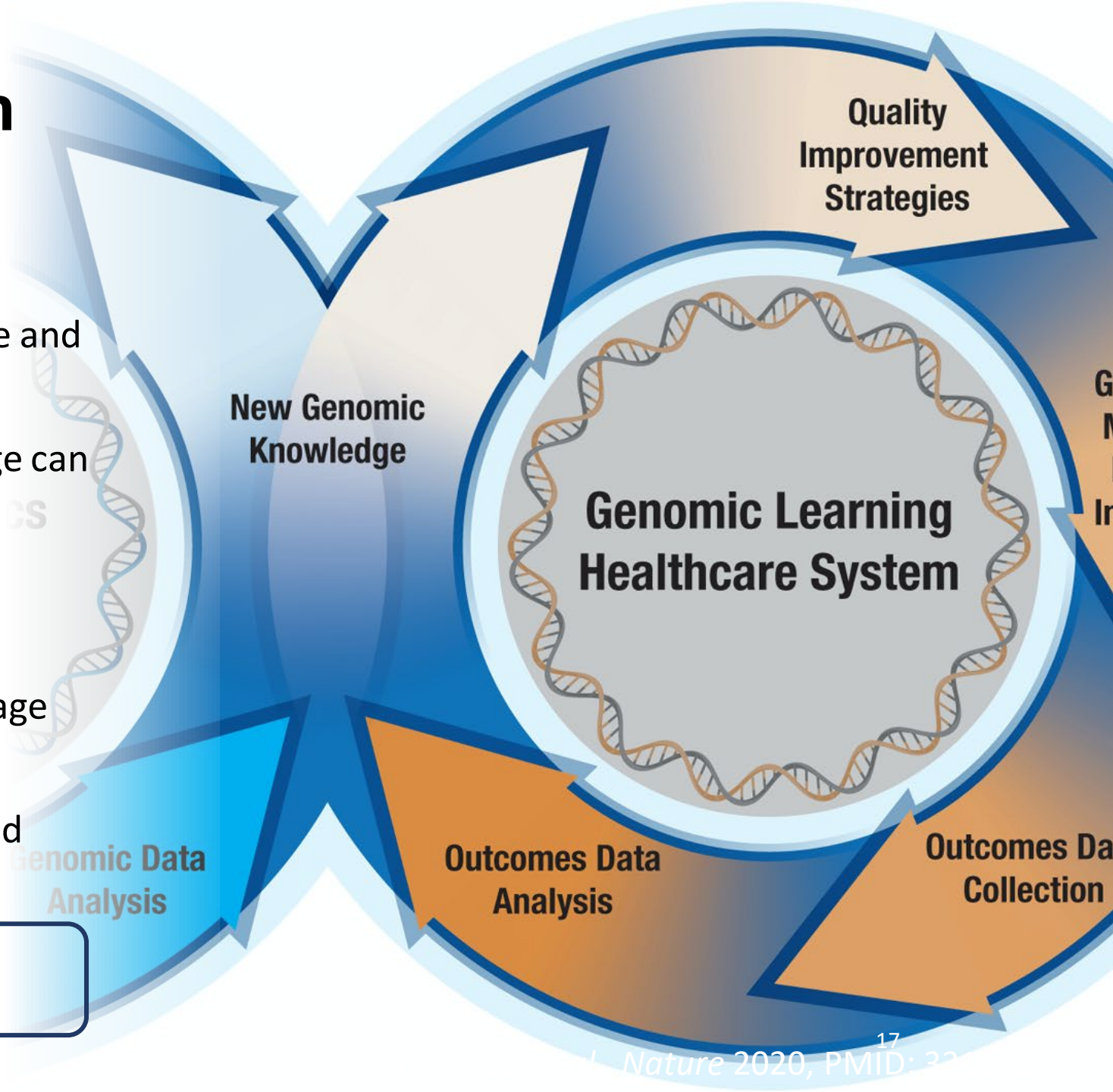
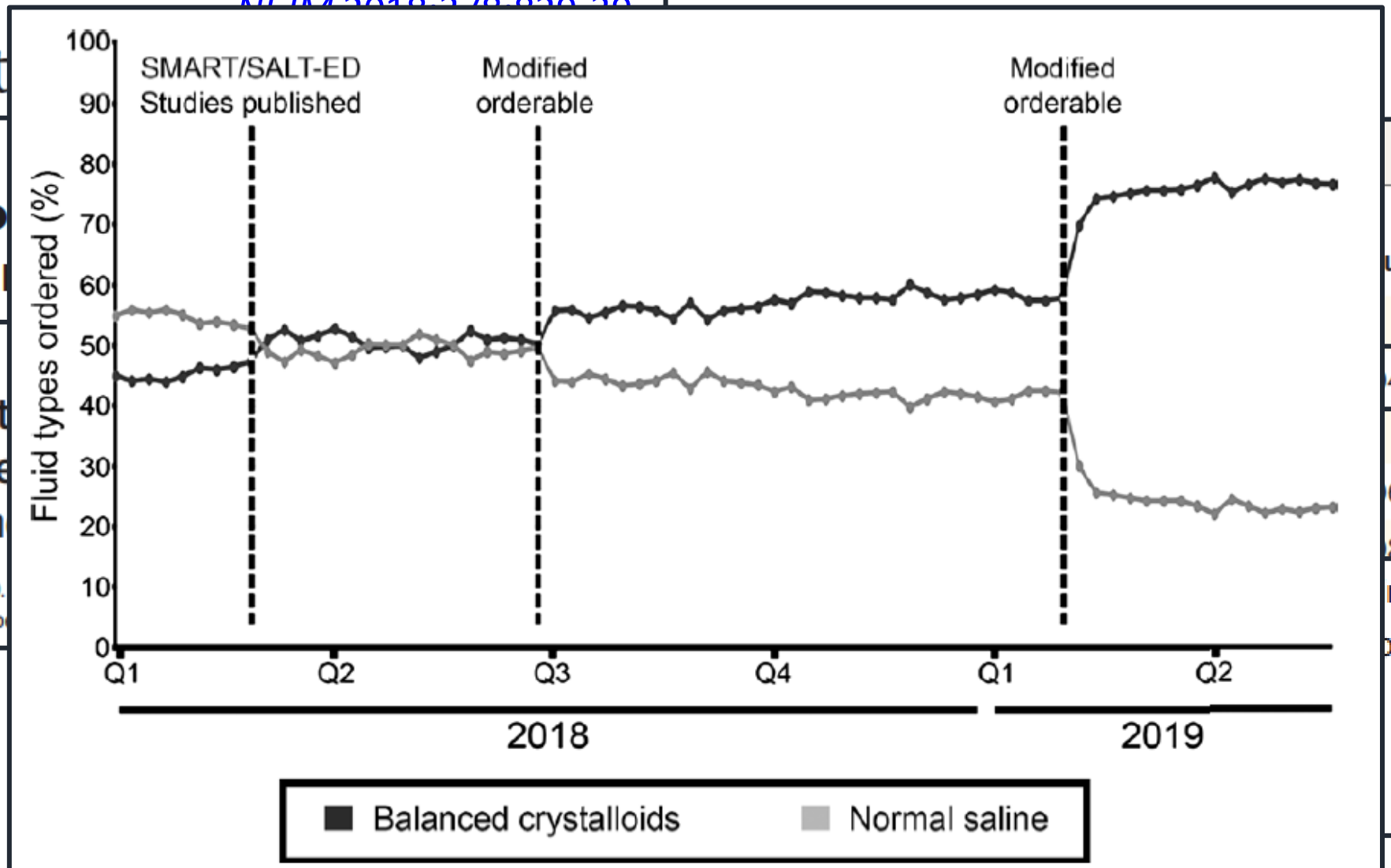
What is a learning health system (LHS)?

- One in which internal data and experience are systematically integrated with external evidence and resulting knowledge is put into practice
- Fundamental principle: Generalizable knowledge can be captured from every patient encounter and provided to clinicians to improve practice
- Examples:
  - Early introduction of palliative care in end-stage liver disease reduces readmissions
  - Testing and de-labeling patients with reported penicillin allergy is safe and effective
  - Balanced crystalloids reduce adverse kidney outcomes in critically ill patients



Nature 2020, PMID: 32

LHS Model of Implementation, Evaluation, Re-implementation



Gaps that gLHS could address

- Slow uptake of several evidence-based genomic medicine interventions:
  - Actionable conditions (HBOC, FH, Lynch Syndrome testing)
  - Time-sensitive genome sequencing in critically ill infants
  - Pharmacogenomic testing to reduce adverse drug reactions
- Limited incorporation of genomics in LHS
- Need for improved exchange of genomic information across health systems
- Limited dissemination of gLHS approaches, tools, resources

Courtesy R Rowley, NHGRI

Department of Health and Human Services

Part 1. Overview Information

Funding Opportunity Title	Network of Genomics-Enabled Learning Health Systems (gLHS)
Notice	
Compa	

Objective: Establish Network of institutions with track record of using gLHS approaches in their health system, including in resource limited communities

- Refine and develop these practices into implementation resources
- Identify 2-4 Network-wide implementation projects
- Implement the 2-4 implementation projects Network wide
- Use implementation projects to increase system-wide and across health systems interoperability and refine resources for broader sharing
- Establish validated tools and resources for sites implementing a gLHS

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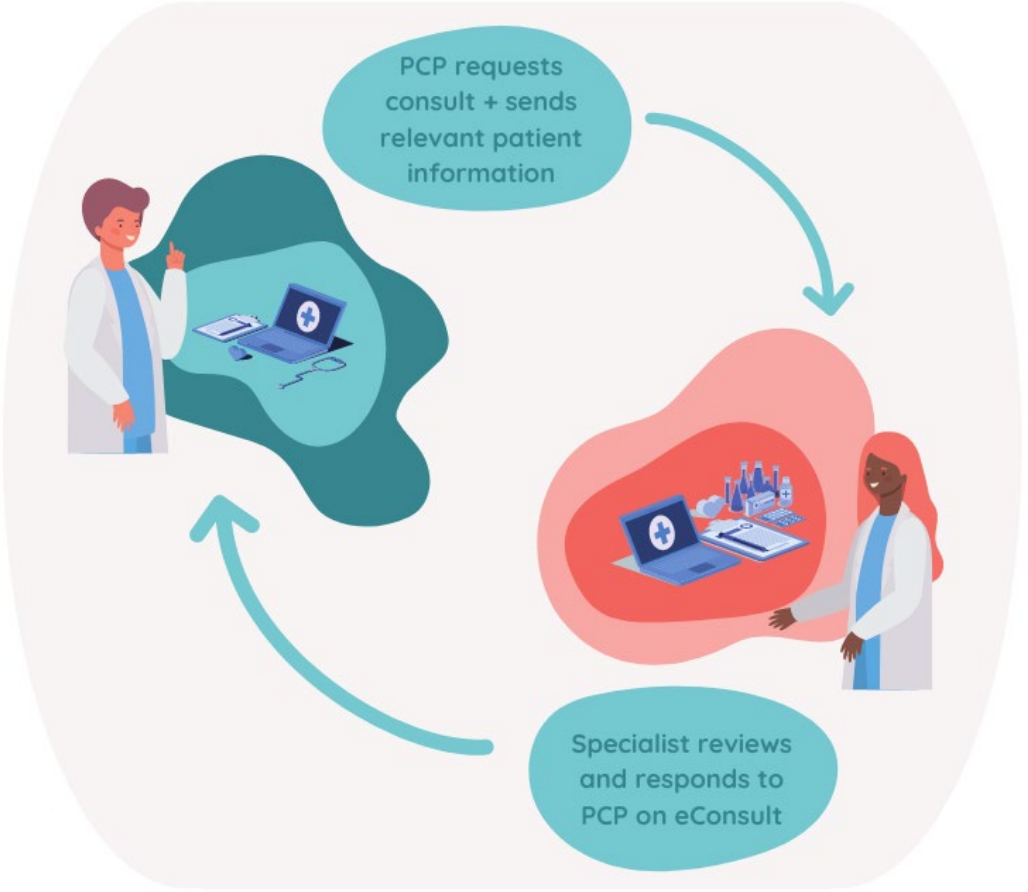
Peer to Peer “eConsults”

- eConsult = clinician-to-clinician support
- Currently utilized by many specialties
  - Provide actionable recommendations
  - Increase primary care providers ability to provide care, decreasing specialty referrals
  - Reduce wait times
  - Decrease patient burden
  - Increase health equity
- Most are within a single institution
- Few genomic medicine eConsult services

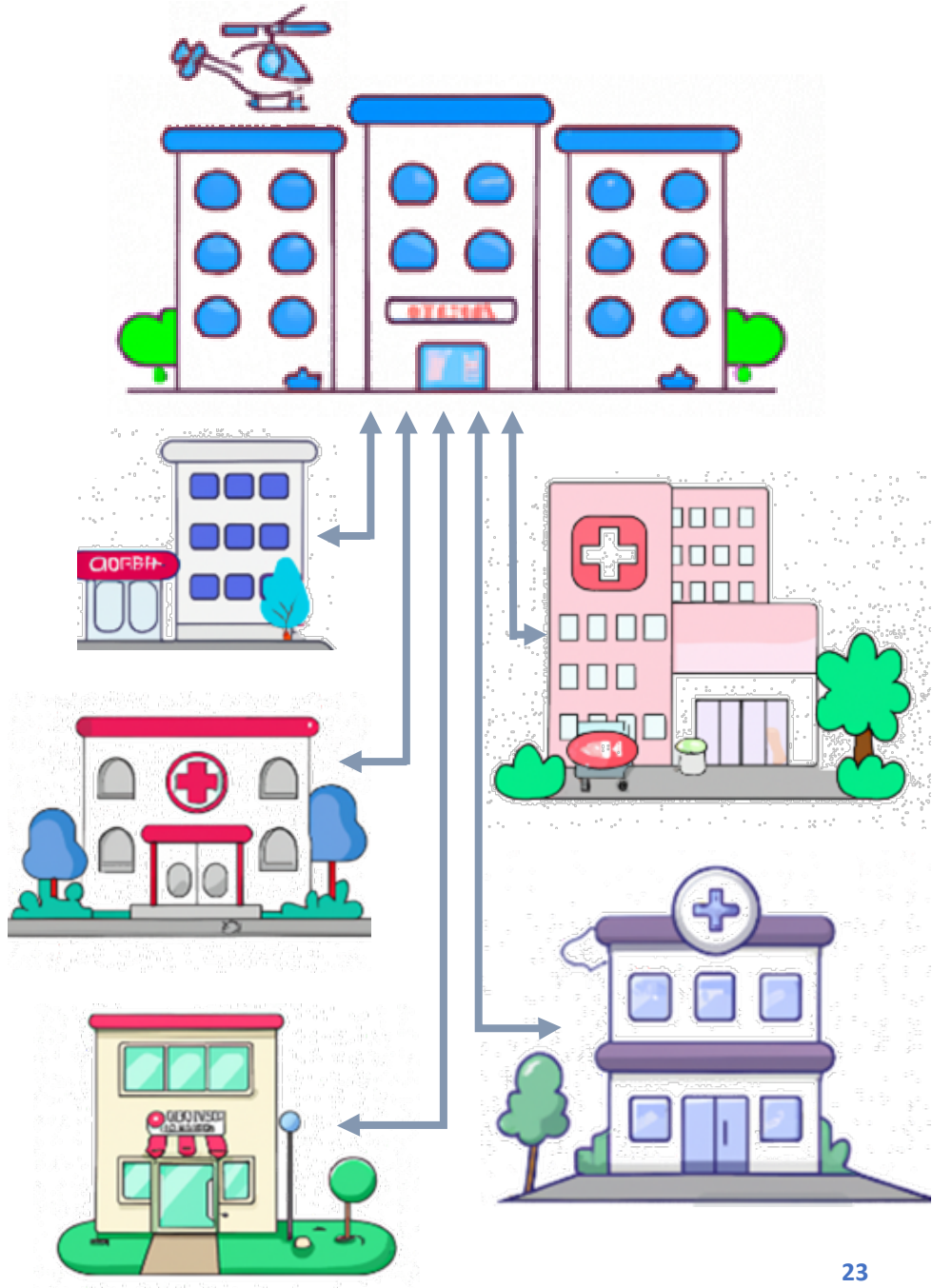


PCP = Primary Care Provider

Courtesy R Rider, NHGRI

Multi-institution eConsult Service

- Specialist(s) at one institution provide support to clinicians at other institutions
  - Including those outside of their system
- Allows clinicians without specialists at their institution to get patient specific recommendations
- Increase health equity, in groups such as:
  - Frail elderly
  - Long-term care residents
  - Rural patients
  - Transgender patients



Courtesy R Rider, NHGRI

Research Questions

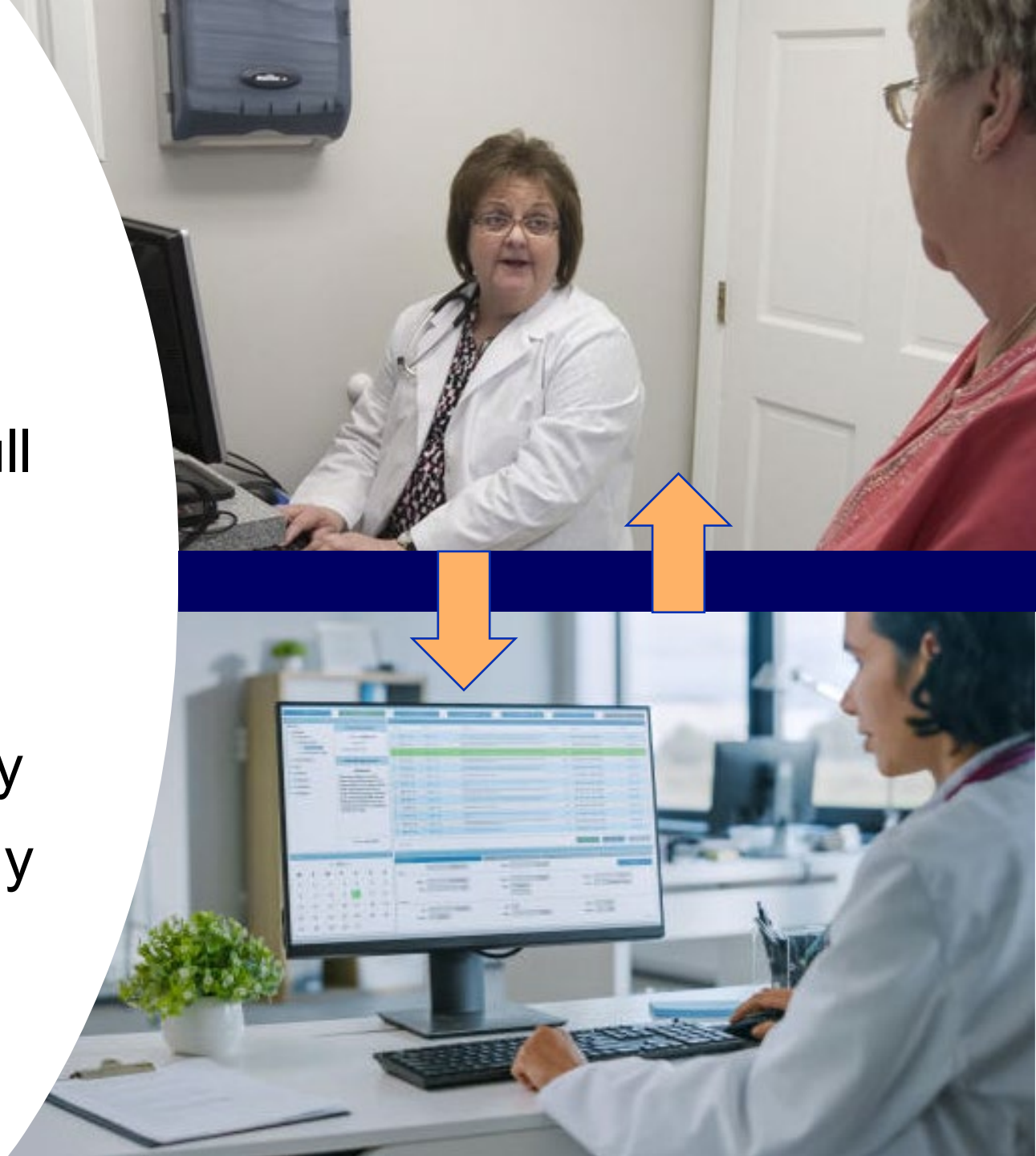
1. What impact do genetic eConsult services have when they are implemented at the regional level?
2. How can regional genetic eConsult services be implemented and sustained?
3. Can tools be created and shared with others who are creating regional eConsult services?

Courtesy R Rider, NHGRI

Genomic Medicine eConsult Network

Expected to:

- Respond to questions from the full breadth of specialties that utilize genomic medicine
- Advise on a wide range of topics
- Assess success and sustainability
- Share tools and resources broadly



NIH NHGRI

Courtesy R Rider, NHGRI

Department of Health and Human Services

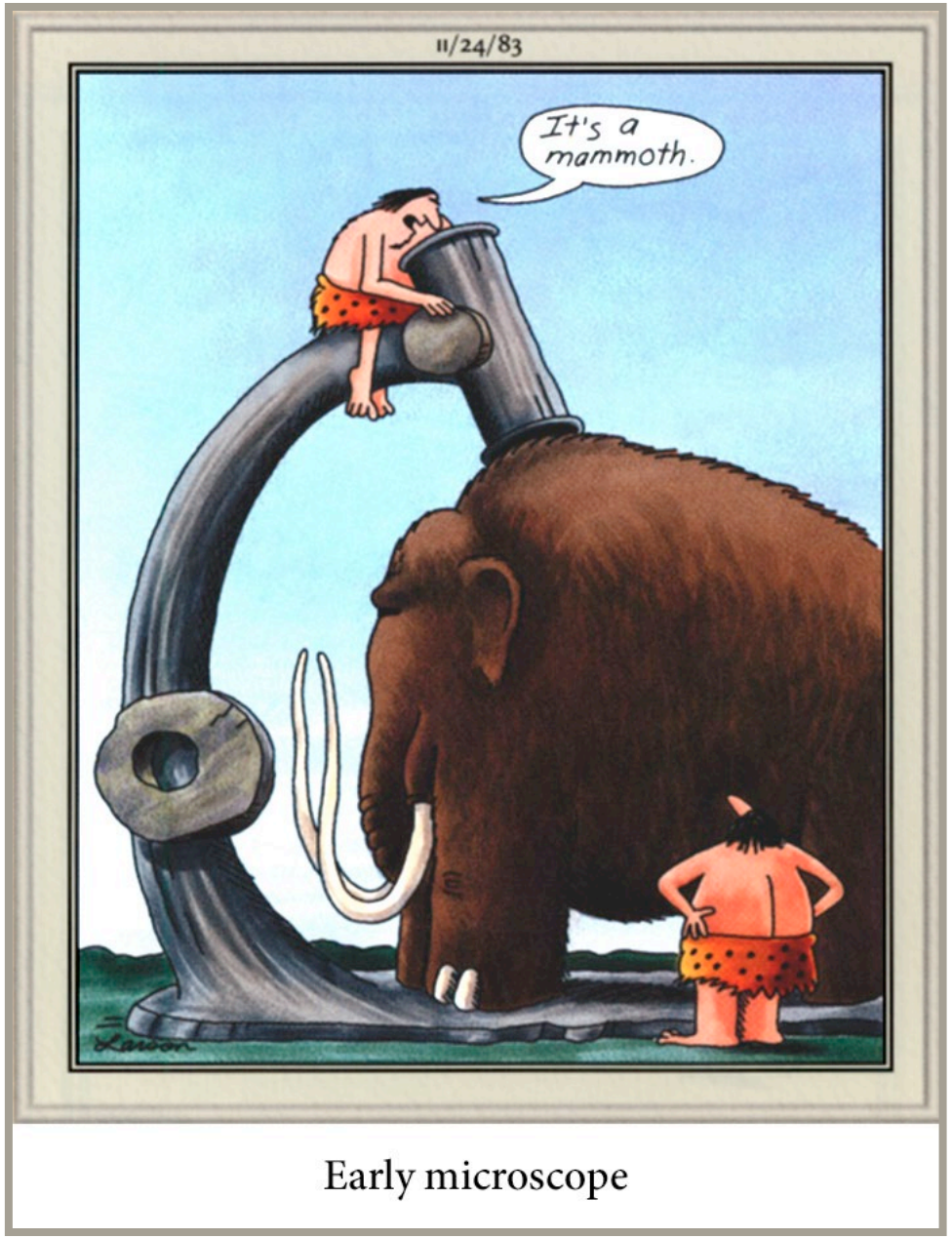
Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	Research on the Impact of [illegible]nting Regional Genomic Medici[illegible] Clinical Trials Optional)

Applications due May 30, 2024

Objective: Conduct re[illegible]t of and methods for implementing regional cli[illegible]nomic medicine eConsult services

- Resear[illegible] best design, implement, and sustain regional genomic medicine eConsult services
- Provide outreach to potential users, including those at underserved settings
- Assess impact on key stakeholders while developing successful implementation strategies and resources that can be broadly shared and adopted.



Early microscope

Larson, G. *The Complete Far Side*. 2003.

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Nov 8-9, 2023, Bethesda

Genomic Medicine XV: Genomics and Population Screening

Planning Group: Jonathan Berg, Gail Jarvik, Bruce Korf, George Mensah

Objectives:

- Review the current state of population genomic screening in the U.S.
- Examine obstacles and opportunities for expanded screening and available evidence of the impact of screening on outcomes and cost
- Identify research directions to inform expanded screening as appropriate

Genomic Medicine XV Recommendations – Screening Pilot

Engagement and equity

- Pilot studies for near-Tier 1 conditions, including engagement of prevention research community
- Approaches to ensure equitable implementation of population screening and follow-up

Data management and analysis

- Methods for storage, access, and transportability of screening data within and among health systems, research enterprise
- Development or improvement of evidence-based models for evaluating genomic screening tests
- Development of a probabilistic model for adding genes for screening, similar to Richards criteria – ACMG effort beginning to address
- Improved estimates of numbers needed to screen, penetrance, natural history of conditions

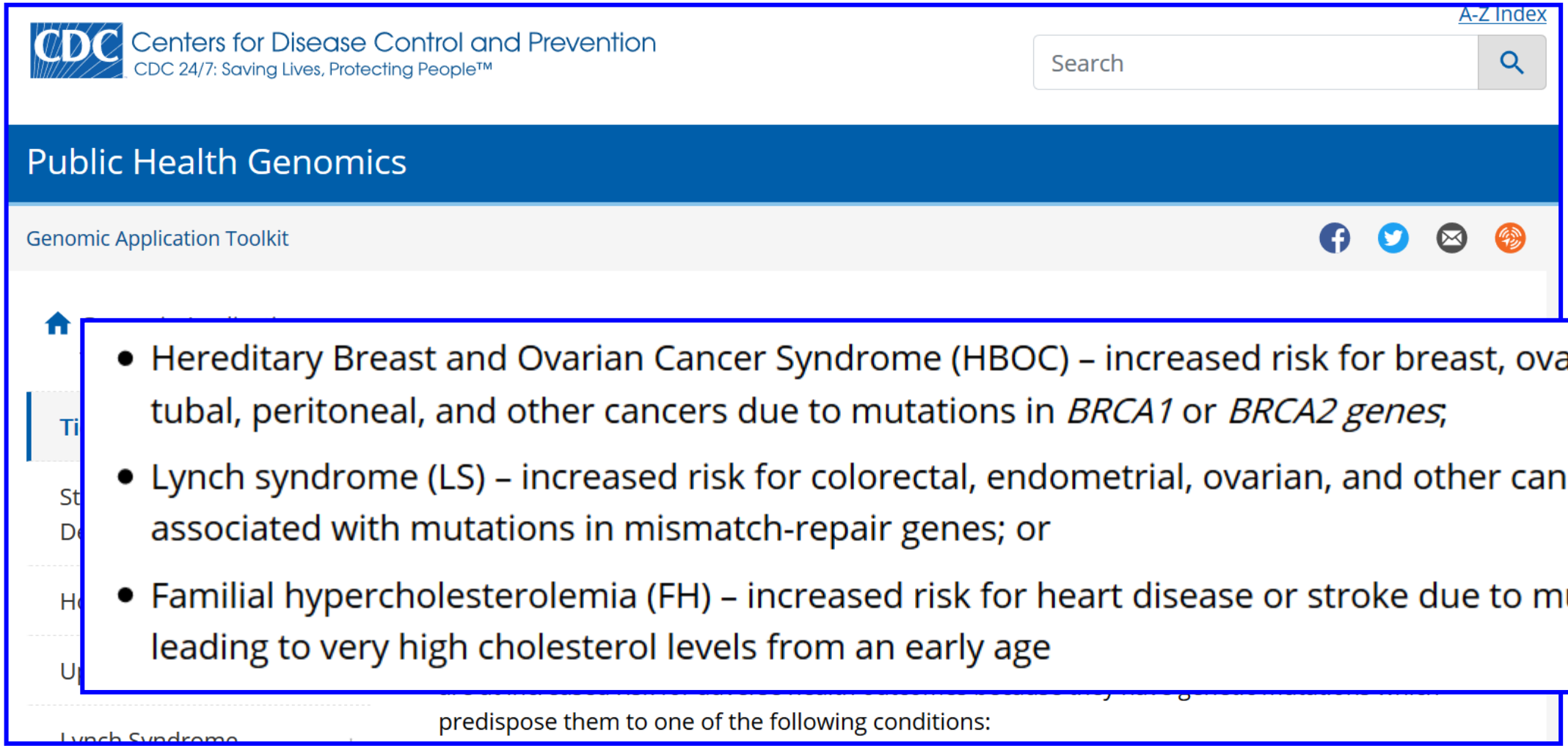
Genomic Medicine XV Recommendations – Screening Pilot

Clinical workflow and communication

- Methods to reduce complexity of, and standardize, pre-test consent and ordering
- Methods enabling
  consultation and
- Potential roles fo
  screening results
- Methods to supp
  as genomic know
- Approaches for setting realistic expectations for genomic screening, mitigating risks of false reassurance, and facilitating accurate communication of results within families

To avoid the risks of false reassurance, individuals with a negative screening result (>98% of all who are tested) should receive effective communication that standard cancer and cardiovascular screening tests are still recommended. – Guzauskas *et al., Ann Intern Med* 2023, PMID: 37155986

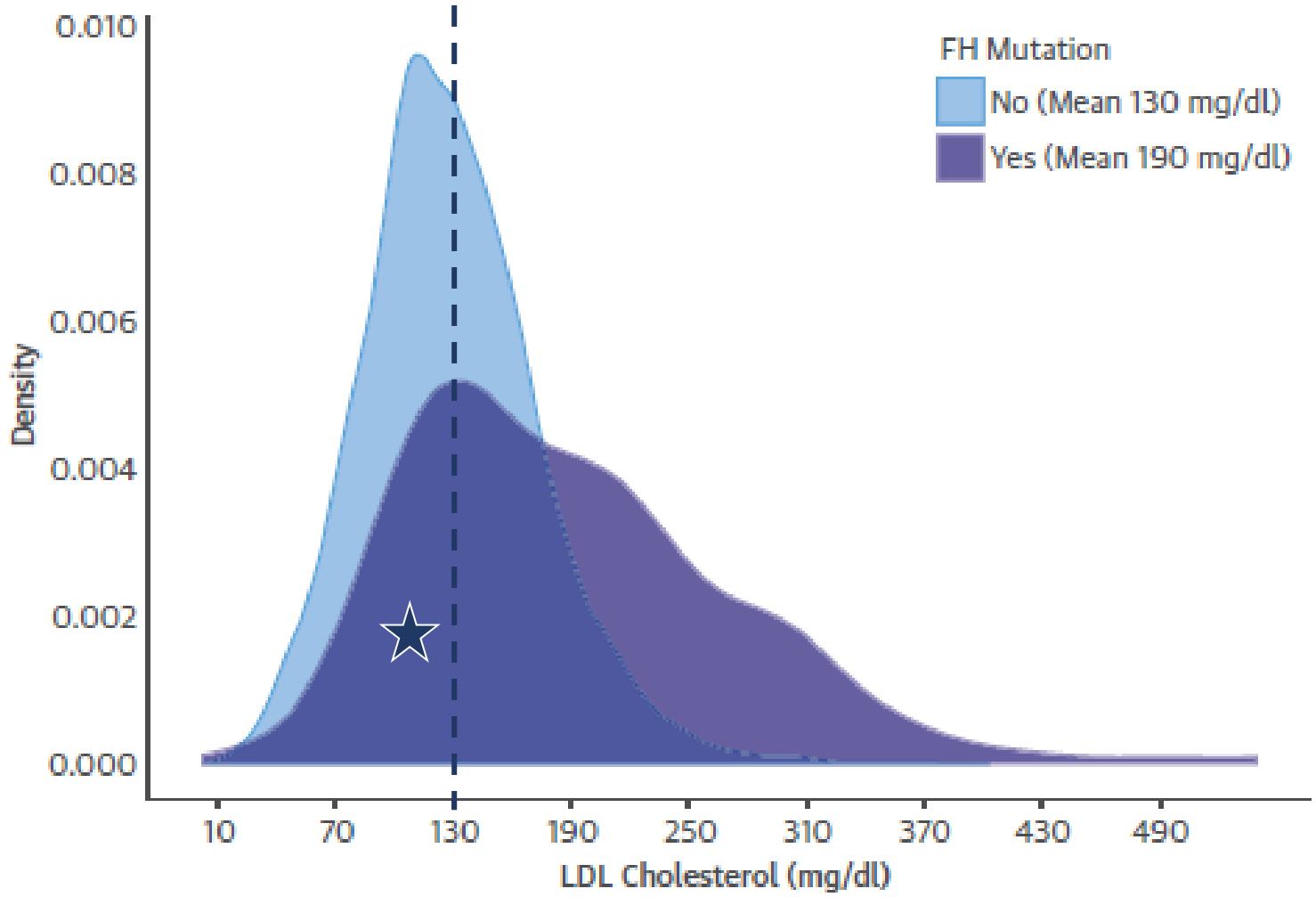
U.S. Centers for Disease Control and Prevention Tier 1 Genomic Applications



- Hereditary Breast and Ovarian Cancer Syndrome (HBOC) – increased risk for breast, ovarian, tubal, peritoneal, and other cancers due to mutations in *BRCA1* or *BRCA2 genes*;
- Lynch syndrome (LS) – increased risk for colorectal, endometrial, ovarian, and other cancers associated with mutations in mismatch-repair genes; or
- Familial hypercholesterolemia (FH) – increased risk for heart disease or stroke due to mutations leading to very high cholesterol levels from an early age

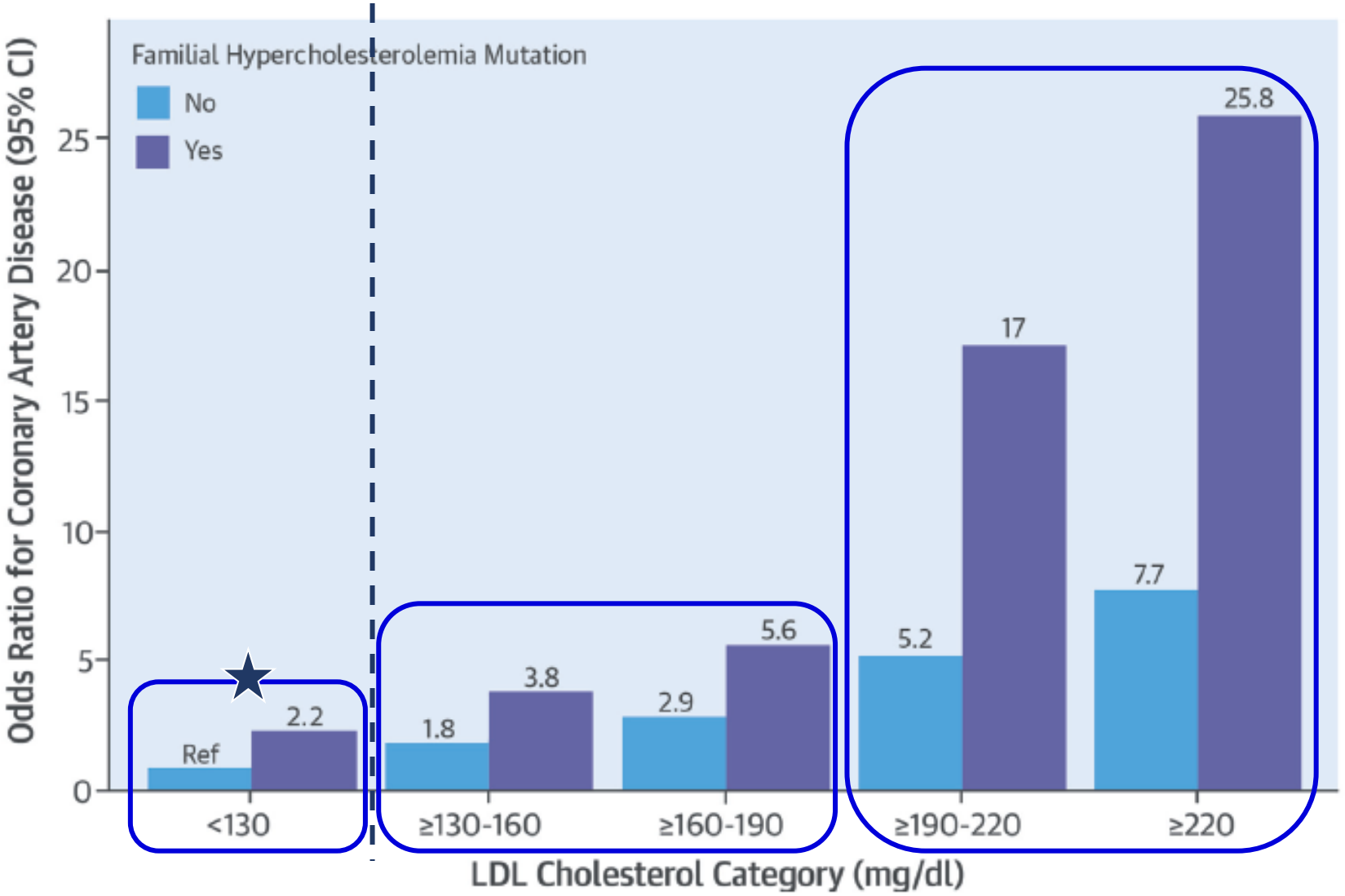
https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm

Overlap of LDL-C Levels in 26,025 Persons with and without FH Mutations



Khera A *et al., J Am Coll Cardiol*. 2016;67(22):2578–89.

FH Variants and CAD Risk by LDL-C Level



Khera A *et al., J Am Coll Cardiol*. 2016;67(22):2578–89.

Population Screening in Primary Care (Feb 2024)

Proposing 3 RFAs for an implementation and evidence generation pilot program of population screening for common, actionable genomic conditions predominantly in the primary care setting. Specifically:

1. Select, implement, and evaluate screening for 4-8 genomic conditions in diverse populations and clinical settings;
2. Use established strategies for meaningful community engagement to design, conduct, and evaluate outcomes of screening; and
3. Develop effective strategies for connecting patients screening positive to follow-up care.

Courtesy S Volpi, NHGRI

Tier 1 Genomic Conditions and Primary Care

- "Tier 1" defined by CDC: HBOC, Lynch syndrome (LS), and FH.
- Are at present poorly ascertained by US healthcare and patients are often unaware of them until they present with late-stage disease.
- Primary care providers (PCPs) are typically "first line" for managing preventive care.
- Gaps in genomic screening for this workforce include efficiency measures, knowledge, confidence, and a robust informatics infrastructure to support analysis.
- Address by picking a few high-value, high-evidence screening tests that are straightforward to implement and understand

Courtesy S Volpi, NHGRI

Community Engagement

Community engagement is critical to conducting successful genomic research and providing effective care.

Incorporate community values, concerns, and aspirations into decision-making and establishing meaningful, ongoing partnerships.

Apply implementation science principles and methods.

Courtesy S Volpi, NHGRI

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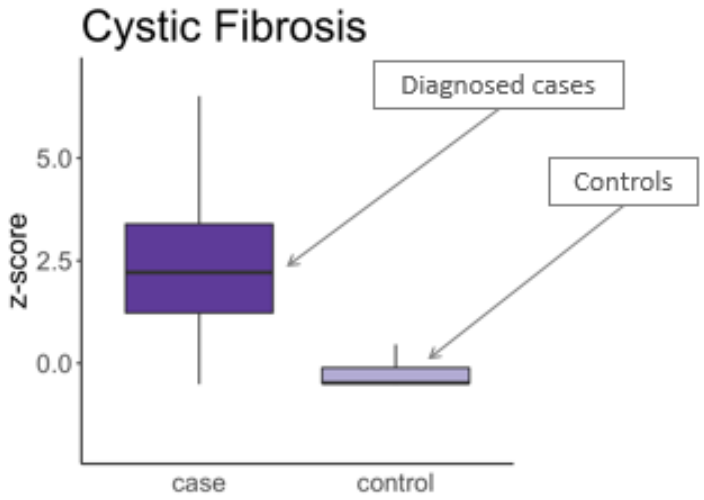
HUMAN GENOMICS

Phenotype risk scores identify patients with unrecognized Mendelian disease patterns

Lisa Bastarache,[1] Jacob J. Hughey,[1] Scott Hebbring,[2] Joy Marlo,[1] Wanke Zhao,[3] Wanting T. Ho,[3] Sara L. Van Driest,[4,5] Tracy L. McGregor,[5] Jonathan D. Mosley,[4]

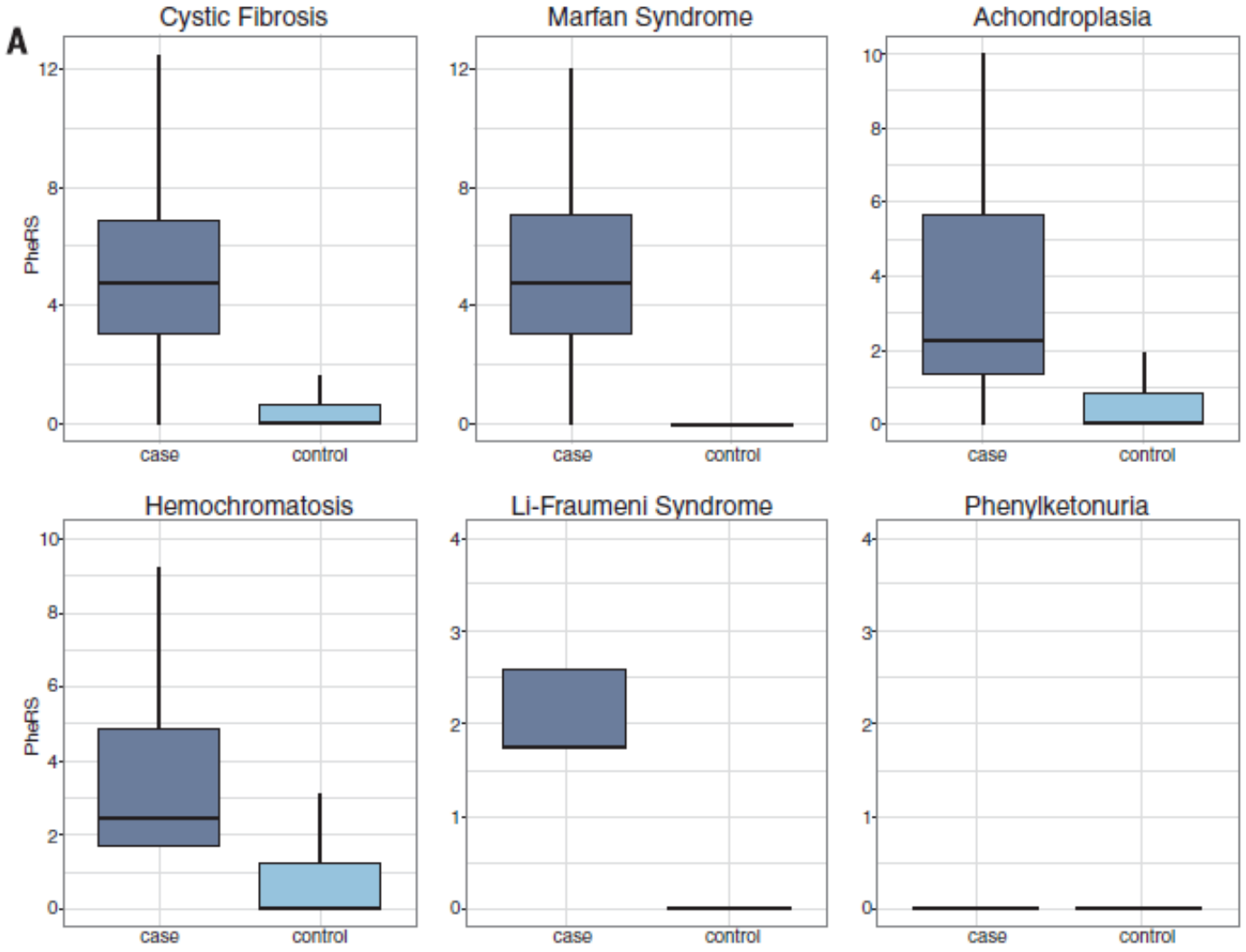


Courtesy L Bastarache, Vanderbilt U

- Leverage phenotypic patterns of Mendelian diseases
- Map clinical manifestations of Mendelian disease to phenotypes extracted from EHR
- Compute “phenotype risk score” (PheRS) expressing overlap of patient’s findings with Mendelian disease
- Weighted aggregation of genetically related phenotypes

CYSTIC FIBROSIS; CF

PheRS of Clinically Recognized Cases and Controls for Six Mendelian Diseases



Expansions of PheRS

- Why couldn't approach be expanded to complex diseases, particularly those difficult to diagnose?
  - Major psychoses
  - Rheumatologic disorders
  - Cognitive decline/dementia
  - Chronic kidney disease
- Other important phenotypes?
  - What diseases are most likely to be undiagnosed?
  - What diseases are most important to diagnose?

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NHGRI Extramural Training Mission

Prepare a diverse and talented genomics workforce that is operating at the forefront of genomics to accelerate scientific and medical breakthroughs to improve human health.

Career transitions

Loan repayment

Fellowships

Career development

Educational activities

Institutional training

Courses/curricula

Mentored research experiences

Administrative supplements: diversity, re-entry, re-integration, continuity, retention

Courtesy H Colley, NHGRI

NHGRI Scientific Mission

NHGRI supports resources, approaches, and technologies that accelerate genomic research focused on:

- structure and biology of genomes
- genomics of disease
- implementation and effectiveness of genomic medicine
- computational genomics and data science
- impact of genomic technology, advances, and implementation on health disparities and health equity
- ethical, legal, and social issues related to genomic advances

NHGRI supports studies that provide **generalizable** methods and knowledge.

Courtesy H Colley, NHGRI

https://grants.nih.gov/grants/guide/parent_announcements.htm

Institutional Training Award: T32

- Supports predoctoral and postdoctoral research training
- Combination of didactic and hands-on research training
- Mentoring and career development components
- NHGRI supports T32 programs in these areas:
  - Implementation and effectiveness of genomic medicine
  - Structure and biology of genomes
  - Computational genomics and data science
  - Health disparities in genomics
  - Ethical, legal, and social issues related to genomic advances

Full list of NHGRI-supported T32 training programs

Courtesy H Colley, NHGRI

Individual Awards: Training and Career Development

Graduate / predoctoral

- Fellowships (F30, F31)
- Fellowships, diverse backgrounds (F31-D)
- Predoc to postdoc transition, diverse backgrounds (F99/K00)

Postdoctoral

- Fellowships (F32)
- Postdoc to faculty (K99/R00)
- Postdoc to faculty, diverse backgrounds (MOSAIC K99/F00)

Early and mid-stage investigator

- Workforce diversity (R01)
- Mentored research scientist (K01)
- Loan repayment program (LRP)

Specific professional focus

- Clinical scientist (K08)
- Quantitative Scientist (K25)

Diversity, re-entry, and re-integration administrative supplements

Courtesy H Colley, NHGRI

NHGRI Training and Career Development for MDs

- **Individual Fellowship for Students at Institutions Without NIH-Funded Institutional Predoctoral Dual-Degree Training Programs (Parent F30)**
  - Target audience: MD/PhD students
- **Mentored Clinical Scientist Research Career Development Award (Parent K08)**
  - Target audience: MDs and other clinical doctorate degree holders
- **Short Term Mentored Research Career Enhancement Award to Promote Diversity (K18)**
  - Target Audience: faculty members from diverse backgrounds

Training/career development for practitioners

- **Genomic Curriculum Development for Medical Students (R25)**
  - Target Audience: Medical Students
- **NHGRI Short Courses for Genomics-Related Research Education (R25)**
  - Target Audience: Scientists, including clinicians

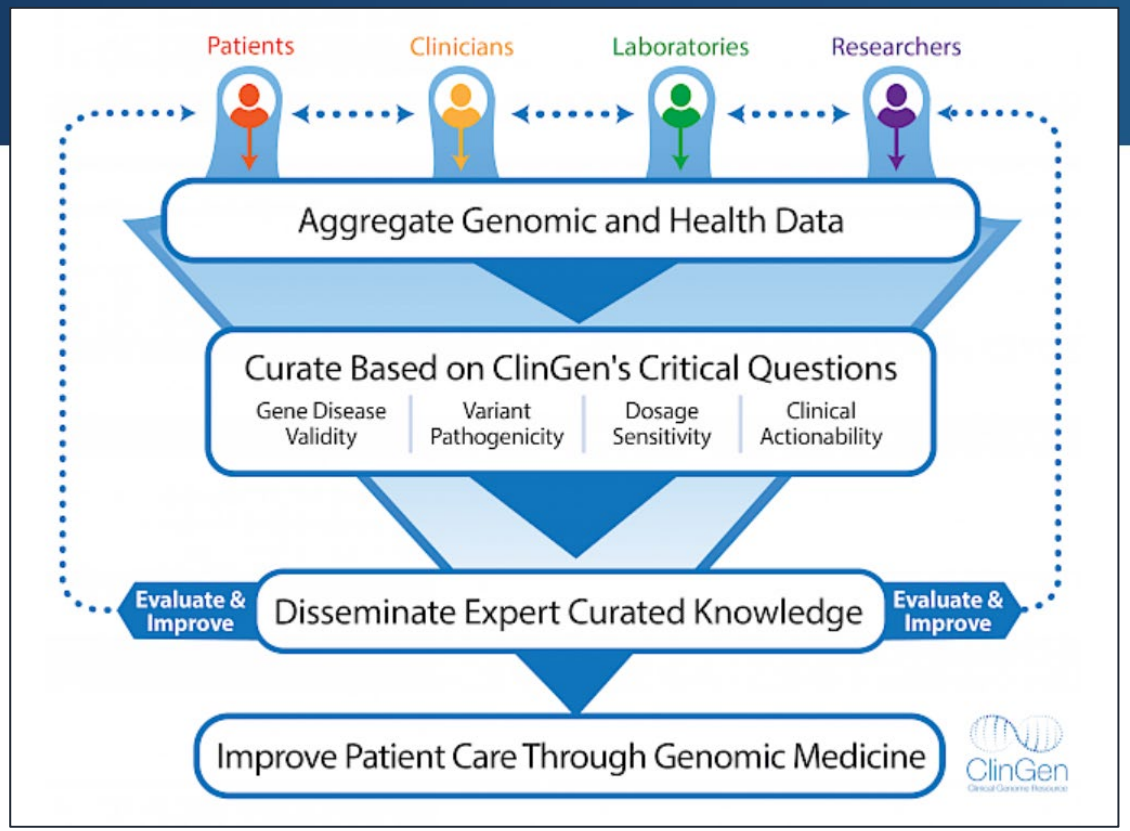
Curriculum development

Courtesy H Colley, NHGRI

ClinGen - Clinical Genome Resource

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

ClinGen
Clinical Genome Resource



The ClinGen Network

ClinGen Working Group and Expert Panels from 2,421 investigators across 65 countries

Updated October 2023 by Zo Bly

119 ClinGen Variant and Gene Curation Expert Panels

- *ABCA4*
- *ACADVL*
- Alport Syndrome
- Aminoacidopathy
- Amyotrophic Lateral Sclerosis Disorders
- Antibody Deficiencies
- Arrhythmogenic RV Cardiomyopathy
- *BCR:ABL1*-like B-lymphoblastic Leukemia/Lymphoma
- Brain Malformations
- Breast/Ovarian Cancer
- Brugada Syndrome
- Cardiomyopathy
- Catecholaminergic Polymorphic VT
- *CDH1*
- Cerebral Creatine Deficiency
- Cerebral Palsy
- Charcot-M
- Coagulati
- Colon Can
- Compleme
- Congenital
- Congenital
- Congenital
- Congenital Myopathies
- Craniofacial Malformations
- Desmosomal Cardiomyopathy
- DICER1 and miRNA-Processing Gene
- Dilated Cardiomyopathy
- Dopa Decarboxylase
- ENIGMA *BRCA1* and *BRCA2*
- Epilepsy
- Epilepsy Sodium Channel
- Familial Hypercholesterolemia
- Fatty Acid Oxidation Disorders
- *FBN1*
- Fibroblast Growth Factor Receptor Mutations
- *FLT3* (Fms Related Receptor Tyrosine Kinase 3)
- Galactosemia
- General
- General Inborn Errors of Metabolism
- Glaucoma and Neuro-Ophthalmology
- Glaucoma
- Glomerulopathy
- Glucose-6-phosphate dehydrogenase
- GRIN Disorders
- Hearing Loss
- Hemoglobinopathy
- Aortic Aneurysm and Dissection
- Histone H3 Somatic Cancer
- Hypertrophic Cardiomyopathy
- InSiGHT Hereditary Colorectal Cancer/Polyposis
- Intellectual Disability and Autism
- Interstitial Lung Disease
- *KCNQ* Channel Brain Disorders
- Kidney Cystic and Ciliopathy Disorders
- Leber Congenital Amaurosis/early onset Retinal Dystrophy
- Leukodystrophy and Leukoencephalopathy
- Limb Girdle Muscular Dystrophy
- Long QT Syndrome
- Lysosomal Diseases
- Malignant Hyperthermia Susceptibility
- *MAPK/ERK* Pathway Somatic Cancer
- Nuclear and Mitochondrial Mitochondrial Diseases
- Monogenic Autoinflammatory Diseases
- Monogenic Diabetes
- Monogenic Systemic and Incomplete Lupus Erythematosus
- Motile Ciliopathy
- Muscular Dystrophies and Myopathies
- Myeloid Malignancy
- Neurofibromatoses and Schwannomatosis
- NTRK Fusions Somatic Cance
- Optic Ne
- Immune Regulatory Disorders
- *PTEN*
- Pulmonary Hypertension
- RASopathy
- Retina
- Rett and Angelman-like Disorders
- SCID-CID
- Severe Combined Immunodeficiency Disease
- Short QT Syndrome
- Skeletal Disorders
- Syndromic Disorders
- Thrombosis
- *TP53*
- Tubulopathy
- Urea Cycle Disorders
- *VHL*
- von Willebrand Disease
- X-linked Inherited Retinal Disease

Volunteer to Curate

Please take a brief survey to tell us more about your interests and desired level of involvement so we can pair you with an appropriate curation activity and/or Expert Panel.

Future Directions in Genomic Medicine

- NHGRI's approaches to identifying future directions
- Implementation research (UDN/NICU/PGx)
- Genomic Learning Health Systems
- eConsults
- Population screening
- Hidden Mendelians – Phenotype Risk Scores (PheRS)
- Training opportunities and priorities
- Not covered: Data sharing, genomic influences on lab values, AI methods, multi-omic technologies
- Already in care: Non-invasive prenatal testing, liquid biopsy, somatic sequencing for cancer therapies

Many Thanks…

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Genomic Medicine Program Investigators and Participants