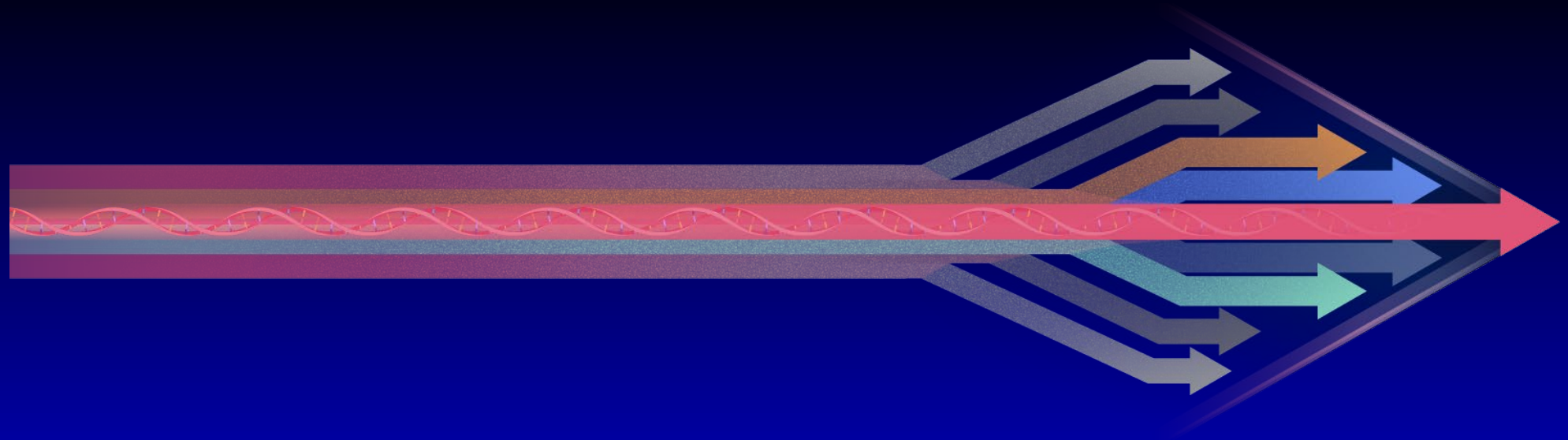


The Forefront of Genomics®

Genomic Medicine Working Group Update

Teri Manolio, M.D., Ph.D.

National Advisory Council for Human Genome Research
February 13, 2023

National Human Genome Research Institute

The Forefront of Genomics®

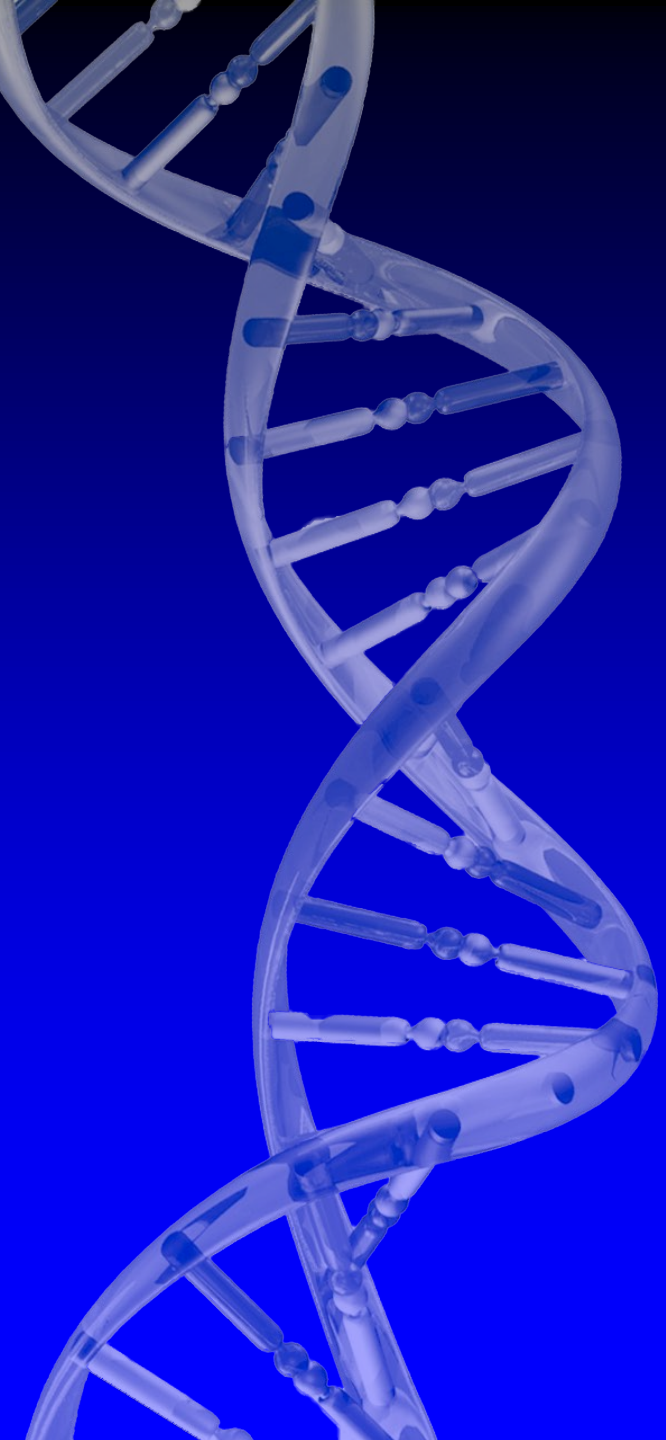
NACHGR Genomic Medicine Working Group Members

Carol Bult	Jackson Labs
Rex Chisholm	Northwestern
Pat Deverka	Veranex
Geoff Ginsburg	All of Us Res Prog
Gail Jarvik	U Washington
George Mensah	NHLBI
Mary Relling	St. Jude
Dan Roden	Vanderbilt
Marc Williams	Geisinger

NHGRI

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

Accomplishments in Genomic Medicine

Search for specific publications by title, author, category and/or by date range. For an explanation about the categories, see the **list of categories and their definitions**.

Search Title or Author | Category: - Any - | From: mm/dd/yyyy | To: mm/dd/yyyy | Search

2023

January 04, 2023 - **Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG)**

First Author: Dungan JS

Category: Educational Resource

Accomplishments in Genomic Medicine: Categories and Definitions

The criteria for **notable accomplishments**:

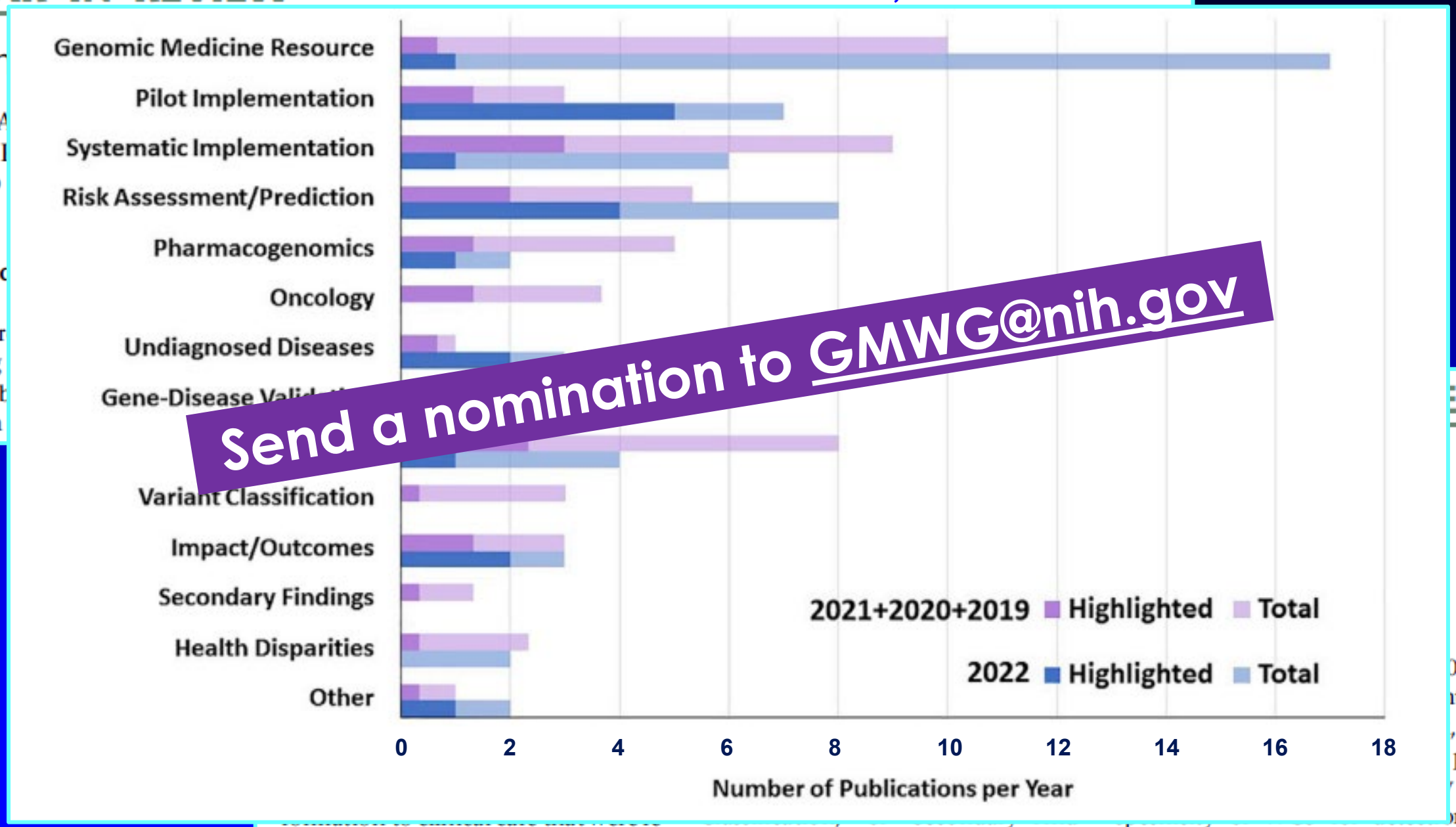
- Involve use of patients' genomic variant information in clinical decision making.
- Demonstrate impact of direct clinical implementation on health outcomes or behaviors.
- Demonstrate the potential for clinical implementation.
- Are likely to be generalizable beyond original setting.
- Are likely to have implications for healthcare systems or practice guidelines.
- Are important considerations for diversity and health equity.
- Are sufficiently large and rigorous to overcome sampling error and other bias.
- Are broadly representative of the field beyond NHGRI-sponsored or US-funded programs.

Categories:

- Genomic Medicine Resource
- Pilot Implementation
- Systematic Implementation
- Risk Assessment/Prediction
- Pharmacogenomics
- Oncology
- Undiagnosed Diseases
- Gene-Disease Validation
- Sequencing
- Variant Classification
- Impact/Outcomes
- Secondary Findings
- Health Disparities
- Other

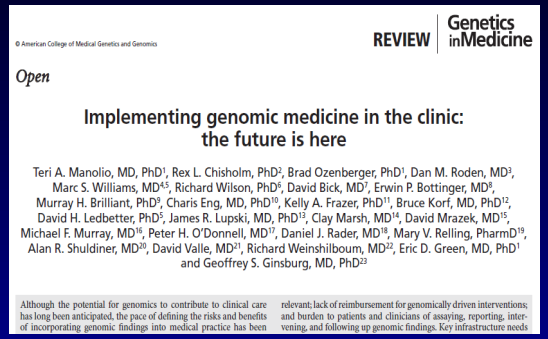
YEAR IN REVIEW

Am J Hum Genet 2019;105:1072-75.



Send a nomination to GMWG@nih.gov

Genomic Medicine Colloquium, June 2011

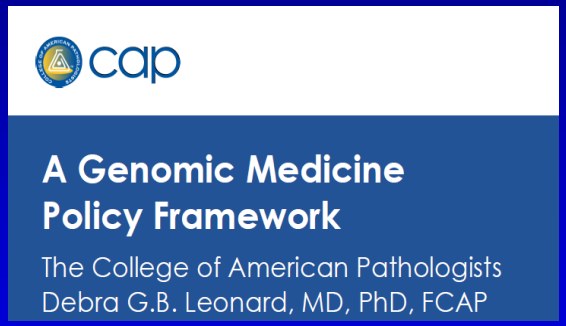


GM II: Forming Collaborations, Dec 2011

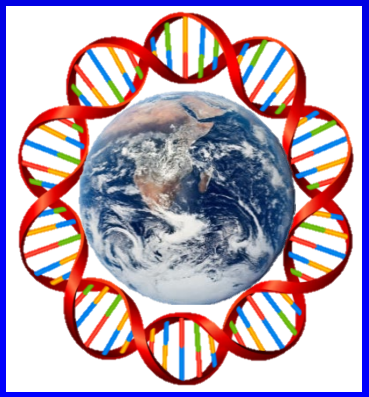
GM III: Stakeholders, May 2012

GM IV: Physician Education, Jan 2013

GM V: Federal Strategies, May 2013



GM VI: Global Leaders, Jan 2014

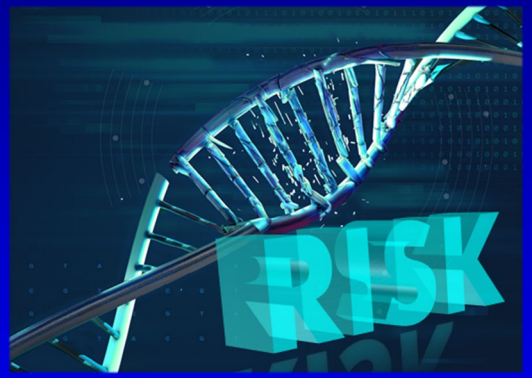


GM VII: Genomic CDS, Oct 2014

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

GM XI: Clinical Implementation, Sept 2018

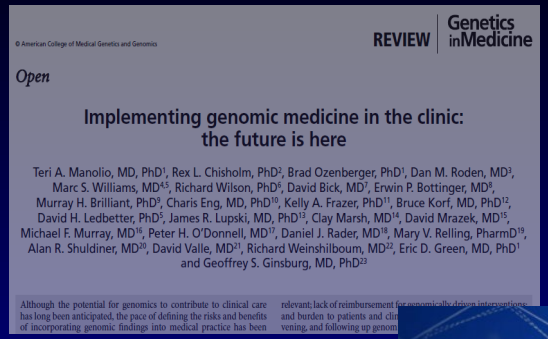
GM XII: Genomics and Risk Prediction, May 2019



GM X: PGx Implementation, May 2017

GM IX: Bedside Back to Bench, April 2016

Genomic Medicine Colloquium, June 2011



GM II: Forming Collaborations, Dec 2011

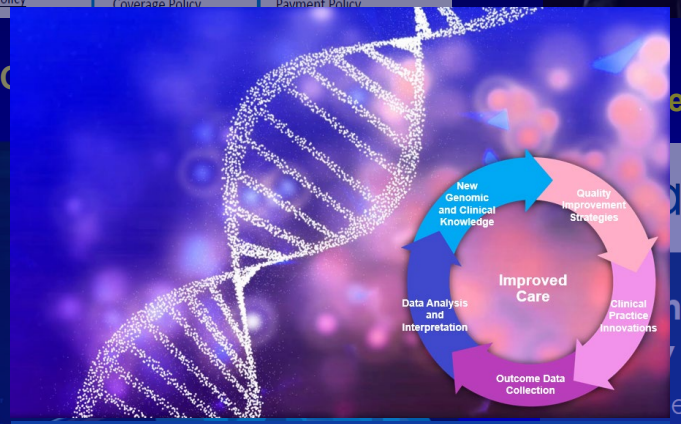
GM III: Stakeholders, May 2012

GM IV: Physician Education, Jan 2013

GM X: PGx Implementa… May 2017

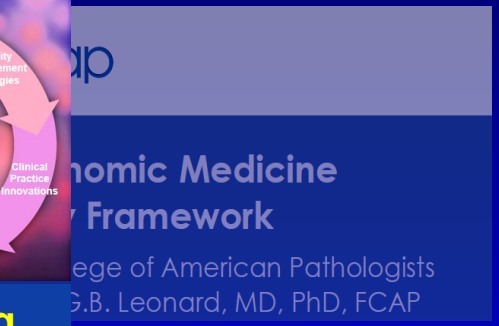
GM XIII: Clinical Informatics Research Agenda, Feb 2021

…ation,



GM XIV: Genomic Learning Healthcare Systems, Aug 2022

…ederal Strategies, May 2013



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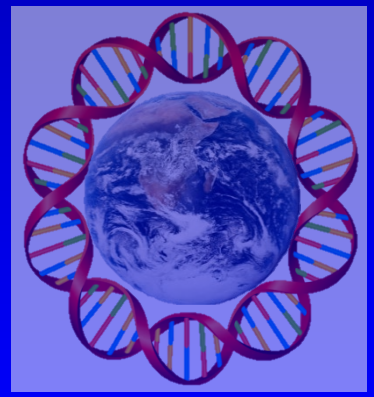

Products of GM Meetings

GenCC
The Gene Curation Coalition
A global effort to harmonize gene-level resources.
Home | GenCC Database | About | Members | News & Articles

PA-18-867
Variant, Function, Disease

NOT-HG-20-020
Genomic Medicine Modules

ISCC

Employers Health and Wellness

NOT-HG-22-011
Patient-Centered Informatics

STAY TUNED

ClinGen
Clinical Genome Resource

Clin Action

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

emerge network
Genomic Risk Assessment

PRIMED consortium

eMERGE PGx

IGNITE
Implementing GeNomics In pracTicE

G2MC

IHCC

Genomic Medicine Meetings

NIH National Human Genome Research Institute

Begin your search here

About Genom

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NIH National Human Genome Research Institute

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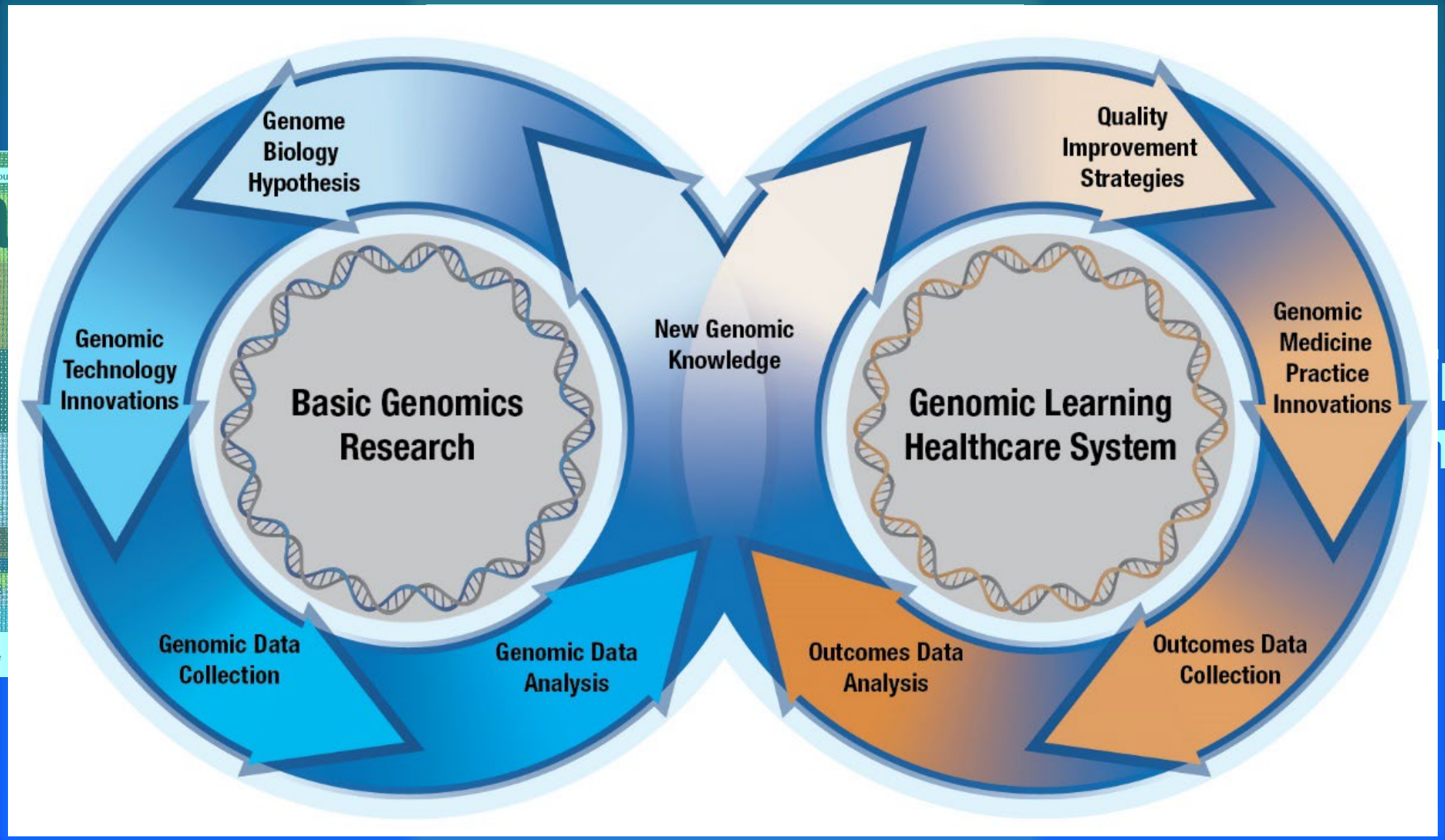
Genomic M

Genomic Medicine XIV: Genomic Learning Healthcare Systems

Summaries

- Executive Summary
- Meeting Summary

2020 NHGRI Strategic Vision



Green E. *et al*., Strategic research priorities and opportunities for improving human health at The Forefront of Genomics. *Nature*, 2020; 586:683-92.

Aug 31 – Sep 1, 2022, Virtual

Genomic Medicine XIV: Genomic Learning Healthcare Systems

Co-Chairs: Pat Deverka, Renee Rider

Objectives:

- Explore real-world examples of how gLHS apply cycles of genomic medicine implementation, evaluation, adjustment, and updated implementation practices across delivery systems.
- Examine barriers and identify potential solutions, focusing on lessons learned from effective gLHS and their potential transportability to other settings.
- Determine ways to develop and share solutions and form collaborations to facilitate research on implementing gLHS.

Genomic Medicine XIV: Genomic Learning Healthcare Systems

Key recommendations:

- Build on and extend interoperability of methods for integrating genomic data into care and exchanging with other systems.
- Create a national LHS network to gather data from collaborating gLHS on genomic medicine practice, quality improvement, and benchmarking.
- Create a consult service or expert panel to help educate clinicians about genetic test orders, interpretation, and determining next steps.
- Develop “learning community of practice” listserv to provide information and updates, potentially supplemented with a panel of experts.
- Promote equity of implementation in low-resourced and underserved settings.

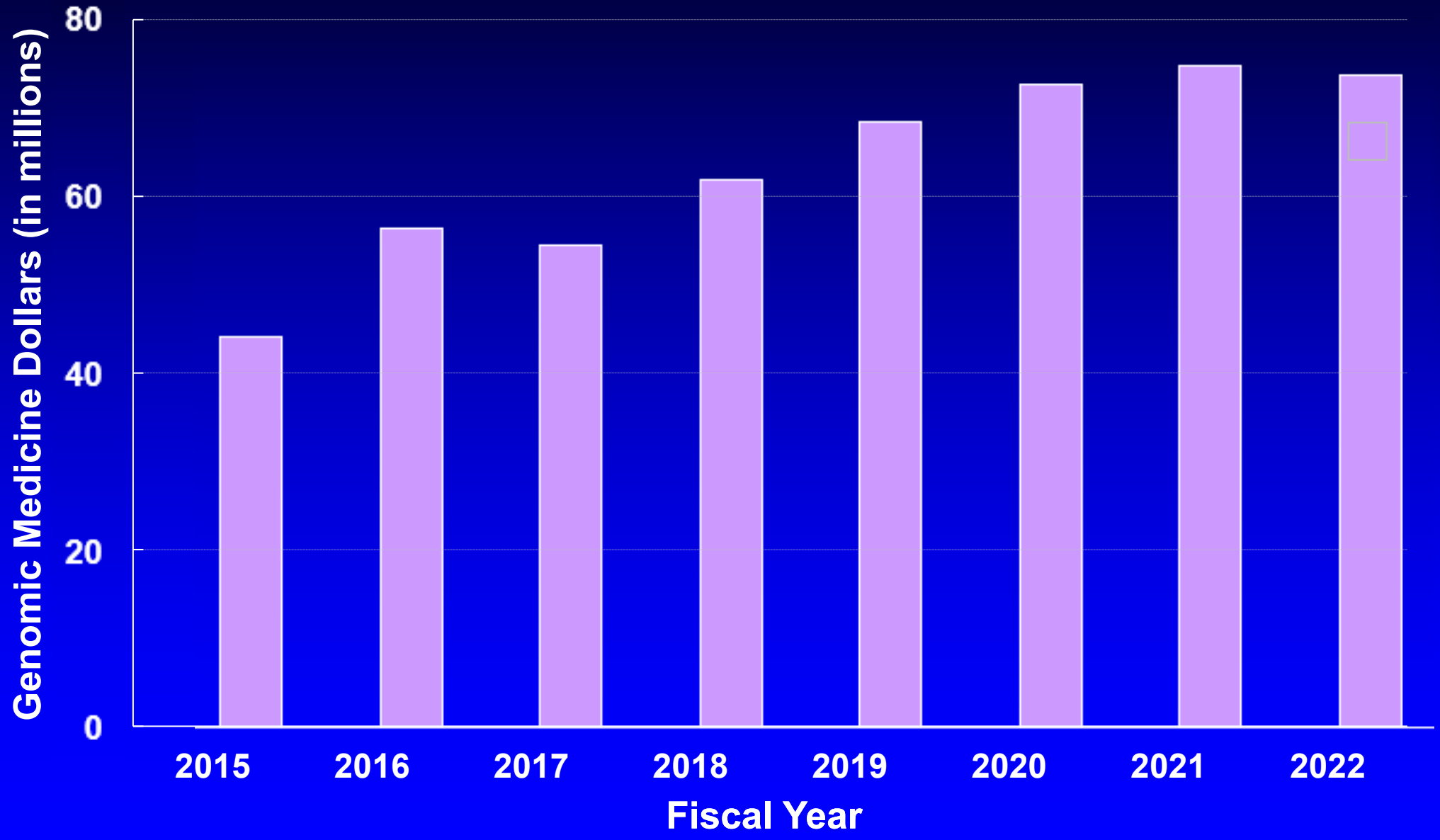
NHGRI's Genomic Medicine Program and FY22 Funds, 9/30/2022

Program	Goal	$M	Years
UDN[1]	Diagnose rare and new diseases by expanding NIH's Undiagnosed Diseases Program	16.4	FY13-22
CSER[2]	Generate evidence of clinical utility of sequencing in diverse clinical settings	0	FY12-21
eMERGE	Develop, implement, and disseminate multi-ethnic genomic risk assessment and management tools for clinical use	19.6	FY07-24
IGNITE	Conduct pragmatic clinical trials of genomic interventions (*APOL1* testing and PGx for pain and depression treatment)	9.9	FY13-24
ClinGen[2]	Develop and disseminate consensus information on genes and variants relevant to clinical care	13.7	FY13-24
ClinGen[3] Curation	Establish expert panels for genes and variants relevant to participating NIH Institutes and Centers	5.6	FY20-
Investigator -Initiated	Advancing genomic medicine research, genetic counseling processes, dissemination and implementation	25.6	FY15-
Training	Institutional training grants, fellowships, career development	5.2	FY16-

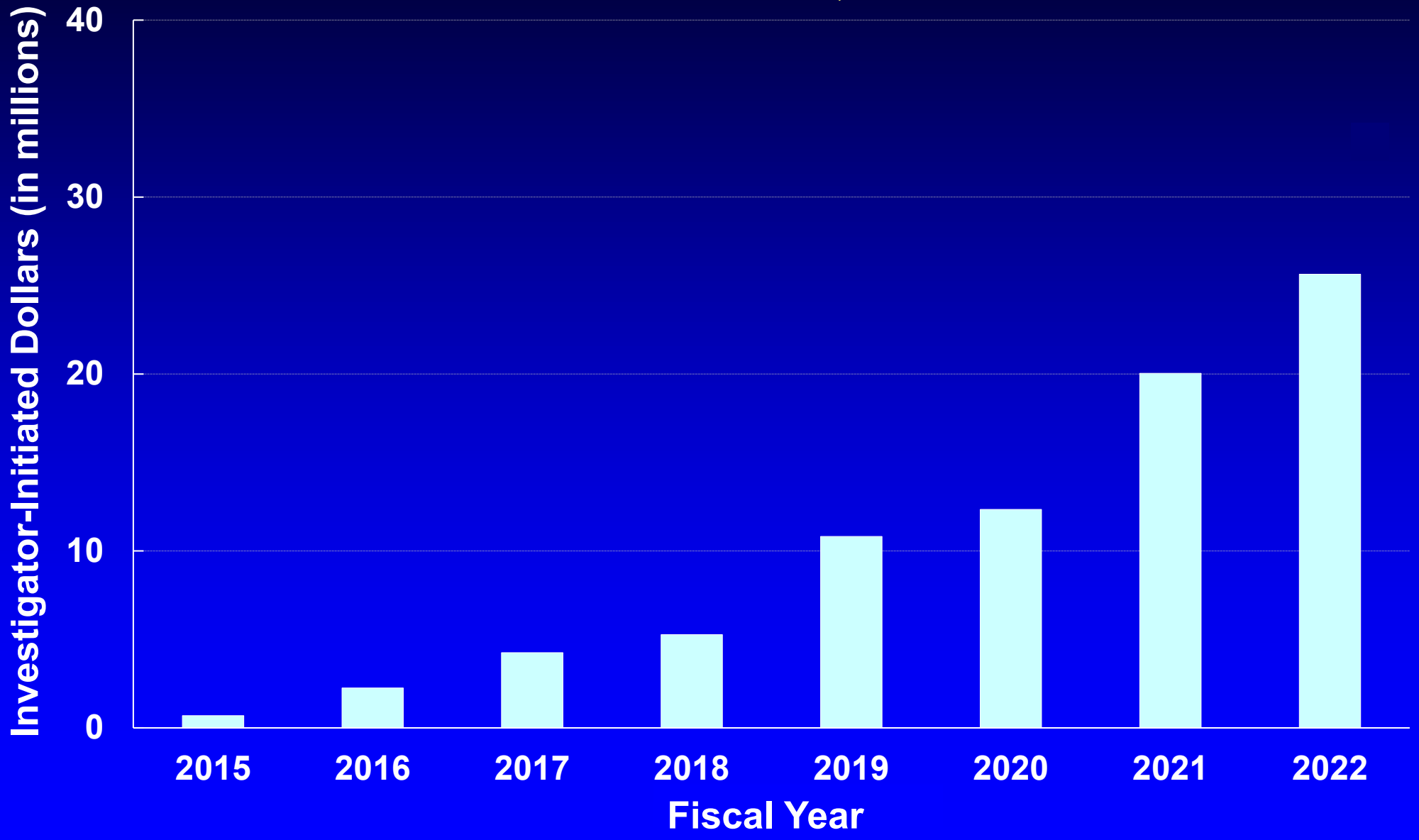
FY22 total genomic medicine funding $96M; $74M NHGRI (35% investigator-initiated)
FY22 total extramural budget $441M (~18% genomic medicine)

[1]NIH Common Fund; [2]Co-Funded by NCI. [3]Funded by 5 partner ICs .

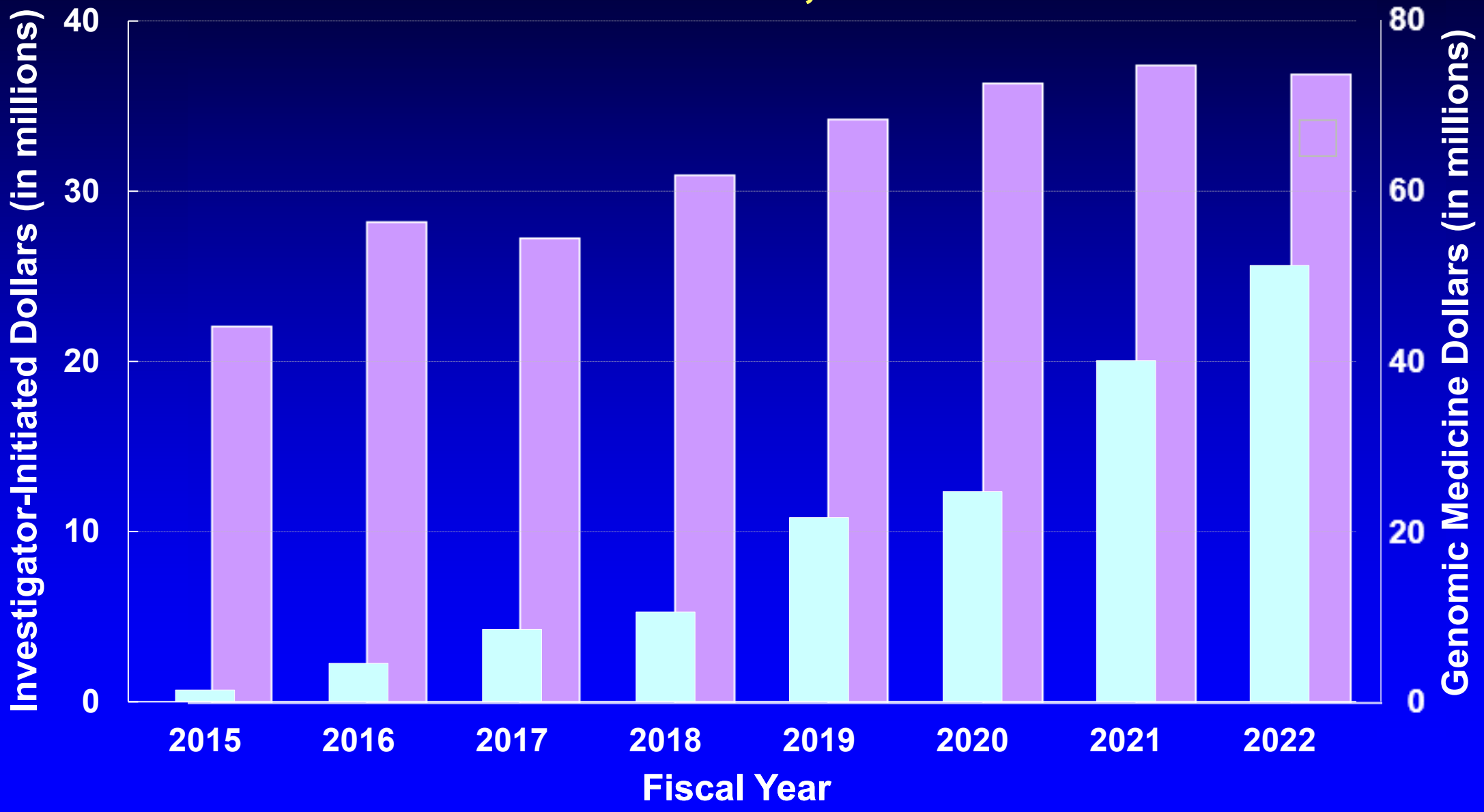
NHGRI Funding for Genomic Medicine Research, 2015-2022



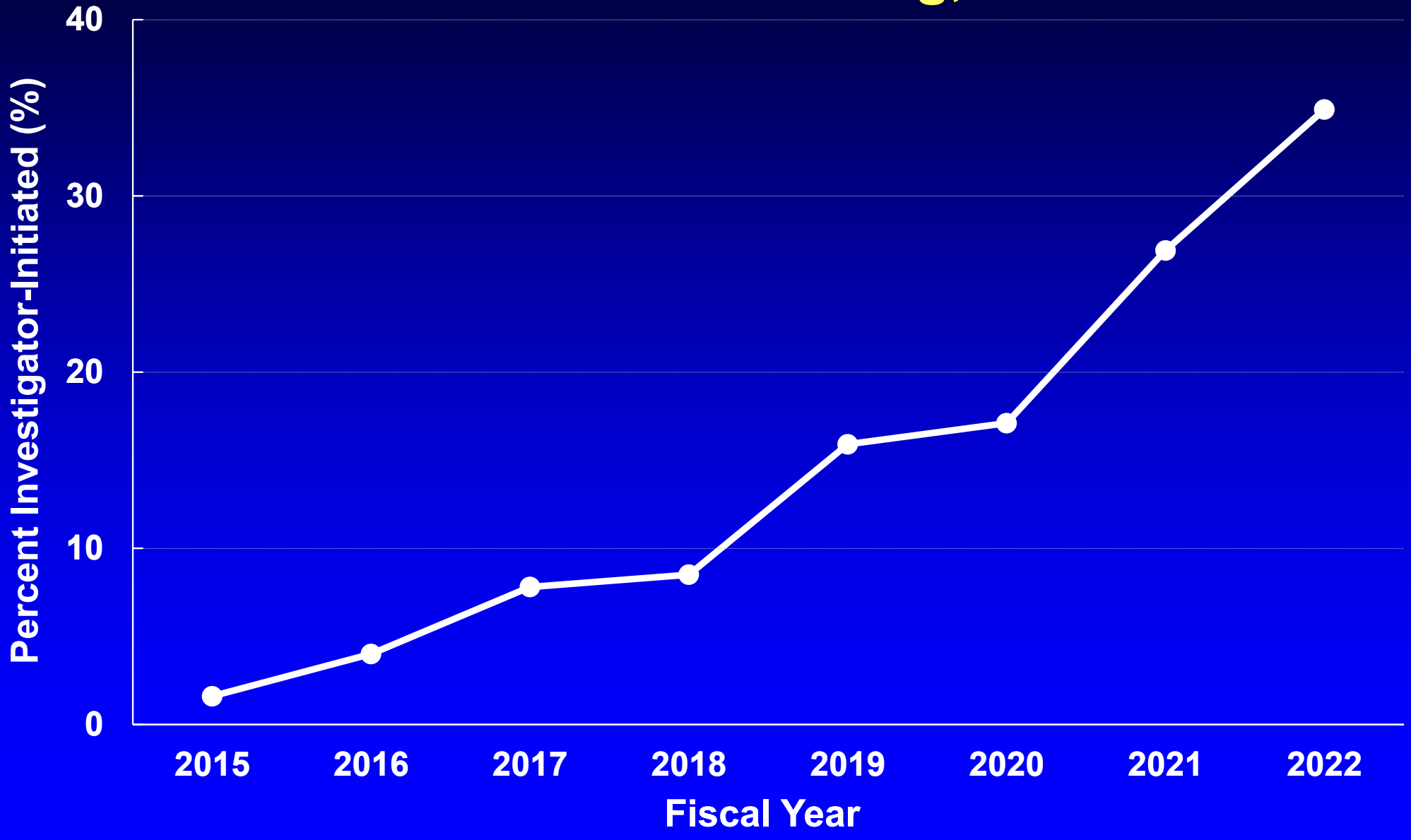
NHGRI Funding for Investigator-Initiated Genomic Medicine Research, 2015-2022



NHGRI Funding for Investigator-Initiated Genomic Medicine Research, 2015-2022



Investigator-Initiated Funding as Percent of Genomic Medicine Research Funding, 2015-2022

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