National Advisory Council for Human Genome Research
February 13-14, 2023
Concept Clearance for RFA

Advancing Genomic Medicine Research (R01, R21)

Purpose:
The renewal of these RFAs will solicit proposals that stimulate innovation and advance understanding of when, where and how best to implement the use and sharing of genomic information and technologies in clinical care in all persons irrespective of ancestral origins or sociodemographic status. The funding announcements will encourage scientific research studies focused on genomic medicine, defined as using genomic information about an individual as part of their clinical care (e.g., for screening, diagnostic, or therapeutic decision-making) and the health outcomes and policy implications of that clinical use. Projects will be broadly applicable to genomic medicine as a field; applications focusing on a particular disease or organ system should make the case for generalizability to multiple diseases and organ systems. To promote progress in the field and encourage broad adoption of successful approaches, awardees will be expected to budget for and participate in an annual meeting with other awardees and NHGRI staff. A pre-application webinar will be held to promote interest and answer applicant questions.

Background:
The past decade has seen a growth in the implementation of genomics in clinical practice. NHGRI has primarily funded genomic medicine research through multi-disciplinary consortia, which provide rich opportunities for collaboration or ancillary projects and have produced valuable data resources and tools for independent genomic medicine research (For more information about these programs, see https://www.genome.gov/about-nhgri/Division-of-Genomic-Medicine#two). As the field grows, opportunities for focused research projects outside large-scale coordinated consortium approaches are growing as well.

This funding opportunity is a renewal of HG-20-036 and HG-20-037, and builds upon the Investigator-Initiated Genomic Medicine Research R01 and R21 PARs. There are no significant changes proposed in this renewal compared to the previous RFAs, although funding decisions in this and other NHGRI programs will continue to emphasize programmatic balance across previously funded awards and emerging scientific priorities. Appendix I lists awards made under the previous RFAs in this program in FY21 and FY22.

Proposed Scope and Objectives:
This concept centers on addressing research gaps related to the use of genomic information to advance the application of genomics in clinical care.

Investigators new to the field of genomic medicine will be encouraged to apply. Genomic medicine research is a multidisciplinary field, and research teams may include experts from multiple disciplines, including but not limited to the fields of clinical genetics including genetic counseling, nursing, genetic epidemiology, biostatistics, data science, public health, implementation science, health outcomes research, health economics, health equity and disparities, health policy, and molecular genetics. Studies addressing or incorporating health disparities are encouraged, defined as health differences that adversely affects disadvantage populations (https://www.nimhd.nih.gov/about/strategic-plan/nih-strategic-plan-definitions-and-parameters.html). For NIH, populations that experience health disparities may include racial and ethnic minority groups, people with lower socioeconomic status, underserved rural communities, and sexual and gender minority groups. Studies that take place outside

academic research settings or can demonstrate the ability for findings to be transferrable to other settings are also encouraged. Where applicable, investigators are strongly encouraged to consider various models for community engagement in research and plan and budget for the implementation of meaningful approaches across various stages of proposed research projects (e.g., generation of research questions and hypotheses, interpretation and translation of findings, dissemination of findings).

In the context of their relevance to genomic medicine, the following are some examples of the areas of research studies that would be appropriate for these FOAs, grouped by category:

Implementing genomic medicine

Implementation research projects would elucidate whether use of genomic information about an individual improves clinical care and/or health outcomes, or how genomic medicine should be implemented.

Facilitating analysis of clinical genomic data

The pace and volume of genomic data being generated presents challenges and opportunities for methods and tools that facilitate clinical analysis.

Improving clinical access and sharing of genomic data

Relationship to Ongoing Activities:

This concept is intended to nurture and expand genomic medicine research by enhancing interactions among grantees and promoting sharing of successful approaches and resulting data. Related applications might also be received through the NIH Parent R01 and R21 announcements, as well as the Dissemination and Implementation PARs (PAR-22-105, PAR-22-106 and PAR-22-109), the Ethical, Legal, and Social Implications (ELSI) of Genomics Research PARs (PA-20-254, PAR-20-255 and PAR-20-257), and Investigator-Initiated Research in Computational Genomics and Data Science PARs (PAR-21-254 and PAR-21-255). Although these FOAs might receive some applications with relevance to genomic medicine research, none specifically calls for genomic medicine research projects. Genomic medicine projects specifically focusing on the development and implementation of clinical informatics tools to enhance patients' use of genomic information should refer to NOT-HG-22-011 for additional guidance.

Mechanism of Support and Funds Anticipated:

R01 (Research Project) up to $600K DC/year, project period of up to 5 years. Total 4 R01s.

R21 (Exploratory/Developmental Research) up to $250K DC/year, project period up to 3 years. Total 4 R21s.

NOSI for Small Business R43/R44 (PAR-22-176 and PAR-22-177) following standard receipt dates.

Total cost of program $5.2/year ramping to $23.1M/yr. Total first five years = $74M.

One receipt date per year; July 2023, July 2024, July 2025.

ICs with interest in funding genomic medicine research would be contacted for the possibility of signing-on.

Appendix 1: Awards under previous rounds of FOA (showing only FY21 and FY22 award amounts)

RFA HG-020-036, HG-020-037

Grant	Activity code	Contact PI	Title	2021	2022
HG011792	R01	Phillips, Kathryn	Building the Evidence Base for Appropriate and Efficient Implementation of Emerging Genomic Tests for Disease Management and Screening	$953,132	$937,434
HG011794	R01	Orlando, Lori	Deploying a genomic-medicine risk assessment model for diverse primary care populations and settings	$759,738	$721,508
HG011795	R01	Bellen, Hugo	Genomic medicine and gene function implementation for an underserved population	$962,213	$962,213
HG011798	R01	Agrawal, Pankaj	VIGOR: Virtual Genome Center for Infant Health	$1,097,885	$1,073,077
HG011799	R01	Williams, Marc	Real-time genetic diagnosis at the point of care	$975,703	$917,779
HG011800	R01	Duarte, Julio	Preemptive pharmacogenetic testing in medically underserved populations	$749,991	$822,805
HG012262	R01	Peterson, Joseph	Rational Integration of Polygenic Risk Scores (RIPS)		$999,221
HG012271	R01	Berg, Jonathan	Age-based genomic screening in newborns, infants, and children: a novel paradigm in public health genomics		$933,000
HG012286	R01	Marth, Gabor	Calypso: a web software system supporting team-based, longitudinal genomic diagnostic care		$914,518
HG012655	R01	Liu, Cong	RESCUE: Rare Disease Detection and Escalation Support via a Learning Health System		$1,019,964
HG012657	R01	Bastarache, Lisa	Translating the Clinical Knowledge of Mendelian Diseases to Real-world EHR Data to Improve Identification of Undiagnosed Patients		$1,027,598
HG012670	R01	Nathanson, Katherine	Using Behavioral Economics and Implementation Science to Advance the Use of Genomic Medicine Utilizing an EHR Infrastructure across a Diverse Health System		$924,042
HG011802	R21	Brownstein, Catherine	Navigating Online Patient Experiences of Genomic Medicine: Identifying and Overcoming Obstacles for African American Communities	$318,487	$292,975
HG011803	R21	Boerwinkle, Eric	ImplementatioN Sclence for Genomic Health Translation (INSIGHT)		$443,629
HG011805	R21	Gallagher, Renata	Toward DNA Sequencing as a Primary Newborn Screen for Treatable Disorders not Amenable to Current Screening	$323,000	$323,000
			Total	**$6,140,149**	**$12,312,763**