National Advisory Council for Human Genome Research
September 18, 2023
Concept Clearance for RFA

Research on the Impact of and Methods for Implementing Regional Genomic Medicine eConsult Services

Purpose:
NHGRI proposes an RFA to conduct research on the impact of and methods for implementing regional clinician-to-clinician genomic medicine eConsult services. Specifically, 2-3 sites will be funded to research how to best design and implement regional genomic eConsult services, as well as how to provide outreach to potential users, including those at underserved settings. At each stage, they will assess the impact on key stakeholders and create tools that could be disseminated to others starting similar eConsult services.

Background:
Electronic consults (eConsults) have been defined as "electronic messages sent between clinicians about general or patient-specific questions" (American Association of Medical Colleges 2016). These services provide clinician-to-clinician support and do not directly involve the patient. NHGRI is defining "regional" eConsult services as those providing services to multiple unaffiliated healthcare organizations.

Non-regional eConsult services (*i.e.*, those within a single organization or system) have been established by many institutions in non-genomic specialties. They've been shown to reduce specialty clinic appointment wait times (Chen 2103), healthcare costs and utilization (Newman 2019), and patient travel costs and burden (Kirsh 2015), and to improve primary/specialty communication (Keely 2013). Previous research has demonstrated the feasibility of eConsult services (Kirsh 2015) and identified factors promoting successful implementation including compatibility with existing workflows, clarity of communications, technologic simplicity, and protected time for consultants (Stevenson 2018). In addition to the benefits seen in non-regional services, regional eConsult services have successfully provided specialty care in large, disadvantaged populations with historically poor access to specialty care (Barnett 2017). Regional services can promote equity of access (Liddy 2019).

The use of genomic testing in primary and specialty care remains scant, in part due to lack of understanding and confidence with genomics among non-genetic clinicians (Aday 2019) and lack of access to genetic specialists (Haga 2013). Yet, genomic medicine eConsult services are rare despite the potential support that these services could provide.

The August 2022 *Genomic Medicine XIV Meeting* on Genomic Learning Health Systems highlighted the need for consult services to support clinicians in providing genomic care, especially in lower-resourced settings that do not have genetic experts. The subsequent *NHGRI Genomic Consultation Research Working Group* explored the current status of genomic eConsult services at their institutions and research needed to successfully implement these services elsewhere.

First-year results from one genomic medicine eConsult service demonstrated rapid turnaround times (median 2 days), high rates of actionable recommendations (80%), and 82% follow-through on recommendations (Folkerts 2023). More extensive experience in Ontario showed that genomic eConsults enabled PCPs to provide care without referring the patient in 36% of cases and that 86% of referring PCPs perceived the eConsult as valuable for patient management (Carroll 2022). Over half of these PCPs also agreed that their eConsult question "addressed an important clinical problem that should be incorporated into

upcoming continuing education events." Unfortunately, such services are currently targeted only to the clinicians within these institutions, so clinicians in locations without access to genomic specialists lack access to genomic medicine eConsults as well.

While regional genomic medicine eConsult services have been shown to be effective in Ontario (Carroll 2022), similar services are not available in the U.S. A diagnostics company recently funded eConsult services to Federally Qualified Health Centers in five metropolitan areas but included no genomic medicine. An eConsult network set up for San Francisco's safety net system includes only cancer genetics. Indeed, an attempt to provide genomic medicine eConsults through the New England Regional Genetic Network was stymied by barriers such as inconsistent licensure laws and lack of consultants. Implementation research is needed to determine how to provide genomic eConsults to clinicians in locations without genomic expertise and to determine the impact of providing those services.

Proposed Scope and Objectives:
NHGRI proposes to fund 2-3 sites to conduct research to determine the impact of and methods for implementing regional clinician-to-clinician genomic medicine eConsult services. The overarching research questions would be: 1) what impact do genomic eConsult services have when they are implemented at the regional level; 2) how can regional genomic eConsult services be implemented and sustained; and 3) can tools be created and shared with others who are creating regional eConsult services?

Each site would establish a separate regional eConsult service and test diverse approaches to implementation, potentially addressing different barriers and facilitators unique to their situations. Each would use an implementation science framework, such as the Practical Implementation Sustainability Model (PRISM), RE-AIM (Reach, Effectiveness, Adoption, Implementation, Maintenance), or the Consolidated Framework for Implementation Research (CFIR). Because this proposal focuses on both impact and implementation, investigators may choose an effectiveness-implementation hybrid design (Curran 2012). Common metrics would be proposed and adopted by the sites in collaboration with NHGRI.

This initiative would support research on regional genomic medicine eConsult services that are clinician-to-clinician, providing advice to healthcare professionals and not to patients or the lay public. Each site would create a network of consultants with relevant genomic expertise and could research differing service models of referral and compensation for the consultants. Other expertise could include genetic counseling, implementation science, informatics, insurance and reimbursement, health disparities and access to care, liability and licensure, health economics, communications and marketing, and health education. Each service could create their own eConsult infrastructure or use existing infrastructure on commercial platforms that provide options to receive, triage, manage, and respond to eConsult requests. The sites and NHGRI would share organizational activities such as organizing joint calls and meetings.

To maximize the applicability of this research, sites would provide genomic medicine eConsults for a breadth of medical disciplines. Each site would be expected to cover a population anticipated to generate at least 1000 consults per year. Ideally, each site's population would include a variety of settings, such as academic medical centers, community hospitals, private practices, and safety net services; use multiple Electronic Health Record (EHR) platforms; and serve a diversity of communities including at least two medically underserved populations/areas. In addition to exploring models for receiving and responding to eConsults, each site would examine methods for publicizing the availability of the service to a wide range of potential users, including resource-limited settings. Options for sustainability after NIH support would also be studied.

To assess the impact of the eConsult service, sites would agree on common metrics such as uptake and use of the service, usefulness of recommendations and whether they are followed, specialty clinic appointment wait times, healthcare costs and utilization, patient travel costs and burden, primary/specialty communication, and equity of access. To assess the implementation of the service, sites could study methods for triaging eConsults and urgent requests and for using informatics to increase the efficiency of eConsults services. They could also evaluate factors such as ease and burden of use for both the requestor and consultant, appropriateness and completeness of requests, and timeliness of responses.

A major focus of this initiative would be to study how to create effective outreach and referral networks. Outreach efforts should focus on making sure that clinicians in lower-resourced settings know about the availability of genomic medicine eConsults, their potential benefit, and when and how the eConsult should be ordered. These outreach efforts should continue for the duration of the grant, and be designed in a way that their impact and implementation can be measured. Establishing the referral network should focus on the infrastructure and administrative structure needed for conducting the eConsult service and assessing its impact.

In all phases of the project, sites should consider what tools could be developed for dissemination to other institutions that may implement eConsult services. For example, after researching best methods for outreach, they could produce a guide for conducting outreach with ideal target audiences and sample advertisements or content for continuing education for clinicians. Or, after determining common referral questions, they could produce a set of templated responses.

Consultation with key stakeholders should begin in the first year and continue throughout the program to enhance availability and applicability to underserved settings, optimize efficiency, promote feedback, and evaluate impact.

Sites will be encouraged to establish a leadership team with diverse expertise and an advisory panel of stakeholders that would meet regularly. Collaboration between the awardees and NHGRI would involve regular meetings to establish common metrics, discuss obstacles, brainstorm and share solutions, review stakeholder input, and monitor progress.

Relationship to Ongoing Activities:
This RFA will be NHGRI's and NIH's first initiative focused on genomic medicine eConsult services. The Maternal and Child Health Bureau of HRSA has supported the Regional Genetics Networks to increase access of patients and clinicians to genetic services for several years; for the past three, NHGRI has supplemented these programs to evaluate the implementation and outcomes of its services. Discussions regarding additional collaborations with HRSA are ongoing.

Mechanism of Support:
A single RFA will support 2-3 U01 cooperative agreements to establish regional genomic medicine eConsult services.

Funds Anticipated:
The amount of funding would increase as the volume of eConsults increases (Year 1: $3.0M, Year 2: $3.2M, Years 3 - 5: $3.5M total costs), for a total of $16.7M over 5 years. NHGRI staff will seek collaboration and funding from other NIH Institutes and federal agencies to increase the number of awards, expand the scope of service, and/or speed the expansion and evaluation of the service and its outreach activities.