# The Implementation of Clinical Genomics at CUIMC

## Introduction

This report summarizes the recommendations of the VP&S/CUIMC taskforce on the Implementation of Clinical Genomic Medicine convened by the Dean of the Vagelos School of Medicine on July 21, 2022. Taskforce members represent leaders in clinical genomic medicine and genomic research at CUIMC (See appendix). Dean Armstrong's charge was to develop recommendations focused on Columbia University's approach to integrating genomic medicine into clinical practice. The objective is to prepare for the eventuality that every patient will be sequenced, and data is in charts; to ensure equitable access to genomic medicine for the patients served at CUIMC; to develop genomic medicine infrastructure to support ongoing discovery and the full realization of precision medicine incorporating genomics, epigenomics, environmental exposure, and other data in clinical practice.

## Goals

- Ensure the equitable implementation of clinical genomics for patients at CUIMC-NYP.
- Create integrated clinical genomics systems, structures, platforms, and governance to support patients, clinicians, the lab, and researchers.
- Leverage investments, including partnerships with industry, in clinical genomics to advance genomic research, thereby driving the development of new genomic applications and other precision medicine approaches.
- Train the next generation of genomics professionals (clinicians and scientists).
- Build infrastructure that supports the clinical implementation of future precision medicine discoveries.

The taskforce's recommendations call for a governance and leadership structure under the Precision Medicine Initiative Director's oversight and a newly created Chief Clinical Genomics Officer to coordinate communication and facilitate decision-making across clinical domains, to develop robust, sustainable infrastructure, and to integrate clinical workflows using EPIC/EHR and a genomics data warehouse for clinical care and research discovery. Additionally, the taskforce's recommendations emphasize the importance of education for students, trainees, and clinicians (MD, NP, RN).

## Taskforce process

Beginning July 2022, the taskforce reviewed the current state of genomic medicine in the clinical areas, discussed present obstacles and challenges to the implementation of clinical genomics, reviewed tools to support genomics ordering, reporting, clinical decision support, and education in the clinic; contacted peer institutions, and included invited speakers to present on the clinical genomics lab services (PGL, PGM, NYGC), NYP Precision Medicine/EPIC Genomics current status and plans, and workforce development and education.

The taskforce focused on the steps to integrate genomic medicine into clinical practice and considered the following questions:

- How are patients identified, screened, and referred for testing?
- How are patients educated and counseled about testing?
- How is testing selected/ordered and results returned?
- How are test results incorporated into clinical decision-making?
- How can we strategically position our genetics expertise across the healthcare system?
- How can we improve the knowledge and skills of non-genetics providers?
- How do we create leadership, governance, and structure that integrates with our research infrastructure to ensure the ongoing discovery, translation, and implementation in genomic medicine?
- How can we create a financially sustainable model to support genomic medicine?

## Meeting topics:

I. Introductions, Dean's charge to the taskforce, and discussion
II. Genomics Landscape at CUIMC report back from clinical domains and peer institutions
III. Discussion about the taskforce's focus to develop recommendations for governance and structure, equity/access for our patients, infrastructure to support genomics and other precision medicine approaches
IV. Clinical Sequencing at CUIMC – PGL, PGM, and NYGC laboratories - presentation by Vaidehi Jobanputra, PhD
V. NYP Precision Medicine Updates – EPIC (ordering, results integration, CDS) and Datawarehouse – presentation by Che Martin, PhD
VI. Genomics Workforce Development – presentation by Amanda Bergner, MD, CGC
VII. Discussion of draft recommendations

## The discussion revealed:

- Implementation of genomic medicine is uneven.
- Clinical programs at CUIMC range from prenatal testing to diagnosis of unexplained syndromes.
- Accessibility to data (raw and reports) and analysis tools is uneven.
- Different models (including testing approaches) are being used and may be appropriate for other clinical areas.
- Genetic counselors and genetic specialists are widely endorsed, with many areas having specialized genetic clinics.
- The use of educational approaches outside of genetic counseling is uneven.
- Tests are sent to various outside labs primarily driven by clinician preference.
- Financial barriers are substantial and contribute to the use of outside labs that cover patient costs.
- There is limited integration into the EHR, which inhibits effective CDS as well as discovery research.
- There is a strong interest in linking clinical care to discovery and innovation.

To develop these recommendations, the taskforce members' comments and suggestions, and information from the presentations were organized to address the needs, opportunities, improvements, strengths, and exceptions in the themes of leadership and governance, clinical workflow, laboratory, data storage, and data management, EHR and Clinical Decision Support, workforce development and education, and financial issues.

- **Needs:** What is needed to achieve the widespread adoption of clinical genomic medicine at CUIMC?
- **Opportunities:** What external factors offer opportunities to implement genomic medicine? Peers who are leading in this space to learn/share best practices? Are there unexplored areas that provide new opportunities?
- **Improvements:** How must we adjust to achieve needs and prepare to take advantage of opportunities?
- **Strengths:** What are we doing well currently? How do we measure success?
- **Exceptions:** Of the four above subjects, what is already present? List all factors regardless of their current impact.

## I. Leadership

**Needs:**

- Establish leadership and organizational structure to support the broad implementation of clinical genomic services, to create sustainable infrastructure to support the implementation of future research discoveries into clinical practice.

**Opportunities**:

- Succession of CU PMI leadership

**Improvements:**

- Organizational structure to facilitate collaboration and support the implementation of guideline-based care and development of new best practices in genomic medicine
- Update the vision for CU PMI to align innovation, clinical care, and research

**Strengths:**

- Genomics expertise and leadership in clinical departments
- National/international leaders in genomics at CUIMC

**Exceptions:**

- Well-established, well-organized specialty departmental clinics and multi-specialty genomics clinics
- InterDepartmental Genetic Counseling Program (IDGCP- cross-department)
- CUIMC genetic counseling consortium – cross-departmental

**RECOMMENDATIONS**:

- Identify the next **Columbia University Precision Medicine Initiative** Director and establish a new vision to advance the broad implementation of genomic medicine.
- Harness the **Precision Medicine Initiative** to create a cohesive program for the implementation of genomic medicine, a strong genomics curriculum for trainees and clinicians, and robust infrastructure to support clinical genomics and genetic research. Within the Precision Medicine Initiative, establish a new role to coordinate and oversee the implementation of clinical genomics (**Chief Clinical Genomics Officer**) who will chair a new committee (Clinical Genomics Steering Committee (CGSC)) to coordinate and oversee the implementation of clinical genetics/genomics across clinical services. The CGSC will include representatives from each of the major clinical areas engaged in the implementation of genomic medicine with the goal of establishing best practices, developing shared resources, and overcoming common implementation barriers.

## II. Clinical Workflow

**Needs:**

- Access to effective and efficient support for implementing/optimizing genomic clinical workflow, including:
    - Patient identification
    - Guidance for test and laboratory selection
    - Clinical decision support tools in the EHR
    - Access to genetic counseling and testing services
    - New tools to educate patients on genetics/genomics

**Opportunities**:

- NYP's commitment to leading in precision medicine with:
    - Access to all patients for genomic testing regardless of location
    - Standardized genomics operations with centralized and scalable infrastructure
    - Clinical Decision Support Tools (CDS) with updated knowledge
    - Diverse communities served by our institution
- Chief Medical Information Officer's (ColumbiaDoctors) commitment to serve as a liaison to support the implementation of EPIC genomic module and clinical genomics workflows

**Improvements:**

- Provide guidance, support, consent, and appropriate testing options across all clinical areas – outpatient Columbia Doctors and ACN practices and inpatient clinical services.
- Implement new technologies to increase access to genetic testing and genetic counseling: telemedicine for genetic counseling services and family-based testing services; AI/Chatbot, videos, and content developed (culturally appropriate)
- Inpatient team to facilitate rapid diagnostic testing

**Strengths:**

- Successful examples in some clinical areas – i.e., neurogenetics specialty clinic, "Destination Center" for diagnosing complex and previously undiagnosed conditions – Undiagnosed Disease Network Site.
- Genetic counselors and genetic specialists are widely endorsed, with many areas having specialized genetic clinics.

**Exceptions:**

- InterDepartmental Genetic Counseling Program (IDGCP) provides genetic counseling support in areas previously not staffed and has increased the capacity of GC services in high-volume areas.
- Center for Precision Medicine and Genomics (CPMG), Clinical Genetics in Pediatrics, Reproductive Genetics, and specialty clinics are established.
- Implementation of EPIC and telemedicine increasing access to genetic services
- Ongoing implementation of EPIC genomics module

**RECOMMENDATIONS**:

- Harness the **Precision Medicine Initiative** to create a cohesive program for the implementation of genomic medicine, a strong genomics curriculum for trainees and clinicians, and robust infrastructure to support clinical genomics and genetic research
- Within the Precision Medicine Initiative, establish a new role to coordinate and oversee the implementation of clinical genomics (**Chief Clinical Genomics Officer**) who will chair a new committee (Clinical Genomics Steering Committee) to coordinate and oversee the implementation of clinical genetics/genomics across clinical services.
- Unify the clinical consent of patients to allow for ongoing data storage, reanalysis, recontact, and research.
- Standardize access to genetic testing services across the institution, facilitate ordering and data storage via Epic or another interface to allow data access for reanalysis, recontact, and research.
- Maximize the integration of clinical genomics services with genetic counseling and other educational programs to facilitate the expansion of a well-trained and effective clinical genomics workforce.
- Coordinate development within and across clinical domains.

## III. Laboratory — Genomic Testing

**Needs:**

- Accurate and affordable genetic testing that is widely accessible to patients
- Access to raw data from testing for reanalysis and research
- Pathology/laboratory training opportunities

**Opportunities:**

- Established partnerships with NYP, NYGC, and Industry
- CU labs (PGM and PGL) and the NYGC have capacity and can support additional testing volume.
- CU labs (PGM and PGL) and the NYGC have the infrastructure and clinical pathology/genomics expertise and there are opportunities to combine services to reduce redundancies.

**Improvements**:

- Standardize consent (content and process).
- Standardized test ordering algorithms working with clinical experts to determine the standard of care.
- Create and maintain a list of appropriate internal and external laboratories for testing services in each clinical area.
- Ensure all labs (CU and external) provide high-quality (Rapid and Standard turn-around-times (TAT)) and cost-effective genomic testing.
- Ensure all labs share raw data and that data is stored in the genomics data warehouse for reanalysis and research.
- Build infrastructure to support re-analysis of sequencing results– integrated results, data warehouse.
- Create infrastructure for family testing - cascade testing and rapid CLIA validation of variants detected in research settings.
- Develop financial models that maximize access to testing and support internal laboratories to advance innovation, training, and clinical services.

**Strengths:**

- Two internal clinical CU labs, PGL and PGM, and NYGC have extensive capabilities and underutilized sequencing capacity.
- NYGC and WCW are well-established partners.

**Exceptions:**

- CUIMC lab, PGL, PGM, and the New York Genome Center, offer clinical sequencing – WES and WGS with NYS/CLIA approval.

**RECOMMENDATIONS:**

- Create a **Clinical Genomics Steering Committee** chaired by the Chief Clinical Genomics Officer to oversee and support genomic testing at CU, working collaboratively with WCM and NYP.
    - Establish working groups reporting to the genomic testing committee to identify appropriate genomic testing algorithms and laboratories.
- Create a short-term taskforce to determine the optimal model for supporting sequencing and analysis.
    - Consider organizing and integrating current CU sequencing and analysis resources, including PGL/PGM.
    - Consider coordinating clinical sequencing across CUIMC, WCM, and NYGC to maximize throughput, clinical offerings, and turnaround time and to minimize inefficiency/redundancy.
    - Use one laboratory to coordinate CLIA confirmations of research discoveries.
- Develop a financial strategy to maximize the support for genomic testing at CU labs, including improving reimbursement and exploring potential partnerships.
- Establish connections between clinical laboratories and the research-omics cores to facilitate the translation of research advances to the clinical lab and the patient.

## IV. Data Storage and Management (DSM)

**Needs:**

- Enable all genomic test data, including raw and processed files, to be stored for reanalysis, clinical decision support, and research

**Opportunities:**

- Existing patients with clinical sequence data available
- Systems available with relevant institutional genomics DSM solutions
- NYP PM vision includes NGS Datawarehouse

**Improvements**:

- A central repository of human genetic data: all sequences generated at CUIMC (research and clinical sequencing).
- Clinical information linked to the human genetic data: de-identified clinical information via linkage to EHR and local clinical research databases
- The ability to link/query/access clinical (EHR), genomic sequencing, variants, biorepository, and research data.
- Platform which ensures accessibility of data and certain analytic/filtering tools to diverse stakeholders to minimize the need for gatekeeping (e.g. bioinformatic expertise)

**Strengths:**

- NYP CDW and Columbia University Biobank

**Exceptions:**

- Well-established Tripartite Request Assessment Committee (TRAC) governance process for data access

**RECOMMENDATIONS:**

- Create a genomics data repository to store genomic test results (internal and external labs to support reanalysis, CDS, and seek options also to support research)
- Unify consent to allow for variant storage, linkage to EHR, and ability to re-contact for clinical and research purposes

## V. EHR and Clinical Decision Support

**Needs:**

- EHR with the ability to receive discrete genetic results
- Implement real-time, integrated clinical decision support for genomic medicine
- Access to raw data for clinical analysis and research purposes

**Opportunities:**

- NYP commitment to lead in Precision Medicine
- Leadership of ColumbiaDoctors CMIO
- NYP investment in the EPIC Genomics Module
- CUIMC is an NIH-funded eMERGE site with resources and expertise to guide CDS genomics
- Ongoing implementation of EPIC genomics module to facilitate e-consent, orders, genomic results integration, and clinical decision support (CDS)

**Improvements:**

- NYP-TRAC (Tripartite Request Assessment Committee) process for prioritizing and implementing genomic medicine functions in EPIC
- Updated knowledge bases and CDS workflows
- Completion of EHR genomics infrastructure to support the implementation of genomic clinical decision support
    - Integrate genomic laboratories prioritized by a genomic testing committee, including PGM, GeneDX, Ambry
    - Implement EPIC Genomics enhancements, including algorithms to identify patients eligible for genomic testing and variant updates
- Provide training and tools for clinicians and researchers

**Strengths:**

- Genomic and clinical informatics expertise in DBMI and NIH eMERGE project leadership at CUIMC.
- Existing clinical genomics expertise in cardiology, cancer, neurology, immunology, nephrology, and genetic counseling to guide EHR/Genomics priorities

**Exceptions:**

- Ongoing EPIC Genomics implementations and enhancements led by the NYP team
    - The lab integration process in progress for PGM, GeneDX
    - Pharmacogenomics BPAs implemented (ARUP, Quest)
    - EPIC pedigree module
- NYP governance process for EHR/Genomics requests, including lab integrations and CDS implementations

**RECOMMENDATIONS:**

- The Clinical Genomics Steering Committee will guide EPIC Genomics/PM priorities and inform the established NYP Tri-institute Governance process.
- Priorities will include:
    - Integrate internal and reference labs – orders, results (discrete results for CDS, raw data for data warehouse, reports)
    - Improve access to genetic test results – organization/location in the e-chart for integrated and nonintegrated lab results.
    - Create functionality to link results in pregnancy to fetus to child
    - Prioritize CDS/BPA - develop alerts/notifications of patients to may benefit from genetic testing and/or be at risk of genetic disease
    - Improve existing tools (EPIC family history) or develop/procure new tools to support genomic medicine
    - Develop Epic-embedded CDS pipelines/paths for all major clinical genetic disorders (committee overseen)
- Leverage existing CU research resources to enhance the genomics EHR tools, e.g., eMERGE.

## VI. Workforce development and education

**Needs:**

- Increase the cFTEs of genetic counselors and other genetic expertise across the clinical enterprise
- Expand GC services with GCAs to alleviate GC administrative workload
- Develop a career pathway for genetic counselors (faculty and non-faculty positions)
- Improve the genomics knowledge and skills (and confidence) of non-genetics providers (NGP)

**Opportunities:**

- CU Genetic Counseling Masters training program
- Using GCAs to increase genetic expertise productive and a pipeline for the genetic counseling program
- Faculty appointments for GCs in the training/education program

**Improvements:**

- Utilize genetic counselors for their clinical and genomic training expertise, functioning independently in their scope of practice
- Create scope of practice guides for genetic counselors and genetic counseling assistants and minimize overlap between genetic experts
- Guide departments in hiring practices of genetic counselors and career path development to address recruitment and retention issues
- Develop education and training for NGP
- Develop education and training for medical and nursing students, and especially MDs/NPs etc – required training and link to credentialing

**Strengths:**

- Genetic Counseling Master's program
- ~40FTE of genetic counselors (~21cFTE)

**Exceptions:**

- Genomic and Precision Medicine course offerings are available
    - Precision Medicine Course for medical students
    - Precision Medicine: Biology & Sociology
    - Genetic Counseling curriculum
    - Irving Institute TRANSFORM and PMR

**RECOMMENDATIONS:**

- Create a Genetic Counseling Services Program, a "professional home" for CUIMC genetic counselors, to coordinate and integrate genetic counseling across the clinical enterprise and improve the retention of genetic counselors by creating career paths and advancement opportunities
- Hire genetic counseling assistants to increase genetic counselor productivity, assist genetic experts in administrative tasks, and support specialty clinics.
- Ensure genetic counselors can work independently by advancing NYS licensure efforts, credential GCs and ensure ordering privileges, and creating Scope of Practice for GC and GCAs
- Organize and coordinate genetic and genomics education across the educational and clinical enterprise
    - Establish formal clinical genomics education for NGPs
    - Support and expand current educational commitments.
    - Organize and coordinate genomics/genetics education for medical students, residents, fellows and GC students
        - Comprehensive integration of genetics throughout the medical school curriculum.
    - Organize and coordinate genomics/genetics education, training and credentialing for all clinical practitioners (MD, NP, RN)
    - Community health workers training in genetics

## VII. Financial

**Needs:**

- Best practices to create financially sound genetics and clinical genomics service
- A financial model to ensure the stability of an integrated clinical genomics service

**Opportunities:**

- NYP-CUIMC partnerships
- Partnerships between CU and NYGC – Reproductive Genetics, ALS
- Philanthropy to support
- Industry partnerships
- NYP Global Services Program

**Improvements:**

- Standardize billing practices for genetic counseling and testing
- Billing for telehealth and out-of-state visits
- Payer reimbursement for recommended genomic testing algorithms
- Investment in the development of novel genomic testing applications

**Strengths:**

- ColumbiaDoctors contracting

**Exceptions:**

- CRO manages CU preauthorization for genomic/genetics testing
- Reproductive genetics has credentialed its genetic counselor and is billing for services

**RECOMMENDATIONS:**

- Work with NYP, Pathology, and ColumbiaDoctors Clinical Revenue Office, Compliance, Contracting, and Credentialing offices to create uniform policies for counseling, evaluation, and genetic and genomic testing
    - Enable all GCs to bill for independent services
    - Work with payers to ensure reimbursements for genetic services and genomic testing
    - Provide patients with assistance for pre-authorization and appeals for denied services and tests
- Create a financial model to support genomic testing recommended by the Clinical Genomics Steering Committee prior to being reimbursed by payers.
- Leverage philanthropic interest in precision medicine to advance the implementation of clinical genomics.

## VIII. Other

- Clinical and research space
- Communications (including social media)
- Development staff for fundraising

## Appendix: Taskforce Membership

### Chair

**Katrina Armstrong, MD**
Chief Executive Officer of Columbia University Irving Medical Center, Executive Vice President for Health and Biomedical Sciences, and Dean of the Faculties of Health Sciences and Medicine.

### Administrator and co-chair

**Jennifer Williamson, MS, MPH**
Associate Vice Dean for Research, VP&S

### Members

**Adam Bass, MD**
Professor of Medicine (in the Herbert Irving Comprehensive Cancer Center); Department of Medicine, Division of Hematology/Oncology; Director of the Columbia Center for Precision Cancer Medicine

**Amanda Bergner, MS, CGC**
Associate Professor of Genetic Counseling (in Genetics and Development) at CUMC; Director, Columbia Genetic Counseling Graduate Program

**Wendy Chung, MD, PhD**
Kennedy Professor of Pediatrics (in Medicine), Department of Pediatrics; Chief, Division of Clinical Genetics; Medical Director, Columbia Genetic Counseling Graduate Program; co-Director of Precision Medicine Resource in the Irving Institute

**Ali Gharavi, MD**
Jay Meltzer Professor of Nephrology and Hypertension (in Medicine), Department of Medicine; Chief, Division of Nephrology; Director, Center for Precision Medicine and Genomics (CPMG); Interim Director, Institute for Genomic Medicine

**Matt Harms, MD**
Associate Professor of Neurology; Department of Neurology, Neuro-Genetics

**Eldad Hod, MD**
Associate Professor of Pathology and Cell Biology; Vice Chair of Laboratory Medicine; Director of the Automated Laboratory; Director of the Center for Advanced Laboratory Medicine (CALM)

**Josh Milner, MD**
Professor of Pediatrics; Department of Pediatrics; Director, Division of Pediatric Allergy, Immunology, and Rheumatology

**Ron Wapner, MD**
Professor of Obstetrics & Gynecology at CUMC; Department of Obstetrics and Gynecology; Director, Reproductive Genetics; Medical Director, Columbia Genetic Counseling Graduate Program

**Muredach Reilly, MD**
Professor of Medicine; Director, Irving Institute for Clinical and Translational Research; Associate Dean for Clinical and Translational Research; Director, Cardiometabolic Precision Medicine Program