

COLUMBIA PRECISION MEDICINE INITIATIVE

PRECISION MEDICINE

ANNUAL NEWSLETTER 2020 - 2021



COLUMBIA | PRECISION MEDICINE

Dear Colleagues,

Over the past year, the Columbia Precision Medicine Initiative (CPMI) has continued to grow with the engagement and collaboration of faculty and leadership throughout the University. With more critical recruitments, conferences, and pilot awards, the Columbia Precision Medicine Initiative is moving forward.

We are delighted to welcome Dr. Adam Bass, MD, a physician-scientist in the field of cancer genomics and gastrointestinal cancer. He will join the Herbert Irving Comprehensive Cancer Center as the founding director of the Center for Precision Cancer Medicine and director of gastrointestinal oncology. He will also serve on the faculty of the Vagelos College of Physicians and Surgeons as professor of medicine. Adam will work closely with the Precision Oncology and Systems Biology Research Program, co-led by Dr. Andrea Califano, PhD and Dr. Richard Carvajal, MD. A key initiative within the center will be a program to sequence patients' tumors during treatment, building a vast array of data that researchers will use to further refine their understanding of cancer's defense mechanisms and develop new ways to counteract them.

The recently launched Herbert and Florence Irving Institute for Cancer Dynamics (IICD), directed by Simon Tavaré is making excellent progress. IICD is an interdisciplinary institute located on the Morningside Heights campus and is focused on the interplay between STEM disciplines and cancer research.

In addition, the Center for Precision Medicine and Genomics in the Department of Medicine has been making excellent progress. Under the leadership of Dr. Ali Gharavi, the center is building multidisciplinary expertise in Precision Medicine for each subspecialty of Internal Medicine.

We are currently reviewing applications for the Roy and Diana Vagelos Precision Medicine Pilot Awards. This program, now in its fourth year, has stimulated collaboration across all three Columbia campuses, led to external funding and high visibility publications. The Initiative also supported four awards for the Vagelos Mouse Genome Editing with the objective of funding the generation of mouse models of human disease. A new call for Mouse Genome Editing awards will be issued soon.

The Precision Medicine and Society program, chaired by Paul Appelbaum, MD and Gil Eyal, PhD, sets Columbia apart from other precision medicine initiatives by focusing on ethical and equity issues in the impact of precision medicine. One of the highlights of the previous year was our panel on Covid-19 and public health, and a paper to be published on the relevance of precision medicine in the age of Covid. The second conference will take place in the Spring.

Another highlight was our fourth academic conference, **Advances in Precision Medicine: Harmonization of Clinical and Genomic Data**, which provided a full day of high impact international speakers covering basic and applied science in this field of precision medicine. We look forward to hosting our fifth conference on April 7, 2020, which will focus on the genetics of neurodevelopmental disorders.

I would like to take this opportunity to thank Dr. Roy Vagelos for his continuing scientific and medical leadership in precision medicine, and his generous gift to the Precision Medicine Initiative. The gift is being used to fund a number of critical recruitments in precision medicine research, and the infrastructure required to advance basic science.

A more detailed description of progress in the Precision Medicine Initiative during the past year and further details of the activities during the coming year is provided in this newsletter.

Tom

Tom Maniatis, Ph.D.
Director of Columbia University Precision Medicine Initiative; and the
Isidore S. Edelman Professor in the Department of Biochemistry & Molecular Biophysics

In this issue:

Letter from Dr. Tom Maniatis

4th Annual CPMI Conference: Advances in Precision Medicine: Harmonizing Clinical and Genomic Data

Precision Medicine Scholars Day

Faculty Announcements

Updates

- Institute for Genomic Medicine
- Irving Institute for Clinical and Translational Research
- BRIDGE BioBank
- Precision Medicine Publications
- Targeted Research and Exploration Advancing Trial Models, Editing, and Next-generation Therapies (TREATMENT)
- EpicTogether
- Electronic Medical Records and Genomics (eMERGE) network
- Precision Genomics Laboratory (PGL)
- Laboratory of Personalized Genomic Medicine (PGM)
- Center for Precision Medicine and Genomics (CPMG)
- Precision Oncology and Systems Biology (POSB) Program
- Irving Institute for Cancer Dynamics (IICD)

Precision Medicine Awards

- Roy & Diana Vagelos Precision Medicine Pilot Awards
- Herbert Irving Comprehensive Cancer Center Award
- Irving Institute for Clinical and Translational Research Award
- Mouse Genome Editing Awards

Precision Medicine & Society

- Precision Medicine: Ethics, Politics and Culture (PMEPC)
- Publications
- Events
- Seminars & Conferences
- Seed Grants

Educational Initiatives

- The Economics of Precision Medicine

Advances in Precision Medicine Conference: Harmonizing Clinical and Genomic Data

One of the challenges facing us today is how to leverage the huge amounts of genomic data generated every day through regular patient care? How can this data be integrated into electronic health records in order to generate knowledge in a clinically useful way?

At the 4th Annual CPMI conference: Advances in Precision Medicine: Harmonizing Clinical and Genomic Data we learned about the most recent strategies to integrate large amounts of genomic data into electronic health records, to generate clinically-useful knowledge in these presentations. Please find the links to each talk below and also in our video library here.

[David Ledbetter, Ph.D., Geisinger](#)

[Dan Roden, M.D., Vanderbilt University School of Medicine](#)

[Marylyn Ritchie, Ph.D., University of Pennsylvania](#)

[Rex L. Chisholm, Ph.D., Northwestern Feinberg School of Medicine](#)

[Chunhua Weng, Ph.D., Columbia University](#)

[Nancy J. Cox, Ph.D., Director, Vanderbilt University](#)

[David Madigan, Ph.D., Columbia University](#)

[Nicholas Tatonetti, Ph.D., Columbia University](#)

The 4th Annual CPMI conference: Advances in Precision Medicine: Harmonizing Clinical and Genomic Data was held on April 24, 2020 via Zoom.

SAVE THE DATE

**5th Annual Advances in Precision Medicine Conference:
Genetics of Neurodevelopmental Disorders
Friday, April 7, 2021**

Precision Medicine Scholars Day

On October 23, 2020, the CPMI, Irving Institute for Clinical and Translational Research and the Herbert Irving Cancer Research Center presented Precision Medicine Scholars' Day, an annual half-day symposium that serves as a platform for Precision Medicine fellows and pilot award recipients to share results of their research projects with the Columbia University community.

Event Program can be downloaded [here](#).

Faculty Announcements

CHRISTOPHER MAKINSON, PhD



Christopher Makinson, PhD is a newly recruited scientist from Stanford University who will be joining Columbia University's Institute for Genomic Medicine in June of 2020. Educated at Wake Forest University and Emory University, Dr. Makinson completed his postdoctoral training at Stanford University where he became an expert in interrogating neural circuits in epilepsy using state-of-the-art electrical and optical physiology methods. At Columbia, Dr. Makinson uses rodent models and human brain organoids to develop novel avenues for personal/precision medicine. His goal is to increase our understanding of how neural networks give rise to normal and pathological conditions, with an emphasis on elucidating the genetic underpinnings of neurological diseases.

HEMALI PHATNANI, PhD

Hemali Phatnani, PhD, assistant professor of neurological sciences, in the Division of Neuromuscular Medicine, the Center for Motor Neuron Biology and Disease, and the Columbia Translational Neuroscience Initiative. Dr. Phatnani received her PhD in Biochemistry and Molecular Biology at Duke University, and a BSc degree in Life Sciences from Bombay University. As a postdoctoral fellow in the laboratory of Dr. Tom Maniatis at Harvard and Columbia Universities, Dr. Phatnani studied ALS disease mechanisms using stem cell-derived motor neurons and genomic profiling methods to study the complex interplay between motor neurons and astrocytes in ALS disease progression. Dr. Phatnani will join the Eleanor and Lou Gehrig ALS Center team, and will retain a joint appointment at the New York Genome Center as the Director of the Center for Genomics of Neurodegenerative Disease. In that role, Dr. Phatnani established the NYGC ALS Consortium involving clinicians, scientists, geneticists, computational biologists and industry partners, providing a framework to apply clinical and functional genomics together with bioinformatics to study ALS.



BRADLEY MILLER, MD, PhD



Bradley Miller is an Assistant Professor in the Department of Psychiatry at Columbia University. He received his MD/PhD from Washington University. Dr. Miller then joined Columbia University to pursue clinical training in psychiatry followed by a research fellowship with René Hen and Sander Markx. Dr. Miller's lab now focuses on two areas aimed at bringing precision medicine to the treatment of major depression. The first builds on a discovery he made with Dr. Markx that a rare mutation in a receptor called GPR156 increases the risk of major depression threefold. Dr. Miller's laboratory is investigating how this mutation alters cell signaling, gene expression, and neuronal activity in the brain and leads to depression. This could lead to novel interventions to treat depression. Dr. Miller's second area of research is investigating the function of the serotonin, the target of the most widely used antidepressants. Clarifying how serotonin release regulates emotional behaviors could lead to improvements on current serotonergic antidepressant medications.

Updates

Institute for Genomic Medicine



Over the past year, the Institute for Genomic Medicine (IGM), directed by Dr. David Goldstein, has continued to make precision medicine a reality across the medical center. In partnership with several clinical departments, the IGM is breaking new ground toward the clinical application of genomics across lifespans of individuals and toward investigations of disease-causing mutations in the laboratory to provide personalized medicine in the clinic.

Results of one major clinical and research collaboration were published in September of this year in the New England Journal of Medicine. This study identified novel genes that cause stillbirth in approximately one of every ten cases studied. Using standard and advanced analysis techniques, this study demonstrates the importance of diagnostic sequencing in all cases of unexplained stillbirth. The work highlights major ways that genomic analyses can contribute to unraveling the complex biology of stillbirth. The study was conducted in partnership with the Department of Obstetrics and Gynecology at the Vagelos College of Physicians and Surgeons.

In another new study led by the IGM, researchers found that lowering testosterone could reduce the severity of COVID-19 disease by preventing the novel coronavirus from entering lung cells. The study sought to identify compounds that were capable of regulating expression of genes that encode key viral entry proteins. Their work showed that TMPRSS2 represented a promising viral entry protein target, and that lowering testosterone would be the best way to lower expression of TMPRSS2, and potentially reduce viral entry. Based on these findings, a [clinical trial](#) to test the effects of reducing testosterone in COVID-19 patients is set to begin at three Veterans Affairs hospitals in New York City, Los Angeles and Seattle.

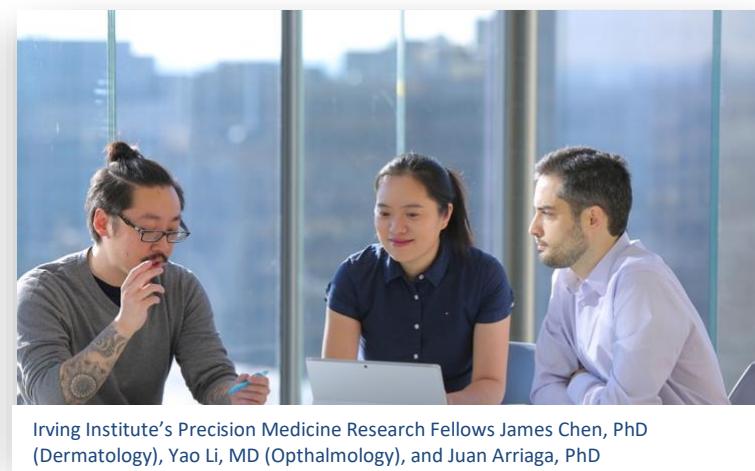
The IGM and the Vagelos College of Physicians & Surgeons Department of Neurology successfully recruited Christopher Makinson, PhD as an Assistant Professor in the Department of Neurology and in the IGM. His research focuses on how neural networks give rise to different brain states including sleep and arousal as well as disease states such as epilepsy. Chris is on a tenure track appointment and was also appointed as a CTNI Scholar in the Columbia Translational Neuroscience Initiative. The CPMI, funded by Roy and Diana Vagelos' generous gift, will provide \$1M of support to the development of Chris' research program.

Another key joint recruit for the IGM, and in partnership with Columbia's Department of Pediatrics, was Joshua Milner, MD. Joshua will serve as the Chief of the Division of Allergy, Immunology, and Rheumatology in the Department of Pediatrics and Professor of Pediatrics in IGM. He will also serve as Chief of Allergy, Immunology, and Rheumatology services at New York-Presbyterian Morgan Stanley Children's Hospital. Joshua is a leader in the field of allergy, immunology, and rheumatology whose research led to the discovery of genetic bases of allergic diseases in children.



Irving Institute for Clinical and Translational Research

In the past year under the leadership of Wendy Chung, MD, PhD, Krzysztof Kiryluk, MD, Ronald Wapner, MD, David Goldstein, PhD, and Gary Miller, PhD, the [Precision Medicine Resource](#) team of the Irving Institute continued establishing new and supporting existing programs focused on providing funding opportunities, research services, and workforce development in the emerging domain of patient-centered healthcare. In collaboration with the CPMI and Herbert Irving Comprehensive Cancer Center (HICCC), the Resource selected five new interdisciplinary teams (PIs: Richard Carvajal, MD; Thomas Nikolas, MD; Lynn Petukhova, PhD; Samuel Sternberg, PhD; Chaolin Zhang, PhD) to receive one-year [Precision Medicine Pilot awards](#) to supporting research projects focused on tailoring medical care to the individual patient's needs in a wide range of basic, pre-clinical and clinical precision medicine domains. On October 23, 2020, research accomplishments of past and current award recipients were celebrated at the annual university-wide [symposium - "Precision Medicine Scholars Day"](#), hosted in the virtual format jointly with the CPMI and HICCC teams. The one-semester Vagelos College of Physicians and Surgeons [graduate course "Introduction to Precision Medicine"](#), offered by the Resource for the fourth consecutive year, allowed medical, genetic counseling, and other health sciences scholars to gain insights into diverse precision medicine topics, such as genomic medicine, digital health, exposomics, and others. Now in its sixth year, a monthly seminar series ["Advances in Precision Medicine"](#) continued to bring to campus thought leaders in the field. To enable direct communication, sharing, and collaboration among current (and future) NIH NCATS Clinical and Translational Science Awards Program hubs with a Precision Medicine focus, the Resource created a [Precision Medicine Discussion Forum](#). Past summer the team also supported the launch of the [Irving Institute's YouTube channel](#) to disseminate its provider-, researcher-, and patient-facing precision medicine content. In addition, the Resource played a key role in several other university precision medicine initiatives, including the establishment of Columbia University Biobank, publication of a review series of "Cases in Precision Medicine" in the Annals of Internal Medicine journal, establishment of the Precision Medicine team within the EpicTogether tri-institutional collaboration, the launch of the fourth phase of the NHGRI-funded eMERGE project, and many others.



Irving Institute's Precision Medicine Research Fellows James Chen, PhD (Dermatology), Yao Li, MD (Ophthalmology), and Juan Arriaga, PhD (Pharmacology) (from left to right)

BRIDGE Biobank

[Columbia University Biobank \(CUB\)](#) was quickly launched in April 2020 in response to the urgent needs surrounding the coronavirus pandemic. Since then, CUB has collected biospecimens from Columbia University Irving Medical Center/NewYork-Presbyterian Hospital (CUIMC/NYP) patients who tested positive for SARS-CoV-2, and re-distributed samples to researchers across campus. Currently we have over 90,000 biospecimens banked from more than 14,000 patients (both COVID-positive and -negative) including DNA, RNA, serum, plasma, urine, and other tissue samples. Given the number of patients treated for COVID-19 at CUIMC/NYP and the diversity of our patient population, Columbia's COVID-19 biobank is arguably the largest and most distinct in the United States. Additionally, CUB has consented more than 2,400 patients for genomic research and future research studies to further advance our understanding of COVID-19 and its devastating clinical consequences. Whole exome sequencing data has been generated on more than 1,000 patients diagnosed with SARS-CoV-2 and is currently available in AWS for download with the appropriate IRB and CUB approvals. Host and virome RNA sequencing and microbiome data generation is ongoing and will soon be available to investigators with CUB approval. We are also excited to announce that Dr. David Goldstein has partnered with Vanda Pharmaceuticals to perform whole genome sequencing on 1,000 CUB COVID+ patients. Once generated, this data will also be available to CUIMC researchers for their own analyses. Dr. Krzysztof Kiryluk in the Department of Medicine is spearheading an effort to generate SARS-CoV-2 genomic phenotypes, which will then be combined with sequencing data generated by the Institute for Genomic Medicine, into a single dataset that will be available centrally through CUB. The CUB Clinical Phenotype Committee (CPC), led by Dr. Mitch Elkind, has been established to oversee efforts related to the generation of broader COVID-19 clinical phenotypes. The CPC is multidisciplinary working group which includes experts from the Departments of Anesthesiology, Surgery, Pediatrics, Emergency Department, Biomedical Informatics, Cardiology, Obstetrics and Gynecology, and Medicine (Pulmonary, Nephrology and Transplant). The CPC has focused initial efforts on standardizing, validating, and refining core SARS-CoV-2 related phenotype definitions, which will be made widely available when completed. As of

October 2020, over 140 requests to the biobank were reviewed by the CUB Sample and Data Committee, and more than 22,000 samples have been disbursed to support a wide range of research studies across the CUIMC community. With assistance from Drs. Kevin Roth, Eldad Hod, and Susan Whittier in the Department of Pathology and Cell Biology, we are able to continue residual clinical sample collections, which will be critical as the coronavirus continues to spread. While CUB has initially focused on COVID-19 due to obvious needs, it continues to look past the pandemic and begin University-wide recruitment efforts towards the ultimate goal of enrolling all CUIMC/NYP patients. To this end, we are excited to be partnering with the IGM's *All of Us* team. The Columbia University Biobank is now being offered to *All of Us* participants during their enrollment and retention activities. CUB looks forward to applying the lessons learned from the *All of Us* Research Program to the CUB recruitment efforts. The pilot initiative, [Cardiometabolic Precision Medicine Program](#) (executive leaders - Muredach Reilly, MBBCh, MSCE; Steven Marx, MD; Wendy Chung MD, PhD; program manager - Sheila M. O'Byrne, PhD), is ongoing and recruiting in the Division of Cardiology/Department of Medicine, but will be integrated with CUB in the coming year. We encourage you to apply to use these research resources through the CUB [website](#).

Precision Medicine Publications

Led by Krzysztof Kiryluk, MD, the Resource team joined forces with a diverse group of Precision Medicine experts across the CUIMC to publish 10 articles in the Annals of Internal Medicine on various topics in Precision Medicine including a series of 9 case studies, each dealing with a common clinical issue regarding precision medicine.

A current list of publication from the series:

[Precision Medicine in Internal Medicine: Overview of the series](#)

[The Role of Pharmacogenetics in Precision Prescribing](#)

[When Patients Present With Direct-to-Consumer Genetic Test Results](#)

[Should You Participate in a Study Involving Genomic Sequencing of Your Patients?](#)

[Genetic Assessment After a Sudden Cardiac Death in the Family](#)

[The Role of Tumor and Germline Genetic Testing in Breast Cancer Management](#)

[APOL1 and Genetic Testing in the Evaluation of Chronic Kidney Disease and Potential Transplant](#)

[Concerns About Privacy and Discrimination After Genomic Sequencing](#)

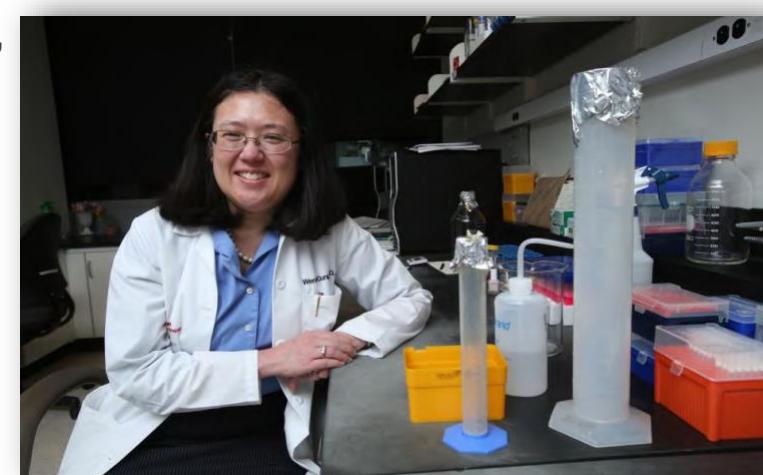
[A Personalized Approach to Stroke and Cardiovascular Risk Assessment in Women](#)

[Cases in Precision Medicine: The Role of Polygenic Risk Scores in Breast Cancer Risk Assessment](#)

Targeted Research and Exploration Advancing Trial Models, Editing, and Next-generation Therapies (TREATMENT)

The Targeted Research and Exploration Advancing Trial Models, Editing, and Next-generation Therapies (TREATMENT) program was established by Dr. Wendy Chung to expand our capacity to care for patients with rare genetic diseases, understand the natural history and molecular mechanisms of genetic diseases, and develop new treatments for these conditions. The program was established to make Columbia a destination medical center for a growing international network of patients and families and serve as a nucleus of physicians and scientists in academia and industry working together toward cures for rare genetic diseases.

We are developing novel molecular methods of treatment including the use of antisense oligonucleotides, gene edition, and gene therapy. We have a Research Collaboration with Ovid for the development of treatments for patients with neurogenetic disorders, including KIF1A associated neurological disorder. Our work has been featured in the Ken Burns documentary, *The Gene* (<https://www.pbs.org/kenburns/the-gene/>). We are currently participating in 6 clinical trials of new treatments for genetic disorders and have patients from around the United States participating in these trials. TREATMENT unites families, patients and families and provides them with hope for a brighter future.



EpicTogether

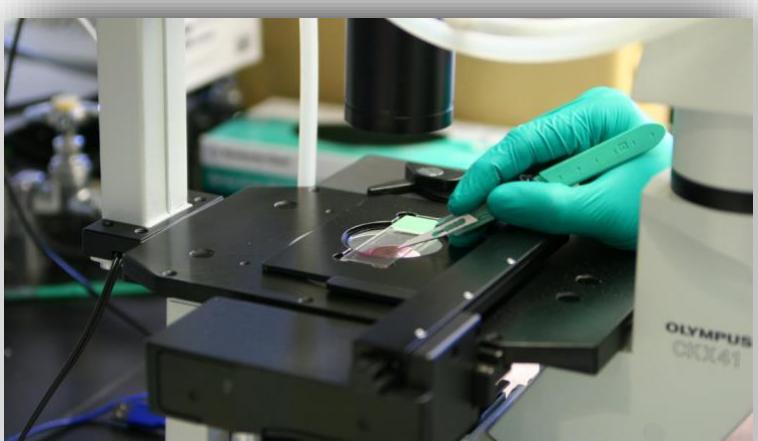


[EpicTogether](#), the operational managing team for New York Consortium Epic implementation, has established a Precision Medicine team with dedicated analysts to work on Epic's new Genomic module and other activities around genomic data. A collaborative group representing the 3 institutions (NYP, Columbia, Cornell) is developing the infrastructure to identify genomic lab tests, convert test results to genomic clinical indicators, and develop appropriate Best Practice Advisory alerts. The initial focus

was on Clinical Pharmacogenetics Implementation Consortium (CPIC) recommended guidelines for actionable pharmacogenetic results, with a recent expansion to include three CDC Tier 1 genomic applications with evidence-based guidelines. Additionally, a genomic variant entry tool was implemented to enable documentation of discrete variant data, in essence creating a hospital-wide repository of genomic variants. This feature in combination with Epic's reporting capabilities will allow providers to create custom, on-demand queries of the hospital-wide variant data. The team will continue to champion, set goals and develop procedures for Precision Medicine data capture and alerting in Epic EHR.

Electronic Medical Records and Genomics (eMERGE) network

On July 1, 2020, the National Institutes of Health announced that Columbia University was selected to be one of ten clinical and enhanced diversity sites from around the United States to form a Genomic Risk Assessment and Management Network as part of the fourth phase of the [Electronic Medical Records and Genomics \(eMERGE\)](#) project. Having been part of this initiative since 2011, Columbia eMERGE team co-led in this phase by investigators Chunhua Weng, PhD, Wendy Chung, MD, PhD, George Hripcak, MD, and Krzysztof Kiryluk, MD (project manager – Alexander Fedotov, PhD) will establish protocols and methodologies for improved genomic risk assessments in diverse groups and their integration in clinical care. As part of this effort, the team will also spearhead efforts on the recruitment of participants from racial and ethnic minority populations, underserved populations, and populations who experience poorer medical outcomes, to conduct and validate genomic risk assessment for a number of common complex diseases of public health importance.



Precision Genomics Laboratory (PGL)

The Precision Genomics Laboratory, a joint initiative of the Department of Pathology and Cell Biology and the Institute for Genomic Medicine, continues to apply advanced genomic science in a clinically actionable setting to improve the diagnosis and treatment of human disease. This CLIA/CLEP certified and CAP accredited laboratory currently offers clinical cystic fibrosis screening and Sanger sequencing of individual variants. The latter test is crucial to our local genomic sequencing initiatives because it ensures that high quality, clinical grade variant reports

are transmitted to the electronic medical record, thereby translating laboratory-based precision medicine technologies into clinical decision-making tools.

In the past year, the PGL's multidisciplinary clinician scientist teams have submitted several assays/ testing platforms to the NYS Department of Health. The PGL now has approval to use the Global Screening Array, a high throughput genotyping platform for clinical tests. Please contact Dr. Vimla Aggarwal to learn more about this offering.

The PGL has also been an integral part of the COVID-19 biobank at CUIMC. By performing DNA extraction from residual buffy coats and nasopharyngeal swab specimen, the PGL has been supporting institution wide COVID-19 research efforts.

The PGL plans to begin offering whole exome sequencing and a ~400 gene expanded carrier screening panel as clinical tests upon approval by the NYS Department of Health.

The PGL also employs a team of genetic counselors to act as liaison between clinical care providers and the laboratory in order to ensure that the genomic information generated by the PGL is used to inform and guide personalized, patient-focused healthcare for the CUIMC community and beyond.

Laboratory of Personalized Genomic Medicine (PGM)

The Laboratory of Personalized Genomic Medicine (PGM) in the Department of Pathology and Cell Biology is a state-of-the-art diagnostic laboratory that performs cutting-edge tests in the areas of genetics, neurogenetics, oncology, cytogenomics, and molecular microbiology. The CLIA-accredited laboratory, directed by Mahesh Mansukhani, MD, is accredited by the College of American Pathologists (CAP), and the Clinical Laboratory Evaluation Program of the New York State Department of Health (NYS-DOH). PGM offers multiple clinical molecular oncology and constitutional genomics assays, including single gene assays, small cancer panels, a large 467-gene cancer panel, as well as whole-exome and whole-transcriptome sequencing. PGM has received NYS-DOH approval for Darwin OncoTarget™/OncoTreat™ analysis of transcriptomes, a powerful and novel systems biology approach that assesses activity of potentially targetable master regulators (developed in collaboration with Dr. Andrea Califano in the Department of Systems Biology), which will further advance precision oncology efforts at CUIMC. The laboratory has recently completed validation of a 50-gene myeloid panel and a 28 gene circulating DNA panel. In 2019, PGM performed nearly 60,000 clinical tests including nearly 9,000 constitutional genetics assays, over 5,000 oncology assays, and nearly 45,000 molecular microbiology and virology assays.



In recent years, the PGM has developed a national presence in the field of molecular oncology laboratory testing. In August 2018, PGM was selected to participate as a CLIA-certified laboratory for the NCI-Molecular Analysis for Therapy Choice (NCI-MATCH) precision medicine trial. NCI-MATCH is the largest trial to date that seeks to determine whether therapies targeting specific gene mutations will be effective regardless of cancer type. Tumor gene testing by a designated lab is the only pathway for patients to enroll into the trial. In 2020, PGM will partner with the Herbert Irving Comprehensive Cancer Center (HICC) and Department of Pediatrics in the upcoming ComboMATCH and NCI-COG Pediatric MATCH clinical trials, which employ combinations of precision medicine agents to treat a variety of cancers in the pediatric and adult patient populations. In addition, during the COVID-19 emergency, the laboratory of PGM completed verification of the first clinical SARS-CoV-2 test performed at the Medical Center, and an FDA emergency use authorization (EUA) was obtained by the PGM laboratory for a SARS-CoV-2 assay developed at the Center for Infection and Immunity of the Mailman School of Public Health.

PGM faculty and staff members participate in pediatric and adult molecular tumor boards at CUIMC and nationally in the American Society of Clinical Oncology's Targeted Agent and Profiling Utilization Registry (TAPUR) study molecular tumor board. Additionally, the PGM bioinformatics team has led CUIMC involvement in Project GENIE, an American Association for Cancer Research program for aggregation of cancer genomics and clinical outcome data in a HIPAA compliant registry with the goal of catalyzing clinical and translational cancer research. Faculty and trainees at the Laboratory of Personalized Genomic Medicine lead multidisciplinary tumor boards of the Precision in Pediatric Sequencing (PIPSEQ) and the Columbia Precision Oncology Initiative (CPOI) of the Department of Pediatrics and HICC, respectively. The Laboratory of Personalized Genomic Medicine is committed to supporting and enhancing clinical and research initiatives among the CUIMC precision oncology community.

Center for Precision Medicine and Genomics (CPMG)

The success of the CPMI requires a transformation of our clinical and translational paradigms and infrastructures and extensive education of our faculty. A collaboration between the Department of Medicine (DoM) and the Institute for Genomic Medicine (IGM), the newly created Center for Precision Medicine and Genomics (CPMG) in the Department of Medicine brings together physicians, scientists and other health professionals to deliver the promise of Precision Medicine for adult constitutional disorders. Led by Dr. Ali Gharavi, MD, the Center builds on the existing collaborations between the DoM, the IGM, and virtually every clinical and basic science departments at CUIMC. The Center aims to develop a clinically oriented Precision Medicine program in each subspecialty of Internal Medicine. In addition, CPMG is dedicated to educational programs for clinicians and physician-scientists. The broad range of programs developed to date includes summer internships for undergraduate and medical students, bimonthly genetics case studies series, monthly seminars by prominent scientists, and a two-day CME course on genomic medicine for healthcare providers. In addition, CPMG leads a vibrant multidisciplinary research program and develops bioinformatics tools that will enhance our capacity for Precision Medicine across the entire campus. For more information on these activities, please visit the CPMG website: <http://www.columbiamedicine.org/cpmg/>

Precision Oncology and Systems Biology (POSB) Program

Cancer therapies have traditionally been based on the tumor site, like breast cancer or lung cancer. More recently, researchers have found that cancers across tumor sites have common genetic mutations, and those mutations can be targeted with specific drugs.

Precision cancer medicine is a rapidly developing field that goes beyond studying cancer by tumor site, using genetic sequencing techniques and novel approaches to uncover a patient's specific tumor mutations and molecular composition in order to deliver more personalized treatment. In January 2021, the Herbert Irving Comprehensive Cancer Center (HICCC) will welcome Adam Bass, MD, a leading physician-scientist in the field of cancer genomics and gastrointestinal cancer, as the founding director of the Center for Precision Cancer Medicine and director of gastrointestinal oncology. He also will serve on the faculty of the Columbia University Vagelos College of Physicians and Surgeons, proposed as Irving professor of medicine in the Division of Hematology and Oncology.



Dr. Bass joins the Precision Oncology and Systems Biology (POSB) Research Program – one of four research programs supported by the HICCC. He will lead an actively growing program of physician-scientists working at the interface of cancer biology and the development of new cancer diagnostics and therapies. The new center will coalesce investigators across Columbia and NewYork-Presbyterian in a 360-degree approach, not only bringing discoveries from the lab to patients' bedsides, but also incorporating research in real time, allowing researchers to understand how cancer evolves and adapts in response to therapies. A key initiative within the center will be a program to sequence patients' tumors during treatment, building a vast array of data that researchers will use to further refine their understanding of cancer's defense mechanisms and develop new drugs and drug combinations to counteract them. Columbia's Department of Systems Biology, one of the leading programs in the country, will collaborate to apply its pioneering algorithms that process and interpret these complex data.

In addition to Dr. Bass, the HICCC welcomes a number of new recruits working across the cancer research continuum to understand cancer from the basic biology to new approaches in personalized cancer prevention and treatment. These new recruits include:

Elham Azizi, PhD (*from Memorial Sloan Kettering Cancer Center*), an assistant professor of biomedical engineering who is combining cutting-edge single-cell genomic technologies with machine learning

techniques, to characterize complex populations of interacting cells, and their dysregulated circuitry, in the tumor microenvironment.

Chia-Wei Cheng, PhD (*from Koch Institute at Massachusetts Institute of Technology*), an assistant professor of genetics and development focused on the interface of nutritional and transcriptional regulatory networks in adult stem cells, with a particular interest in the signaling (non-energetic) role of metabolites in regulating lineage specification and cellular plasticity.

Eunhee Choi, PhD (*from University of Texas Southwestern Medical Center*), an assistant professor of pathology and cell biology whose research focuses on how systemic signaling communicates with cell division to maintain physiological homeostasis and chromosome integrity, and to elucidate the pathogenesis of cancer and metabolic diseases.

Elena Elkin, PhD (*from Memorial Sloan-Kettering Cancer Center*), Professor of Health Policy and Management, whose research focus is on evaluating determinants of cancer screening, treatment and outcomes using population-based observational data analysis, decision analysis and patient surveys, primarily in breast cancer.

Robyn Gartrell, MD (*from Columbia University Irving Medical Center*), assistant professor of pediatrics, who is using multiplex platforms and immunogenomics to help determine which patients will respond to immunotherapy and find the right combinations of treatments to improve immune responses in patients who are resistant to these therapies.

Nobuko Hijiya, MD (*from Northwestern University*), a professor of pediatrics focused on pediatric leukemias, development of new drugs utilizing a translational approach in collaboration with basic researchers, a broad range of ancillary studies, and cancer survivorship.

Lauren Houghton, PhD (*from Mailman School of Public Health, Columbia*), assistant professor of epidemiology, who is studying the role of androgens in breast cancer etiology, how they relate to early life risk factors such as pubertal timing and psychosocial stress, and whether androgen patterns track across the lifecourse.

Ben Izar, MD PhD (*from the Dana-Farber Cancer Center*), an assistant professor of medicine working to understand the biology of melanoma development and to develop and implement technologies that enable high-resolution single-cell profiling and imaging of patient tissues.

Shawn Liu, PhD (*from the Whitehead Institute at Massachusetts Institute of Technology*), assistant professor of physiology and cellular biophysics focused on the role of epigenome in normal physiology and human diseases, and the functional contribution of epigenetic mechanisms during the progression of cancer.

Alexander Melamed, MD, MPH (*from Massachusetts General Hospital*), assistant professor of medicine, gynecological oncology focused on health services, comparative effectiveness research and surgical care of women with gynecologic cancer.

Catherine (Katie) Spina, MD (*from Columbia University Irving Medical Center*), assistant professor of radiation oncology, who seeks to understand how radiation behaves as an immune modulator of the tumor microenvironment and to leverage this mechanistic insight to develop targeted therapies for combination with radiation therapy and other immunotherapies to improve outcomes for cancer patients,

Alison Taylor, PhD (*from the Dana-Farber Cancer Center*), an assistant professor of pathology and cell biology who is working to understand how specific aneuploidy events contribute to cancer initiation, progression, and treatment response.

Aaron Viny, MD PhD (*from Memorial Sloan-Kettering Cancer Center*), an assistant professor of pathology and cell biology with a focus on pathophysiologic effects of altered chromatin structure and DNA methylation and on the functional effects and potential reversibility of chromatin structure on normal and malignant hematopoiesis.

Jianlong Wang, PhD (*from Mount Sinai School of Medicine*) a professor of medical sciences who is dissecting the novel transcriptional and epigenetic mechanism underlying breast cancer metastasis and dormancy.

Xuebing Wu, PhD (*from the Whitehead Institute at Massachusetts Institute of Technology*), assistant professor of medicine and systems biology who is focused on network models that integrate phenotypic information of all diseases and protein interaction data of all genes to understanding how genetic information is encoded in the genome and how genetic defects cause human diseases, including cancer.

Xiao Zhao, MD MTR (*from the University of Pennsylvania*), assistant professor of medicine, whose work uses the zebrafish model to understand hepato-biliary cell biology in the context of diseases that arise therein.

Irving Institute for Cancer Dynamics (IICD)

The Herbert and Florence Irving Institute for Cancer Dynamics (IICD), directed by Simon Tavaré, was launched in July 2018. IICD is an interdisciplinary institute located on the Morningside Heights campus and focused on the interplay between STEM disciplines and cancer research. IICD collaborates across disciplinary boundaries to support research and develop technology that can improve our understanding of cancer biology, origins, evolution, treatment, relapse and prevention. The IICD is housed in newly renovated space in Schermerhorn Hall.

The Institute appointed its first endowed chair, Elham Azizi, Assistant Professor of Biomedical Engineering and Herbert and Florence Irving Assistant Professor of Cancer Data Research, in January 2020. Dr Azizi's research uses single-cell genomic technologies to study the heterogeneity in the tumor microenvironment and underlying mechanisms of response to immunotherapies in cancer patients. IICD also expanded its research themes by recruiting three Associate Research Scientists who will develop independent research projects: Dr David Tourigny (cancer metabolism), Dr Sanket Rane (immunology) and Dr Karol Nowicki-Osuch (esophageal cancers), and recruited several Postdoctoral Research Scientists. The Institute also counts 14 Affiliates and Associate Members that are supporting the Institute's mission.

IICD is also developing a strong outreach program by providing seed grant funds and supporting working groups, summer internships and seminar series. During the fall, IICD partnered with the Probability and Society Initiative to invite two stellar speakers, Amaury Lambert (professor at Sorbonne Université in Paris) and Alison Etheridge (Professor of Probability and Head of the Department of Statistics, University of Oxford) for a mini series of six virtual lectures on probabilistic modeling in biology.

IICD is also supporting instrumentation throughout the campus and NYC. The Institute partnered with the NYGC to build a Direct Library Preparation Plus+ (DLP+) device, a scalable single cell whole genome sequencing system. Additionally, IICD will build in the ZMBBI a serial two-photon tomography system, together with an automated collector, to image and annotate tumor biopsies. We built a Virtual Reality suite in the IICD space that will allow tumors to be visualized in 3D when in-person research fully resumes. The Institute also supported the purchase of a 10X chromium system and a MiSeq DNA sequencer on the Morningside Heights campus, in partnership with the department of Biological Sciences.



Precision Medicine Awards

Five teams of researchers from the Vagelos College of Physicians and Surgeons and the Columbia University Irving Medical Center have been awarded pilot grants to fund a diverse set of precision medicine research.

Jointly awarded by the CPMI, the Herbert Irving Comprehensive Cancer Center (HICCC), and the Irving Institute for Clinical and Translational Research (Irving Institute), the Precision Medicine Pilot Awards underscore Columbia's commitment to supporting research targeting the promise of precision medicine, across multiple diseases. The five teams will each receive \$100,000 in funding for one year.

The Roy and Diana Vagelos Precision Medicine Pilot Awards are a cornerstone of the CPMI mission: to establish world class academic research centers of excellence to build precision medicine as a basic and applied science at Columbia. Seeding basic research in precision medicine with these awards is an efficient way of converting this money to external research grants and we look forward to this return on investment in due course.

The three winning Vagelos proposals reflect the high standard and the broad base of precision medicine basic science research being conducted and conceived at Columbia. They cover research into the role of the vaginal microbiome in premature births; the skin disease hidradenitis suppurativa; and a high-throughput screening strategy to identify splicing-regulatory elements for any gene.

Roy and Diana Vagelos Precision Medicine Pilot Awards:

“Mechanistic Investigation of the Vaginal Microbiome in Different Manifestations of Spontaneous Preterm Birth”; Lead Investigator: Tal Korem, PhD; Co-PIs: Anne-Catrin Uhleman, MD, PhD; George Gallos, MD; Joy-Sarah Vink, MD

Spontaneous preterm birth (sPTB) is a leading cause of neonatal morbidity and mortality. The vaginal microbiome is associated with sPTB, but the underlying mechanisms are largely unknown. This stems from low taxonomic resolution attainable from 16S rRNA amplicon sequencing, and from the oversimplified clinical profiling of sPTB, which ignores the heterogeneity in its pathophysiology. Dr. Korem and his lab will optimize methods for bacterial DNA extraction and perform metagenomic sequencing of vaginal microbiome samples from a deeply-phenotyped cohort of pregnant women. They will study host-microbiome interactions in the context of sPTB and its underlying etiologies, using microbiome analysis methods which raise mechanistic insights regarding microbial growth rates, genomic variation, and predicted metabolite production. They intend to validate promising hypotheses in vitro and by metabolomic analysis of a subset of samples, and their aim is that this research will lead to novel insights regarding the involvement of the microbiome in different manifestations of sPTB, addressing a critical gap in the field.

“Deciphering Monogenic and Polygenic Etiologies of a Longitudinal Multi-Ethnic Hidradenitis Suppurativa Cohort”; Lead Investigator: Lynn Petukhova, PhD; Co-PI: Suzanne Leal, PhD

Drs. Petukhova and Leal are investigating the chronic skin disease, hidradenitis suppurativa (HS), aiming to find better ways to manage and hopefully prevent it. HS, which typically appears after puberty, causes painful lumps to form deep within the skin. The condition can persist for many years and get worse over time. There is currently a lack of therapies and understanding of HS, causing patients' needs to remain unmet. The researchers believe that HS has a genetic architecture that is similar to other chronic inflammatory diseases. They will be studying a multi-ethnic group of participants with HS, with a goal of garnering new knowledge about the biological drivers of disease.

“Unbiased Screen of Proximal and Distal Splicing Regulatory Elements Towards Drug Discovery.”; Lead Investigator: Chaolin Zhang, PhD; Co-PI: Samuel Sternberg, PhD

Numerous Mendelian diseases are caused by mutations that disrupt individual genes and could potentially be treated by modulating gene expression to restore normal protein production. A level of molecular regulation called alternative splicing occurs ubiquitously in human genes and frequently generates a combination of RNA isoforms that code for proteins or are noncoding. Modulation of alternative splicing using synthetic genetic strings called antisense oligonucleotides (ASOs) to target splicing regulatory elements has recently emerged as a powerful means of increasing gene expression levels. For example, SPINRAZA is an FDA-approved ASO drug that targets the SMN2 gene to treat spinal muscular atrophy. A critical challenge, however, is pinpointing the most effective regulatory RNA elements that can be targeted to modulate splicing.

Drs. Zhang and Sternberg are proposing a high-throughput screening strategy to do just that—to exhaustively identify splicing-regulatory elements for any gene.

Herbert Irving Comprehensive Cancer Center Award:

"Biological and Therapeutic Relevance of Exosomes in Uveal Melanoma"; Lead Investigator: Richard Carvajal, MD; Co-PIs: Alex Rai, MD; Grazia Ambrosini, PhD

Dr. Carvajal, alongside Drs. Rai and Ambrosini, are working towards identifying a treatment strategy that can prevent the development of metastatic uveal melanoma (UM). UM is a rare melanoma that is distinct from those that start in the pigment producing cells of the skin. Recent analyses of UM patients have shown an increase of proteins contained with exosomes, small vesicles or blisters released from the cell. Cancer-derived exosomes contribute to cancer development and progression, making them both a potential indicator of disease and an opportunity for intervention. The researchers will further assess the role of the exosomes in UM disease progression. The end goal is to identify one or more lead treatment strategies to prevent the development of metastatic disease and devise a clinical trial for patients at high risk for disease recurrence.

Irving Institute for Clinical and Translational Research Award:

"A microRNA Approach to Identify Renal Osteodystrophy Sub-Type"; Lead Investigator: Thomas Nickolas, MD, MS; Co-PIs: Stavroula Kousteni, PhD; Krzysztof Kiryluk, MD, MS

Together, with his collaborators Drs. Kousteni and Kiryluk, Dr. Nickolas is tackling renal osteodystrophy (ROD), a disorder that weakens the skeleton, resulting in bone loss, fractures, and cardiovascular complications. ROD can be classified based on changes in bone turnover rates as high-turnover ROD (markedly elevated) or low-turnover ROD (markedly suppressed). Currently, treatment of ROD focuses on stopping high-turnover ROD, while also avoiding the development of low-turnover ROD that can occur through excessive use of these treatments. There currently is a strong need for a better system of diagnosing bone turnover rate in patients in order to better manage disease treatment. The team believes circulating fragments of cellular RNA called microRNAs (miRNAs) can assess turnover types in ROD. They are looking to identify miRNA profiles in order to test them as biomarkers of ROD turnover-type, positively impacting the diagnosis and management of ROD.

Roy and Diana Vagelos Mouse Genome Editing Awards

The supplemental funding for mouse models is made possible by the generous gift of Roy and Diana Vagelos, to the CPMI, and it is intended to support ground-breaking research in the field of precision medicine.

G protein signaling in a new neurodevelopmental disorder: a molecular and neuronal investigation of GNB1 Encephalopathy. **David Goldstein**, Professor, Director, Institute for Genomic Medicine

DDX10as a Precision Medicine target for the Acute Respiratory Distress Syndrome (ARDS). **Jahar Bhattacharya, MD, DPhil**, Professor of Medicine Division of Pulmonary, Allergy and Critical Care Medicine, Department of Medicine

"Novel mouse models of human cardiomyopathy and skeletal myopathy based on a human FLNC c.7416_7418delGAA mutation" **Wendy K. Chung, MD, PhD**, Kennedy Professor of Pediatrics; **Carrie Welch, PhD**, Assistant Professor of Medical Sciences

The role of the RNA binding protein Pumilio1 in two different neurological diseases, **Vincenzo A. Gennarino, Ph.D.**, Genetics & Development, Pediatrics and Neurology



Precision Medicine and Society

In the past year, Columbia faculty have continued to explore the impact of precision medicine on diverse fields, including economics, law, the humanities, and sociology as part of Columbia's Precision Medicine and Society (PM&S) program within the University's overall precision medicine initiative. The program is directed by a Steering Committee of faculty chaired by Paul Appelbaum, MD and Gil Eyal, PhD.

Precision Medicine: Ethics, Politics and Culture (PMEPC)

Jointly sponsored by the Center for the Study of Social Difference and the PM&S program, PMEPC funds a number of graduate fellowships and a series of scholarly events. During the 2019-20 academic year, Maya Sabatello, JD, PhD and Gil Eyal, PhD invited four scholars to the University to give public lectures and hold workshops with PMEPC fellows on topics ranging from the use of polygenic risk scores in education to the debate about the relevance of genomics in racial classification in South East Asia.

Prof. Sabatello is co-leading the series with Prof. Eyal in 2020-21. To date, PMEPC has sponsored a presentation by Alison Bateman-House, PhD, MPH, MA, on ethical issues related to non-trial preapproval access to gene therapies. This year's program will include additional presentations by Catherine Bliss, PhD, on the relevance of genomic medicine to precision public health; Melanie Myers, PhD on assisting teens in decision-making about genomic results; Vence Bonham, Jr., JD, on how genetics is reckoning with the history of systemic racism; and Dan Navon, PhD, about social factors in the emergence of gene-first disease classification. You can find details on the series, including the dates of the 2019-20 and 2020-21 events [here](#).

After a successful 2019-20 program in which PMEPC sponsored six Graduate Fellows, we circulated a call for the 2020-21 PMEPC Graduate Fellowships at the beginning of the academic year and received 26 applications across departments and campuses. This high demand and the impressive quality of applications led us to rethink the number of positions we should offer. We subsequently decided to increase the number of Graduate Fellows from six to eight (with some additional support from the PM&S program). The 2019-20 Graduate Fellows were:

- Sonia Mendoza-Grey (Sociomedical Sciences)
- Larry Au (Sociology)
- Diana Garofalo (Public Health)
- Amy Weissenbach (Sociology)
- Irina Kulichenkova (Narrative Medicine)
- Sunny Jones (Systems Biology)

The 2020-21 Graduate Fellows are:

- Colby Lewis (Biostatistics)
- Bella Horton (Sociomedical Sciences)
- Sarah Adelman (Population and Family Health)
- Lulu Chen (Medicine)
- Bree Martin (Genetic Counseling)
- Ari Gaiper (Sociology)
- Clare Casey (Anthropology)
- Supriya Kapur (Sociomedical Science)

All of the Fellows are participating in our series of public talks and small group meetings. In addition, as was the case last year, each of Graduate Fellows will develop a publishable research paper relating to PM&S.

This combination of public talks, working group discussions and publications will encourage extensive engagement in PM&S issues among students in the upcoming year.

Precision Medicine & Society Publications

In addition to the previously published paper (in *Genetics in Medicine*) considering the likely impact of precision medicine on the physician-patient relationship, the members of the PM&S Steering Committee collaborated on a paper considering the role and implications of precision medicine in the Covid-19 pandemic, now out for journal review. Members of the Steering Committee also published articles addressing genetic stigmas in South Africa, ethical responsibilities for periodic reinterpretation of genetic data, the vulnerability of genetic laboratories to malpractice claims, perspectives of individuals with disabilities about receiving genetic test results, and attitudes toward genetic screening in the Latino and Ashkenazi Jewish communities. These publications can be found [here](#).

Precision Medicine & Society Events

Center for Research on Ethical, Legal & Social Implications of Psychiatric, Neurologic & Behavioral Genetics

During the 2019-20 academic year, the Center hosted nine scholarly presentations. Presenters included Eva Kittay, PhD, Dept. of Philosophy, Stony Brook; Gary Marchant, JD, College of Law, Arizona State; Josephine Johnston, LLB, MBHL, The Hastings Center; Angela Bradbury, MD, Dept. of Medicine, UPenn; Aaron Panofsky, PhD, Institute for Society and Genetics, UCLA; Pamela Sankar, PhD, Dept. of Medical Ethics and Health Policy, UPenn; Kathryn Tabb, PhD, Dept. of Philosophy, Bard College.

This year, the Center has sponsored two presentations to date and will present eight additional talks on a wide range of topics. You can see a listing of upcoming events below:

2021

- January 11: Kostas Kampourakis, PhD, Section of Biology, University of Geneva, Switzerland
- February 22: Susan Domchek, MD, Basser Center for BRCA, University of Pennsylvania
- March 15: Steven Joffe, MD, Depts. of Medical Ethics & Health Policy and Pediatrics, University of Pennsylvania
- April 19: David Veenstra, PhD, School of Pharmacy, University of Washington
- May 17: Daniel Geschwind, MD, Dept. of Neurology, UCLA
- June 14: Bettina Meiser, PhD, Psychosocial Research Group, University of New South Wales, Sydney, Australia (Note: this seminar will take place from 4-5pm)

All talks will take place online during 2020-21, 12:00 - 1:00pm. To receive a link to each talk, interested individuals should send an email to janee.frankel@nyspi.columbia.edu.

Precision Medicine & Society Seminars and Conferences

Following a successful two-day conference in April 2019 on Precision Medicine: Its Impact on Patients, Providers and Public Health, in May 2020, PM&S sponsored its second major conference focused on Precision Medicine & Society: International Perspectives. Scholars from around the world presented their views on precision medicine in their societies, the impact of precision medicine on health disparities and global health and economic, legal and regulatory issues. You can view the program, including the names and biographies of the speakers [here](#), and recordings of each panel from the 2020 conference [here](#).

In October, 2020, the PM&S program hosted a virtual seminar covering the topic: Is Precision Medicine Relevant in the Age of Covid-19? The four speakers were Dr. Scott Gottlieb, former Commissioner of the FDA, Dr. Teri Manolio, Director of Genomic Medicine at NHGRI, Professor James Heath, President of the Institute for Systems Biology, and Dr. Amy Zhou, Assistant Professor

in the Department of Sociology, Barnard College. The seminar was moderated by John Rowe from the Mailman School of Public Health. You may view the seminar program, including the biographies of the speakers [here](#), and the recording of the seminar in our video library [here](#).

Planning is under way for another major conference at Columbia in the Spring of 2021 that will feature the work of young investigators and emerging scholars on precision medicine and its impact on healthcare and society.

Precision Medicine & Society Seed Grants

During 2019-20, the PM&S program solicited applications jointly with the Center for Research on Ethical, Legal & Social Implications of Psychiatric, Neurologic & Behavioral Genetics for seed grants to support research on the social, legal, political and ethical dimensions of precision medicine. A sub-committee of the Steering Committee reviewed six applications and selected two to be funded; two others were funded by the Center. Funded projects focused on the potential harms of polygenic scores for educational attainment and on developing a legal framework for neurodata privacy protection in the context of genomic data regulation.

Funded projects:

Genomic Data Regulation: A legal framework for neurodata privacy protection. Rafael Yuste, Biological Sciences; George Hripcsak, Biomedical Informatics

On the potential harms of polygenic scores for educational attainment. Lucas J Matthews, Psychiatry

We are currently in the process of selecting projects to be supported in 2020-21 jointly by the PM&S program and the Center.

Educational Initiatives

Economics of Precision Medicine

Professor Bhaven Sampat developed and taught a new course on the Economics of Precision Medicine. The course drew about 25 students and will be offered again in the future.

Course description: Precision Medicine, typically defined as the targeting of therapies according to individual characteristics, has generated considerable enthusiasm and drawn significant public and private investments. Tailored approaches to treatment (and prevention) are increasingly common, and many argue they are enabling a new forms of healthcare treatment and delivery and ushering in a new era of medicine. Whether the promise of precision medicine is realized, and its ultimate impact on health, will depend not only on scientific and technological advances, but also economic incentives. This course provides an overview of the basic economics of precision medicine, drawing on literature on precision medicine as well as lessons from the broader literature on the economics of pharmaceuticals, healthcare, and biomedical innovation. We will bring an economic perspective to understanding the incentives for developing precision medicines, their impact on costs, access, and healthcare outcomes, and to examine how public policies (public funding, FDA regulation, intellectual property, insurance, pricing) can shape the rate and direction of advances in precision medicine.

