The Faculty of Medicine of Harvard University
Curriculum Vitae

Date Prepared:	June 14, 2024
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Education:

05/1990	BA	Biochemistry	Cornell University Ithaca, NY
05/1996	PhD	Genetics (Dr. Rudolph Leibel)	Rockefeller University New York, NY
05/1998	MD	Medicine	Cornell University Medical College Ithaca, NY

Postdoctoral Training:

6/96- 5/97	Postdoctoral Fellow	*Genetics of obesity in rodents and man* (Dr. Rudolph L. Leibel)	Laboratory of Human Behavior and Metabolism, Rockefeller University
7/98-6/99	Intern	Pediatrics	Columbia Presbyterian Medical Center (CPMC)
7/99-6/00	PGY2 Resident	Pediatrics	CPMC
7/00-6/02	Fellow	Clinical Genetics	Division of Clinical Genetics, Department of Pediatrics, CPMC
7/02-6/03	Fellow	Molecular Genetics	Division of Clinical Genetics, Department of Pediatrics, CPMC

Faculty Academic Appointments:

1997-1998	Lecturer	Pediatrics (Molecular Genetics)	Columbia University
1998-2002	Associate Research Scientist	Pediatrics (Molecular Genetics)	Columbia University
1998-2002	Guest Investigator	Laboratory of Human Behavior and Metabolism	Rockefeller University
2002-2013	Assistant Professor of Pediatrics in Medicine	Pediatrics (Molecular Genetics)	Columbia University
2013-2015	Associate Professor of Pediatrics in Medicine with tenure	Pediatrics	Columbia University
2015-2017	Kennedy Family Associate Professor of Pediatrics in Medicine, with tenure	Pediatrics	Columbia University
2017-2023	Kennedy Family Professor of Pediatrics in Medicine, with tenure	Pediatrics	Columbia University
2023-2024	Faculty	Pediatrics	Harvard Medical School
2024-	Professor, without limit of time	Pediatrics	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions:

2002-2023	Attending Physician	Pediatrics and Medicine	New York Presbyterian Hospital
2007-2009	Consultant	Pediatric Genetics	The Valley Hospital Ridgewood, NJ
2023-	Chief of Pediatrics	Pediatrics	Boston Children's Hospital
2023-	Associate Member		Broad Institute

Other Professional Positions:

2001-	Member, Medical Advisory Board	Children's Cardiomyopathy Foundation	8 hours / year
2006-2018	Member, Medical Advisory Board	Association for Glycogen Storage Disease	

2012-2015	Member, Genetics in Primary Care Institute Quality Improvement Innovation Network	American Academy of Pediatrics	
2012-2016	Member	NIGMS Human Genetic Cell Repository (HGCR)	
2012-2016	Member, Genomics & Society Working Group (GSWG)	National Advisory Council for Human Genome Research (NACHGR)	
2012-2023	Director of Clinical Research	SFARI Simons Foundation	
2013-	Member, Scientific Advisory Board	Regeneron Genetics Center	4 hours / year
2014-	Member, Research Council	von Hippel Lindau Alliance	4 hours / year
2014-	Member, Clinical Council	von Hippel Lindau Alliance	4 hours / year
2014-	Member, Advisory Council	Hastings Center	4 hours / year
2017-	Member, Scientific Advisory Board	Jordan's Guardian Angels	8 hours / year
2017-	Affiliate Member	New York Genome Center	1 hour / month
2017-	Member, Scientific Advisory Board	PACS1	2 hours / year
2017-	Member, Scientific Advisory Board	HNRNPN2	4 hours / year
2018-	Member, Scientific Advisory Board	CSNK2A1	8 hours / year
2023-	Member, Life Science Advisory Committee	Empire State Development (ESD)	4 hours / year
2024-	Member, Scientific Advisory Board	Hope for Rare Foundation	4 hours/ year
2024-	Member	Genetic Metabolic Diseases Advisory Committee in the Center for Drug Evaluation and Research at the U.S. Food and Drug Administration	16 hours/year

Major Administrative Leadership Positions:

Local

2001-2023	Section Organizer, Medical Genetics (Science Basic to the Practice of Medicine)	Columbia University
2002-2023	Course Director, Genetics (Science Basic to the Practice of Medicine) Columbia University	Columbia University
2002	Course Director, Molecular Genetics for the Practicing Clinician (CME)	Columbia University
2003-2013	Director, Clinical Genetics	Columbia University
2003-2013	Chief, Division of Clinical Genetics	Columbia University Medical Center (CUMC)
2003-2023	Director, Clinical Cancer Genetics	Columbia University
2004	Course Director, How to Integrate Advances in Genetics into your Clinical Practice (CME)	Columbia University
2004, 2007	Course Director, Neonatology: Recent Advances in Neonatal Intensive Care Unit	Columbia University
2005-2009	Section Organizer, Biochemistry/metabolism (Science Basic to the Practice of Medicine)	Columbia University
2006-2017	Director, Molecular and Cytogenetics Fellowship Program	Columbia University
2008	Course Director, Fetal Diagnosis and Treatment, 6th Annual Sloane Conference (CME)	Columbia University
2010-2023	Co-Director, Medical Genetics Training Fellowship	Columbia University
2014-2023	Resource Director, Precision Medicine, Irving Institute for Translational Research	Columbia University
2015-2023	Course Director, Precision Medicine	Columbia University
2016-2018	Director, TL1 Training Program in the Clinical and Translational Science Awards (CTSA) Program	Columbia University
2016-2023	Co-Director	NY Obesity Research Center (NYORC) Molecular Biology Core
2019-2023	Associate Director for Education, Herbert Irving Comprehensive Cancer Center	Columbia University

2019-2023	Medical Co-director, Genetic Counseling Graduate Program	Columbia University
2020-2023	Chief, Clinical Genetics	Columbia University Medical Center

National

2022-	Chair, Data and Safety Monitoring Board (DSMB)	IGNITE (Implementing Genomics in Practice)

Committee Service:

Local

2002-	Naomi Berrie Diabetes Center	Columbia University Member
2003-2023	Continuing Medical Education Advisory Committee	Columbia University Member
2004-2006	Committee for Medical Education in Genetics	Columbia University Member
2004-	Herbert Irving Cancer Center	Columbia University Member
2005-	Motor Neuron Center	Columbia University Member
2005-2013	MSCHONY Laboratory Committee	Columbia University Chair
2005-2023	Molecular Diagnostics Laboratory Committee	New York Presbyterian Hospital (NYPH) Member
2006	Committee on Genetic Testing	Columbia University Medical Center (CUMC)
2006-	Center for Human Genetics	Columbia University Member
2006-2023	Innovative Diagnostics and Therapeutics	NYPH Member
2006-	Center for Bioethics Steering Committee	Columbia University Member
2006-2023	Pharmacogenetic Committee	NYPH Member
2007-2023	First Year Medical Student Faculty Committee	Columbia University Member
2007-2023	Department of Pediatrics, Recruitment Committee	Columbia University Member
2007-2023	Medical Advisory Board	Columbia IVF Center

		Member
2007-2023	CTSA Advisory Committee	Columbia University Member
2009-2023	P&S Evaluation Subcommittee	Columbia University Member
2009-2023	P&S Fundamentals Faculty Committee	Columbia University Member
2009-2023	AOA committee	Columbia University Member
2009-2023	Advisory Board for the Patient-Oriented Research Master's Program (MS/POR) of the Mailman School of Public Health	Columbia University Member
2010-2023	Center for the Study of Science and Religion	Columbia University Board of Advisors Member
2012	Strategic Planning Committee for Research	Columbia University Member
2013	Director of Personalized Medicine Search Committee	Columbia University Member
2014-2016	Precision Medicine Planning Committee	Columbia University Member
2014-2023	Columbia Medical Review	Columbia University Advisor
2019-2021	Search Committee for Dean of Columbia Medical School	Columbia University Member
2019-2023	Institutional Conflict of Interest Committee	Columbia University Member

National

2019-2022	National Human Genome Research Institute (NHGRI) Council	NIH Member
2020-2022	NHGRI Extramural Training and Career Development Program	NIH Research Training Expert Panel (RTEP)
2021-	Newborn Screening Translational Research Network Steering Committee (NBSTRN)	NICHD Co-chair
2021-	All of Us	NIH Research Program Advisory Panel
2023-	Committee on Autism Care Demonstration Program	National Academies of Science, Engineering, and Medicine
2024-	Newborn Screening Outreach and Dissemination	National Academies of Science, Engineering, and Medicine

Professional Societies:

1990-	American Association for the Advancement of Science	Member
1993-2013	American Diabetes Association	Member
1998-	American Society of Human Genetics 2022 2022-	Member Treasurer-elect Member, Board of Directors
2003-	American College of Medical Genetics	Member
2008-2023	Glenda Garvey Teaching Academy	Member
2010-	Society for Pediatric Research	Member
2012-2023	Virginia Apgar Academy of Educators	Member
2014-	American Society of Clinical Investigation	Member
2020-	National Academy of Medicine	Member
2021-	Association of American Physicians	Member
2021-	American Pediatric Society	Member

Grant Review Activities:

2005-2015	Genetic grants	American Heart Association Ad hoc
2010-2015	Genomic medicine	National Human Genome Research Institute Ad hoc
2010-2015	Genetic grants	Qatar National Research Fund Ad hoc
2018-2023	Training grants and loan repayment grants	National Cancer Institute Ad hoc
2023	Birth defects grants	Center for Disease Control

Editorial Activities:

- **Ad hoc Reviewer**

  American Journal of Human Genetics
  American Medical Journal of Genetics
  Clinical Genetics
  Circulation
  Circulation Research
  Genetics in Medicine
  JCI Insight
  Journal of Clinical Endocrinology and Metabolism

Journal of Inherited Metabolic Disease
Journal of the Academy of Clinical Cardiology
Journal of the American Medical Association
Leukemia Research
Neurogenetics
Neurology
New England Journal of Medicine
Obesity
Obesity Research
Prenatal Diagnosis
Proceedings of the National Academy of Science
Public Health Genomics

- **Other Editorial Roles**

2015-2024	Editorial Board Member	Molecular Case Studies
2015-	Board of Consulting Editors	JCI Insight
2020-2022	Editorial Board Member	The American Journal of Human Genetics
2022-	Advisory Board	Cell Genomics
2024-	Editorial Advisory Committee Member	Journals of the American College of Medical Genetics and Genomics

Honors and Prizes:

1986	Westinghouse Science Talent Search, 1st place	
1986	National Merit Scholar	
1986-1990	Cornell Scholar; Dean's List	Cornell University
1988	Summer training grant	National Science Foundation (NSF)
1990	Phi Beta Kappa Outstanding College Students of America Phi Kappa Phi Golden Key Honor Society The National Dean's List	Cornell University
1992	Outstanding Student Research Award	American Institute of Nutrition
1994	Louis Gibofsky Memorial Prize	Cornell University Medical College
1995, 1998	Dean's Research Award	Cornell University Medical College

2001	Young Investigator Research Grant Award	American Academy of Pediatrics
2005	Best Translational Research	Columbia University Department of Pediatrics Assistant Professor Research Symposium
2008	Charles W. Bohmfalk Award for Distinguished Contributions to Teaching in the Clinical Years	Columbia University Medical College
2008	Distinguished Lecturer, Class of 2011	Columbia University
2008	Medical Achievement Award	Bonei Olam
2009	Presidential Award for Outstanding Teaching	Columbia University
2010	Distinguished Lecturer of the Year, Class of 2013	Columbia University
2011	Distinguished Lecturer of the Year, Class of 2014	Columbia University
2012	Inductee, Dade County Hall of Fame	Dade County
2014	Dean's Distinguished Lecture in the Clinical Sciences	Columbia University
2014	Best Paper in 2013	Science Unbound Foundation
2014	Samberg Scholars in Children's Health	New York Presbyterian Hospital
2017	Best Grand Rounds of the Year	Department of Pediatrics, Columbia University
2018	Fundamentals Outstanding Teacher Award, Class of 2021	Columbia University
2018	Medal for Distinguished Contributions in Biomedical Science	New York Academy

2019	Mentor of the Year Award	College of Dental Medicine, Columbia University
2019	2019 Rare Impact Award	National Organization for Rare Disorders (NORD)
2019	The Robyn Barst Lecture Award	Pulmonary Hypertension Association
2022	Quality and Patient Safety Recognition Award Columbia Doctors	Columbia University
2024	BCRP Equity, Diversity, and Inclusion Honor Roll	Boston Children's Hospital

Report of Funded and Unfunded Projects

Past

2001-2002 Characterization of a new murine neurological mutant Hcn2^{ap}
American Academy of Pediatrics: Young Investigator's Research Award
PI ($50,000)
The goal of this project is to electrophysiologically characterize a new murine neurological mutant *Hcn2^{ap}*.

2001-2002 Identification of Novel Genes and Pathways in Type 2 Diabetes Using N-Ethyl-N-Nitrosurea (ENU) Pilot Project
New York Obesity Research Center (NYORC)
PI ($50,000)
The major goal of this project is to screen ENU mutagenized mice for diabetes and determine if mice the hyperglycemia is transmitted as a monogenic trait for the eventual purpose of diabetes gene identification.

2001-2004 Children's Health Research Center
NIH – NICHD P30 HD34611
PI ($100,000)
The goal of this project is to electrophysiologically characterize a new murine neurological mutant Hcn2^{ap}.

2002-2008 BMPR2 Mutations in Pulmonary Hypertension
NIH - NHLBI R01 HL060056
PI ($250,000)
The goal of this study is to characterize the nature and frequency of mutations in BMPR2 in pulmonary hypertension, correlate genotype with phenotype, and determine if genotype is correlated with response to therapy.

2003-2004 Naomi Berrie Diabetes Research Fellow
Naomi Berrie Diabetes Center, Columbia University
PI ($100,000)

The goal of this project is to genetically characterize obese subjects for genes for obesity.

2003-2004 BRCA Founder Mutations among Jewish Participants of the Long Island Breast Cancer Study Project
Women at Risk**,** Columbia Presbyterian Medical Center
Co-PI (PI: R. Senie - $10,000)
The goal of this project is to test all participants in the Long Island breast cancer study for the three Ashkenazi BRCA1 and BRCA2 founder mutations to determine if this is responsible for the increased incidence of breast cancer in this cohort.

2003-2004 The role of genetic polymorphisms in the regulation of cardiac hypertrophy
Office of Clinical Trials, Columbia University
Co-PI (PI: S. Mital - $30,000)
The goal of this project is to determine if there are genetic factors that are responsible for the differential response of patients with congenital heart disease and Cardiomyopathy to cardiac hypertrophy.

2003-2004 BRCA Founder Mutations among Jewish Participants of the Long Island Breast Cancer Study Project
Herbert Irving Comprehensive Cancer Center Pilot Funding Awards (PI: Chung)
PI **(**$20,000)
The goal of this project is to test all participants in the Long Island breast cancer study for the three Ashkenazi BRCA1 and BRCA2 founder mutations to determine if this is responsible for the increased incidence of breast cancer in this cohort.

2003-2007 Genetics Core Laboratory for the Pediatric Heart Disease Clinical Research Network
Pediatric Heart Network
PI ($142,875)
The goal of this project is to serve as the genetics core for the multi-site pediatric heart network that was established to study cardiac disease unique to children. The genetics core will bank DNA samples on all study participants and perform genotypic analysis relevant to the clinical studies.

2003-2008 Cloning of a Type 2 Diabetes Modifier in Obese Mice
NIH – NIDDK DK066518
Co-PI (PI: Leibel - $375,000)
The goal of this project is to clone a quantitative trait locus that predisposes mice with monogenic obesity due to mutations in *leptin* to type 2 diabetes.

2004-2004 How to Integrate Advances in Genetics into Clinical Practice
March of Dimes Birth Defects Foundation, Grant No. 4-FY04-43
PI ($5,000)
The goal of this project is for continuing medication education for medical professionals on "How to Integrate Advances in Genetics into Clinical Practice."

2004-2007 Irving Center for Clinical Research, Irving Scholars Program
Columbia University
PI ($60,000/year)
The goal of this project is to identify novel human genes predisposing to early onset type 2 diabetes in Dominicans.

2004-2009 Cardiovascular Development and Disease in the Young

NIH NHLBI 1T32 HL076116
Co-I (PI: Rosen - $557,100)
Training grant for pediatric cardiology post-doctoral fellows.

2004-2022 Pediatric Neuromuscular Clinical Research Network for SMA Clinical Trials
Spinal Muscular Atrophy Foundation
Co-I (PI: DeVivo $210,556)
The goal of this project is to establish a clinical research network that clinically and molecularly characterizes patients with spinal muscular atrophy at baseline and establishes methods of monitoring clinical efficacy in preparation for SMA clinical trials.

2005-2008 Identification of Novel Germline Breast Cancer Susceptibility Genes in High Risk Ashkenazi Jewish Families
Manhasset Women's Coalition Against Breast Cancer
PI ($100,000)
The goal of this research is the identification of a novel breast cancer susceptibility gene in Ashkenazi Jewish families with hereditary breast cancer who do not harbor mutations in BRCA1 or BRCA2.

2005-2009 Identification of a Novel Breast Cancer Susceptibility Gene in the Ashkenazi Jewish Population
Fuirst Foundation
PI ($650,000)
The goal of this project is to initially map and then clone a novel gene for breast cancer susceptibility by testing in Ashkenazi Jewish families with hereditary breast cancer who do not harbor mutations in BRCA1 or BRCA2.

2006-2007 Identification of Genetic Modifiers of *BRCA1* or *BRCA2* in Ashkenazi Mutation Carriers
Women at Risk, Columbia Presbyterian Medical Center
PI ($15,000)
The goal of this project is to test the effect of polymorphisms in genes involved in DNA repair with founder Ashkenazi mutations that may interact with BRCA1 and BRCA2 to modify the risk of cancer.

2006-2008 Studies of Pharmacological Modulation of Survival Motor Neuron in a Mouse Model of Spinal Muscular Atrophy PGI Mouse Studies
Spinal Muscular Atrophy Foundation
PI ($55,157)
The goal of the project is to test pharmacological agents *in vivo* in mouse models of spinal muscular atrophy for clinical efficacy and effect on biomarkers of survival motor neuron modulation including *SMN* gene expression and protein production.

2006-2008 CD36: A Putative Taste Receptor for Dietary Fat in Humans
New York Obesity Research Center (NYORC) Pilot Project
Co-I (PI: Keller - $15,000)
The goal is to genetically determine the CD36 genotypes and haplotypes in subjects with varying taste preference for dietary fats.

2006-2009 Survival Motor Neuron Protein assay
Westat RFP 8079-05-03
PI ($17,863)

This is a Phase I/II clinical trial of phenylbutyrate for the treatment of spinal muscular atrophy. This proposal is to serve as the core facility to measure the biomarker, survival motor neuron protein from blood in this clinical trial.

2006-2010 Genome Scans in Congenital Heart Disease using ROMA
NIH – NHLBI HL080146-02
Co-I (PI: Warburton - $570,761)
The goal of this project is to genomically characterize subjects with hypoplastic left heart syndrome or conotruncal heart defects using Representational Oligonucleotide Microarray Analysis (ROMA) to determine the locations of novel genes associated with these types of congenital heart disease and develop methods of improving prognostication for outcomes and associated birth defects and neurocognitive deficits in subjects with congenital heart disease.

2006-2011 Metropolitan New York Registry of Breast Cancer Families
NIH - NCI U01CA069398
Co-I (PI: Terry - $453,607/year)
The goal of the project is to collect and study families with multiple cases of breast and/or ovarian cancer and to study genetic and environmental factors influencing cancer susceptibility, clinical outcomes, identify high risk individuals for prevention trials, and study health behaviors.

2007-2008 Views and Approaches toward Pre-implantation Genetic Diagnosis (PGD) and Barriers to Its Use Among Providers and Patients
Co-I (PI: Klitzman - $50,000)
The goal of this project is to understand utilization of preimplantation genetic diagnosis as a reproductive option and identify barriers to implementation.

2007-2008 GATHER: Genetic Arrythmia Testing Helping Evaluate Risk
Columbia CTSA Pilot grant
Co-I (PI: Hickey - $25,000)
The goal of this project is to develop screeners to identify patients most likely to benefit from genetic testing for inherited arrhythmias.

2007-2009 Conversations in Genetics: Development of Educational DVDs to Teach Medical Genetics
Glenda Garvey Testing Academy at Columbia University
PI ($15,400)
The goal of this project is to develop an educational library of videotapes of patients who have and are undergoing genetic testing for a variety of disorders to teach medical, dental, and nursing students how to effectively educate and counsel patients about genetic testing.

2007-2010 Copy number variation in *SMN1* and *SMN2* in humans and murine models of ALS
Motor Neuron Center
PI ($80,000)
To assess *SMN1* and *SMN2* genotype in modifying ALS age of onset and severity of disease.

2007-2010 Doris Duke Charitable Foundation Clinical Scientist Development Award
PI ($135,000)

The goal of this research is the identification of a novel breast cancer susceptibility gene in Ashkenazi Jewish families with hereditary breast cancer who do not harbor mutations in BRCA1 or BRCA2.

2008-2008 Effects of the Histone Deacetylase Inhibitor LBH589 on *In Vitro* Transcription and Translation of Survival Motor Neuron in Spinal Muscular Atrophy
PI ($100,000)
The goal of this research project is to test the effects of the novel histone deacteylase inhibitor LBH589 on SMN production in fibroblasts from patients with spinal muscular atrophy.

2008-2008 Studies of Pharmacological Modulation of Survival Motor Neuron in a Mouse Model of Spinal Muscular Atrophy
Families of Spinal Muscular Atrophy (#34646)
PI ($15,000)
The goal of the project is to test pharmacological agents *in vivo* in mouse models of spinal muscular atrophy for clinical efficacy and effect on *SMN* gene expression and protein production.

2008-2009 Optimization of the A2 scaffold, which upregulates SMN protein
SMA Foundation
Collaborator (PI: Stockwell - $100,000/year)
The goal of this research project is to translate a hit that emerged from a screen into a drug lead for SMA.

2008-2009 John M. Driscoll, Jr. Children's Research Fund
Columbia University
PI ($40,000)
The goal of this research is to characterize the underlying genetic basis for cardiomyopathy in children.

2008-2009 Provider and Patient Views and Approaches Toward PGD Use
Greenwall Foundation
Co-I (PI: Klitzman)
The goal of this research project is to understand patient and medical providers' views about preimplantation genetic diagnosis and identify barriers and facilitators of its use.

2008-2013 Identification of Novel Genes for Congenital Diaphragmatic Hernia by Characterizing Genetic Copy Number Alterations
NIH NICHD R01 HD057036-01A1
PI ($512,270/year NCE)
The goal of this study is to identify genes causing congenital diaphragmatic hernia by assessing genetic copy number on a genome wide basis of oligonucleotide arrays.

2008-2013 Diabetes and Endocrine Research Center
NIH - NIDDK P30 DK063608-10
Co-I (PI: Accilli - $901,002)
Molecular Biology/Molecular Genetics Core. The goal of this project is to establish a research center with common interests and expertise in diabetes and endocrinology.

2009-2010 Irving Institute Collaborative and Multidisciplinary Pilot Research (CaMPR) Award: An Interdisciplinary Collaboration to Create a Biobank to Enable Personalized Medicine at Columbia University

PI ($125,000)
The goal of this research is to pilot a biobank in cardiology as a model for a Columbia University biobank.

2009-2011 CNV Atlas of Human Development
NIH – NICHD RC2 HD064525-01
Co-I (PI: Ledbetter/Wapner - $259,603)
The goals of the two year project are to develop the processes and infrastructure for ongoing collection of a large number of high-quality genome wide array data and the associated phenotypic findings. In the process of developing these processes we will contribute genotypic and phenotypic data on 4,000 prenatal cases and 10-15,000 pediatric cases.

2009-2012 Spinal Muscular Atrophy (SMA): Disease Phenotype and Mechanisms
U.S. Department of Defense
Co-PI (Co-PI: Henderson - $2,925,000 total)
To assess the pathology of specific muscle groups in SMA patients.

2009-2013 Molecular Genetic Analysis of Human Obesity
NIH – NIDDK DK52431-16
Co-PI (Co-PI: Leibel - $333,099)
The goal of the project is to identify the genes that mediate susceptibility to obesity in humans.

2010-2013 Genetics of the Brain and Behavior
Center for ELSI Research on Psychiatric, Neurologic, and Behavioral Genetics
NIH – NHGRI P20 HG005535-01
Co-I (PI: Appelbaum - $160,000/year)
The goal of this project is to plan a center of ELSI scholars to conduct research and train the next generation of research related to genetic conditions involving the brain and behavior.

2011-2012 Clinical and Translational Science Award Supplement
NIH – NCRR 3UL 1RR024156-06S2
PI ($300,000)
Establishment of a Biobank to facilitate translational research.

2011-2013 Identification of Novel Genes for Infantile Cardiomyopathy
Children's Cardiomyopathy Foundation
PI ($100,000/year)
The goal of this study is to identify novel genes for cardiomyopathy presenting in infancy.

2011-2013 Challenges of Informed Consent in Return of Data from Genomic Research
NIH - NHGRI R21 HG006596
Co-I (PI: Appelbaum)
The goal of this study is to assess models for consent in genetic research studies that allows for return of individual genetic results.

2011-2015 Impact of Return of Incidental Genetic Test Results to Research Participants in Genomic Studies
NIH – NHGRI 5R01 HG006600-03
PI ($375,417)

The goal of the study is to understand how to consent and return incidental research results to participants in genetic research studies.

2011-2015 LEGACY: A cohort of youth in families from the Breast Cancer Family Registry
NIH - NCI 5R01 CA138822 -05 $ 392,954
Co- I (PI: Terry)
The goal of this study is to identify risk factors during childhood and adolescence that confer lifetime risk of breast cancer.

2011-2016 A twin study of obesity pathogenesis using fMRI (PI: Schur); (Leibel subcontract PI)
NIH- NIDDK R01 DK089036-04
Co-I (PI: Schur - $250,000); (Leibel subcontract PI - $28,959)
Studies how the brain regulation of appetite may be altered by genetic and/or environmental risk factors for obesity.

2011-2017 Genes, Environment, and Breast Cancer Risk: The 15-year follow-up of the Breast Cancer Family Registry
NIH – NCI 1R01CA159868-05
Co-I (PI: Terry, Hopper - $1,968,016)
The goal of this study is to gather long term longitudinal clinical data including new cancer diagnoses and outcomes in a large cohorts for breast cancer.

2011-2022 Gene Mutation and Rescue in Human Diaphragmatic Hernia
NIH/NICHD 1P01HD068250 Program Project
Co-I (PI: Donahoe)
The goal is to uncover the mutations causing Congenital Diaphragmatic Hernia by linkage, gene expression, bioinformatic prioritization of genes and proteins, exome/genome sequencing of probands & trios, and functional work in multiple animal models.

2012-2017 Psychosocial Impact of Genetics in Epilepsy
NIH – NINDS R01NS078419-04
Co-I (PI: Ottman - $343,783)
The goal of the study is to understand the psychosocial impact of establishing a genetic diagnosis for epilepsy in a longstanding research cohort.

2012-2017 Prenatal Cytogenetic Diagnosis by Array-based copy number Analysis
NIH – NICHD 5 U01 HD055651-09
Co-I (PI: Wapner - $ 1,572,369)
The goal of this study is to assess the diagnostic yield and methods to implement cytogenomics in the prenatal setting.

2012-2017 Hormonal, Metabolic and Signaling Interactions in Pulmonary Arterial Hypertension
NIH -NHLBI HL108800-04
Co-I (PI: Loyd - $1,782,360; $54,151 subcontract)
The goal of the study is to understand the genetic and hormonal factors contributing to pulmonary arterial hypertension risk.

2012-2017 Breast Cancer Family Registry Cohort
NIH –NCI UM1 CA164920
Co-I (PI: Terry, Hopper, Andrulis, Daly, John - $447,837)
The goal of this study is to gather long term longitudinal clinical data including new cancer diagnoses and outcomes in a large cohort for breast cancer.

2012-2023 Breast Cancer Family Cohort
NIH/NHLBI U01CA1649204
Co-I (PI: Terry)
The goal of this study is to gather long term longitudinal clinical data including new cancer diagnoses and outcomes in a large cohorts for breast cancer.

2013-2018 Center for Research on the Ethical, Legal and Social Implications of Psychiatric, Neurologic and Behavioral Genetics
NIH - NHGRI 1P50HG007257-01
Co-I (PI: Appelbaum - $716,651)
The goal of this project is to establish a center of ELSI scholars to conduct research and train the next generation of research related to genetic conditions involving the brain and behavior.

2013-2018 Functional imaging and eating behavior among FTO genotypes in pre-obese children
NIH – NIDDK R01 DK097399
Co-I (PIs: Rosenbaum and Mayer - $399,670)
The goal of this study is to understand the ingestive behavior for individuals with a single genetic risk factor for obesity, FTO genotype.

2014-2017 Newborn screening for Spinal Muscular Atrophy
Biogen Idec
PI ($758,000)
The goal is to pilot a newborn screening study for SMA in New York state.

2014-2018 Returning Genetic Research Panel Results for Breast Cancer Susceptibility
NIH - NCI/NHGRI R01CA190871-02
Subcontract PI/Co-I (PI: Bradbury - $398,917)
The goal is to return genetic results for hereditary cancer to a research cohort.

2014-2020 PVDOMICS: Defining the Future Fingerprints of Pulmonary Vascular Disease
NIH – NHLBI U01 HL125218 -05
Co-I (PIs: Berman-Rosenzweig and Horn - $260,813)
The goal of this pulmonary vascular disease (PVD) NOMICS study to systemically characterize WHO Groups 1- 5 pulmonary hypertension (PH) patients utilizing clinical, biochemical, imaging, physiological and pathological assessments combined with genomic and RNA technology to improve our mechanistic and pathobiological understanding of the pulmonary vascular disease process.

2015-2018 Goals and Practices for Next Generation Prenatal Testing
NIH- NHGRI 1R01 HG008805-01A1
Co-I (PI: Johnston - $264,157; subcontract to Columbia - $23,540)
The goal is to learn how to implement prenatal genetic testing using DNA sequencing.

2015-2019 Genomic analysis of congenital diaphragmatic hernia
NIH –NHLBI 1X01 HL132366-01
HL136998-01
HL140543
Co-PI (Co-PI: Shen -in kind sequencing, no funds)
The goal is to elucidate the underlying genomic architecture of CDH by performing whole genome sequencing on parent child trios and RNA sequencing of diaphragm

tissue in a clinically well characterized cohort to identify *de novo* mutations and inherited rare variants.

2015-2020 Columbia GENIE (GENomic Integration with EHR)
NIH - NHGRI 1U01HG008680-01
Co-I (PI: Weng, Hripcsak, Gharavi - $540,000)
This project uses genomic knowledge for disease prevention and health improvement.

2016-2017 Strengthening Public Health Infrastructure for Improved Health Outcomes
Columbia/Cornell/Harlem Hospital Precision Medicine Initiative HPO
1UG3OD023183-01
Co-I (PIs: Goldstein, Rubin, Hripcsak, Gharavi, Kaushal, Ross - $3,716,357)
The goal of this project is to build a research cohort to enroll 10,000 subjects in the national PMI biobank.

2016-2019 NRSA Training Grant
NIH 1TL1TR001875-01
PI ($530,464)
Goal: to provide training stipends to research fellows in precision medicine with a goal to recruit, train, support and nurture the next generation of clinical and translational investigators in multi- and interdisciplinary team science environments.

2016-2019 The Virome of Manhattan: A Testbed for Radically Advancing Understanding and Forecast of Viral Respiratory Infections (DARPA)
BAA-US Army
Co-I (PI: Shaman - $11,998,963)
The goal is to develop a method for viral surveillance for respiratory infections.

2016-2021 Molecular Genetic Analysis of Human Obesity
NCE NIH/NIDDK R01 DK52431-23
MPI (Leibel and Chung - $302,145)
The major goal of this project is to identify the genes that mediate susceptibility to obesity in humans.

2016-2021 Molecular approaches to gene identification in congenital heart disease
NIH – NHLBI U01 HL098163-01
PI ($85,000)
The goal of the project is to identify the genes that mediate susceptibility to congenital heart disease in humans.

2016-2021 Clinical and Translational Science Award U54
NCATS/NIH U54 TR00187 3-01
Co-I (PI: Ginsberg/Reilly - $8,261,483)
The goal of the Irving Institute CTSA is to transform the culture of biomedical research enabling CUMC investigators to develop new treatments faster and deliver those treatments to patients more efficiently, and safely than ever before; to utilize medical research advances to benefit patients and the community, converting knowledge into practice; and to recruit, train, support and nurture the next generation of clinical and translational investigators in multi- and interdisciplinary team science environments.

2017-2018 The Impact of Genetic Testing for Cardiomyopathies in Children and Their Families
Children's Cardiomyopathy Foundation

PI ($7,250)
The goal of this study is to understand the families' perspectives in genetic testing for children with a personal or family history of cardiomyopathy.

2017-2018 PTEN-CFTR interactions regulate pulmonary inflammation in Cystic Fibrosis – a Potential Target for Therapy
Irving Institute/Integrating Special Populations (ISP) Pilot Award
Co-I (PI: Prince - $40,000)
The goal is to understand the role of PTEN in disease pathogenesis of cystic fibrosis.

2017-2020 Decision Support for BRCA Testing in Ethnically Diverse Women
ACS RSG-17-103-01-CPPB
Co-I (PI: Kukafka - $280,736)
The objective of this proposal is to expand genetic testing for HBOC to a broader population of high-risk women by prompting appropriate referrals from the primary care setting with the use of an electronic health record-embedded breast cancer risk navigation (BNAV) tool.

2018-2021 Health Care Provider Responses to Receiving Unsolicited Genomic Results (HCP) proposal
NIH 1 R01 HG010004-01A1
Co-I (PI: Holms - $23,482)
The goal of this study is to understand the perspective of health care providers when results of a genetic research study are returned to their patients.

2018-2023 Clinical Characterization of PPP2R5D Mutations
UNIVERSITY OF CALIFORNIA, DAVIS/UCAL CU17-2559
PI ($77,589)
The research and the development and dissemination of information is related to the mutation in gene PPP2R5D.

2018-2023 Psychosocial Impact of Genetics in Epilepsy
5R01NS104076-03
Co-I (PI: Ottman - $499,819)
Goal: This study focuses on understanding the psychosocial impacts of genetic causal attribution in the epilepsies.

2018-2023 Deep Phenotyping in Electronic Health Records for Genomic Medicine
NLM/NHGRI 1R01LM012895-01
Co-I (PI: Weng/Wang)
The goal of this project is to develop data science and informatics methods to accelerate deep phenotyping using the unstructured data in electronic health records for genomic diagnostic decision support and genomic knowledge discovery.

2019-2021 Integrate Gene Expression Data to Characterize the Contribution of Rare Genetic Risk Factors to Structural Birth Defects
NIH R03HL147197
Co-I (PI: Shen - $100,000)
Goals: This project aims to discover new risk genes and elucidate the genetic architecture of structural birth defects. We propose to use cross-disease genetic

analysis of both protein-coding and noncoding variants and integrate gene expression data to prioritize candidate risk genes.

2019-2022 Newborn screening for Duchenne Muscular Dystrophy
Parent Project Muscular Dystrophy
Co-I (PI: Caganna - $200,000)
Goal is to pilot newborn screening for Duchenne Muscular Dystrophy.

2019-2022 Identification of genes for congenital heart disease in a consanguineous community
Saving Tiny Hearts Society
PI ($75,000)
Congenital heart disease gene identification in Palestinian families.

2020-2021 A novel biomarker to improve risk-prediction in familial breast cancer patients
DOH01-C34925GG-3450000
Co-I (PI: Dalerba - $359,899)
The goal of this study is to elucidate the clinical utility of this novel biomarker in familial breast cancer patients (n=737) from the New York site of the Breast Cancer Family Registry (BCFR). The study envisions three specific aims: Aim-1: to test whether, among BRCAX patients, high levels of biomarker expression associate with functional inactivation of BRCA1; Aim-2: to test whether, among BRCAX patients, high levels of biomarker expression associate with increased risk of second tumors and reduced survival; Aim-3: to test whether, among BRCAX patients, high levels of biomarker expression can be used to improve the predictive accuracy of clinical algorithms used to estimate the risk of second tumors.

2020-2022 AADC patient identification
PTC Therapeutics GT, Inc.
PI ($32,213)
Goal: This project is to use electronic health records (EHR) to identify previously undiagnosed AADC patients and recontact and test the identified suspicious AADC patients.

2020-2023 CURE Spinal Muscular Atrophy
Co-I (PI: De Vivo)
To confirm the SMN1 and SMN2 genotypes on all individuals in the biorepository either through review of medical records or by directly assessing genotype.

2021-2022 Muscular Dystrophies Diagnostic Decision Support Using EHR
Sarepta Therapeutics, 2020-RMS-GRT-1303
Co-I (PI: Weng $29,530)
Goal: This project aims to increase healthcare provider awareness and decrease the time to diagnosis of muscular dystrophies by using a systematic process to develop, validate, and deploy augmented intelligence tools identifying potential MD patients from electronic health records (EHR) and provide clinical decision support (CDS) to physicians in the form of educational materials, evidence-based guidelines for screening recommendations for specialist referral.

2021-2023 A Multi-site Observational Study of Post-Acute Sequelae of SARS-CoV-2 Infection in Pediatric Populations
NIH/NHLBI OT2HL161847
Co-I (MPI: Stockewell, Berman-Rosenzweig, Millner)

The goal is to characterize the long term clinical symptoms of SARS-CoV-2 infection in children.

2021-2023 Clinical and Translational Science Award
NCATS/NIH 2UL1TR001873-07
Co-I ($8,261,483)
The goal of the Irving Institute CTSA is to transform the culture of biomedical research enabling CUMC investigators to develop new treatments faster and deliver those treatments to patients more efficiently, effectively, and safely than ever before; to utilize medical research advances to benefit patients and the community, converting knowledge into practice; and to recruit, train, support and nurture the next generation of clinical and translational investigators in multi- and interdisciplinary team science environments.

2016-2023 The Molecular Genetic Analysis of human obesity
NIH/NIDDK / 5R01DK052431-25 – NCE
PI ($295,225)
The goal of this project is to identify the genes that mediate susceptibility to obesity in humans.

2017-2023 Gene Mutation and Rescue in Human Diaphragmatic Hernia
NICHD 1P01HD068250-06A1 (Donahue)
PI ($150,000 annual)
Goal: Genomic and gene expression analyses to discover human CDH genes and pathways.

2018-2023 Center for Research on the Ethical, Legal and Social Implications of Psychiatric, Neurologic and Behavioral Genetics
NHGRI 2RM1HG007257
Co-I (PI: Appelbaum - $704,918)
The goal is to support a center to promote research and training on ELSI issues in psychiatric, neurologic and behavioral genetics.

2018-2023 Development of Recommendations and Policies for Genetic Variant Reclassification
NIH 1 R01 HG010365
PI ($545,145)
The goal is to identify the relevant ethical principles and their potential impact on the formulation of an approach to variant reinterpretation, and countervailing considerations that may shape the nature of an ethical duty.

2020-2023 Treatments for neurogenetic disorders
Ovid Therapeutics
PI ($925,926)
The goal is to establish a supported collaboration program for the development of treatments for patients with neurogenetic disorders.

2020-2024 COVID Recovery Corps
Chan Zuckerberg Initiative Foundation CZIF2020-004123
PI ($1,300,462)
Goal: To support COVID Recovery Corps research: a research study and registry to engage COVID-19 survivors directly in research through structured surveys, widespread antibody testing, return of individual results, and ongoing educational outreach and feedback.

2020-2025 Cancer Center Support Grant
NIH/NCI P30 CA013696

Associate Director for Education and Training (PI: Rustgi)
Goal: To support the NIH-designated Herbert Irving Comprehensive Cancer Center (HICCC).

2010-2023 Simons Foundation Powering Autism Research for Knowledge
Simons Foundation 337701
PI ($165,764 Chung study site) ($6,700,000/year across all centers)
The goal of this project is to characterize patients with genetic causes of autism and neurodevelopmental disorders.

Current

2016-2026 New York Obesity Research Center
NIH – NIDDK P30 DK026687
Co-I (PI: Leibel - $749,848) ($143,626 Molecular Biology Core)
This core provides assistance to qualified investigators in the application of molecular biology & molecular genetic techniques to studies of energy metabolism in animals and man. Responsible for the supervising genotyping and sequencing within the molecular genetics core and providing consultation to investigators in study design and consultation and instruction to users.

2016-2027 Developmental Mechanisms of Trachea-Esophageal Birth Defects
NIH/NICHD 5P01P01HD093363-01 (Zorn)
PI ($110,656 annual)
The goal is to coordinate activities of the CARE study including recruitment and clinical characterization of esophageal atresia/tracheoesophageal fistula in patient in the CARE network and analyze and interpret genomic data.

2018-2023 Prenatal Genetic Diagnosis by Genomic Sequencing
NCE NIH/NICHHD/ELSI R01 HD055651
Co-I (PI: Wapner, Chung - $1,572,369)
Goal: to continue investigations of the use of molecular cytogenetic testing by array copy number analysis in prenatal diagnostic testing.

2018-2026 Center for Identification and Study of Individuals with Atypical Diabetes Mellitus
NIH/NIDDK U54DK118612
Site Co-I (PI: Phillipson - $75,587)
The goal is to identify genetic causes of atypical diabetes and characterize the clinical phenotypes.

2018-2023 Screening for Cardiac Amyloidosis with Nuclear Imaging in Minority Populations
NCE NIH/NHLBI R01 HL139671 SCAN-MP
Co-I (PI: Maurer - $1, 181,768)
The goal is to identify the frequency of TTR mutations in African Americans and to determine the penetrance of amyloidosis in TTR mutation carriers.

2019-2024 ELSI.hub: National Center for ELSI Resources and Analysis
NIH/NHGRI 1U24HG010733
Co-I (PI: Cho/Lee - $947,376)
Major goals: To support a center that will serve as a locus for resource sharing and community building to enhance the production, sharing, and use of research on the ethical, legal, and social implications of genetics and genomics (ELSI research), using

the "knowledge to action" conceptual framework which highlights facilitators of and barriers to knowledge sharing and use.

2019-2024 EHR-based Genomic Risk Assessment and Management for Diverse Populations
NIH 2U01HG008680
MPI (PI: Weng, Hripcsak, Kiryluk, Chung-$945,000)
The goal is to develop and clinically implement genomic integrated risk scores for 10 common conditions in adults in 4 common conditions in children and assess how participants and providers utilize this information.

2020-2024 Role of the Kinesin KIF1A in Neurological Disease (MPI: Vallee, Chung)
NIH/NINDS 1R01NS114636
PI responsible for all human studies (MPI: Vallee, Chung)
Goal is to understand KIF1A neurodevelopmental disorders and test novel therapeutic strategies.

2020-2024 Impact of receiving Alzheimer's Disease Genetic Risk information among Latinos in northern Manhattan
NIH/NIA R01 AG062528
Co-I (PI: Ottman, Chung - $1,591,612)
Goal: To assess the psychosocial, behavioral, and cognitive impact of receiving personal risk information about Alzheimer's disease based on APOE genotypes among Latinos residing in northern Manhattan.

2020-2024 Disability, Diversity and Trust in Precision Medicine Research: Stakeholders' Engagement
NIH/ NHGRI R01HG010868-04
Co-I ($3,190,962)
Goal: To study trust in and trustworthiness of precision medicine research among disability and scientific communities.

2020-2025 Identifying and applying genetic variation relevant to clinical outcomes for individuals with congenital heart disease
NHLBI U01HL131003
PI (MPI: CU: Chung, Shen /MSSM: Gelb - $100,000)
The goal of this study is to determine the genetic contributions to clinical outcomes in individuals with CHD and to begin to use this information in clinical care.

2020-2025 Identifying and applying genetic variation relevant to clinical outcomes for individuals with congenital heart disease
1U01 HL153009
MPI (MPI: CU: Chung, Shen /MSSM: Gelb - $275,000)
The goal of this study is to determine the genetic contributions to clinical outcomes in individuals with CHD and to begin to use this information in clinical care and to design better clinical trials of treatments for CHD.

2021-2024 Simons Variation in Individuals Project (Simons VIP)
Simons Searchlight-225718
PI ($397,634)
Goal: The 16p11.2 deletion is the most common genetic disorder associated with autistic spectrum disorder (ASD).

2021-2024 ClinGen Expert Curation Panel for Severe Structural Anomalies and Stillbirth

NIH U24HD104588
Co-I (PI: Wapner - $1,068,752)
Birth defects are a leading cause of perinatal, infant, and childhood morbidity and mortality. Recent advances in ultrasound imaging now identify these anomalies in utero and increasingly more sophisticated genomic testing such as sequencing allows for increased understanding of the underlying etiology and improved options for care.

2021-2026 Molecular approaches to gene identification in congenital heart disease
NIH U01HL131003
Co-I ($70,000 annual)
The goal of this study is to determine the genetic contributions to CHD.

2022-2026 Prenatal air pollution and neurodevelopment: a longitudinal neuroimaging study of mechanisms and early risk for ADHD in Puerto Rican children
NIH/NIEHS 1R01 ES032870-01A1
Co-I ($3,940,710)
This study seeks to understand the relationship between prenatal maternal air pollution exposure and offspring risk for ADHD and alterations in neurodevelopment in an intergenerational cohort of Puerto Ricans and examine two potential -modifiable- mechanisms: prenatal maternal inflammation and offspring sleep problems.

2022-2027 Rescue: Rare Disease Detection and Escalation Support via a Learning Health System
1R01HG012655-01
Co-I
In this study, we will build a SMART-on-FHIR based Rare Disease Detection and Escalation Support (RESCUE) CDSS. It will use a centralized informatics approach to identify suspected rare disease patients from clinical data warehouse (CDW) and send alerts to physicians with escalation support including phenotype summarization, genetic/genomic test requisition and research opportunity discovery.

2022-2027 Prospective Genetic Risk Evaluation and Assessment (PROGRESS) in Autism
NIH /NICHD P50HD109879
MPI Chung and Venstra-Vanderweele ($11,734,750)
Goal: The goal of the autism center grant is to identify and study a diverse, population-based cohort of infants with monogenic risk for autism to evaluate the impact of early life identification of genomic risk variants on parent experience, neurodevelopmental trajectories, and prediction of autism diagnosis.

2023-2028 Breast Cancer Family Registry
NCI 2U01 CA164920-11
Co-I ($11,191,964)
Goal: The Breast Cancer Family Registry (BCFR) Cohort is an international cohort in the U.S., Canada and Australia comprised of multi-generational families (33,037 women and 6,992 men from 15,056 families) that started in 1995. We will strengthen and continue to provide to the research community an important and unique long-term family cohort with extensive epidemiologic and molecular data to address cutting-edge and clinically important research questions on breast cancer susceptibility, outcomes, survival and survivorship with the overall goal of advancing knowledge of the biology of breast cancer development and progression so as to reduce the cancer burden and cancer disparities.

2023-2028 Fair Phenotype Annotation and Genomic Reinterpretation

NHGRI R01 HG013031
MPI (PI: Weng. Chung. Wang - $886,418)
Our overarching goal is to design a scalable and sustainable informatics framework to support continuous genomic reanalysis for symptomatic patients with non-diagnostic exome or genome sequencing in diverse populations.

2023-2024 Etiology and pathogenesis of lethal lung developmental disorders in neonates
NIH /NHLBI 1R01 HL165301
Co-investigator (PI: Stankiewicz - $793,392)
This study aims to identify the genetic basis of developmental lung disorders and describe the clinical and pathological features.

2024 GUARDIAN 2 planning grant
CHAN ZUCKERBERG INITIATIVE Project Number: 332495
PI: Chung $250,000
This planning grant is to plan for GUARDIAN newborn screening in the Commonwealth of Massachusetts

2024-2026 SPARK and Simons Searchlight Cohorts
Simons Foundation
PI: Chung $24,000,000
This grant supports the SPARK cohort for autism and the Simons Searchlight cohort for rare neurogenetic conditions.

2024 GUARDIAN (Genomic Uniform-screening Against Rare Disease In All Newborns)
Ultragenyx
PI: Chung $25,000.00
This grant supports the GUARDIAN study providing expanded newborn screening to diagnose conditions for early treatment to improve outcomes.

Training Grants and Mentored Trainee Grants

1978-2028 Postdoctoral Training in Arteriosclerosis Research
NHLBI 2T32HL007343-46
Faculty mentor
Provide training to postdoctoral fellows in arteriosclerosis research.

1980-2025 Translational Research Training in Child Psychiatry
NIMH 5T32MH016434-43
Faculty mentor
The mission of our training program is to train investigators in the methods and techniques of contemporary, multidisciplinary research that will improve knowledge of the causal pathways that produce psychiatric disorders in children, and how to use that knowledge to develop and deliver interventions that more effectively prevent, manage, or cure those disorders, and thereby improve the mental and emotional well-being of children and their families.

1981-2026 Short Term Training Grant
NHLBI 5T35HL007616-42

Faculty mentor
Provides training to medical students over the summer for research experiences.

1989-2027 Obesity Research Center Training Grant
2T32DK007559-32
Faculty mentor
This T32 post-doctoral training program, now in its 26th year, provides 2-3 years of fellowship designed to prepare physicians and PhDs for investigative careers in the area of obesity.

1990-2025 Graduate Training in Nutrition
NIDDK 5T32DK007647-33
Faculty mentor
Provide training to graduate students in nutrition

1992-2027 Training in Biomedical Informatics at Columbia University
NLM 2T15LM007079-31
Faculty mentor
Columbia University's biomedical informatics training program seeks to advance the discipline of biomedical informatics by providing a broad and rigorous formal course exposure paired with intense research training in a strong health-focused environment.

1996-2026 Postdoctoral Training in Cardiovascular Disease
NHLBI 5T32HL007854-27
Faculty mentor
This application requests funding for the second competitive renewal of a postdoctoral training program in cardiovascular diseases. Initially the training program was designed for surgical residents, to prepare for an investigative career in cardiovascular sciences.

2004-2025 Training Grant in Pediatric Endocrinology, Diabetes and Metabolism
NIDDK 5T32DK065522-18
Faculty mentor
This program provides training to fellows in Pediatric Endocrinology, Diabetes and Metabolism at Columbia University, College of Physicians & Surgeons.

2009-2024 BEST-DP: Biostatistics & Epidemiology Summer Training Diversity Program
NHLBI 5R25HL096260-15
Faculty mentor
The BEST (Biostatistics and Epidemiology Summer Training) Diversity Program provides research opportunities in the quantitative health sciences of biostatistics and epidemiology, as applied to heart, lung, blood, and sleep (HLBS) research. Our target audience comprises undergraduates who are under-represented in biomedical research (those from disadvantaged backgrounds, racial and ethnic minorities, and individuals with disabilities), and who will contribute to a more diverse research workforce in the future.

2009-2024 Multidisciplinary Training in Translational Gastrointestinal and Liver Research
NIDDK 5T32DK083256-14
Faculty mentor
The program's mission is to train MD and MD/PhD trainees to become independent basic, clinical and translational researchers in gastroenterology and hepatology.

2012-2027 Training Medical Students in NIDDK Research
NIDDK 5T35DK093430-12
Faculty mentor
Training medical students to do biomedical research.

2013-2028 Brief Research In Aging and Interdisciplinary Neurosciences (BRAIN)
NIA 2T35AG044303-11
Faculty mentor
The Department of Neurology at Columbia University Medical Center serves as a rich site for multidisciplinary neurological research, with particular focus on disorders associated with the aging nervous system. In this proposal, the Brief Research in Aging and Interdisciplinary Neurosciences (BRAIN) program, we have developed a comprehensive approach to develop a formal research program for predoctoral students early in developing careers in biomedical, behavioral and clinical research.

2014-2023 TRAINING IN CARDIOVASCULAR TRANSLATIONAL RESEARCH
NHLBI 5T32HL120826-10
Faculty mentor
This application requests funding for a pre-doctoral (4 slots) and post-doctoral (4 slots) training grant entitled, 'Training in Cardiovascular Translational Research'. This training grant application is uniquely designed to train future CV scientists who will have expertise in bringing basic discoveries from the laboratory into clinical practice through development of novel therapeutics.

2016-2026 Clinical and Translational Science Award (NRSA Training Core)
NCATS 5TL1TR001875-07
Faculty mentor
Our goal is to establish the TRANSFORM TL1 Precision Medicine (PM) Program to provide training and mentoring in the methods and applications of PM to pre-docs, post-docs, junior faculty, and a wide range of research personnel.

2016-2026 Molecular Oncology Training Program
NCI 5T32CA203703-07
Faculty mentor
This is a new proposal to establish a training program at Columbia University focused on training physicians in research techniques that will form the basis of careers in translational investigation of cancer biology, diagnosis and treatment.

2021-2026 Hormones: Molecular Mechanism of Action and Functions
NIDDK 5T32DK007328-42
Faculty mentor
Provide training to postdoctoral fellows in endocrinology about hormone function.

2021-2026 Genetic Approaches to Development and Disease
5T32GM141882-02
Faculty mentor
This proposal describes a new PhD training program, Genetic Approaches to Development and Disease (GADD) at Columbia University Irving Medical Center (CUIMC), which trains young scientists in the use of modern genetics to address major challenges in biomedical research.

2022-2027 Training in Cellular, Molecular and Biomedical Studies (CMBS)
NIGMS 1T32GM145766-01

	Faculty mentor The Integrated Program in Cellular, Molecular and Biomedical Studies (CMBS) is an umbrella program that presents students with a unique opportunity to obtain individualized training in all aspects of biomedical sciences, including basic cell and molecular biology, microbiology, structural biology, biophysics, genetics, immunology, neurobiology, systems and computational biology, as well as translational biomedical disease-related research.
2022-2027	Training in Health Equity, Highlighting Environmental Inequities, & Growing neighborHood Teachers and Students (YES in THE HEIGHTS) NCI 1R25CA274180-01 Faculty mentor The mission of this program at the Herbert Irving Comprehensive Cancer Center (HICCC) is to reduce the cancer burden and cancer health inequities in the HICCC Catchment Area (CA) through training and mentoring of students and teachers to increase the diversity of future cancer researchers.
2023-2027	Child Health Research Career Development Award (CHRCDA) Program (K12) 5K12HD052896-17 Faculty mentor The goal of the Boston Children's Hospital K12 CHRCDA Program is to develop independent pediatrician physician-scientists who will decipher the pathobiology of childhood disease and develop transformative new therapies.

Report of Local Teaching and Training

Teaching of Students in Courses:

2002-2023	Elective in Medical Genetics 4th year medical students	Columbia University 20 hours/year
2003	Breast Cancer Graduate students	School of Public Health, Columbia University 2 hours/year
2003-2014	Medical Genetics in Pediatrics 3rd year medical students	Columbia University 5 hours/year
2003-2015	Ethics in Medical Genetics 4th year medical students	Columbia University 2 hours/year
2004	Genetic Approaches to Biological Problems Graduate students in Genetics and Development	Columbia University 2 hours/year
2005-2012	Teratology, Human Development 1st year medical students	Columbia University 2 hours/year
2005-2015	Oncogenetics Human genetics clinical training program	Columbia University 2 hours/year

2006-2014	Incorporating Genetics into Advanced Nursing Practice, Nursing N8290.001 Cardiac genetics/ Diabetes genetics Nursing students offered spring and summer semesters	Columbia University 5 hours/year
2007-2009	Ethics and Experimentation Graduate students	Columbia University 2 hours/year
2007-2009	Ethics in Genetics Research Graduate students	Columbia University 2 hours/year
2007-2014	Practicum in Genetics, Nursing N8165 Genomic medicine Nursing students	Columbia University 20 hours/year
2008-2010	Mechanisms of Human Disease Graduate students	Columbia University 3 hours/year
2009-2012	Molecular Nutrition Master's students in Institute of Human Nutrition	Columbia University 2 hours/year
2010-2012	Nutrition; Genetics of Diabetes lecture Nutrition graduate students	Columbia University 2 hours/year
2010-2023	Pharmacology Journal Club Pharmacology graduate students	Columbia University 2 hours/year
2010-2023	Pharmacogenetics Pharmacology graduate students	Columbia University 2 hours/year
2012-2015	Ethics and genetics Graduate students	Columbia University 1 hour/year
2013-2023	Genetics and the Law Law students	Columbia University 2 hours/year
2017-2023	Human Genetics Graduate students	NY Genome Center 3 hours/year
2017-2023	Human Genetics and Development Graduate students	Columbia University 2 hours/year
2017-2023	Precision Medicine Graduate students	Columbia University 15 hours/year
2017-2023	BIOL G4305 Seminar for the MA in Biotechnology Master's students	Columbia University 8 hours/year

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs):

2002-2015	Conference on pediatric genetics House staff	Columbia University 2 lectures/year
2002-2023	Conference on cancer genetics	Columbia University

	Oncology fellows	2 lectures/year
2002-2023	Conference on cardiac genetics Cardiology fellows	Columbia University 2 lectures/year
2002-2023	Conference on neurogenetics Neurology fellows'	Columbia University 1 lecture/year
2002-2023	Conference on psychiatric genetics Psychiatry fellows	Columbia University 1 lectures/year

Clinical Supervisory and Training Responsibilities:

2002- 2023	Supervision in the genetics clinic Residents and fellows	Columbia University 8 hours/week

Formally Mentored Harvard Students (Medical, Dental, Graduate, and Undergraduate):

Columbia University medical student mentoring

2007-2008	Wendy Chang, Research fellow 2 publications resulting from research
2008-2009	Kelly Burke, Research fellow 2 publications resulting from research
2008-2009	Laura Brenner, Doris Duke fellow 2 publications resulting from research
2011-2012	Christian Rose, Doris Duke fellow
2014-2015	Alexandra Coromilas, scholarly project 2 publications resulting from research
2015	Emily Webster 1 publication resulting from research
2015-2016	Stephanie Bronfman, scholarly project
2015-2016	Heidi Lumish, scholarly project 3 publications resulting from research
2016	Ian Halim 1 publication resulting from research
2016	Stefano Iantorno
2016	Vlad Velicu
2016	Akshay Save
2016	Christopher Dambrosia
2016	Anoushka Sinha 1 publication resulting from research
2016	Diana Stern Publication resulting from research

2017	Brigitte Kazzi 2 publications resulting from research
2017	Talia Weitz Publication resulting from research
2017	Linda Wang Publication resulting from research
2017	Phillip Allen Publication resulting from research
2018	Michael Artin Publication resulting from research
2018	Jonah Tischler Publication resulting from research
2018	Ronald Laracuente Publication resulting from research
2018	Andrew Thorton
2018	Alice Mei
2018	Joseph Grimes Publication resulting from research
2018	Anne Reed-Weston Publication resulting from research
2018	Kirsten Craddock Publication resulting from research
2018	Mary Nattakom Publication resulting from research
2018	Sonya Besagar
2019	Sam Bruce
2019	Lily Lao
2019	Jonathan Tiao
2020	Ashley Kahenkashani
2020	Saundra Albers
2020	Juliana Nitis
2020	Sarah Wyckoff
2020	Catherine Jennings Publication resulting from research
2020	Ayla Safran
2020	Abigayle Dolmseth
2021	Kimberly Peloza

2021	Bethany Onyirimba
2021	Rebecca Weitz
2021	Allison Rosenbaum 2 publications resulting from research
2021	Amy Lipman Publication resulting from research
2021	Alice Tao Publication resulting from research
2021-2022	Catherine Kernie, scholarly project Publication resulting from research
2022	Alina Andrews
2022	Joseph Ryu
2023	Ryan J Cohen
2023	Akina Sanyang
2023	Sneha Sharma

Columbia University dental student mentoring

2016	Maria Fontana Publication resulting from research
2016	Tomer Madar
2016	David Holland Publication resulting from research
2017	Anna Szentirmai
2018	Deanna Noble Publication resulting from research
2018	Josue Diaz-Melendez
2019	Nikita Chintalapudi
2019	Emily Horowitz
2019	Jennifer Shahar
2019	Parker Green
2019	Bobby Lin Publication resulting from research
2019	Ashley Kahen Publication resulting from research
2020	Dana Dobrowski

2020 Madison Garrity

2020 Leelah Weitz

2021 Goldi Weiser

2022 Julian Mis

2022 Shukran Babkir

2022 Neil Ming
Publication resulting from research

2023 Tongzhuo Chang

2023 Parina Bhuva

2023 Gursimran Grewal

2023 Andrew Manahan

2023 Micole Zeidman

2023 Jay Patel

Columbia University graduate student mentoring

2002-2005 Marija Dokmanovich, Institute of Human Nutrition doctoral thesis committee
Publication resulting from research

2000-2005 Loan Phan, Institute of Human Nutrition doctoral thesis committee
Publication resulting from research

2003-2004 Rachel Dominguez, Sarah Lawrence College, genetic counseling master's student thesis advisor

2003-2005 Sara Bretschger, Institute of Human Nutrition doctoral thesis committee
Publication resulting from research

2006 Elaine Budreck, MD PhD student: Clinical Contact During the Lab Years

2006-2007 Mariko Welch, Institute of Human Nutrition, graduate student thesis advisor
Publication resulting from research

2006-2007 Ashley Wilson, Sarah Lawrence College, genetic counseling master's student thesis advisor
Publication resulting from research

2007 Jeffrey Douglass, nursing master's student, Clinical Genetics mentor

2007 Anne O'Donnell, MD PhD student: Clinical Contact During the Lab Years mentor. Continued mentorship and now a medical geneticist and researcher.

2007-2008 David Malito, TRANSFORM mentor
Publication resulting from research

2010-2012	Kelly Ruggles, TRANSFORM mentor, Institute of Human Nutrition, doctoral thesis committee
2010-2012	Pelisa Charles-Horvath, Department of Pharmacology, doctoral thesis committee
2010-2015	Richard Gill, School of Public Health, thesis advisor Publication resulting from research
2012	Jacqueline McCray, Master's student Biotechnology, Department of Biological Sciences, thesis advisor.
2012-2014	Justin Lee, Integrated Program, Qualifying exam committee chair.
2013	Ettie Lipner, Genetic Epidemiology, Thesis committee.
2014	Sindhuri Prakash, MD PhD student, Integrated program, TRANSFORM mentor
2014-2017	Michael Bohnen, MD PhD student, Integrated program, thesis committee. Publication resulting from research
2015-2020	Alexander Hsieh, Systems Biology, graduate student Publication resulting from research
2016-2021	Bryan J. Gonzalez, graduate student, Institute of Human Nutrition thesis committee
2016-2021	Lia Boyle, Integrated Program, thesis advisor Publication resulting from research
2018-	Bulat Ziganshin, Genetics and Development, thesis advisor
2020	George Timmins, master's student, School of Public Health Publication resulting from research
2020	Siying Chen, graduate student, Systems Biology, thesis committee
2021	Archana Kumar, Master of Biotechnology, thesis advisor
2021-2022	Jessica de Voest, George Washington University, PhD thesis committee Publication resulting from research
2021-2024	Yige Zhao, Systems Biology, thesis committee Publication resulting from research
2022	Marek Svoboda, MD PhD program, Dartmouth, thesis committee
2022-	Guojie Zhong, Systems Biology, thesis committee Publication resulting from research

Other Mentored Trainees and Faculty:

Fellow mentoring

2005-2008	Sheila Carroll, MD / Associate Professor, Cornell University Career stage: Cardiology fellow. Mentoring role: Fellowship advisor. Accomplishments: 1 publication.

2007-2008 Amy Jean, MD / Assistant Professor of Pediatrics, University of Pittsburgh School of Medicine
Career stage: Endocrine fellow. Mentoring role: Fellowship advisor.
Accomplishments: 1 publication, 1 first author.

2007-2013 Teresa Lee, MD / Assistant Professor, Columbia University
Career stage: Genetics and cardiology fellow. Mentoring role: Fellowship mentor.
Accomplishments: 15 publications, 5 first author.

2008-2009 Rushika Conroy, MD / Associate Professor, University of Massachusetts
Career stage: Endocrine fellow. Mentoring role: Fellowship advisor.
Accomplishments: 1 publication.

2008-2013 Aimee Lucas, MD / Associate Professor Mount Sinai
Career stage: GI fellow. Mentoring role: Fellowship advisor. Accomplishments: 3 publications, 3 first author.

2009-2011 Rachelle Gandica, MD / Assistant Professor, Columbia University
Career stage: Endocrine fellow. Mentoring role: Fellowship advisor.
Accomplishments: 1 publication, 1 first author

2011-2013 Casey Overby, PhD / Assistant Professor of Medicine and Biomedical Engineering, Johns Hopkins University
Career stage: Post-doctoral fellow. Mentoring role: Mentor.
Accomplishments: 2 publications, 1 first author.

2012-2013 Lea Tuzovic, MD / Obstetrician-Gynecologist, New Haven, CT
Career stage: Clinical genetics fellow. Mentoring role: Fellowship advisor.
Accomplishments: 2 publications, 2 first author.

2012-2014 Katrina Celis, MD / Associate Scientist, University of Miami
Career stage: Human genetics fellow. Mentoring role: Mentor.
Accomplishments: 3 publications, 1 first author.

2013-2014 Joanne Chiu, MD / Instructor, Harvard Medical School
Career stage: Pediatric cardiology fellow. Mentoring role: Fellowship advisor.
Accomplishments: 1 publication, first author.

2013-2016 Emily Breidbart, MD / Assistant Professor, NYU
Career stage: Endocrinology fellow. Mentoring role: Fellowship advisor.
Accomplishments: 1 publication, first author.

2014-2016 Preti Jain, PhD / Researcher, Hudson Alpha Institute for Biotechnology, Huntsville, AL
Career stage: Molecular genetics fellow. Mentoring role: Fellowship advisor.
Accomplishments: 2 publications.

2014-2016 Joseph Picoraro, MD / Assistant Professor, Columbia University
Career stage: Pediatric gastroenterology fellow. Mentoring role: Fellowship research mentor. Accomplishments: 4 publications, 1 first author.

2014-2016 Matthew Lewis, MD/ Assistant Professor, Columbia University
Career stage: Cardiology fellow. Mentoring role: Research mentor.
Accomplishments: 3 publications, 1 first author.

2017-2019 Abigail Carey, MD / Instructor, Yale University

Career stage: Pediatric intensive care fellow. Mentoring role: Fellowship research mentor. Accomplishments: 1 publication, first author.

2017-2020 Shannon Nees, MD / Assistant Professor, Nemours Children's Health
Career stage: Cardiology fellow. Mentoring role: Fellowship research mentor. Accomplishments: 4 publications.

2018-2019 Stephanie Kochav, MD, MHS / Cardiologist, Valley Health System
Career stage: Cardiology fellow. Mentoring role: Fellowship research advisor. Accomplishments: 2 publications.

Faculty mentoring

2007-2010 Vaidehi Jobanputra, PhD / Professor of Pathology and Cell Biology Columbia University
Career stage: Assistant professor. Mentoring role: Mentor K award. Accomplishments: 8 publications.

2009-2011 Kathleen Hickey, PhD / deceased
Career stage: assistant professor. Mentoring role Robert Wood Johnson Fellowship mentor. Accomplishments: 1 publication.

2010-2012 Susan Carnell, PhD. Associated Professor, Johns Hopkins University
Career stage: postdoctoral fellow. Mentoring role: K award mentor. Accomplishments: 1 presentation.

2010-2013 Jonathan Lu, MD PhD Translational Medicine and Early Clinical Development Head, Saliogen Therapeutics
Career stage: assistant professor. Mentoring role: K award mentor. Accomplishments: 1 presentation.

2010-2013 Mat Maurer, MD Professor, Columbia University
Career stage: associate professor. Mentoring role: K award mentor. Accomplishments: 5 publications.

2010-2013 Douglass Sproule, MD MSc, Chief Medical Officer ML Bio Solutions
Career stage: assistant professor, Mentoring role: K23 award mentor. Accomplishments: 9 publications.

2010-2013 Amanda Pong, MD Neurologist, Adventist HealthCare
Career stage: assistant professor. Mentoring role: K23 award mentor. Accomplishments: 2 publications.

2010-2013 Roy Alcalay, MD Associate Professor, Tel Aviv Soursky Medical Center
Career stage: assistant professor. Mentoring role: Brookdale Leadership in Aging Fellowship, K award mentor. Accomplishments: 14 publications.

2013-2023 Teresa Lee, MD Assistant Professor, Columbia University
Career stage: assistant professor. Mentoring role: K award mentor. Accomplishments: 16 publications.

2013-2023 Sharon Jones-Eversley, PhD Associate Professor, Towson University
Career stage: assistant professor. Mentoring role: NIH PRIDE and Diversity Supplement. Accomplishments: 1 grant submission.

2015-2023 Sylvie Goldman, PhD assistant professor Columbia University

Career stage: Assistant Professor. Mentoring role: Simons Searchlight research mentor. Accomplishments: 2 publications.

2018-2023 Jennifer Bain, MD PhD assistant professor, Columbia University
Career stage: Assistant Professor. Mentoring role: Simons Searchlight research mentor. Accomplishments: 5 publications.

Formal Teaching of Peers (e.g., CME and other continuing education courses):

No presentations below were sponsored by 3rd parties/outside entities

2002	Molecular Genetics for the Practicing Clinician (CME)	Single presentation Columbia University
2002-2023	Pediatrics	2 lectures / year Columbia University
2002-2023	Medicine	3 lectures / year Columbia University
2002-2023	Genetics	5 lectures / year Columbia University
2002-2023	Cardiology	1 lecture / year Columbia University
2002-2023	Oncology	1 lecture / year Columbia University
2002-2023	Gastroenterology	1 lecture / year Columbia University
2002-2023	Surgery	1 lecture / year Columbia University
2004	How to Integrate Advances in Genetics into your Clinical Practice (CME)	Single presentation Columbia University
2004, 2007	Neonatology: Recent Advances in Neonatal Intensive Care Unit	6 lectures American Austrian Foundation
2004, 2007	Genetics	6 lectures American Austrian Foundation
2008	Fetal Diagnosis and Treatment, 6th Annual Sloane Conference (CME)	Single presentation Columbia University
2012-2024	PRIDE: Genetic Epidemiology Faculty students	Single presentation Columbia University

Local Invited Presentations:

No presentations below were sponsored by 3rd parties/outside entities

2009 Monogenic forms of diabetes / Invited presentation
New York Obesity Research Center, New York, NY

2013 Advances in genetics of breast cancer / Invited presentation
Columbia University

2014 Advances in Neurogenetics / Grand Rounds
CUMC Neurology, Columbia University

2014 Developments in Genetics and Genomics in Neurology / Invited presentation
Genetic Testing in Neurological Disorders 2014: Developments and Dilemmas, Center for Excellence in ELSI Research, Annual Meeting, Columbia University New York, NY

2014 Utilizing Genomic Sequencing in Rare Diseases to Guide Clinical Care and Develop New Therapies / The Dean's Distinguished Lecture in the Clinical Sciences.
Columbia University

2014 Lessons Learned from Monogenic Forms of Diabetes. Frontiers in Diabetes Research: Next Wave Science in Diabetes and Obesity /Invited presentation
Columbia University

2015 New Frontiers in Pulmonary Hypertension and ECMO: Personalized medicine in PH: How genetics may change the field / Invited presentation
Columbia University

2016 Prenatal Testing and PNB Traits today. Which Variants Associated with PNB Traits are being Detected Prenatally and Returned to Prospective Parents, and what is on the Horizon? Detecting Variants Associated with PNB Traits at a Moment When Prenatal Testing and Newborn Screening May Be Converging / Invited presentation
Columbia Medical Center

2016 Genetics, Biomarkers, and Connective Tissue Disorders / Invited presentation
Aortovascular Summit 2016: A Multidisciplinary Team Approach. Columbia University

2016 Precision Medicine for Cystic Fibrosis / The 38th Stephanie Lynn Kossoff Memorial Lecture
Columbia University

2017 Genetic "Dark Matter" of Human Energy Homeostasis: Gene Finding and Gene Vetting. Frontiers in Diabetes Research: Advances and Challenges in the Neuroscience of Ingestive Behaviors / Invited presentation
College of Physicians and Surgeons, Columbia University

2020 Pediatric genomic medicine. Inivted presentation
Genomic medicine series, Columbia University

2020 Cell and Gene Therapy: The Next Generation of Personalized Medicine /Invited presentation
16th Annual Healthcare Conference, Columbia Business School

2020 The Future of Genetics is Now. Integrating Genetics into Medical Practice / Invited presentation
Columbia University

2020 Frontiers in Diabetes Research Obesity, Diabetes and COVID-19: Elucidating Bidirectional Links / Inivted presentation
Columbia University

2021	Common Genetic Disorders Encountered in Internal Medicine / Practical Course. Columbia University
2022	The Future of ELSI: Genetics Interventions for Neurodevelopmental Disorders / Invited presentation Columbia University Irving Medical Center
2022	What zebras can teach us about horses: studies of rare genetic diseases / Invited presentation Tissue Talks, Columbia University. Online.

Report of Regional, National and International Invited Teaching and Presentations

Those presentations below sponsored by outside entities are so noted and the sponsor(s) is (are) identified.

Regional

2002	Nature or Nurture: The Role of Genes in Determining Adiposity / Invited presentation Naomi Berrie Fourth Annual Frontiers in Diabetes Research, New York, NY
2003	Addressing the Issues–How to Integrate Clinical Genetics into your Pediatric Practice / Invited presentation New York City Society of Nurse Practitioners, New York, NY
2003	Genes, Genetics, and the Human Genome Project Abyssinian / Seminar Baptist Church, New York, NY
2003	The Genetics of Breast Cancer / Seminar Weill Medical College of Cornell University, New York, NY
2003	Genetics for the General Practitioner / Invited presentation CHONY Pediatrics In Review The Fifth Annual Seminar, New York, NY
2004	Genetics in your Pediatric Practice / Pediatric Grand Rounds St. Barnabas Hospital, Bronx, NY
2004	Putting it all together: Case studies, diagnostic and therapeutic challenges in genetics / Pediatric Grand Rounds Roosevelt Hospital, New York, NY
2004	Medical Genetics for the General Pediatrician / Pediatric Grand Rounds Wyckoff Heights Medical Center, Brooklyn, NY
2004	Incorporating Genetics into Clinical Practice / Invited presentation Executive Health Examination, New York, NY
2004	Management of the High-Risk Patient: The Role of Genetics / Invited presentation Breast Cancer Controversies: Emerging Data, Evolving Strategies, New York, NY
2004	DNA Testing for VHL / Invited presentation VHL Family Alliance Membership, Annual Meeting, New York, NY
2004	Molecular Genetics: An introduction for the pediatrician / Pediatric Grand Rounds St. Barnabas Hospital, Bronx, NY
2005	Genetics of Syndromic and Monogenic Obesity / Pediatric Grand Rounds

Maimonides Medical Center, New York, NY

2005 Genetics and Congenital Heart Disease / Invited presentation
NYPH Adult Congenital Heart Association Seminar, New York, NY

2005 Genetics of Hereditary Breast Cancer / Invited presentation
Breast Cancer in Women of Color: Dispelling Myths, Learning the Facts, New York, NY

2005 A Geneticist's Perspective on the Electronic Medical Record as a Critical Investigative Research Tool. A Roundtable Discussion on the Opportunities and Challenges at the Crossroads of Health Information Technology and Biomedical Research / Invited presentation
United Hospital Fund, New York, NY

2006 How to integrate genetics into clinical practice to tailor care / Medicine Grand Rounds
St. Barnabas Hospital, Bronx, NY

2006 Genetic Research / Invited presentation
IRB Educational Conference: IRB Challenges and Practical Solutions, New York, NY

2006 Physicians Speak Out. Breast Cancer: Survive and Conquer / Invited presentation
Susan G. Komen Breast Cancer Foundation, New York, NY

2006 How to Use Genetic Testing for Breast Cancer to Tailor Women's Breast Care.
Breast Cancer: Survive and Conquer / Invited presentation
Susan G. Komen Breast Cancer Foundation. New York, NY

2006 Genetic Research Involving Newborns, Children, and Adolescents. Ethics of Genetics in Research: Perils and Promises / Invited presentation.
Genetics Task Force, New York, NY

2006 Genetic Basis of Inherited Arrhythmias / Invited presentation
Heart to Heart Cardiac Arrhythmia Research and Education Foundation, New York, NY

2006 Genetic Basis of Cardiac Disease in Children / Pediatric Grand Rounds
St. Vincent's Hospital, NY

2006 Diagnosis of Metabolic Cardiac Disease / Invited presentation
Metabolic Disorders and Heart Disease, New York, NY

2006 Breast cancer genetics. Breast cancer in the young woman: It's the same, but different / Invited lecture
Lighthouse International Conference Center, New York, NY

2007 Advances in Genetic Medicine / Medicine Grand Rounds
New York Downtown Hospital, New York, NY

2007 Genomic Imbalances in Birth Defects / Pediatric Grand Rounds
St. Barnabas Hospital, New York, NY

2007 Integration of Genetics into Medical Practice. Current Clinical Issues in Primary Care / Invited presentation
Pri-Med Conference, New York, NY

2007 Clinical Trials in Spinal Muscular Atrophy / Invited presentation

NYS Genetics Task Force, New York, NY

2007 Integration of Genetics into Medical Practice / Medicine Grand Rounds
St. John's Episcopal Hospital, Bloomfield, NJ

2007 Lessons from Monogenic Forms of Obesity / Invited presentation
Frontiers in Diabetes Research. Naomi Berrie Diabetes Center, New York, NY

2007 Update on Prenatal Diagnosis / Invited presentation
Pediatrics in Review: The Ninth Annual National Seminar, New York, NY

2008 Innovations in Genetics and Utilization in Your Practice / Pediatric Grand Rounds
New York University (NYU), New York, NY

2008 Genetic Evaluation of Sudden Cardiac Death / Invited presentation
Clinical Management of Children with Congenital Heart Disease: From Genetics to Transplantation, New York, NY

2008 Genetics of Pediatric Cardiomyopathy / Pediatric Grand Rounds
St. Barnabas Hospital, Bronx, NY

2008 The Genetics of Syndromic Obesities / Pediatric Endocrinology Symposium
Pediatric Endocrine Society, Newark, NJ

2008 Genetic Syndromes Associated with Congenital Heart Disease. Clinical Management of Children with Congenital Heart Disease: From Genetics to Transplantation / Invited presentation
American Heart Association, New York, NY

2008 Genetic Syndromes Associated with Congenital Heart Disease / Invited presentation
Sixth Annual Sloane Conference, New York, NY

2008 Manage Your Cancer Risk: Hereditary Breast and Gynecological Cancer Syndromes / Invited presentation
Sixth Annual Sloane Conference, New York, NY

2008 The Genetics of Basal Cell Nevus Syndrome / Invited presentation
Basal Cell Carcinoma Nevus Syndrome Life Support Network, New York, NY

2008 Advances in Genetic Medicine and Integration into Obstetric and Pediatric Practice / Grand Rounds
Valley Hospital, Ridgewood, NJ

2008 Mitochondrial Inherited Diabetes and Deafness / Endocrinology Grand Rounds
Downstate Medical Center, Brooklyn, NY

2009 Monogenic forms of diabetes / Endocrinology Grand Rounds
Downstate Medical Center, Brooklyn, NY

2009 Genetics of cardiac disease: Advanced Heart Failure / Invited presentation
New York Academy of Sciences, New York, NY

2009 Cardiovascular genetics for hypertrophic cardiomyopathy / Invited presentation
Management of Advanced Heart Failure, New York, NY

2009 The high risk breast cancer patient / Invited presentation
Breast Cancer Management 2009, New York, NY

2009 What's new in cardiac genetics? / Invited presentation

New technologies and techniques in pediatric cardiology. New York, NY

2010 Monogenic forms of diabetes identify common beta cell deficiencies / Seminar
St. Luke's Hospital, New York, NY

2010 Role of Cardiovascular Genetic Testing for Patients and Families / Invited presentation
Genetics of Cardiac Arrhythmias Symposium, New York, NY

2010 Breast and Ovarian Cancer Syndrome / Invited presentation
Genetic and Heritable Syndromes Involving Pancreatic Cancer, New York, NY

2010 The Hype and the Hope of Personalized Medicine / Invited presentation
Princeton University, Princeton, NJ

2010 Genetics for the Primary Pediatric Practice / Invited presentation
PriMed Conference, New York, NY

2010 Ethical considerations of comprehensive genomic analysis in clinical practice and research / Invited presentation
Personal Genomes, Cold Spring Harbor. NY

2010 Genetics and family planning. Recent Advances in SMA and Other Pediatric Neuromuscular Diseases / Invited presentation
Muscular Dystrophy Association, New York, NY

2010 Genetic Causes of Heart Failure / Invited presentation
Advanced Heart Failure and Cardiac Transplant, New York, NY

2010 Advances in genetics: how to incorporate them into your practice / Invited presentation
New Concepts in Neonatal Intensive Case: A Collaborative Conference, New York, NY

2010 Advances in Genetics for the Pediatric Practice / Pediatric Grand Rounds
Nyack Hospital, Nyack, NY

2011 Inherited cardiac disease: the role of genetic testing / Medicine Grand Rounds
Lenox Hill Hospital, New York, NY

2011 Inherited cardiac disease: the role of genetic testing / Pediatric Grand Rounds
Morristown Memorial Hospital, Morristown, NJ

2011 Optimizing Care: Lessons Learned / Invited presentation
Genetic Diseases of Children, New York, NY

2011 The ABCs of DNA and EKGs / Medicine Grand Rounds
Bronx-Lebanon Hospital, Bronx, NY

2011 The ABCs of DNA and Advances in Molecular Genetic Testing / Invited presentation
Pri-Med Conference, New York, NY

2011 New developments in genetics for the pediatrician / Pediatric Grand Rounds
Morristown Memorial Hospital, Morristown, NJ

2011 Insights into Diabetes Pathogenesis from Rare Monogenic Forms / Invited presentation
Mt. Sinai Hospital, NY

2011 Genetics of Cardiomyopathies / Invited presentation
Controversies in Pediatric Heart Diseases, New York, NY

2011 Advances in genetics for your practice / Invited presentation
National Association of Pediatric Nurse Practitioners (NAPNAP), New York, NY

2011 The ABCs of DNA in cardiology / Cardiology Grand Rounds
University of Medicine and Dentistry of New Jersey (UMDNJ), Newark, NJ

2011 Advances in genetics for your practice / Pediatric Grand Rounds
Bridgeport Hospital, CT

2012 How to effectively use genetics in your pediatric practice / Pediatric Grand Rounds
Summit Hospital, Summit, NJ

2012 Medical characteristics of patients with 16p11.2 deletions and duplications / Invited presentation
Simons Foundation, New York, NY

2012 Studying ASD through the context of an identified recurrent genetic event. Systems biology of autism: from basic science to therapeutic strategies / Invited presentation
Cold Spring Harbor Laboratories, Cold Spring Harbor, NY

2012 Advances in Genomic Testing to Diagnose Pediatric Diseases and Refine Treatment / Pediatric Grand Rounds
Weill Cornell Medical College, New York, NY

2013 The Clinical Utility of Exome Sequencing / Invited presentation
New Jersey Genetics Association, Rutgers University, Newark, NJ

2013 Simons VIP: A Genetic First Approach / Invited presentation
SFARI Annual Meeting, New York, NY

2014 Utilizing Genomic Sequencing in Rare Diseases to Guide Clinical Care and Develop New Therapies / Invited presentation
New York Genome Center, New York, NY

2014 Genetics of Neuropsychiatric Disorders in Children / Visiting Professor
Child Mind Institute, New York, NY

2014 Advances in Neurogenetics / Neurology Grand Rounds
NYU, New York, NY

2014 Advances in Genomic Testing in Neurology / Neurology Grand Rounds
Downstate Medical Center, Brooklyn, NY

2014 Return of Research Results / Invited presentation
Institutional Review Board 8th Annual Educational Conference, New York, NY

2016 Integration of Genomics into Clinical Care for Precision Medicine / Medicine Grand Rounds
Cornell University, New York, NY

2016 Participant Rights to their Sequence Data: Positive, Precautionary and Pragmatic Views on Returning the Incidental Genome / Invited presentation
Biology of Genomes, CSHL, New York, NY

2016 Genetic Counseling and Testing in Breast Cancer / Invited presentation

Advances in Breast Cancer Treatment, NYP-Hudson Valley Hospital, White Plains, NY

2016 Future of Medicine: A Conversation / Invited presentation
NYSCF Conference, New York, NY

2016 Contributions of Germ Line Variations to Carcinogenesis / Invited presentation
New York Cancer Genomics Research Network Monthly Meeting, New York, NY

2017 Autism Research: Where Are We Now? / Invited presentation
Autism Science Foundation, New York, NY

2017 Everything You Wanted to Know About Genetic Testing in Vascular Anomalies Patients / Invited presentation
Key Topics and Case Scenarios. Cases and Controversies in Vascular Anomalies, New York, NY

2017 How Genomics Differentiates Broken Hearts / Invited presentation
Leonard Steinfield Research Symposium. New York, NY

2017 Genetic Causes of Broken Hearts and Other Birth Defects / Invited presentation
Neonatal Care Symposium: Improving the Care and Outcomes for the High Risk Pre-Term Infant, Flushing, NY

2017 Genetics and the Role of Genetic Testing in Pediatric Pulmonary Hypertension / Invited presentation
PHA Pediatric Preceptorship Program: A Collaborative Approach for Pediatric Clinicians on the Front Line, New York, NY

2017 Genomic & Precision Medicine / Invited presentation
On Call: Health + Medicine. THIRTEEN. New York, NY. 12/7/17.
http://www.thirteen.org/blog-post/tune-health-medicine-tri-state-area/

2018 The Hype, The Hope and the Reality of Genomic Medicine / Invited presentation
Pharmacology and Physiology and Cellular and Molecular Pharmacology and Physiology Program, University of Rochester Medical Center, Rochester, NY

2018 The Present and Future of Genomic Medicine / Invited presentation
Third Annual MidAtlantic Bioinformatics Conference, Philadelphia, PA

2019 Genomic Medicine in Children. Jacobi Medical Center Pediatric Grand Rounds. Bronx, NY

2019 Genetic Causes of Broken Hearts and Associations with Outcomes / Keynote Lecture
2019 Joint Conference: Advances in Pediatric Cardiovascular Disease Management, New York, NY

2020 Opportunities in Genomic Medicine / Invited presentation
Icahn School of Medicine at Mount Sinai, New York, NY

2020 Genetic basis of monogenic diabetes / Invited presentation
NYU, New York, NY

2020 Pediatric genomic medicine / Invited presentation
NYU, New York, NY

2020 The genetics of autism and family planning implications / Invited presentation
Cornell University, New York, NY

2020 Precision Medicine / Grand Rounds
Hackensack University Medical Center, Hackensack, NJ

2021 Precision Medicine / Medicine Grand Rounds
Stonybrook University, Stonybrook, NY

2021 Personalized Genomics / Invited presentation
Regional Genetics Network (NYMAC). Online

2021 Pediatric Genomic Medicine / Seminar
Pediatric Surgical Seminar Series. Online

2021 Newborn Screening for Neurodevelopmental Disorders / Invited presentation
Simons Foundation, New York, NY

2021 SPARKing Research in Autism / Invited presentation
Autism New Jersey. Online.

2021 Challenges and Opportunities for Scaling Genomic Medicine / Medicine Grand Rounds
NYU, New York, NY

2022 Updates in Genomic Medicine / Medicine Grand Rounds
Lincoln Hospital, Bronx, NY

2023 Genomic Medicine in Pediatrics / Pediatric Grand Rounds
New York Hospital, Queens, NY

2023 SPARKing Research Advances in Autism and Neurodevelopmental Conditions / Invited presentation, Neuroscience Lecture
New York Genome Center, New York, NY

2023 The Future of Genomic Medicine At Scale for the Brain and Behavior in Children
Translational Neuroscience Seminar
Boston, MA

2023 GUARDIAN: Challenges, Successes, Preliminary Results
Genetic Risk for Early Childhood Cancer: Should newborns undergo genomic cancer risk screening?
Harvard Radcliffe Institute
Boston, MA

2024 Using Genomics to Stratify and Understand Complex Human Behavior Including Autism
Massachusetts General Hospital, Neurology Grand Rounda
Boston, MA

National

2004 Genetics 101: A Primer on Dysmorphology / Invited lecture
Pediatrics in Review: The Sixth Annual National Seminar, New York, NY

2005 Role of Academic Medical Centers in the Translation of Research into Clinical Practice / Invited lecture
President's Cancer Panel, National Cancer Institute, Bethesda, MD

2005 What's New in Newborn Screening / Invited lecture
Pediatrics in Review: The Seventh Annual National Seminar, New York, NY

2006 Genetic Evaluation of Pediatric Cardiomyopathy / Invited lecture
Pediatric Cardiomyopathy: A New Paradigm, Bethesda, MD

2006 Advice from the Experts / Invited lecture
Association for Glycogen Storage Diseases, Orlando, FL

2007 Predictive Genetic Testing for Pediatric Cardiomyopathies / Invited lecture
NHLBI Conference. Idiopathic and Primary Cardiomyopathy in Children, Bethesda, MD

2007 Monogenic Syndromes Associated with Obesity in Children / Invited lecture
Midwest Pediatric Endocrine Society Meeting, Chicago, IL

2007 Genetics of Obesity: Preventive Pediatric Cardiology in Children, Adolescents and Young Adults / Invited lecture
Denver, CO

2007 Genomic Approaches to Congenital Diaphragmatic Hernias / Invited lecture
The Congenital Diaphragmatic Hernia Study Group, Houston, TX

2007 Genetics of Spinal Muscular Atrophy / Invited lecture
SMA Family Meeting, Orlando, FL

2007 Genetic Cancer Syndromes / Invited lecture
University of Miami, Miami, FL

2008 Genetics and Genomics of Pulmonary Arterial Hypertension / Invited lecture
Fourth World Symposium on Pulmonary Hypertension, Dana Point, CA

2008 Genetics of Pulmonary Hypertension / Invited lecture
University of Miami, Miami, FL

2008 Genetics of Pulmonary Arterial Hypertension / Invited lecture
Pulmonary Hypertension Association Eighth International Conference, San Diego, CA

2008 Clinical Evaluation of Glycogen Storage Disease III / Invited lecture
American College of Medical Genetics, Consensus Conference for Glycogen Storage Disease III, Chicago, IL

2008 Clinical Management of Glycogen Storage Diseases / Invited lecture
American Glycogen Storage Disease Meeting, Chicago, IL

2008 Advances in Molecular Genetic Testing for Cardiomyopathies / Invited lecture
National Society for Genetic Counselors, Nashville, TN

2008 Advances in Genetic Medicine and Integration into Pediatric Practice / Invited lecture
Pediatric Grand Rounds. Richmond University Medical Center, Richmond, VA

2008 Use of Chromosome Microarrays in Clinical Diagnosis of Hematological Malignancies / Invited lecture
American Society of Hematology, San Francisco, CA

2010 Advances in the Genetic Basis of Cardiovascular Disease / Invited lecture
Vanderbilt University, Nashville, TN

2010 Novel Gene Discovery in Pediatric Cardiomyopathy / Invited lecture

Second International Conference on Cardiomyopathy in Children, Washington, DC

2010 Genetics of Pulmonary Hypertension / Invited lecture
Ninth International Pulmonary Hypertension Conference, Garden Grove, CA

2010 Medical management of Glycogen Storage Disease type I / Invited lecture
Association of Glycogen Storage Disease Annual Conference, Durham, NC

2011 The genetics of Glut1 deficiency syndrome: Glut1 Deficiency Syndrome / Invited lecture
Glut1 deficiency syndrome Scientific Meeting, New Orleans, LA

2011 Genetics in Pediatric Care: The future is now / Invited lecture
AAP: The future of Pediatrics, Chicago, IL

2011 How to Interpret the Interpretation-Finding meaning in new genetic tests. AAP: The future of Pediatrics, Chicago, IL

2011 Genetics and Etiology of Treacher-Collins Syndrome / Invited lecture
Open Forum on Cleft, Craniofacial and Pediatric Oral and Maxillofacial Surgery, Philadelphia, PA

2011 Genetics First: Insights into the Brain from 16p11.2 / Invited lecture
SFARI, Washington, DC

2011 The Utility of Chromosome Microarrays in the Prenatal Setting / Invited lecture
American College of Medical Genetics, Ontario, CA

2012 Return of Genetic Test Results to Research Participants / Invited lecture
Return of Results Consortium. NIH, Bethesda, MD

2012 Genetics of Cardiac Disease / Invited lecture
Contemporary Issues of Cardiovascular Disease, Louis F. Albright Cardiology Symposium, Boston, MA

2012 Myths of Primary Care Providers, Patients, and Families Regarding Genetics. Time Out for Genetics / Invited lecture
Genetics in Primary Care Institute, American Academy of Pediatrics, Chicago, IL

2012 The ABC of DNA and New Genetic Testing Options / Invited lecture
Maxwell Bogin Lecture, Yale University, New Haven, CT

2013 Applied OMICS-what to tell families about the –omics expedition / Invited lecture
Tenth Annual Dialogues in Neonatal-Perinatal Medicine, Duke University, Durham, NC

2013 Bench to Bassinette: A paradigm for collaborative research / Invited lecture
NICHD Birth Defects Meeting, Bethesda, MD

2013 Are we ready for GATTACA to become a reality? / Invited lecture
TEDMED, New York, NY

2013 A genetics first approach to the study of autism. Advances in Autism Research and Treatment / Invited lecture
Geisinger Health, Lewistown, PA

2013 Advances in Genomic Testing to Diagnose Pediatric Diseases and Refine Treatment / Invited lecture
Riley Hospital for Children: Pediatric Conference, Indianapolis, IN

2013 It's in the Genes-Genetic Components of Common Conditions / Invited lecture
AAP: Dive into the Gene Pool: Integrating Genetics and Genomics into your Pediatric Primary Care Practice, Chicago, IL

2013 Whole Exome Sequencing: How changes in sequencing technology influence providers and patients / Invited lecture
National Society of Genetic Counselors. Anaheim, CA

2013 Genetics of Congenital Heart Disease: Cardiovascular Genetics in Clinical Practice / Invited lecture
Harvard Medical School. Boston, MA

2013 Informed Consent for Whole Genome Sequencing: Experience and Implications for Practice / Invited lecture
American Society for Human Genetics, Boston, MA

2013 Advances in Genetics of Cardiovascular Disease: New Paradigms in Obstetric and Pediatric Genomic Medicine / Invited lecture
Stamford Hospital, Stamford, CT

2013 Simons VIP: A Genetics First Approach to the Study of Autism / Invited lecture
Autism Consortium, 2013 Symposium, Boston, MA.

2013 Now What Do I Do? Genetics in Primary Care. Institute Quality Improvement Project Learning / Invited lecture
Institute Quality Improvement, Chicago, IL

2013 Targeted and Whole Exome Sequencing in Congenital Heart Disease: Clinical Applications and Pitfalls / Invited lecture
American Heart Association, Houston, TX

2014 Insights From Studying the Monogenic Forms of Obesity/ Invited lecture
American College of Medical Genetics, Salt Lake City, UT

2014 Advances in Cardiac Genomics for Your Practice/ Invited lecture
Char Lecture, University of Arkansas, Little Rock, AK

2014 Advances in Genomic Testing to Elucidate Rare Disorders / Invited lecture
Prevention Genetics, Marshfield, WI

2014 Novel Therapeutic Strategies Emerging from Genetic Studies in Pulmonary Arterial Hypertension / Invited lecture
American Thoracic Society, San Diego, CA

2014 Use of genomic methods to elucidate rare causes of pediatric disease. Genes, Genomes and Pediatric Disease. Children's Hospital of Philadelphia. Philadelphia, PA

2014 Utilizing Genomic Sequencing in Rare Diseases to Guide Clinical Care and Develop New Therapies / Invited lecture
Cincinnati Children's Hospital, Cincinnati, OH

2014 Participant Preferences and Reactions to Return of Results from WES / Invited lecture
ASHG/ASBH Joint Satellite Symposium: From Clinical to Community Sequencing: Emerging Ethical, Legal and Social Issues in Genomics, San Diego, CA

2014 The role of genetics in autism / Invited lecture
Saward Lecture, Kaiser Permanente Portland, OR

2015 Clinical Exome Sequencing. / Invited lecture
ACMG Short Course. Salt Lake City, UT

2015 SFARI's Genetic Research Initiatives / Invited lecture
The Wendy Klag Center for Autism & Developmental Disabilities, Johns Hopkins Bloomberg School of Public Health, Baltimore, MD

2015 Genomics as a Tool to Understand the Brain and Behavior in Autism / Invited lecture
The Help Group Summit 2015. Advances and Best Practices in Autism, Learning Disabilities, ADHD, Skirball Cultural Center, Los Angeles, CA

2015 The Future of Pediatric Precision Medicine / Invited lecture
Precision Pediatrics, New York, NY

2015 Precision Medicine: The Intersection of Genomics, Personalized Medicine, and Humanistic Care / Invited lecture
Humanity at the Heart of Health Care, 2015 AMSA Conference, New York, NY

2016 Translational Considerations in Genomic Sampling / Invited lecture
TransCEER Workshop to Explore the Ethical, Legal and Social Implications (ELSI) of Inclusivity and Representation in Precision Medicine: What Will Success Look Like? Bethesda, MD

2016 Genetic Testing: The Toolbox in the Clinical Setting / Invited lecture
Cardiology 2016: 19th Annual Update on Pediatric and Congenital Cardiovascular Disease: Bringing Science to Clinical Practice, Orlando, FL

2016 Roundtable 1: Genetics in Congenital Heart Disease: Case-Based Presentations / Invited lecture
Cardiology 2016: 19th Annual Update on Pediatric and Congenital Cardiovascular Disease: Bringing Science to Clinical Practice, Orlando, FL

2016 Genetic Contributions to Congenital Heart Disease and Related Developmental Disorders / Invited lecture
World Birth Defects Day, University of Arkansas.

2016 The Future Use of Exome Sequencing as the Genetic Test of Choice for Clinical Diagnostics / Invited lecture
Personalized Diagnostics, Tri-Conference. San Francisco, CA

2016 ACMT Tox Mimics in the Critically Ill / Invited lecture
American College of Medical Toxicology, Huntington Beach, CA

2016 Practical Implementation of Genomic Sequencing in Healthcare Settings / Panel
2016 Joint Summits on Translational Science, San Francisco, CA

2016 Is the Future of Medicine in our DNA? / Invited lecture
Jepson Leadership Forum. Richmond, VA

2016 FDA Regulation of Genetic Testing / Invited lecture
Genomics Festival, Boston, MA

2016 Genomic Health Screening: The Hype, Hope and Reality / Invited lecture
Next Generation Dx Summit, Washington DC

2016 Integration of Genetic Medicine into Healthcare / Invited lecture
BioData World USA 2016 Conference, Boston, MA

2016 Genetics of Cardiovascular Defects / Invited lecture
Advances in Fetology 2016, Chicago, IL

2016 Update on DHREAMS / Invited lecture
Advances in Fetology 2016. Chicago, IL

2016 Hype, Hope and Reality of Genomic Testing / Invited lecture
The Precision Health Forum, Chicago, IL

2017 A Complete Understanding of the Genetics of congenital Heart Disease? / Invited lecture
Cardiology 2017, Orlando, FL

2017 The Genetics of Pulmonary Hypertension / Invited lecture
Cardiology 2017, Orlando, FL

2017 Precision Pediatrics Powered by Genomics / Invited lecture
Health Sciences Research Week 2017, University of Iowa, Iowa City, IA

2017 Precision Pediatrics / Invited lecture
College of Human Medicine (CHM), Michigan State University, East Lansing, MI.

2017 The Hype, the Hope, and the Reality of Genomic Medicine / Invited lecture
AGBT Precision Health. Scottsdale, AZ

2017 The Challenges and the Opportunities of the Spectrum of Autism / Keynote Session
12 Annual Thompson Center Autism Conference, St. Louis, MO

2017 Seizing the Gene – The Future of Genomic Medicine / Invited lecture
The Precision Health Forum, University of Illinois, Chicago, IL

2017 Genomic Medicine: Maximizing Benefits and Minimizing Risks / Invited lecture
Risk Management Symposium: Emerging Risks, Rosemont, IL

2018 Genetic Testing and Return of Results / Panel presentation
Precision Medicine World Conference 2018, Mountain View, CA

2018 Women in Academia /Panel presentation
10th Annual Women Empowering Women, Leadership Conference, New Haven, CT

2018 The Hype, the Hope, and the Reality of Genomic Medicine / Invited lecture
2018 Genomic and Precision Medicine Forum, Durham, NC

2018 The Future of Genomic Medicine / Invited lecture
Genetic Medicine: a Chan Zuckerberg Initiative, San Francisco, CA

2018 The Hype, the Hope, and the Reality of Genomic Medicine / TED style talk
American College of Medical Genetics, Charlotte, SC

2018 The Role of Genetic Testing in Pediatric PVD / Invited lecture
UCSF's 11th International Conference: Neonatal & Childhood Pulmonary Vascular Disease, San Francisco, CA

2018 SPARKing Partnerships in Autism Research / Invited lecture
UCLA, Los Angeles, CA

2018 SPARK: Catalyzing Autism Research and Elucidating the Genetic Basis for Autism / Invited lecture
UCLA Center for Autism Research and Treatment, Los Angeles, CA

2018 Pulmonary Vasculopathies: From PPH to HHT / Invited lecture
American Thoracic Society International Conference, San Diego, CA

2018 SPARKing New Paradigms in Translational Autism Research Stratified by Genetics / Invited lecture
Neuro Developmental Disorders Symposium, Boston, MA

2018 SPARKing New Paradigms in Translational Autism Research / Invited lecture
Autism Across the Lifespan, Worcester, MA

2018 The ABCs of DNA: How to Ensure Safe, Effective Use of Genetics in Your Practice / Invited lecture
ISMIE Risk Management Symposium, Oak Brook, IL

2018 Autism: Could Genetics Hold the Answers? / Invited lecture
Mind Science Foundation's 2018 Distinguished Speaker Series, San Antonio, TX

2018 Use of Genomics to Understand Broken Hearts and Implications for Clinical Care / Invited lecture
Cardiovascular Research Institute at Baylor College of Medicine Fall 2018 Seminar, Houston, TX

2018 Opportunities in Pediatric Genomic Medicine / Invited lecture
Nationwide Children's: The 2018 Research Retreat, Columbus, OH

2019 What PH Patients Should Receive Genetic Counseling and Testing / Invited lecture
American College of Cardiology's 68th Annual Scientific Session & Expo (ACC.19), New Orleans, LA

2019 Does Genotype Predict Clinical Risk in Pulmonary Vascular Disease / Invited lecture
American College of Cardiology's 68th Annual Scientific Session & Expo (ACC.19), New Orleans, LA

2019 Gene Replacement Therapy in SMA / Invited lecture
The France Foundation's Are You Ready for Gene Replacement Therapy? Example from Spinal Muscular Atrophy, Seattle, WA

2019 Genetics of Pulmonary Vascular Disease in Children / Invited lecture
Robyn Barst Lecture, 2019 Grover Conference, Sedalia, CO

2019 Clinical Genomics / Invited lecture
AGBT 2019 4th Annual Precision Health Meeting, La Jolla, CA

2019 Scaling Diagnosis and Treatment of Rare Genetic Diseases / Invited lecture
Emory University's Department of Human Genetics' Human Genetics Seminar, Atlanta, GA

2019 Lasker Lessons in Leadership lecture / Invited lecture
Albert and Mary Lasker Foundation, Bethesda, MD

2019 Bigger is Better: More Cancer Genes in More Patients / Invited lecture
AMP 2019 Annual Meeting & Expo, Baltimore, MD

2020 Genetic testing for breast cancer / Invited lecture
Best of Breast, Palm Beach, FL

2020 Understanding the human genome and impact on medicine in the future / Invited lecture
CHEMED Health Conference, Woodcliff, NJ

2020 Present and future of genomics / Invited lecture
CHEMED Health Conference, Woodcliff, NJ

2020 The Genetics Hotline: Responsibility and Liability When Handling Unsolicited Patient Communications / Invited lecture
ACMG. Online

2020 Genomics Causes of the Broken Hearts / Invited lecture
NBSTRN Newborn Screening Summit. Online.

2020 Genetics of Common Congenital Anomalies. / Invited lecture
Fetology Chicago: Practice and Discovery Live Virtual Event. Online.

2020 Facing the Legal Barriers to Genomic Research and Precision Medicine / Invited lecture
LawSeq. Online

2021 Precision Pediatrics / Grand Rounds
Stamford Health, Stamford, CT

2021 Of Mice and Men: Genetics of Congenital Diaphragmatic Hernia / Invited lecture .
Monarch Meeting. Online.

2021 Patient-Researcher Partnerships Across Rare Genetic Forms of NDD and ASD.
Gatlinburg Symposium. Online.

2021 Bardet Biedl Syndrome: Genetic Pathophysiology and Clinical Characteristics / Invited lecture
ACMG. Online.

2021 Chromatinopathies: An Expanding Clinical Spectrum / Invited lecture
ACMG. Online.

2021 Precision Pediatrics / Invited lecture
Bridgeport Hospital, Bridgeport, CT

2021 Rare Genetic Diseases: What Zebras Teach Us About Horses / Invited lecture
Dartmouth University, Hanover, NH

2021 Precision Medicine / Invited lecture
American Physician Scientists Association. Online.

2021 Genetics and Pulmonary Arterial Hypertension / Invited lecture
PHA Live, Online.

2021 Genetics' Growing Interaction with the Law / Invited lecture
Genomics Web Series, Online.

2021 Rare Breakthroughs: now and on the Horizon / Invited lecture
NORD, Online.

2021 Genomic Medicine: Opportunities and Challenges, University of Wisconsin, Madison, WI

2021 N of 1 Precision Medicine in the Era of Antisense Oligonucleotide Therapies / Invited lecture

American Society of Human Genetics, Online.

2021 Spinal Muscular Atrophy: Clinical Decision-Making in the Midst of an Unfolding Phenotype / Invited lecture
Stanford University, Online.

2022 Genomic Medicine / Invited lecture
UPMC Children's Hospital of Pittsburgh, Online.

2022 Ethical, Clinical, Legal, and Economic Issues Surrounding Genetic Variant Reinterpretations / Invited lecture
ELSIcon2022, Online.

2022 Genomic medicine: disparities and opportunities to improve health equity / Invited lecture
Dean's Lecture, McGovern School of Medicine, Houston, TX

2022 GUARDIAN / Invited lecture
Newborn sequencing and screening conference, Boston, MA

2022 Raising Healthy Children / Invited lecture
NICHD 60th Anniversary Symposium, Bethesda, MD

2022 Sparking Research to Understand the Complexities of ASD / Invited lecture
AACAP/CACAP Annual Meeting, Research Institute: Child and Adolescent Psychiatry in the Era of Genomics, Toronto, CA

2022 PROGRESS and opportunities to study early brain development / Invited lecture .
Beyond Baby Sibs, Minneapolis, MN

2022 Rapidly Evolving Opportunities for Treatments for Rare Genetic Diseases. 15th Annual Global Science Summit Program: Focus on Clinic Trials. Palm Beach, FL

2022 Genomic Medicine / Invited presentation
Second Annual Conference on Precision Psychiatry, Massachusetts General Hospital. Boston, MA

2023 Opportunities and Challenges in Precision Medicine / Invited lecture - Visiting Professor
Morbidity and Mortality Quality Assurance Case Conference, Medical University of South Carolina, Charlotte, SC

2023 Pilot Sequencing based Newborn Screening in a Diverse Community / Invited lecture
American College of Medical Genetics, Salt Lake City, UT

2023 Setting the Stage for Genomic Sequencing of All Newborns / Invited lecture
R. Rodney Howell Symposium, American College of Medical Genetics, Salt Lake City, UT

2023 SPARKIing New Insight into Autism across the Lifespan / Invited lecture
Gatlinburg Conference, Kansas City, KS

2023 Genomics and Precision Autism / Invited lecture
Autism Symposium, American Academy of Neurology, Boston, MA

2023 Genomic Integrated risk assessment for breast cancer across patients of diverse ancestry: The eMerge experience / Invited lecture
11th Annual Scientific Symposium, Basser Center for BRCA, Philadelphia, PA

2023 Genetics of Structural Birth Defects: Gene Discovery and Mutation Spectrum / Invited lecture
Understanding Developmental Disorders in Genomic Age, Keystone Meeting, Tarrytown, NY

2023 The Spectrum of Genetic Variation Associated with Autism and Related Neurodevelopmental Disorders
Genetics to Mechanisms to Therapeutics: Symposium on Severe Mental Illnesses
Stanley Center Symposium: Boston, MA

2023 Supporting Equity in Precision Pediatrics / Invited lecture
Department of Genetics Seminar & Precision Medicine Pathway lecture, Washington University St. Louis, MO

2023 GUARDIAN Newborn Screening for Rare Diseases / Invited Lecture
Science in Society Annual Meeting
Newport Beach, CA

2023 Fireside Chat About N of 1 Treatments / Invited Lecture
AGBT Precision Medicine
San Diego, CA

2023 N of 1 Treatment for KAND / Invited Lecture
National Organization of Rare Diseases Breakthrough Summit
Dallas, TX

2023 Variant Interpretation Within the Context of Newborn Screening in GUARDIAN
Critical Assessment of Genome Interpretation (CAGI)
Boston, MA

2023 GUARDIAN: Genomic Uniform screening Against Rare Diseases In All Newborns
National Academy of Medicine
Washington DC

2023 The Role of Genomics as a First-line Test in Newborn Screening / Invited Lecture
APHL/ISNS Newborn Screening Symposium
Sacramento, CA

2023 Emerging therapies for cardiomyopathies
Look in the Future: Pediatric Cardiology in 2043
AAP Meeting, Washington DC

2023 Scaling Screening: A Plethora of Perspectives
Expanded newborn screening using first-tier genome sequencing for highly penetrant early onset conditions to increase health equity for children
ASHG Annual meeting
Washington, DC

2023 Newborn Screening by Genomic Sequencing: Opportunities and Challenges / Invited Lecture
Association of Molecular Pathology
Salt Lake City, Utah

2024 GUARDIAN: Newborn Screening by Genomic Sequencing
UCLA Department of Pediatrics
Los Angeles, CA

2024 GUARDIAN: Newborn Screening by Genomic Sequencing / Invited Lecture
SFARI Investigators Meeting
New York, NY

2024 The future role of medical geneticists
National Human Genome Research Institute
Bethesda, MD

2024 Genetics Diagnoses for Patients with Autism and Neurodevelopmental Disorders
SUNY Downstate
Brooklyn, NY

2024 Genetics of Pediatrics Pulmonary Hypertension / Invited Lecture
17th International Conference Neonatal & Childhood Pulmonary Vascular Disease
San Francisco, CA

2024 Advances in genomic medicine: from N of 1 to population health / Invited Lecture
ASCI
Chicago, IL

2024 N of 1 Therapies to Population Health / Invited Lecture
NINDS Genetics Strategies to Treat the Epilepsies Workshop
Online via Zoom

International

2002 Inherited Lipodystrophic Syndromes / Invited lecture
North American Association for the Study of Obesity, Brazil

2004 Counseling the parents of a Neonate with a Genetic Disease **/** Invited lecture
Introduction to Medical Genetics / Invited lecture
Newborn Screening / Invited lecture
Interpreting Genetic Testing / Invited lecture
Prenatal Diagnosis of Genetic Diseases / Invited lecture
Neonatal Metabolic Emergencies / Invited lecture
Neonatology: Recent Advances in Neonatal Intensive Care Unit, American Austrian Foundation Conference, Salzburg, Austria

2006 Genetics of Syndromic Obesity / Invited lecture
Advances in Pediatrics. Hallym University. Seoul, South Korea

2007 Maternal and Infant Health: High-risk Obstetrics, Fetal and Neonatal Medicine / Invited lecture
American Austrian Foundation Conference. Salzburg, Austria

2009 Evaluation of Suspected Monogenic Forms of Obesity in Childhood / Invited presentation
European Society of Pediatric Endocrinology and Lawson Wilkins Pediatric Endocrine Society, New York, NY

2010 Personalized Medicine and the Age of Genomic Health / Invited lecture

Arab Health Conference. Dubai, UAE

2010 Advances In Genetic Testing: When and What to Order / Invited lecture
Recent Advances in Perinatal and Neonatal Medicine. Dubai, UAE

2010 Evaluation of the Infant with Suspected Genetic Disease / Invited lecture
Recent Advances in Perinatal and Neonatal Medicine. Dubai, UAE

2010 Insight from Monogenic Forms of Obesity / Invited lecture
Eleventh International Conference on Long Term Complications of Treatment of Children and Adolescents for Cancer, Williamsburg, VA

2011 Clinical Characterization of 16p11.2 deletions/duplications: a model for translational CNV studies / Invited lecture
International Standards for Cytogenomic Arrays Consortium, Washington, DC

2016 Legal, Regulatory & Ethical Issues in the Secondary Use of Genomics Data / Invited lecture
PRISME Forum Technical Meeting: Understanding Disease through Mining Clinical Trial Data, Prague, Czech Republic

2017 Going from an N of 1 to Population Based Screening and Treatment of Rare Genetic Disorders / Invited lecture
12th Annual ICORD Conference: 6th China Rare Disease Summit, Beijing, China

2018 Scaling Discovery, Care, and Treatment for Rare Genetic Disorders / Invited lecture
The 7th China Rare Disease Summit, Shanghai, China

2018 Precision Medicine in Immune Related Diseases / Invited lecture
Primary Immunodeficiencies and Immune Dysregulation: From Translational Immunology to Personalized Medicine, Santiago, Chile

2019 Genetic Basis of Congenital Anomalies / Invited lecture
HGSA 43rd Annual Scientific Meeting, Wellington, New Zealand

2019 Future of Genomic Medicine / Invited lecture
HGSA 43rd Annual Scientific Meeting. Wellington, New Zealand

2019 Spark Patient Partnerships Enabling Research in Autism / Invited lecture
Rare Disease Summit, Shenzhen, China

2020 Horses morphing into zebras: hundreds of rare monogenic diseases masquerading as common diseases / Keystone Symposium
Beyond a Million Genomes: From Discovery to Precision Health, Online.

2021 Rare Causes of Common Conditions and Building Rare Disease Communities / Invited lecture
Sanger Center. Online.

2021 The role of genetics in clinical care and future research / Invited lecture
Fifth International Conference on Cardiomyopathy in Children, Online.

2021 Pulmonary Hypertension Gene Curation: ClinGen Gene-Disease Clinical Validity Framework / Invited lecture
Pulmonary Vascular Research InstituteSymposium. Online.

2021 Genetics of pulmonary arterial hypertension: What we can learn by studying children and without congenital heart disease / Live Interactive Webinar Series

PVRI. Online.

2022 Update from ClinGen Task Force on PAH genes / Invited lecture
3rd International Consortium for Genetic Studies in Pulmonary Arterial Hypertension (PAH). Online.

2022 SPARKing Research to Understand the Complexities of Autism / Invited lecture
2022 Peking University Health Science Conference on Autism Spectrum Disorders-Etiology, Family and Support. Peking, China

2022 Genetics Conditions in Children / Invited lecture
BioTechX, Basel, Switzerland

2022 Precision Pediatric Medicine / Invited lecture
International Symposium on Precision Medicine and Cancer Prevention. Zhengzhou University, Zhengzhou, China

2023 GUARDIAN: Genomic Uniform-screening Against Rare Diseases in All Newborns / Invited lecture
2023 BIO Asia-Taiwan 2023, Taipei City, Taiwan

2023 Whole Genome Sequencing (WGS) for Newborn Screening: GUARDIAN / Invited lecture
Newborn Screening Ontario Symposium, Ontario, Canada
GUARDIAN Study. ICoNS Meeting. London, UK

2024 GUARDIAN: Newborn Screening by Genomic Sequencing / Invited Presentation
American College of Medical Genetics
Toronto, Canada

2024 GUARDIAN: Genomic Newborn Screening / Invited lecture
Japan Pediatric Society. Fukuoka City, Japan

2024 Genetically Based Newborn Screening for Cardiomyopathy / Invited Lecture
6th International Conference on Cardiomyopathy in Children, Virtual Conference.

Report of Clinical Activities and Innovations

Past and Current Licensure and Board Certification:

1999	New York Medical License, active
2002-2022	American Board of Medical Genetics-Clinical Genetics
2005-2025	American Board of Medical Genetics-Molecular Genetics
2006	New Jersey Medical License, active
2023	Massachusetts Medical License, active

Practice Activities:

2002-2023	Outpatient consultation	Genetics clinic, DISCOVER program, CUMC	10 hours / week

2002-2023	Outpatient diagnosis and treatment	Cancer Genetics clinic, VHL center, CUMC	5 hours / week
2002-2020	Inpatient diagnosis and evaluation	Genetic inpatient coverage, CUMC	8 hours / month
2023-	Outpatient consultation	Genetics clinic	8 hours / week

Clinical Innovations:

2013	Original plaintiff in the Association for Molecular Pathology et al v. Myriad Genetics Supreme Court Case that overturned gene patents With elimination of gene patents to read out an individual's genetic information, medical genetic and genomic diagnostics including exome/genome sequencing was transformed and became less expensive, more innovative, and more comprehensive. As an example, genetic testing for hereditary breast cancer was transformed from a test of 2 genes for $3600 and a turn around time of 4 weeks to a panel of 59 genes for $300 and a turn around time of 2 weeks.

Report of Teaching and Education Innovations

2001-2002	Developed and directed course in human genetics for medical and dental students Curriculum in use at Columbia Physicians and Surgeons.

Report of Education of Patients and Service to the Community

No presentations below were sponsored by 3rd parties/outside entities

Activities

2019 Podcast: The Beagle Has Landed

2020 Ken Burns Documentary: The Gene: An Intimate History

2014 The truth about autism. TED talk, Vancouver, Canada
https://www.ted.com/talks/wendy_chung_autism_what_we_know_and_what_we_don_t_know_yet?language=en

2021 Podcast: People Behind the Science: Hunting Down Genes that Cause Human Disease

2022 Podcast: Whole genome newborn screening and other big opportunities in genomic medicine

2022 Podcast: Once upon a gene - A Focus On Patient Advocacy - Participation In Research and the Importance of an Engaged Patient Advocacy Group

2022 Podcast Columbia Invents

2023 Podcast: Revolutionizing Healthcare: Wendy Chung's Precision Medicine Journey

2023 Podcast: Doctor's Podcasts

2023 Podcast: A career chat with Wendy Chung

2023 Podcast: Peter Attia: The importance of newborn screening

2024 Podcast: The Geneticist's Journey

Recognition:

2012-2019 Top Doctors Castle Connolly

Report of Scholarship

Peer-Reviewed Scholarship in print or other media:

Research Investigations

1. Dammerman, M., Sandkuijl, L.A., Halaas, J.L., **Chung, W.K.,** Breslow, J.L. An apolipoprotein CIII haplotype protective against hypertriglyceridemia is specified by promoter and 3' untranslated region polymorphisms. Proc Natl Acad Sci USA. 1993 May 15;90(10):4562-4566. PMID: 8099442. PMCID: PMC46552.
2. Chua, S.C., **Chung, W.K.,** Wu-Peng, X.S., Zhang, Y., Liu, S.M., Tartaglia, L., Leibel, R.L. Phenotypes of the mouse diabetes and rat fatty due to mutations in the OB (leptin) receptor. Science. 1996 Feb 16;271(5251):994-996. doi: 10.1126/science.271.5251.994. PMID: 8584938.
3. **Chung, W.K.,** Goldberg-Berman, J., Power-Kehoe, L., Leibel, R.L. Molecular mapping of the murine tubby (*tub*) mutation on chromosome 7. Genomics. 1996 Mar 1;32(2):210-217. doi: 10.1006/geno.1996.0107. PMID: 8833147.
4. **Chung, W.K.,** Power-Kehoe, L., Chua, M., Liebel, R.L Mapping of the OB receptor (OBR) to 1p in a region of non-conserved gene order from mouse and rat to human. Genome Res. 1996 May;6(5):431-438. doi: 10.1101/gr.6.5.431. PMID: 8743992.
5. Chua, S.C., White, D.W., Wu-Peng, X.S., Liu, S.M., Okada, N., Kershaw, E.E., **Chung, W.K.,** Power-Kehoe, L., Chua, M., Tartaglia, L.A., Leibel, R.L. Phenotype of fatty due to Gln269Pro mutation in the leptin receptor (Lepr). Diabetes. 1996 Aug;45(8):1141-1143. doi: 10.2337/diab.45.8.1141. PMID: 8690163.
6. Norman, R.A., Leibel, R.L., **Chung, W.K.,** Power-Kehoe, L., Chua, S.C., Knowler, W.C., Thompson, D.B., Bogardus, C., Ravussin, E. Absence of linkage of obesity and energy metabolism to markers flanking homologues of rodent obesity genes in Pima Indians. Diabetes. 1996 Sept;45(9):1229-1232. doi: 10.2337/diab.45.9.1229. PMID: 8772727.
7. Duggirala, R., Stern, M.P., Mitchell, B.D., Reinhart, L.J., Shipman, P.A., Uresandi, O.C., **Chung, W.K.,** Leibel, R.L., Hales, C.N., O'Connell, P., Blangero, J. Quantitative variation in obesity-related traits and insulin precursors linked to OB gene region on human chromosome 7. Am J Hum Genet. 1996 Sep;59(3):694-703. PMID: 8751871. PMCID: PMC1914918.
8. **Chung, W.K.,** Power-Kehoe, L., Chua, M., Lee, R., Leibel, R.L. Genomic structure of the human OB receptor and identification of two novel intronic microsatellites. Genome Res. 1996 Dec;6(12):1192-1199. doi: 10.1101/gr.6.12.1192. PMID: 8973914.
9. Rose, P.M., Lynch, L.S., Frazier, S.T., Fisher, S.M., **Chung, W.,** Battaglino, P., Fathi, Z., Leibel, R., Fernandes, P. Molecular genetic analysis of a human neuropeptide Y receptor. The human homolog of a murine "Y5" receptor may be a pseudogene. J Biol Chem. 1997 Feb 7;272(6):3622-3677. doi: 10.1074/jbc.272.6.3622. PMID: 9013614.
10. Chagnon, Y.C., Perusse, L., Lamothe, M., Chagnon, M., Nadeau, A., Dionne F.T., Gagnon, J, **Chung, W.K.,** Liebel, R.L., Bouchard, C. Suggestive linkages between markers on human 1p32-p22 and body fat and insulin levels in the Quebec Family Study. Obes Res. 1997 Mar;5(2):115-121. doi: 10.1002/j.1550-8528.1997.tb00651.x. PMID: 9112246.

11. Echwald, S.M., Sorensen, T.D., Sorensen, T.I., Tybjaerg-Hansen, A., Andersen, T., **Chung, W.K.,** Leibel, R.L., Pedersen, O. Amino acid variants in the human leptin receptor: lack of association to juvenile onset obesity. Biochem Biophys Res Commun. 1997 Apr 7;233(1):248-252. doi: 10.1006/bbrc.1997.6430. PMID: 9144432.

12. **Chung, W.,** Chua, S.C., Lee, G.H., Leibel, R.L. Polymerase chain reaction-restriction fragment length polymorphisms (PCR-RFLP) and electrophoretic assays for the mouse obese (Lep^{ob}) mutation. Obes Res. 1997 May;5(3):183-185. doi: 10.1002/j.1550-8528.1997.tb00291.x. PMID: 9192391.

13. **Chung, W.K.,** Zheng, M., Chua, M., Kershaw, E., Power-Kehoe, K., Tsuji, M., Wu-Peng X.S., Williams, J., Chua, S.C., Leibel, R.L. Genetic modifiers of Leprfa associated with variability in insulin production and susceptibility to NIDDM. Genomics. 1997 May 1;41(3):322-344. doi: 10.1006/geno.1997.4672. PMID: 9169130.

14. **Chung, W.K.,** Power-Kehoe, L., Chua, M., Chu, F., Aronne, L., Huma, Z., Sothern, M., Udall, J.N., Kahle, B., Leibel, R.L. Exonic and intronic sequence variation in the human leptin receptor gene (LEPR). Diabetes. 1997 Sep;46(9):1509-1511. doi: 10.2337/diab.46.9.1509. PMID: 9287054.

15. Chua, S.C., Koutras, I.K., Han, L., Liu, S.M., Kay, J., Young, S.J., **Chung, W.K.,** Leibel, R.L. Fine structure of the murine leptin receptor gene: splice site suppression is required to form two alternatively spliced transcripts. Genomics. 1997 Oct 15;45(2):264-270. doi: 10.1006/geno.1997.4962. PMID: 9344648.

16. Silver, K., Walston, J., **Chung, W.K.,** Yao, F., Parikh, V.V., Andersen, R., Cheskin, L.J., Elahi, D., Muller, D., Leibel, R.L., Shuldiner, A.R. The Gln223Arg and Lys656Asn polymorphisms in the human leptin receptor do not associate with traits related to obesity. Diabetes. 1997 Nov;46(11):1898-1900. doi: 10.2337/diab.46.11.1898. PMID: 9356043.

17. **Chung, W.K.,** Belfi, K., Chua, M., Wiley, J., Mackintosh, R., Nicolson, M., Boozer, C.N., Leibel, R.L. Heterozygosity for Lep^{ob} and $Lepr^{db}$ affects body composition and leptin homeostasis in adult mice. Am J Physiol. 1998 Apr;274(4):R985-990. doi: 10.1152/ajpregu.1998.274.4.R985. PMID: 9575960.

18. Walder, K., Norman, R.A., Hanson, R.L., Schrauwen, P., Neverova, M., Jenkinson, C.P., Easlick, J., Warden, C.H., Pecqueur, C., Raimbault, S., Ricquier, D., Silver, M.H., Shuldiner, A.R., Solanes, G., Lowell, B.B., **Chung, W.K.,** Leibel, R.L., Pratley, R., Ravussin, E. Association between uncoupling protein polymorphisms (UCP2-UCP3) and energy metabolism/obesity in Pima Indians. Hum Mol Genet. 1998 Sep;7(9):1431-1435 doi: 10.1093/hmg/7.9.1431. PMID: 9700198.Otabe, S., Clement, K., Dubois, S., Lepretre, F., Pelloux, V., Leibel, R.L., **Chung, W.K.,** Boutin, P., Guy-Grande, B., Froguel, P., Vassuer, F. Mutation screening and association studies of the human uncoupling protein 3 gene in normoglycemic and diabetic morbidly obese patients. Diabetes. 1999 Jan;48(1):206-208. PMID: 9892245.

20. Chagnon, Y.C., **Chung, W.K.,** Perusse, L., Chagnon, M., Roy, S., Leibel, R.L., Bouchard, C. Linkages and associations between the leptin receptor (LEPR) gene and human body composition in the Quebec Family Study (QFS). Intl J Obes Relat Metab Disord. 1999 Mar;23(3):278-286. doi: 10.1038/sj.ijo.0800809. PMID: 10193873.

21. **Chung, W.K.,** Luke, A., Cooper, R.S., Rotini, C., Vidal-Puig, A., Rosenbaum, M., Gordon, D., Leal, S.M., Caprio, S., Goldsmith, R., Andreau, A.L., Bruno, C., DiMauro, S., Heo, M.,

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Scholarship without named authorship

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Narrative Report

Introduction

I am a physician scientist specializing in human genetics. After completing my training in June 2002 at Columbia University, I joined the faculty at Columbia University and was promoted through the ranks to my final position in 2017 as the tenured Kennedy Family Professor of Pediatrics in Medicine. In July 2023, I joined Boston Children's Hospital and HMS where I provide administrative leadership to the Department of Pediatrics, provide clinical care, supervise trainees, and conduct research on human genetics of several diseases (autism, neurodevelopmental disorders, KIF1A associated neurological disorder, congenital heart disease, congenital diaphragmatic hernia, esophageal atresia), pulmonary hypertension, obesity, diabetes, and newborn screening to enhance our knowledge and the quality of patient care. I am a member of the National Academy of Medicine. I am seeking appointment to the rank of Professor with Investigation as my Area of Excellence with Significant Supporting Activities of Clinical Expertise and Administration and Institutional service. I devote 30% of my time to research, 15% to clinical practice, 5% to teaching, and 50% to administration.

Area of Excellence - Investigation

I have been conducting clinical research on human genetic diseases for over 30 years. I have elucidated the genetic basis of many human diseases including contributions from rare de novo genetic variants, inherited rare variants, and common variants and demonstrated associations with clinical outcomes based upon the underlying genetic etiology. I have identified over 50 novel genetic conditions and characterized the clinical phenotype. Three of these conditions bear my name. I have translated advances in genetic diagnostics including the development of multigene panels by clinical indication, implemented exome and genomic sequencing for clinical diagnostics in prenatal and pediatric medicine, and how am assessing the ability to use genomic integrated risk assessment for common conditions in adults and children. I have performed pilot studies for newborn screening for SMA, Duchenne muscular dystrophy, and now am using genome sequencing in GUARDIAN to screen for ~250 genetic conditions simultaneously and have demonstrated synergies with traditional newborns screening and am working toward population based genomic screening. I have begun to develop treatments for some of these conditions including an ASO strategy currently in clinical trial for selection knock down of a dominant negative heterozygous mutation in KIF1A**.** I have received funding for my research from several institutes at NIH including NICHD, NHGRI, NHLBI, NIDDK, NINDS, NCI, and private foundations including the Simons Foundation and CZI. All of my research aims to determine the genetic basis for human disease, tailor care based upon genomic information, and implement genomics into medical care in an ethical, cost effective, and equitable manner. I lead many large national and international consortia including DHREAMS, CARE, SPARK, and Simons Searchlight.

Teaching

I have been actively involved in teaching and supervision of medical students, dental students, residents, fellows, postdocs, and junior faculty since I joined the Columbia faculty. I have personally mentored over 100 students in my research laboratory including thesis advisor, scholarly project advisor, thesis committee, and fellowship mentor. I was responsible for designing the course in human genetics for the first year medical and dental students at Columbia and have been the course director for the last 20 years and consistently receive outstanding assessments for the course. I have served as the fellowship director for our Molecular Genetics and Cytogenetics ABMGG training program. I was the Associate Director for Training in our NCI Comprehensive Cancer Center and the Medical Director for our genetic counseling graduate program. I frequently teach in CME courses. I have received numerous awards for teaching including the Presidential Award for Outstanding Teaching, Columbia's highest teaching award, and the Women's mentorship award from the AMA. I have written over 80 major chapters in textbooks and reviews in the literature. I also teach the public and

patients through numerous videos online describing our research findings and including major TED talks with over 4 million views.

Significant Supporting Activity - Clinical expertise

My clinical interests and expertise revolve around the practice of genetics. Since 2002 I have been actively involved in clinical care, providing treatment for patients with genetic conditions; I have led the neurofibromatosis center of excellence, the von Hippel Lindau center of excellence, founded and directed the DISCOVER program for undiagnosed diseases, and the TREATMENT program to develop new treatments. I receive referrals from colleagues locally, regionally, nationally, and internationally. Patients around the world seek me out to diagnose rare undiagnosed conditions. Patients also seek me out for my expertise in clinical conditions I have described. The programs that I created have grown and now evaluate and treat ~1000 patients each year and serve as a core training experience for medical students, residents, and fellows.

Significant Supporting Activity - Administration and Institutional Service

At Columbia, I served as the Director of Clinical Genetics and the Director of Cancer Genetics. I served as the Director of the Precision Medicine Resource of our Irving Institute of Clinical Research. I have served as a national leader in human genetics as a member of council at the National Human Genome Research Institute, the scientific advisory board of All of Us, a member of the board of the American Society of Human Genetics and position as Treasurer elect, organizer for the 2023 Gordon Conference on Human Genetics and Genomics, and as the original plaintiff in the Supreme Court case that overturned gene patents.

Summary

Since my appointment as Chief, Department of Pediatrics at BCH at HMS I have endeavored to provide excellent clinical care, administrative oversight, and teaching, while also conducting research and clinical trials in rare genetic conditions. Through my lecturing (at a regional, national and international level), my written works with an h index of 125, my clinical research, and my involvement with professional societies I have sought to improve the care for patients with genetic conditions I look forward to working on identification of the genetic basis for human disease, characterizing rare genetic conditions, and implementing genomic medicine at scale to improve health.