The Faculty of Medicine of Harvard University

Curriculum Vitae

Date Prepared: September 23, 2026

Name: Nina Beth Gold

Office Address:

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Education:

08/2005 - 05/2009	B.A. Summa cum laude and distinction in major	Psychology	Colby College
08/2010 - 05/2014	M.D.	Medicine	Harvard Medical School
08/2023 - 05/2025	M.S.	Medical bioinformatics	Harvard Medical School

Postdoctoral Training:

06/2014 - 06/2018	Resident	Combined Residency in Pediatrics and Medical Genetics	Boston Children's Hospital
06/2017 - 06/2018	Chief resident	Harvard Medical School Genetics Training Program	Boston Children's Hospital
07/2018 - 07/2019	Fellow	Medical Biochemical Genetics	Children's Hospital of Philadelphia

Faculty Academic Appointments:

2019 - 2022	Instructor	Pediatrics	Harvard Medical School
2022 - Present	Assistant Professor	Pediatrics	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions:

2019 - Present	Attending Physician, Division of Medical Genetics and Metabolism	Pediatrics	Massachusetts General Hospital
2021 - 2026	Associated Personnel	Division of Genetics and Genomics	Boston Children's Hospital

2024 - Present	Unpaid Affiliate	Medical and Population Genomics	Broad Institute

Faculty Membership in Harvard Initiatives, Programs, Centers, and Institutes

2025 - Present	Associate Member	Center for Genomic Medicine	Mass General Brigham

Other Professional Positions:

2009 - 2010	Clinical Research Coordinator, Active Malformations Surveillance Program (PI: Lewis Holmes, MD)	Massachusetts General Hospital and Brigham and Women's Hospital	30 hours / week
2022 - 2026	Consultant	RCG Consulting	2 hours / month

Major Administrative Leadership Positions:

Local

2020 - Present	Associate Director for Research	Mass General Brigham Personalized Medicine Boston, MA
2020 - Present	Director of Prenatal Medical Genetics	Massachusetts General Hospital, Department of Obstetrics and Gynecology Boston, MA
2022 - Present	Massachusetts General Hospital Medical Genetics Residency Director	Massachusetts General Hospital, Division of Medical Genetics and Metabolism Boston, MA
2025 - Present	Director of Metabolism	Massachusetts General Hospital, Division of Medical Genetics and Metabolism, Department of Pediatrics Boston, MA

Committee Service:

Local

2017 - 2018	Pediatric Clerkship Video Curriculum Working Group	OpenPediatrics
2020 - 2022	Perinatal Translational Research Committee	Massachusetts General Hospital

2020 - Present	Combined Pediatrics and Medical Genetics Residency Admissions Committee	Boston Children's Hospital / Harvard Medical School Genetics Training Program
2020 - Present	Harvard Medical School Genetics Training Program Admissions Committee	Harvard Medical School Genetics Training Program
2026 - Present	Genetics Implementation Collective	Harvard Medical School Founding member
2026 - Present	Reproductive Genetics Steering Committee	Mass General Brigham

Regional

2020	Medical/Clinical Follow-up Work Group	New England Metabolic Consortium Workgroup leader

National

2018 - 2020	Genetics and Genomics Residency Committee	Accreditation Council for Graduate Medical Education
2021	Executive Planning Committee for Workshop on Early Diagnostic Strategies	National Institutes of Health
2021 - 2022	Professional Practices and Guidelines Committee	American College of Medical Genetics
2022	Diagnostic Approaches and Access to Testing Working Group	National Organization for Rare Disorders Centers of Excellence Program
2023	Urea Cycle Disorders Variant Curation Expert Panel	ClinGen
2024 - 2026	Prenatal Sequencing Working Group	American College of Medical Genetics
2024 - Present	Molecular and Clinical Genetics Panel	Food and Drug Administration
2025 - 2026	Genomic Newborn Screening Working Group	American College of Medical Genetics
2025 - Present	Gorlin Syndrome Alliance Patient Registry Advisory Board	Gorlin Syndrome Alliance
2026 - Present	Advisory Board	Florida Sunshine Genetics Act Advisor

International

2024 - Present	Gene List Committee	International Consortium on Newborn Sequencing Co-chair

Professional Societies

2020 - Present	American College of Medical Genetics	Fellow
2024 - Present	International Consortium on Newborn Sequencing	Committee Co-chair

Grant Review Activities:

2023	Emerging Innovations in Health	Patient-Centered Outcomes Research Institute (PCORI) Reviewer

Editorial Activities:

- **Ad hoc Reviewer**

American Journal of Medical Genetics – Part A
American Journal of Medical Genetics – Part C
Genetics in Medicine
Human Genetics and Genomics Advances
Journal of Medical Genetics
New England Journal of Medicine
Orphanet Journal of Rare Diseases

Honors and Prizes:

2009	Psi Chi/Erlbaum Award in cognitive science	Psi Chi: International Honor Society in Psychology	Best undergraduate empirical study in cognitive science in United States
2011	Soma Weiss Day student speaker award	Harvard Medical School	Excellence in research
2012	Norfolk District Medical Society Scholarship	Massachusetts Medical Society	Educational scholarship
2017	Outstanding Resident Teaching Award	Harvard Medical School	Medical education
2017	Finalist in Clinical Decision Support Tool Challenge	Boston Children's Hospital Innovation and Digital Health Accelerator	Clinical innovation
2017	Peter Duncan Award	David W. Smith Workshop on Malformations and Morphogenesis	Best platform presentation by a trainee

2018	Shire/Genzyme ACMGF Next Generation Medical Biochemical Subspecialty Genetics Training Award	American College of Medical Genetics Foundation	Clinical training and translational research
2021 - 2022	APS SPR Journeys & Frontiers in Pediatrics Research Program	Pediatric Academic Societies and Society for Pediatric Research	Pediatric research, nominated by MGH Chief of Pediatrics
2023	Anne Klibanski Visiting Scholars Award	Mass General Brigham	Women with exceptional promise as leaders in their field
2023	David Holtzman Award (inaugural recipient)	Massachusetts General Hospital	International award for embodying excellence in science, medicine, innovation, and compassion
2025	Editor's Choice Award	Genetics in Medicine	Selected by the Genetics in Medicine as the #1 Editor's Choice article of 2025, recognizing the year's most impactful and innovative research in medical genetics

Report of Funded and Unfunded Projects

Past

2021 - 2022	Genotype-First Approach to Identification of Individuals with Inherited Errors of Metabolism Eleanor and Miles Shore Faculty Development Awards Harvard Medical School award PI ($15,000) This study used a genotype-first approach to identify individuals with variants associated with inherited metabolic disorders, characterize variable expressivity and penetrance, and assess barriers to diagnosis.
2021 - 2022	Understanding Perspectives on Genomic Research in an Ethnically and Racially Diverse Cohort of Parents Greenwall Foundation Making a Difference in Real-World Bioethics Dilemmas grant Co-Investigator (PI: Ingrid Holm, MD) This study used semi-structured interviews with a diverse cohort of parents to explore beliefs, hopes, and concerns related to genomic newborn screening.
2021 - 2025	Implementation of Whole Genome Sequencing as Screening in a Diverse Cohort of Healthy Infants National Center for Advancing Translational Sciences and Eunice Kennedy Shriver National Institute of Child Health and Human Development U01 U01TR003201-01

Co-investigator (PIs: Robert C. Green, MD, MPH; Ingrid Holm, MD, MPH)
This follow-up to the BabySeq Project evaluates the medical, behavioral, and economic outcomes of implementing genomic newborn screening in underrepresented infant populations across three diverse U.S. clinical sites.

2023 - 2024 — eMERGE Phase IV Clinical Center at Partners HealthCare
National Human Genome Research Institute U01 U01HG008685-05
Co-investigator (Elizabeth Karlson, MD)
This project leveraged a large biobank, electronic health record data, bioinformatics infrastructure, and expertise in genomic result return to support the development, implementation, and evaluation of polygenic risk scores in clinical care.

2024 - 2025 — Assessing Diagnostic Yield of Whole Genome Sequencing in Newborns with Neonatal Encephalopathy
MGB Department of Pediatrics BWH-MGH Newborn Medicine Collaborative Project
Co-investigator (PIs: Anna Duncan, MD, MHS; Richard Parad, MD, MPH)
This study evaluated the diagnostic yield of whole genome sequencing for identifying genetic contributors to neonatal encephalopathy.

2026 — OWLI Dependent Care Travel Award
Center for Academic Development and Enrichment, Mass General Brigham
Work-life integration award
PI ($500)
This institutional award was used to offset caregiver costs during a presentation at the National Academy of Medicine.

Current

2023 - 2028
NCE — Genome-First Approach to Treatable Genetic Conditions in Adults
National Human Genome Research Institute K08 K08HG012811
PI (Mentors: Robert C. Green, MD, MPH; Heidi Rehm, PhD; Pradeep Natarajan, MD, MMSc) ($1,093,802)
This five-year career development award supports a genome-first approach to identifying adults in hospital-based and population biobanks with genomic variants associated with undiagnosed, treatable genetic disorders.

2025 - 2026
NCE — Identification of Undiagnosed Adults at Risk for Treatable Genetic Disorders
National Academy of Medicine NAM Scholars in Diagnostic Excellence
PI ($40,000)
This award supports the identification of adults with undiagnosed inherited metabolic disorders in a hospital-based biobank and evaluates strategies for returning genomic results to biobank participants.

2025 - 2026
NCE — Exploring the Positive Predictive Value of Genomic Screening for Newborns and Children
MGH Department of Pediatrics Pilot and Feasibility award
PI ($25,000)
This pilot study evaluates individuals in a hospital-based biobank with disease-associated variants in over 100 genes linked to treatable inherited metabolic disorders to inform the positive predictive value of genomic screening in newborns and children.

2025 - 2027	Identification of Undiagnosed Adults at Risk for Treatable Genetic Disorders Executive Committee on Research at Massachusetts General Hospital Claflin Distinguished Scholar Awards PI ($120,000) This study identifies adults in a hospital-based biobank with disease-associated variants in treatable inherited metabolic disorder genes and evaluates evidence of previously unrecognized disease.
2025 - 2028	Feasibility of Genomic Newborn Screening Through Public Health Laboratories NIH Common Fund Common Fund Venture Initiative 1OT2OD040029 PI (Multiple PIs: Robert C. Green, MD, MPH; Ingrid Holm, MD, MPH, Melissa Wasserstein, MD) ($14,399,393) This first-of-its-kind contract supports the integration and evaluation of genomic newborn screening across multiple state public health laboratories in the United States.

Projects Submitted for Funding

Submitted 5/2026	AI-Generated Care Plans to Improve Emergency and Longitudinal Care for Children with Inherited Metabolic Disorders MIT-Hood Hub Innovation award Co-PI ($60,000) This pilot study develops and evaluates an AI-assisted, family-accessible clinical care plan platform to improve emergency and longitudinal care coordination for children with inherited metabolic disorders.

Unfunded Current Projects

2025 - Present	Personal Genetic Diagnoses Revealed Through Reproductive Carrier Screening PI This study uses more than 1 million carrier screening samples from two diagnostic laboratories and over 1,000 clinically ascertained cases from academic medical centers to estimate the prevalence and penetrance of personal genetic diagnoses incidentally revealed through reproductive carrier screening.
2026 - Present	Use of Known Genetic Diagnoses in the Electronic Medical Record PI/Mentor This study, being led by an HMS medical student, evaluates how established genetic diagnoses are documented, recognized, and incorporated into clinical care across electronic medical record notes from primary care, emergency medicine, surgical, and specialty settings.

Report of Local Teaching and Training

Teaching of Students in Courses:

2013 - 2014	Patient-Doctor II Second-year medical students	Harvard Medical School 4 hours / week
2016	Pediatrics clerkship tutor 1:1 third-year medical student tutoring	Boston Children's Hospital 1 hour / week

2018	Advanced Integrative Science Course in Human Genetics Second-year medical students	Boston Children's Hospital 1 hour / week
2020	Foundations Course First-year medical students	Harvard Medical School 1 hour / week

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

2020	Can't Miss Genetic Diagnoses Internal Medicine residents	Massachusetts General Hospital 1 hour
2020	Metabolic Emergencies Pediatrics residents	Massachusetts General Hospital 2 hours
2021	Trainee Session: Difficult Conversations Research trainees	Genomes2People, Brigham and Women's Hospital 2 hours
2023	Clinical Cases in Prenatal Genetics Reproductive endocrinology fellows	Massachusetts General Hospital 1 hour
2023	Metabolic Emergencies Pediatrics residents	Massachusetts General Hospital 1 hour
2024	Metabolic Emergencies Pediatrics residents	Massachusetts General Hospital 1 hour
2024	Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List Maternal-Fetal Medicine fellows	Brigham and Women's Hospital 1 hour
2025	Metabolic Emergencies Pediatrics residents	Massachusetts General Hospital 1 hour
2025 - 2026	Careers in Genetics panel Harvard Medical School Genetics Training Program trainees	Boston Children's Hospital 1 hour / year
2026	Colby @ Mass General Hospital College students	Colby College 1 hour

Clinical Supervisory and Training Responsibilities:

2019 - Present	Rotation director for residents (approximately 6 residents per year) Pediatrics residents	Massachusetts General Hospital 30 hours / year
2019 - Present	Director of rotating residents (approximately 8 residents per year) HMS medical genetics residents and medical biochemical genetics fellows	Massachusetts General Hospital 4 hours / week
2020 - 2021	Supervision of fellows in clinic (1 fellow per year) Maternal fetal medicine fellows	Massachusetts General Hospital 6 hours / year

2020 - 2021	Supervision of fellows in clinic (1 fellow per year) Reproductive endocrinology fellows	Massachusetts General Hospital 8 hours / year
2020 - Present	Supervision of residents in continuity clinic (approximately 2 residents per year) Pediatric neurology residents	Massachusetts General Hospital 12 hours / year
2020 - Present	Supervision of senior resident in continuity clinic (1 to 2 residents per year) HMS Genetics Training Program residents	Massachusetts General Hospital 6 hours / week

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

2021 - 2022	Sarina Madhavan, MD Harvard Medical School and Harvard Business School Second-author publication in Human Genetics and Genomics Advances
2026 - Present	Jiachen Sun Harvard Medical School Student-led research project in progress

Other Mentored Trainees and Faculty:

2020 - 2022	Alexa Nitka, CGC / Prenatal screening coordinator, Cedars-Sinai, Los Angeles, CA Served as Ms. Nitka's capstone research mentor for a project examining parental experiences after false positive or inconclusive newborn screening results for Pompe disease and mucopolysaccharidosis type I.
2021 - 2022	Ani Papazian, MD / Pediatrician, Atrius Health, Boston, MA Hosted Dr. Papazian for a month-long research elective during medical school, during which she contributed to multiple components of the BabySeq2 research project.
2022 - 2023	Jaclyn Omorodion, MD / Medical geneticist, Boston Children's Hospital, Boston, MA Served as Dr. Omorodion's research mentor during medical genetics residency for work that was selected for an oral platform presentation at the 2024 ACMG Annual Clinical Genetics Meeting and resulted in a first-author publication in Genetics in Medicine.
2022 - 2024	Lauren O'Grady, MS, CGC / Genetic counselor, Massachusetts General Hospital, Boston, MA Served as Ms. O'Grady's research mentor during her Sanofi-Genzyme–sponsored lysosomal storage disorders fellowship, supporting work that led to two first-author publications in the American Journal of Medical Genetics, one Opitz Award nomination, and a poster presentation at the 2024 ACMG Annual Clinical Genetics Meeting.
2022 - 2024	Sarah Bick, MD / Medical biochemical geneticist, Seattle Children's, Seattle, WA Served as Dr. Bick's T32 research mentor during medical genetics residency for work that was presented at the 2024 ACMG Annual Clinical Genetics Meeting,

	selected for a rapid-fire presentation at the 2024 ICoNS meeting, published as a first-author article in Genetics in Medicine, and named the journal's #1 Editor's Choice article of 2025.
2022 - Present	Thomas Minten, PhD / Medical student, KU Leuven, Leuven, Belgium Served as Dr. Minten's research mentor for work that was presented at the 2024 ACMG Annual Clinical Genetics Meeting, selected for a rapid-fire presentation at the 2024 ICoNS meeting, and published as a first-author article in Genetics in Medicine, now with an NIH RCR of 12.44.
2023 - 2024	Ilham Abbasi, MHSc / Senior genetics analyst, North York General Hospital, Ontario, Canada Served as Ms. Abbasi's research mentor during her Master of Health Science in Medical Genomics at the University of Toronto for work selected as a top abstract and rapid-fire presentation at the 2024 ACMG Annual Clinical Genetics Meeting.
2023 - 2024	Maya del Rosario, MS, CGC / Genetic counselor, Boston Children's Hospital, Boston, MA Served as Ms. del Rosario's capstone research mentor for a survey project evaluating genetic counselors' views on genomic newborn screening, which resulted in a first-author publication in Genetics in Medicine Open.
2024 - 2025	Julie Yeo, BA / Medical student, Georgetown University School of Medicine, Washington, D.C. Mentored Ms. Yeo across multiple research projects, including work on expert consensus guidelines for genomic newborn screening that received the MGH Clinical Research Day Pediatric Departmental Award and was selected for an oral platform presentation at the ACMG Annual Clinical Genetics Meeting.
2025 - 2026	Hannah Uebele, MS / Board-eligible genetic counselor, Boston, MA Served as Ms. Uebele's capstone research mentor for a project examining the symptoms of adults in the Mass General Brigham Biobank with genetic diagnoses of Noonan syndrome.
2025 - 2026	Harini Somanchi, BSPH / Medical student, UCLA David Geffen School of Medicine, Los Angeles, CA Mentored Ms. Somanchi across multiple research projects, resulting in co-authorship on multiple manuscripts and recognition at the May 2026 Mass General for Children Research Day, where she was selected for one of the prestigious Data Blitz presentations and received a Travel Award for best population health project. She was also selected for a Lightning Presentation at the 2026 International Consortium on Newborn Sequencing meeting.
2025 - 2026	Michelle Wang, MD / Fellow, Maternal-Fetal Medicine; Resident, Medical Genetics, HMS Genetics Training Program, Boston, MA Currently mentoring Dr. Wang on a study estimating the prevalence and penetrance of personal genetic diagnoses incidentally revealed through reproductive carrier screening, which has resulted in a prestigious oral presentation at the Society for Maternal Fetal Medicine Global conference.

Local Invited Presentations:

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

2011	MitoCase Conference (series of 6 conferences) / Case presentations

	Mootha Lab, Massachusetts General Hospital, Boston, MA
2014	Penetrance of Actionable Genomic Findings in Exomes from the Framingham Heart Study / Research presentation Department of Genetics, Harvard Medical School, Boston, MA
2016	Case from the Wards: 3-year-old Boy with Eye Pain / Case presentation Department of Medicine, Boston Children's Hospital, Boston, MA
2016	Genetics in Primary Care / Teaching presentation Children's Hospital Primary Care Center, Boston Children's Hospital, Boston, MA
2017	Morbidity and Mortality Conference / Case presentation Division of Genetics, Boston Children's Hospital, Boston, MA
2017	MitoCase Conference / Case presentation Mootha Lab, Massachusetts General Hospital, Boston, MA
2017	Metabolic Emergencies / Teaching presentation Division of Emergency Medicine, Boston Children's Hospital, Boston, MA
2017	Health Equity Rounds / Teaching presentation Department of Pediatrics, Boston Medical Center, Boston, MA
2017	Fellow's Perspective / Teaching presentation Precision Medicine Summit, Boston Children's Hospital, Boston, MA
2017	Culture of the Wards / Panel Harvard Medical School, Boston, MA
2018	Senior Rounds / Case presentation Boston Combined Residency Program Alumni Reunion, Boston, MA
2018	Genetics Grand Rounds / Case presentation Division of Genetics and Metabolism, Children's Hospital of Philadelphia, Philadelphia, PA
2019	Emergency Laboratory Evaluations for Patients with Inborn Errors of Metabolism / Teaching presentation Division of Medical Genetics and Metabolism, Massachusetts General Hospital, Boston, MA
2020	MitoCase Conference / Case presentation Mootha Lab, Massachusetts General Hospital, Boston, MA
2020	Pediatric Diagnostic Criteria for Basal Cell Nevus Carcinoma Syndrome / Research presentation Division of Medical Genetics and Metabolism, Massachusetts General Hospital, Boston, MA
2020	Genetic Causes of Disorders of Sexual Differentiation / Teaching presentation Department of Pediatrics, Massachusetts General Hospital, Boston, MA
2021	Metabolic Emergencies / Teaching presentation Department of Pediatrics, Massachusetts General Hospital, Boston, MA
2021	Emergency Evaluations of Children with Inherited Metabolic Disorders / Grand rounds

	Department of Pediatrics, Massachusetts General Hospital, Boston, MA
2021	Genetic Testing Methodologies for Neurogenetics / Teaching presentation Department of Neurology, Massachusetts General Hospital, Boston, MA
2021	Rare Genetic Causes of Obesity / Teaching presentation Division of Pediatric Endocrinology, Massachusetts General Hospital, Boston, MA
2021	Introduction to Metabolism / Teaching presentation Harvard Medical School Genetics Training Program, Harvard Medical School, Boston, MA
2022	Brief Introduction to Testing Modalities and Overview of Can't Miss Diagnoses / Teaching presentation Department of Medicine, Massachusetts General Hospital, Boston, MA
2022	Genomic Screening Tools: What We Know and How Far We Have to Go / Research presentation Division of Medical Genetics and Metabolism, Massachusetts General Hospital, Boston, MA
2023	Penetrance and Expressivity of Pathogenic and Likely Pathogenic Variants / Teaching presentation Department of Obstetrics and Gynecology, Massachusetts General Hospital, Boston, MA
2023	NEJM Clinicopathological Conference: 50-year-old Woman with Confusion and Falls / Case presentation Department of Medicine, Massachusetts General Hospital, Boston, MA
2023	Genomic Screening Tools: What We Know and How Far We Have to Go / Research presentation Medical and Population Genetics Primer, Broad Institute, Boston, MA
2023	Clinical Dysmorphology: What Utilization and Impact Does It Have Today? / Panel Genetic counseling program, MGH Institute of Health Professions, Boston, MA
2023	Brief Introduction to Testing Modalities and Overview of Can't Miss Diagnoses / Teaching session Department of Medicine, Massachusetts General Hospital, Boston, MA
2023	Reevaluating the "Right Not to Know" in Genomics Research / Teaching presentation Genetic counseling group, Broad Institute, Boston, MA
2024	Annual Holmes Lecture: The Road to Genomic Newborn Screening / Grand rounds Department of Pediatrics, Massachusetts General Hospital, Boston, MA
2024	Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List / Research presentation Division of Medical Genetics and Metabolism, Massachusetts General Hospital, Boston, MA

2024	The Road to Genomic Newborn Screening / Grand rounds Department of Pediatrics, Cambridge Health Alliance, Cambridge, MA
2025	MGH Internal Medicine Residency Noon Conference: Genetics in Internal Medicine / Research presentation Department of Medicine, Mass General Brigham, Boston, MA
2025	Brigham and Women's Genetic Counseling Retreat: Advancing Precision Care in Pregnancy Through an Actionable Fetal Findings List / Research presentation Department of Obstetrics and Gynecology, Mass General Brigham, Boston, MA
2025	Identification of Treatable Genetic Disorders in Fetuses and Newborns / Research presentation (invited faculty speaker at 16th MGfC Pediatric Research Day) Department of Pediatrics, Massachusetts General Hospital, Boston, MA
2025	Metabolism monthly case series: Maple Syrup Urine Disease / Case presentation Division of Endocrinology, Department of Medicine, Mass General Brigham, Boston, MA
2025	Metabolic Emergencies / Teaching presentation Department of Emergency Medicine, Boston Children's Hospital, Boston, MA
2025	Metabolic Emergencies / Teaching presentation Departments of Pediatric Emergency Medicine, satellites of Boston Children's Hospital, Boston, MA
2026	Estimating the Lifelong Penetrance of Inherited Metabolic Disorders / Research presentation (invited faculty speaker at 17th MGfC Pediatric Research Day) Department of Pediatrics, Mass General Brigham, Boston, MA
2026	We Can Only Treat What We First Find: Genomic Screening Across the Lifespan / Research presentation Medical and Population Genomics Rounds, Broad Institute, Boston, MA
2026	BEACONS-NBS: Genomic Screening of Infants Across Seven U.S. States and Territories / Research presentation Division of Medical Genetics and Metabolism, Mass General Brigham, Boston, MA
2026	Newborn Medicine Grand Rounds: How Do We Decide Which Conditions to Include? / Grand rounds Division of Newborn Medicine, Boston Children's Hospital, Boston, MA
2026	Assessing the Prevalence and Penetrance of Genomic Changes Across the Lifespan / Research presentation Genomics Implementation Collective, Mass General Brigham, Boston, MA

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

2009	Anatomic and Etiological Classification of Congenital Limb Deficiencies / Selected oral abstract

Canada-New England-New York (CANNEW) session, Portsmouth, NH

National

2017 Natural History of Nevoid Basal Cell Carcinoma (Gorlin) Syndrome / Selected oral presentation
David W. Smith Workshop on Malformations and Morphogenesis, Stowe, VT

2018 Resident Recruitment Panel / Plenary session
Association of Professors of Human and Medical Genetics Workshop, Santa Fe, NM

2018 Promoting Student Engagement in Genetics / Panel
Association of Professors of Human and Medical Genetics Workshop, Santa Fe, NM

2021 Low Frequency of Treatable Pediatric Disease Alleles in gnomAD: An Opportunity for Future Genomic Newborn Screening / Selected oral abstract
American College of Medical Genetics, Virtual

2023 Clinical Genomics Careers Panel Series: Physician Clinicians / Panel
ClinGen consortium, Virtual

2023 Genomic Newborn Screening: What We Know and How Far We Have to Go / Invited talk
Ambry Genetics EducateNext Webinar, Virtual (Ambry Genetics)

2023 Softening the Curious Gaze (presented in absentia by Dr. Angela Lin) / Selected session
American College of Medical Genetics, Salt Lake City, UT

2023 Genomic Newborn Screening: What We Know and How Far We Have to Go / Invited talk
Baylor College of Medicine, Houston, TX

2023 The Road to Genomic Newborn Screening / Invited talk
Lucile Packard Children's Hospital, Palo Alto, CA

2023 The Road to Genomic Newborn Screening / Invited talk
Children's National Hospital, Washington, D.C.

2024 Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List / Invited talk
University of North Carolina, Department of Genetics, Chapel Hill, NC

2024 The Road to Genomic Newborn Screening / Invited talk
Pacific Northwest Research Institute, Seattle, WA (virtual)

2025 International Consortium on Newborn Sequencing (ICoNS) consensus guidelines for gene selection in genomic newborn screening (presented in absentia by Julie Yeo) / Selected oral presentation
American College of Medical Genetics, Los Angeles, CA

2025 Roundtable Discussion on Newborn Screening / Advisory panel
National Academies of Sciences, Engineering, and Medicine, Washington, D.C.

2025 Center for Precision Health Research Seminar Series: Identification of Treatable Genetic Disorders in Fetuses and Newborns / Invited talk

	National Human Genome Research Institute, Washington, D.C. (virtual)
2025	A Genotype-first Approach Among Adults at Risk for Treatable Monogenic Disorders Reveals Underdiagnosis and Incomplete Penetrance / Selected oral presentation American College of Medical Genetics, Los Angeles, CA
2025	Identification of Treatable Disorders in Fetuses and Newborns / Invited talk Children's Health Symposium hosted by the Wyss Institute Diagnostics Accelerator, Boston, MA
2025	Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List / Keynote speaker Basic Training in Reproductive Medical Genetics Course, Columbia University Irving Medical Center, New York, NY (virtual)
2025	Advancing Precision Care in Pregnancy Through an Actionable Fetal Findings List / Selected oral presentation University of California San Francisco, San Francisco, CA
2026	Works in Progress: BabySeq 3 / Research presentation Genomics Implementation Collective, Mass General Brigham, Boston, MA
2026	Before the Symptoms Start / Keynote speaker HackRare Hackathon, Cambridge, MA
2026	The BEACONS-NBS Study / Invited talk Immune Deficiency Foundation, Virtual
2026	Long-Term Penetrance of Disease Variants in Genes Prioritized for Genomic Newborn Screening: Evidence from Adult Biobanks / Research presentation National Academy of Medicine, Scholars in Diagnostic Excellence, Washington, D.C.

International

2018	Nationwide Variation in the Emergency Laboratory Evaluation of Children with Inborn Errors of Metabolism / Selected oral abstract Pediatric Academic Societies, Toronto, Canada
2024	Using Clinical Cases to Illustrate Preconception Planning Among Women with Inherited Metabolic Disorders / Selected session American College of Medical Genetics, Toronto, Canada
2024	The Blurred Line Between Prenatal Diagnostic Testing and Genomic Screening / Invited talk International Society for Prenatal Diagnosis, Boston, MA
2024	Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List / Invited talk Fetal Sequencing Consortium, Virtual
2024	Gene List Committee Update / Invited talk International Consortium on Newborn Sequencing, New York, NY
2025	Advancing Precision Care in Pregnancy Through a Treatable Fetal Findings List / Invited talk

	33rd Annual Symposium – New Developments in Prenatal Diagnosis & Medical Genetics, Toronto, Canada

Report of Clinical Activities and Innovations

Past and Current Licensure and Board Certification:

2019	Massachusetts medical license
2021	Maine medical license
2021	New Hampshire medical license
2021	Rhode Island medical license

Practice Activities:

2019 - 2021	Medical geneticist	MGHfC Division of Medical Genetics and Metabolism, Boston, MA	30 hours / week (7 clinics/week)
2021 - 2023	Medical geneticist	MGHfC Division of Medical Genetics and Metabolism, Boston, MA	14 hours / week (3.5 clinics/week)
2023 - Present	Medical geneticist	MGfC Division of Medical Genetics and Metabolism, Boston, MA	6 hours / week (1.5 clinics/week)

Clinical Innovations:

Guideline for the Management of Acute Hyperammonemia in Adults (2025 – Present)	Guidelines for MGH pharmacy and inpatient teams to manage the care of adults with hyperammonemia

Report of Education of Patients and Service to the Community

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

Activities

2010	Personal Genetics Education Program / Participation in programs that contribute to K-12 education
2012	Health Professions Recruitment Program / Mentor to a Boston high school student

Educational Material for Patients and the Lay Community:

Books, articles, and presentations in other media

2017	Clinical Genetics 101: When to Refer	Article	Notes: Boston Children's clinical health blog
2017	If I Have the Mutation, Will I Get the Disease? New Research Looks at Genetic Penetrance	Interview	Vector: Boston Children's Hospital's science and clinical innovation blog
2017	In Search of Young Medical Geneticists	Article	Vector: Boston Children's Hospital's science and clinical innovation blog
2019	"Diagnosis": Episode 1	Expert consultant	Netflix series

Recognition:

2025 - Present	Rising Mentor Award nominee	Center for Faculty Development

Report of Scholarship

* denotes equal authorship contribution

** denotes mentored trainee.

Peer-Reviewed Scholarship in print or other media:

Research Investigations

1. **Gold NB**, Westgate MN, Holmes LB. Anatomic and etiological classification of congenital limb deficiencies. Am J Med Genet A. 2011 Jun; 155A(6):1225-35. PMID: 21557466
2. Lieber DS, Calvo SE, Shanahan K, Slate NG, Liu S, Hershman SG, **Gold NB**, Chapman BA, Thorburn DR, Berry GT, Schmahmann JD, Borowsky ML, Mueller DM, Sims KB, Mootha VK. Targeted exome sequencing of suspected mitochondrial disorders. Neurology. 2013 May 07; 80(19):1762-70. PMID: 23596069
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Non-peer reviewed scholarship in print or other media:

Letters to the Editor

1. Vockley J, Brunetti-Pierri N, Chung WK, Clarke AJ, **Gold N**, Green RC, Kagan S, Moroz T, Schaaf CP, Schulz M, De Baere E. Response to Beretich and Beretich. Genet Med. 2023 Oct; 25(10):100903. PMID: 37245089

Professional educational materials or reports, in print or other media:

1. Gold NB, Byler S. Genetics. In: Chiel L, ed. Housestaff Manual 2.0. http://www.thebcrp.com

Local/Unpublished Clinical Guidelines and Reports:

1. Guidelines for the Use of Arginine in Pediatric Patients

Manuscripts Submitted to Preprint Servers

1. **Gold NB**, Zouk H, Yeo J, Lipsitz S, Koyama S, Somanchi H, Perez E, Selvaraj MS, O'Grady L, Miller E, Lewis ACF, Karlson EW, Strong A, Gold JI, Rehm HL, Natarajan P, Green RC. Long-term Penetrance of Disease Variants in Genes Prioritized for Genomic Newborn Screening: Evidence from Adult Biobanks. *medRxiv* [Preprint]. 2026 Jun 12:2026.06.10.26355380. doi: 10.64898/2026.06.10.26355380. PMID: 42326822; PMCID: PMC13278267.

Narrative Report

I am a physician-scientist, medical geneticist, and medical biochemical geneticist whose work focuses on identifying individuals with rare, treatable genetic conditions before irreversible symptoms develop. As I

practicing clinician, I care for and provide consults to pediatric and adult patients with genetic and metabolic disorders, on both inpatient units and outpatient clinics at MGH. My research uses genomic information from individuals across the lifespan— including fetuses, newborns, children, and adults—to evaluate disease risk, understand penetrance, and guide clinical implementation.

My interest in genomic screening began during medical school, when I worked with Dr. Robert Green to study penetrance of actionable genomic findings in population cohorts. That early work shaped the central question of my career: once a genomic variant is identified, how do we determine who is truly at risk, who may benefit from intervention, and how health systems should responsibly return and act on this information?

After completing combined residency training in pediatrics and medical genetics at Boston Children's Hospital, where I served as chief resident of the Harvard Medical School Genetics Training Program, I completed fellowship training in medical biochemical genetics at the Children's Hospital of Philadelphia. This training deepened my focus on inherited metabolic disorders, for which early diagnosis can be lifesaving but delayed diagnosis remains common. My 2025 M.S. in Medical Informatics from Harvard Medical School, completed while I was an attending physician, strengthened my ability to conduct many of my own statistical analyses and gave me a deeper understanding of data science, analytics, and clinical informatics.

Since joining the faculty at Harvard Medical School and Massachusetts General Hospital, I have built an independent research program focused on the prevalence, penetrance, and clinical implementation of genomic screening for treatable genetic disorders. My work is supported by an NHGRI K08 award, the National Academy of Medicine Scholars in Diagnostic Excellence program, the MGH Claflin Distinguished Scholar Award, and an MGH Department of Pediatrics Pilot and Feasibility Award. These studies use genotype-first approaches in hospital-based and population biobanks, paired with electronic health record review and clinical recontact, to identify adults with potentially unrecognized treatable genetic disease. A major goal is to inform the positive predictive value and clinical utility of genomic screening in newborns and children. My clinical and administrative roles as Director of Prenatal Medical Genetics, Director of Metabolism, Medical Genetics Residency Director at Massachusetts General Hospital, and Associate Director for Research for the Mass General Brigham Biobank are closely aligned with this scholarship.

In parallel, I have become a national and international leader in genomic newborn screening. I serve as co-chair of the Gene List Committee for the International Consortium on Newborn Sequencing and helped lead consensus recommendations for applying the Wilson-Jungner screening principles to genomic newborn screening. My work has been recognized by Genetics in Medicine, which selected one of my mentored publications as its most impactful genetics research of 2025.

I now serve as a principal investigator of BRIDGES-NBS, a $14 million NIH Common Fund Venture Initiative project evaluating genomic newborn screening across multiple state public health laboratories. Through this work, I am helping to develop the evidence base, operational infrastructure, and ethical framework needed to determine which conditions should be included in genomic newborn screening and how results should be returned to families and clinicians. I have led the development of the list of nearly 800 conditions to be screened, as well as the corresponding action sheets designed to help primary care pediatricians and other community-based clinicians care for children with positive screening results. Ultimately, this work aims to accelerate the translation of early genomic diagnosis into timely, life-saving, and life-transforming clinical care in the community.

Mentorship and education are also central to my academic identity. I direct the Massachusetts General Hospital rotation for Harvard Medical School genetics trainees, and mentor students, residents, fellows,

genetic counselors, and junior collaborators across institutions. My mentees have led first-author publications, presented at national meetings, and received awards for their work.

Across my research, clinical, and educational roles, my long-term goal is to define how genomic screening can be used responsibly to identify treatable genetic disease early enough to improve outcomes. I hope my work will help clarify which conditions should be prioritized for screening, how to interpret the risk associated with genomic findings, and how to build equitable systems that return results in ways that are useful for patients, families, clinicians, and public health programs.