The Faculty of Medicine of Harvard University
Curriculum Vitae

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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
09/2023-	Masters of Biomedical Informatics (in process)		Harvard Medical School

Postdoctoral Training:

06/14-06/18	Resident	Pediatrics and Medical Genetics (combined program)	Boston Children's Hospital
07/18-07/19	Fellow	Medical Biochemical Genetics	Children's Hospital of Philadelphia

Faculty Academic Appointments:

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

Appointments at Hospitals/Affiliated Institutions:

2019	Assistant	Pediatrics	Massachusetts General Hospital
2021	Associated personnel	Division of Genetics and Genomics	Boston Children's Hospital

Other Professional Positions:

2009-2010	Research assistant Active Malformations Surveillance Program (Lewis Holmes, MD)	Massachusetts General Hospital and Brigham and Women's Hospital	
2022	Consultant	RCG Consulting	2 hours/month

Major Administrative Leadership Positions:

Local

2017-2018	Chief resident	Harvard Medical School Genetics Training Program
2020-	Associate Director for Research	Mass General Brigham Personalized Medicine
2020-	Director of Prenatal Medical Genetics	Massachusetts General Hospital Department of Obstetrics and Gynecology
2022-	Massachusetts General Hospital Medical Genetics Residency Director	Massachusetts General Hospital Division of Medical Genetics and Metabolism

Committee Service:

Local

2017-2018	Pediatric clerkship video curriculum working group	OpenPediatrics
2020-	Harvard Medical School Genetics Training Program admissions committee	Harvard Medical School Genetics Training Program

2020-	Pediatrics residency recruitment	Mass General Hospital for Children
2020-	Pediatrics-genetics combined residency admissions committee	Boston Children's Hospital / Harvard Medical School Genetics Training Program
2020-	Perinatal translational research committee	Massachusetts General Hospital

Regional

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

National

2018-2020	Genetics and Genomics Residency Committee	Accreditation Council for Graduate Medical Education Medical
	2018-2020	Resident member
2021-	Executive Planning Committee for Workshop on Early Diagnostic Strategies	National Institutes of Health
2021-	Professional Practices and Guidelines Committee	American College of Medical Genetics
2022	Diagnostic Approaches / Access to Testing Working Group	National Organization for Rare Disorders Centers of Excellence program
2023-2023	Urea Cycle Disorders Variant Curation Expert Panel	ClinGen

International

2024	Co-chair	International Consortium on Newborn Screening, Gene List committee

Professional Societies:

2020-	American College of Medical Genetics

Grant Review Activities:

2023	Emerging Innovations in Health (EI)	Patient-Centered Outcomes Research Institute (PCORI)
	2023	Grant reviewer (ad hoc)

Editorial Activities:

Ad hoc Reviewer

Human Genetics and Genomics Advances

Journal of Medical Genetics

American Journal of Medical Genetics – Part A

American Journal of Medical Genetics – Part C

Genetics in Medicine

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	Finalist in Clinical Decision Support Tool Challenge	Boston Children's Hospital Innovation and Digital Health Accelerator	Clinical innovation
2017	Outstanding Resident Teaching Award	Harvard Medical School	Medical education

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)	Mootha laboratory	International award for embodying excellence in science, medicine, innovation, and compassion

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 PI ($15,000) We ascertained individuals with genotypes associated with inherited metabolic disorders (IMD) to describe the variable expression and penetrance of IMD and identify barriers to diagnosis of these disorders.
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: Dr. Ingrid Holm) We performed a series of semi-structured interviews with diverse parents of

infants to learn more about their beliefs, hopes, and concerns regarding genomic newborn screening.

Current

2021-2026 Implementation of Whole Genome Sequencing as Screening in a Diverse Cohort of Healthy Infants
NIH grant TR003201
Co-Investigator (PI: Robert Green, MD, MPH and Ingrid Holm, MD, MPH)
This multi-center project aims to develop a recruitment and retention strategy to enroll 500 apparently healthy, ethnically, and racially diverse infants (0-6 months) into a randomized controlled trial of genome sequencing.

2023-2024 eMERGE Phase IV Clinical Center at Partners HealthCare
National Human Genome Research Institute; NIH grant U01HG008685-05
Co-Investigator (PI: Elizabeth Karlson)
To enable the application of PRS development and implementation, eMERGE IV from Partners HealthCare leverages a large biobank, clinical data in the electronic health records, advanced bioinformatics expertise, state-of-the-art genetic analysis, established expertise in returning genomics results, and experience using information technology to transform clinical processes and assessing outcomes.

2023- Genome-first approach to treatable genetic conditions in adults
National Human Genome Research Institute; NIH grant 1K08HG012811-01
PI ($1,093,802)
Five-year research career development program focused on the identification of individuals in a hospital-based biobank with genomic variants associated with undiagnosed treatable genetic disorders.

Projects Submitted for Funding

2024- Identification of deleterious variants in treatable disease genes
PI ($120,000)
Identification of deleterious variants in participants in the MGB Biobank across 300 genes associated with treatable disorders

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
2016-2016	Pediatrics clerkship tutor 1:1 third-year medical student	Boston Children's Hospital 1 hour / week
2018-2018	Advanced Integrative Science Course in Human Genetics Second-year medical students	Boston Children's Hospital 1 hour / week
2020-2020	Foundations course First-year medical students	Harvard Medical School 1 hour / week

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

2016-2018	Noon conference, genetics and metabolism teaching block Pediatrics residents	Boston Children's Hospital 6 hours / year
2017	Senior Rounds Pediatrics residents	Boston Children's Hospital 1 hour / year
2020	Noon conference Internal Medicine residents	Massachusetts General Hospital 1 hour / year
2020	Noon conference Pediatrics residents	Massachusetts General Hospital 2 hours / year
2021-2021	Trainee session Research trainees	Genomes2People, Brigham and Women's Hospital 2 hours / year Two 1-hour presentations
2023	Clinical cases in prenatal genetics Reproductive endocrinology fellows	Massachusetts General Hospital 1 hour / year
2023	Metabolic Emergencies Pediatrics residents	Massachuetts General Hospital 1 hour / year

Clinical Supervisory and Training Responsibilities:

2019-	Supervision of rotating residents (approximately 6 residents per year) Pediatrics residents	Massachusetts General HospitalMassachusetts General

		Hospital 30 hours / year
2019-	Supervision of rotating residents (approximately 8 residents per year) HMS genetics residents	Massachusetts General Hospital 4 hours / week Weekly 4-hour clinic and inpatient consultations when on call (2 months)
2020-	Supervision of residents in continuity clinic (2 residents per year) Pediatric neurology residents	Massachusetts General Hospital 12 hours / year 4-8 hours of supervision in clinic per resident
2020-2021	Supervision of fellows in clinic (1 fellow per year) Maternal fetal medicine fellows	Massachusetts General Hospital 6 hours / year 4-8 hours of supervision in clinic
2020-2021	Supervision of fellows in clinic (1 fellow per year) Reproductive endocrinology fellows	Massachusetts General Hospital 8 hours / year 8 hours of supervision in clinic
2020-	Supervision of senior resident in continuity clinic (1 resident) HMS Genetics Training Program	Massachusetts General Hospital 6 hours / week Weekly 6-hour clinic

Research Supervisory and Training Responsibilities:

2020-2022	Research mentor Alexa Nitka / Genetic counseling student, MGH Institute of Health Professions	Massachusetts General Hospital 20 hours / year
2020-	Research mentor Jacklyn Omorodion, MD / Resident, Combined Pediatrics and Medical Genetics	Massachusetts General Hospital 20 hours / year
2021-2021	Research mentor Sarina Madhavan / MD/MBA candidate, Harvard Medical School and Harvard Business School	Massachusetts General Hospital 15 hours / year 1-month research elective

2021-2021	Research mentor Ani Papazian / MD candidate, Tufts Medical School	Massachusetts General Hospital 10 hours / year 1-month research elective

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

2021-2022 Sarina Madhavan, Harvard Medical School / Harvard Business School
Second author on a publication currently under review

Other Mentored Trainees and Faculty:

2020-2022 Alexa Nitka, CGC / Genetic counselor, UCLA Health
Genetic counseling student at MGH Institute of Health Professions, capstone project mentor
Completed capstone project on parental perspectives of positive newborn screening results for mucopolysaccharidosis type I

2021-2022 Ani Papazian, MD / Pediatrics resident, UC San Diego
Fourth-year medical student at Tufts Medical School, participated in a one-month research elective
Created research resources with other members of the Genomes2People research group

2022- Jaclyn Omorodion, MD / Combined pediatrics and medical genetics resident
Combined pediatrics and medical genetics resident at Boston Children's Hospital, research mentor
Ongoing research project on healthcare disparities in genetic testing for hereditary ovarian cancer

2022- Sarah Bick, MD / Harvard Medical School Genetics Training Program resident
Resident in Harvard Medical School Genetics Training Program, research mentor
Ongoing research project on molecular and biochemical diagnoses of patients followed in Boston Children's Hospital metabolism clinic

Local Invited Presentations:

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

2011	MitoCase conference (series of 6 conferences) Mootha Lab, Massachusetts General Hospital
2014	Penetrance of Actionable Genomic Findings in Exomes from the Framingham Heart Study / Genetics floor meeting presentation Harvard Medical School, Department of Genetics
2016	Case from the Wards: 3-year-old boy with eye pain / Grand Rounds Boston Children's Hospital, Department of Medicine
2016	Genetics in Primary Care / Faculty lecture Children's Hospital Primary Care Center, Boston Children's Hospital
2017	Culture of the Wards / Panel discussion Harvard Medical School
2017	Fellow's Perspective / Precision Medicine Summit Boston Children's Hospital, Division of Genetics and Genomics
2017	Health Equity Rounds / Boston Medical Center Grand Rounds Boston Medical Center, Department of Pediatrics
2017	Metabolic Emergencies / Faculty lecture Boston Children's Hospital, Division of Emergency Medicine
2017	MitoCase conference Mootha Lab, Massachusetts General Hospital
2017	Morbidity and Mortality conference Boston Children's Hospital, Division of Genetics
2018	Genetics Grand Rounds Children's Hospital of Philadelphia, Division of Genetics and Metabolism
2018	Senior Rounds / Boston Combined Residency Program Alumni Reunion Boston Children's Hospital, Department of Medicine
2019	Emergency laboratory evaluations for patients with inborn errors of metabolism Massachusetts General Hospital, Division of Medical Genetics and Metabolism

2020	Genetic causes of disorders of sexual differentiation / Case from the Wards Massachusetts General Hospital, Department of Pediatrics
2020	Mitocase conference Mootha Lab, Massachusetts General Hospital
2020	Pediatric Diagnostic Criteria for Basal Cell Nevus Carcinoma Syndrome Massachusetts General Hospital, Division of Medical Genetics and Metabolism
2021	Genetic testing methodologies for neurogenetics / Division conference Massachusetts General Hospital, Division of Pediatric Neurology
2021	Emergency evaluations of children with inherited metabolic disorders / Grand Rounds Massachusetts General Hospital, Department of Pediatrics
2021	Metabolic emergencies / Noon conference Massachusetts General Hospital / Department of Pediatrics
2021	Rare Genetic Causes of Obesity / Division conference Massachusetts General Hospital, Division of Pediatric Endocrinology
2021	Introduction to metabolism / Harvard Medical School Genetics Training Program Boston Children's Hospital / Teaching session
2022	Brief introduction to to testing modalities and overview of can't miss diagnoses / Teaching session Massachusetts General Hospital, Department of Medicine, Advanced Pathways program
2022	Genomic screening tools: What we know and how far we have to go / Division conference Massachusetts General Hospital, Division of Medical Genetics and Metabolism
2023	Genomic screening tools: What we know and how far we have to go Medical and Population Genetics Primer, Broad Institute

2023	NEJM Clinicopathological Conference (CPC): 50-year-old woman with confusion and falls Department of Medicine, Massachusetts General Hospital
2023	Penetrance and expressivity of pathogenic and likely pathogenic variants Maternal Fetal Medicine, Obstetrics and Gynecology / Massachusetts General Hospital
2023	Clinical Dysmorphology: What Utilization and Impact Does It Have Today? (panel discussion) Empowerment: An anti-oppressive approach to genetic counseling (Sponsored by: MGH Institute of Health Professions' Genetic Counseling Program)
2023	Genomic Newborn Screening: What We Know and How Far We Have to Go Ambry Genetics EducateNext Webinar
2023	Clinical Genomics Careers Panel Series: Physician Clinicians ClinGen
2023	Reevaluating the "right nottoknow" in genomics research Genetic counselors at Broad Institute
2023	Reevaluating the "right nottoknow" in genomics research / Invited Lecture Genetic counselors, Broad Institute
2023	Brief introduction to to testing modalities and overview of can't miss diagnoses / Teaching session Massachusetts General Hospital, Department of Medicine

Report of Regional, National and International Invited Teaching and Presentations

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

Regional

2009	Anatomic and etiological classification of congenital limb deficiencies / Canada-New England-New York (CANNEW) session (selected abstract) Portsmouth, NH

National

2017	Natural history of nevoid basal cell carcinoma (Gorlin) syndrome / Platform presentation (selected abstract) David W. Smith Workshop on Malformations and Morphogenesis, Stowe, VT
2018	Plenary session: Resident recruitment panel / Panel presentation Association of Professors of Human and Medical Genetics Workshop, Santa Fe, NM
2018	Promoting student engagement in genetics / Panel presentation 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 / Platform presentation (selected abstract) American College of Medical Genetics Annual Conference
2023	Softening the curious gaze (selected presentation) / Workshop American College of Medical Genetics conference (presented in absentia by Dr. Angela Lin)
2023	Genomic Newborn Screening: What We Know and How Far We Have to Go / Invited Lecture Baylor College of Medicine Molecular and Human Genetics Seminar Series
2023	The Road to Genomic Newborn Screening / Invited Lecture Lucille Packard Hospital, Division of Medical Genetics
2023	The Road to Genomic Newborn Screening / Invited Lecture Children's National Hospital, Division of Genetics

International

2018	Nationwide variation in the emergency laboratory evaluation of children with inborn errors of metabolism / Presentation (selected abstract) Pediatric Academic Societies Meeting, Toronto, Canada

Report of Clinical Activities and Innovations

Past and Current Licensure and Certification:

2019-	Massachusetts medical license
2021-2022	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	30 hours / week 7.5 clinics/week
2021-	Medical geneticist	MGHfC Division of Medical Genetics and Metabolism	14 hours / week 3.5 clinics/week

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 (pgEd), Consultant
2012	Health Professions Recruitment Program (HPREP)

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	Contributor	Netflix series

Report of Scholarship

ORCID: 0000-0003-0867-722X

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):1225-1235. PMID: 21557466. https://doi.org/10.1002/ajmg.a.33999. Epub 2011 May 09

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 7;80(19):1762-1770. PMID: 23596069. PMCID: PMC3719425. https://doi.org/10.1212/WNL.0b013e3182918c40. Epub 2013 Apr 17

3. Natarajan P,* **Gold NB**,* Bick AG,* McLaughlin H, Kraft P, Rehm HL, Peloso GM, Wilson JG, Correa A, Seidman JG, Seidman CE, Kathiresan S, Green RC. Aggregate penetrance of genomic variants for actionable disorders in European and African Americans. Sci Transl Med. 2016 Nov 9;8(364):364ra151. PMID: 27831900. PMCID: PMC5823271

4. Carapito R, Ivanova EL, Morlon A, Meng L, Molitor A, Erdmann E, Kieffer B, Pichot A, Naegely L, Kolmer A, Paul N, Hanauer A, Tran Mau-Them F, Jean-Marçais N, Hiatt SM, Cooper GM, Tvrdik T, Muir AM, Dimartino C, Chopra M, Amiel J, Gordon CT, Dutreux F, Garde A, Thauvin-Robinet C, Wang X, Leduc MS, Phillips M, Crawford HP, Kukolich MK, Hunt D, Harrison V, Kharbanda M, Deciphering Developmental Disorders Study, University of Washington Center for Mendelian Genomics, Smigiel R, **Gold N**, Hung CY, Viskochil DH, Dugan SL, Bayrak-Toydemir P, Joly-Helas G, Guerrot AM, Schluth-Bolard C, Rio M, Wentzensen IM, McWalter K, Schnur RE, Lewis AM, Lalani SR, Mensah-Bonsu N, Céraline J, Sun Z, Ploski R, Bacino CA, Mefford HC, Faivre L, Bodamer O, Chelly J, Isidor B, Bahram S. ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. Am J Hum Genet. 2020 Jan 2;106(1):137. PMID: 31879022. PMCID: PMC7042478. https://doi.org/10.1016/j.ajhg.2019.11.014. Epub 2019 Dec 24

5. Gubbels CS, VanNoy GE, Madden JA, Copenheaver D, Yang S, Wojcik MH, **Gold NB**, Genetti CA, Stoler J, Parad RB, Roumiantsev S, Bodamer O, Beggs AH, Juusola J, Agrawal PB, Yu TW. Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genet Med. 2020 Apr;22(4):736-744. PMID: 31780822. PMCID: PMC7127968. https://doi.org/10.1038/s41436-019-0708-6. Epub 2019 Nov 29

6. **Gold NB**, Li D, Chassevent A, Kaiser FJ, Parenti I, Strom TM, Ramos FJ, Puisac B, Pié J, McWalter K, Guillen Sacoto MJ, Cui H, Saadeh-Haddad R, Smith-Hicks C, Rodan L,

Blair E, Bhoj E. Heterozygous de novo variants in CSNK1G1 are associated with syndromic developmental delay and autism spectrum disorder. Clin Genet. 2020 Dec;98(6):571-576. PMID: 33009664. https://doi.org/10.1111/cge.13851. Epub 2020 Oct 12

7. Sheppard SE, Campbell IM, Harr MH, **Gold N**, Li D, Bjornsson HT, Cohen JS, Fahrner JA, Fatemi A, Harris JR, Nowak C, Stevens CA, Grand K, Au M, Graham JM Jr, Sanchez-Lara PA, Campo MD, Jones MC, Abdul-Rahman O, Alkuraya FS, Bassetti JA, Bergstrom K, Bhoj E, Dugan S, Kaplan JD, Derar N, Gripp KW, Hauser N, Innes AM, Keena B, Kodra N, Miller R, Nelson B, Nowaczyk MJ, Rahbeeni Z, Ben-Shachar S, Shieh JT, Slavotinek A, Sobering AK, Abbott MA, Allain DC, Amlie-Wolf L, Au PYB, Bedoukian E, Beek G, Barry J, Berg J, Bernstein JA, Cytrynbaum C, Chung BH, Donoghue S, Dorrani N, Eaton A, Flores-Daboub JA, Dubbs H, Felix CA, Fong CT, Fung JLF, Gangaram B, Goldstein A, Greenberg R, Ha TK, Hersh J, Izumi K, Kallish S, Kravets E, Kwok PY, Jobling RK, Knight Johnson AE, Kushner J, Lee BH, Levin B, Lindstrom K, Manickam K, Mardach R, McCormick E, McLeod DR, Mentch FD, Minks K, Muraresku C, Nelson SF, Porazzi P, Pichurin PN, Powell-Hamilton NN, Powis Z, Ritter A, Rogers C, Rohena L, Ronspies C, Schroeder A, Stark Z, Starr L, Stoler J, Suwannarat P, Velinov M, Weksberg R, Wilnai Y, Zadeh N, Zand DJ, Falk MJ, Hakonarson H, Zackai EH, Quintero-Rivera F. Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. Am J Med Genet A. 2021 Jun;185(6):1649-1665. PMID: 33783954. PMCID: PMC8631250. https://doi.org/10.1002/ajmg.a.62124. Epub 2021 Mar 30

8. MacPherson MJ, Erickson SL, Kopp D, Wen P, Aghanoori MR, Kedia S, Burns KML, Vitobello A, Tran Mau-Them F, Thomas Q, **Gold NB**, Brucker W, Amlie-Wolf L, Gripp KW, Bodamer O, Faivre L, Muona M, Menzies L, Baptista J, Guegan K, Male A, Wei XC, He G, Long Q, Innes AM, Yang G. Nucleocytoplasmic transport of the RNA-binding protein CELF2 regulates neural stem cell fates. Cell Rep. 2021 Jun 8;35(10):109226. PMID: 34107259. https://doi.org/10.1016/j.celrep.2021.109226

9. **Gold NB**, Campbell IM, Sheppard SE, Tan WH. Proposed criteria for nevoid basal cell carcinoma syndrome in children assessed using statistical optimization. Sci Rep. 2021 Oct 5;11(1):19791. PMID: 34611197. PMCID: PMC8492651. https://doi.org/10.1038/s41598-021-98752-9. Epub 2021 Oct 05

10. **Gold NB**, Kritzer A, Weiner DL, Michelson KA. Emergency Laboratory Evaluations for Patients With Inborn Errors of Metabolism. Pediatr Emerg Care. 2021 Dec 1;37(12):e1154-e1159. PMID: 31738301. https://doi.org/10.1097/PEC.0000000000001936

11. Strong A, Skraban C, Meyers K, Amaral S, Furth S, Drant S, Hsiao W, Galea L, Gold J, **Gold NB**, Leonard J, Lopez S, Zackai EH, Pyeritz RE. Expanding the phenotypic

spectrum of Mendelian connective tissue disorders to include prominent kidney phenotypes. Am J Med Genet A. 2021 Dec;185(12):3762-3769. PMID: 34355836. PMCID: PMC9888756. https://doi.org/10.1002/ajmg.a.62449. Epub 2021 Aug 06

12. Blout Zawatsky CL, Shah N, Machini K, Perez E, Christensen KD, Zouk H, Steeves M, Koch C, Uveges M, Shea J, **Gold N**, Krier J, Boutin N, Mahanta L, Rehm HL, Weiss ST, Karlson EW, Smoller JW, Lebo MS, Green RC. Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. Am J Hum Genet. 2021 Dec 2;108(12):2224-2237. PMID: 34752750. PMCID: PMC8715145. https://doi.org/10.1016/j.ajhg.2021.10.005. Epub 2021 Nov 08

13. **Gold NB**, Harrison SM, Rowe JH, Gold J, Furutani E, Biffi A, Duncan CN, Shimamura A, Lehmann LE, Green RC. Low frequency of treatable pediatric disease alleles in gnomAD: An opportunity for future genomic screening of newborns. HGG Adv. 2022 Jan 13;3(1):100059. PMID: 35047849. PMCID: PMC8756496. https://doi.org/10.1016/j.xhgg.2021.100059. Epub 2021 Sep 25

14. Sourbron J, Jansen K, Mei D, Hammer TB, Møller RS, **Gold NB**, O'Grady L, Guerrini R, Lagae L. SLC7A3: In Silico Prediction of a Potential New Cause of Childhood Epilepsy. Neuropediatrics. 2022 Feb;53(1):46-51. PMID: 34872132. https://doi.org/10.1055/s-0041-1739133. Epub 2021 Dec 06

15. Gold JI, **Gold NB**, DeLeon DD, Ganetzky R. Contraceptive use in women with inherited metabolic disorders: a retrospective study and literature review. Orphanet J Rare Dis. 2022 Feb 8;17(1):41. PMID: 35135572. PMCID: PMC8822780. https://doi.org/10.1186/s13023-022-02188-x. Epub 2022 Feb 08

16. Omorodion J, Dowsett L, Clark RD, Fraser J, Abu-El-Haija A, Strong A, Wojcik MH, Bryant AS, **Gold NB**. Delayed diagnosis and racial bias in children with genetic conditions. Am J Med Genet A. 2022 Apr;188(4):1118-1123. PMID: 35037400. PMCID: PMC10064482. https://doi.org/10.1002/ajmg.a.62626. Epub 2022 Jan 17

17. Gold JI, **Gold NB**, Strong A, Tully E, Xiao R, Schwartz LA, Ficicioglu C. The current state of adult metabolic medicine in the United States: Results of a nationwide survey. Genet Med. 2022 Aug;24(8):1722-1731. PMID: 35543711. PMCID: PMC9911209. https://doi.org/10.1016/j.gim.2022.04.018. Epub 2022 May 11

18. O'Grady L, Schrier Vergano SA, Hoffman TL, Sarco D, Cherny S, Bryant E, Schultz-Rogers L, Chung WK, Sacharow S, Immken LL, Holder S, Blackwell RR, Buchanan C, Yusupov R, Lecoquierre F, Guerrot AM, Rodan L, de Vries BBA, Kamsteeg EJ, Santos Simarro F, Palomares-Bralo M, Brown N, Pais L, Ferrer A, Klee EW, Babovic-Vuksanovic D, Rhodes L, Person R, Begtrup A, Keller-Ramey J, Santiago-Sim T, Schnur RE, Sweetser DA, **Gold NB**. Heterozygous variants in PRPF8 are associated

with neurodevelopmental disorders. Am J Med Genet A. 2022 Sep;188(9):2750-2759. PMID: 35543142. https://doi.org/10.1002/ajmg.a.62772. Epub 2022 May 11

19. Yu TW, Kingsmore SF, Green RC, MacKenzie T, Wasserstein M, Caggana M, **Gold NB**, Kennedy A, Kishnani PS, Might M, Brooks PJ, Morris JA, Parisi MA, Urv TK. Are we prepared to deliver gene-targeted therapies for rare diseases?. Am J Med Genet C Semin Med Genet. 2023 Mar;193(1):7-12. PMID: 36691939. https://doi.org/10.1002/ajmg.c.32029. Epub 2023 Jan 24

20. Vockley J, Brunetti-Pierri N, Chung WK, Clarke AJ, **Gold N**, Green RC, Kagan S, Moroz T, Schaaf CP, Schulz M, De Baere E. THE EVOLVING ROLE OF MEDICAL GENETICISTS IN THE ERA OF GENE THERAPY: AN URGENCY TO PREPARE. Genet Med. 2023 Apr;25(4):100022. PMID: 36681872. PMCID: PMC10643995. https://doi.org/10.1016/j.gim.2023.100022. Epub 2023 Jan 19

21. **Gold NB**, Adelson SM, Shah N, Williams S, Bick SL, Zoltick ES, Gold JI, Strong A, Ganetzky R, Roberts AE, Walker M, Holtz AM, Sankaran VG, Delmonte O, Tan W, Holm IA, Thiagarajah JR, Kamihara J, Comander J, Place E, Wiggs J, Green RC. Perspectives of Rare Disease Experts on Newborn Genome Sequencing. JAMA Netw Open. 2023 May 1;6(5):e2312231. PMID: 37155167. PMCID: PMC10167563. https://doi.org/10.1001/jamanetworkopen.2023.12231. Epub 2023 May 01

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Other peer-reviewed scholarship

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Narrative Report

I am a K08-funded physician-scientist interested in the identification of individuals with actionable genetic disease. As more therapies for rare disease become available, I seek to understand who will benefit from therapy and how best to screen them for disease.

My fascination with medical genetics began while working as a research assistant to Chief Emeritus of Medical Genetics at Massachusetts General Hospital (MGH), Dr. Lewis Holmes. As a student at Harvard Medical School, I became intrigued by the potential of genomics as a screening tool. Working with Dr. Robert Green at Brigham and Women's Hospital, I identified participants in the Framingham and Jackson Heart Studies with genetic variants related to cancer and heart disease and characterized which of these individuals developed signs of disease. After completing a combined residency in pediatrics and medical genetics, I was drawn to subspecialty training in medical biochemical genetics, which I pursued with an award from the American College of Medical Genetics Foundation and Takeda.

As a medical geneticist and biochemical geneticist at Massachusetts General Hospital, my interest in actionable genetic disease led me to apply for a K08 award from the National Human Genome Research Institute, which is expected to provide funding for my career development from Jan. 2023 to Jan. 2028. I plan to study the prevalence and penetrance of inherited metabolic disorders and other treatable genetic conditions in the MGB and UK biobanks. Relatedly, I work as an Associate Director of Research for MGB Precision Medicine, through which I supervise the return actionable genetic results among participants in the MGB biobank. I am also a co-investigator of BabySeq2, a multi-center randomized controlled trial assessing the utility of whole genome sequencing as a screening technique in healthy infants.

I have a commitment to health equity within medical genetics. I was the co-investigator of a project interviewing parents from diverse backgrounds regarding their views on genomic newborn screening and mentored a resident on a study of non-white children who experienced delays in genetic diagnosis. I also have a longstanding love of medical education and as a trainee, received an Outstanding Resident Teaching award from Harvard Medical School students. I was the resident member of the ACGME residency committee on Genetics and Genomics and am currently the residency director for genetics

trainees at MGH.

I plan to continue devoting my professional efforts to studying genomic newborn screening. Eventually, I hope this work will help shape policies on the most effective and ethical uses of genetic sequencing technology in newborns and children.