



RYAN LEWIS COLLINS



Investigator, Division of Population Sciences, Dana-Farber Cancer Institute
Instructor, Department of Medicine, Harvard Medical School
Affiliate Faculty, Cancer Program, Broad Institute of M.I.T. and Harvard

375 Longwood Ave., LW-662
Boston, MA, 02115
(603) 290-2200
Ryan_Collins@dfci.harvard.edu

Professional status: independent faculty at Dana-Farber Cancer Institute building a research program on germline genetics in cancer risk & tumorigenesis via population-scale genomic data science.

Career objectives: to realize the potential of genomic data science to clarify the central biology of tumorigenesis and improve our clinical ability to anticipate, prevent, and treat cancer & other diseases.

Scientific interests: human genetics; genome biology; cancer; data science; clinical oncology; probability; statistics; gene regulation; machine learning; genomic technologies; cancer initiation.

EDUCATION

DEGREES

- Ph.D.** Harvard University, Cambridge, MA, USA
2016-2022
Division of Medical Sciences: Bioinformatics and Integrative Genomics
Thesis Title *The landscape and consequences of structural variation in the human genome*
Advisor Michael E. Talkowski, Ph.D.
- A.B.** Dartmouth College, Hanover, NH, USA
2009-2013
Department of Biological Sciences: Computational Genetics
Research Focus *Models of genetic epistasis in common disease risk*
Advisor Jason H. Moore, Ph.D.

ACADEMIC APPOINTMENTS

- Investigator** Department of Medical Oncology (DFCI)
2025-Present Dana-Farber Cancer Institute | Boston, MA, USA
- Affiliate Faculty** Cancer Program & Program in Medical and Population Genetics
2025-Present Broad Institute of M.I.T. and Harvard | Cambridge, MA, USA
- Instructor** Department of Medicine (HMS)
2024-Present Harvard Medical School | Boston, MA, USA
- Instructor** Department of Medical Oncology (DFCI)
2024-2025 Dana-Farber Cancer Institute | Boston, MA, USA

RESEARCH EXPERIENCE

- Research Fellow** Laboratories of Eli Van Allen, M.D., Kevin Haigis, Ph.D., and Alexander Gusev, Ph.D. (co-advised)
2022-2025 Dana-Farber Cancer Institute | Boston, MA, USA
- Associated Scientist** Program in Medical & Population Genetics and Cancer Program
2015-2022 Broad Institute of M.I.T. and Harvard | Cambridge, MA, USA
- Ph.D. Candidate** Laboratory of Michael E. Talkowski, Ph.D.
2017-2022 Massachusetts General Hospital | Boston, MA, USA
- Ph.D. Rotation Student** Laboratory of Eric S. Lander, Ph.D. (co-supervised by Jesse Engreitz, Ph.D.)
2017 Broad Institute of MIT and Harvard | Cambridge, MA, USA
- Ph.D. Rotation Student** Laboratory of Daniel G. MacArthur, Ph.D.
2016 Broad Institute of MIT and Harvard | Cambridge, MA, USA
- Bioinformatics Specialist II** Laboratory of Michael E. Talkowski, Ph.D., and MGH Genomics and Technology Core
2013-2016 Massachusetts General Hospital | Boston, MA, USA
- Student Research Assistant** Laboratory of Jason H. Moore, Ph.D.
2011-2013 Geisel School of Medicine at Dartmouth College | Hanover, NH, USA
- Student Research Assistant** Department of Orthopedic Medicine
2010 Dartmouth Hitchcock Medical Center | Lebanon, NH, USA
- Student Research Assistant** Laboratory of Thomas P. Jack, Ph.D.
2009 Dartmouth College | Hanover, NH, USA

PROFESSIONAL ACTIVITIES

TEACHING

- Invited Lecturer** International Molecular Genomic Medicine and Biostatistics Course
2023 International Center for Genetic Disease (Brigham and Women's Hospital & Harvard Medical School) | Bahrain (Virtual)

Teaching Assistant 2020	Clinical Genetics (GEN288; Instructor: David Sweetser, M.D.) Harvard Medical School Boston, MA, USA
Invited Lecturer 2018	Human Genetics (GEN202; Instructor: Matthew Warman, M.D.) Harvard Medical School Boston, MA, USA
Invited Lecturer 2017	Short Course on Rare Variant Interpretation in Mendelian Diseases Broad Institute of MIT and Harvard Cambridge, MA, USA
Teaching Assistant 2012-2013	Gene Expression and Inheritance (BIOL013; Instructor: Thomas Jack, Ph.D.) Dartmouth College Hanover, NH, USA
Volunteer Tutor 2010-2011	Calculus and Advanced Mathematics Hanover High School Hanover, NH, USA

TRAINEES MENTORED

2026-Present	Melissa Franco, Ph.D.	Postdoctoral fellow @ DFCI	Co-mentored with Dr. Riaz Gillani
2025-Present	Kailey Ferger, Ph.D.	Postdoctoral fellow @ DFCI	
2024-Present	Gwen Miller, B.S.	Ph.D. @ Harvard University	Co-mentored with Dr. Eli Van Allen
2024-Present	Jiayi Chen, B.S.	M.S. @ Harvard University	Co-mentored with Dr. Kevin Haigis
2023-Present	Claire Johnson, B.S.	Ph.D. @ Harvard University	Co-mentored with Dr. Eli Van Allen
2025	Doris Fu, B.S.	Ph.D. @ Harvard Univ.	Rotation; co-mentored with Dr. Eli Van Allen
2022-2023	Ellen Chen, B.A.	M.S. @ Harvard University	Co-mentored with Dr. Kevin Haigis
2020-2021	Selin Everett	B.S. @ Middlebury College	Co-mentored with Drs. Michael Talkowski & Harrison Brand

STAFF SUPERVISED

2026-Present	Carter Nakagawa, M.S.	Computational biologist	Co-supervised with Dr. Jackie LoPiccolo
2023-Present	Noah Fields, M.S.	Computational biologist	Co-supervised with Dr. Eli Van Allen
2019-2022	Isaac Wong, B.S.	Computational biologist	Co-supervised with Drs. Michael Talkowski & Harrison Brand

LEADERSHIP

Organizer 2025-Present	Germline Genetics Working Group, Division of Population Sciences Dana-Farber Cancer Institute Boston, MA, USA
Director 2023-Present	Germline Genomics of Cancer (G ² C) Initiative Dana-Farber Cancer Institute Boston, MA, USA
Team Lead 2023-2025	Germline Genetics Subgroup, Clinical Computational Oncology Group Dana-Farber Cancer Institute Boston, MA, USA
Co-Lead 2018-2023	Structural Variation Team, The Genome Aggregation Database (gnomAD) Consortium Broad Institute of MIT and Harvard Cambridge, MA, USA
Team Lead 2018-2022	Population Genetics & Genome Biology Team, Broad-SV Group Broad Institute of MIT and Harvard Cambridge, MA, USA
Organizer 2019-2020	Bioinformatics and Integrative Genomics Student Research Seminar Series Harvard Medical School Boston, MA, USA

Elected Representative
2008-2009
Dresden School Board
School Administrative Unit (SAU) 70 | Hanover, NH & Norwich, VT, USA

ACADEMIC SERVICE

Invited Referee
2020-Present
14 articles for 13 journals, including: *Cell Genomics*; *Genome Research*; *Human Genetics*; *iScience*; *Journal of Clinical Investigation*; *Nature Communications*; *Nature Methods*; *npj Genomic Medicine*; *Nucleic Acids Research*; *Trends in Genetics*; *Science Advances*; *Scientific Data*; *Scientific Reports*

Ad Hoc Referee
2013-Present
16 articles for 13 journals, including: *American Journal of Human Genetics*; *BMC Genomics*; *Cell*; *Clinical Genetics*; *Human Molecular Genetics*; *Molecular Genetics & Genomic Medicine*; *Molecular Psychiatry*; *Nature*; *Nature Communications*; *Nature Genetics*; *Nature Reviews Genetics*; *New England Journal of Medicine*; *PLOS Genetics*
All *ad hoc* referee activities were conducted with Michael Talkowski, Ph.D.

Invited Lecturer
2026
Medical & Population Genetics Primer
Broad Institute of MIT and Harvard | Boston, MA, USA

Invited Panelist
2025
Writing Effective Career Development Plans for Research Instructors
Dana-Farber Office of Faculty Development | Boston, MA, USA

Invited Panelist
2024
Short Course on Applying for an NIH K Award
Dana-Farber Postdoctoral and Graduate Student Affairs Office | Boston, MA, USA

Session Moderator
2018
American Society for Human Genetics Annual Meeting
San Diego, CA, USA

Admissions Interviewer
2018
PhD Student Admissions, Program in Bioinformatics and Integrative Genomics
Harvard Medical School | Boston, MA, USA

Keynote Session Chair
Gordon Research Seminar: Human Genetics and Genomics

2017 Stowe, VT, USA

Invited Panelist The Future of Genome Sequencing in Rare Diseases, NIH Center for Mendelian Genomics
2017 Broad Institute of MIT and Harvard | Cambridge, MA, USA

OUTREACH

Invited Panelist Prospective Graduate Student Panel
2019 Massachusetts General Hospital Research Institute | Boston, MA, USA

Volunteer Reviewer *Journal of Emerging Investigators*
2016-2019 Cambridge, MA, USA

Contributing Author *The Nascent Transcript* (American Society for Human Genetics Periodical)
2016-2017 USA

Contributing Author Science in the News
2016-2017 Harvard University | Cambridge, MA, USA

Invited Panelist Dartmouth College Young Alumni Panel on Biotechnology Careers
2015 Boston, MA, USA

Alumnus Mentor Dartmouth Externship Program for Professional Exploration
2015 Boston, MA, USA

CONSORTIA

Selected representative publications listed below each consortium, where applicable

- 2025-Present** International Alliance for Cancer Early Detection (ACED)
- 2023-2024** Pancreatic Cancer Early Detection (PRECEDE) Consortium
- 2016-2024** Genome Aggregation Database (gnomAD), Broad Institute of MIT and Harvard
In May 2020, seven gnomAD papers were published in *Nature*, *Nat. Med.*, and *Nat. Commun.*, and featured on the cover of *Nature*
- 2015-2022** Core Analysis Group, Autism Sequencing Consortium
Fu *et al.*, *Nat. Genet.* (2022); Satterstrom, *et al.*, *Cell* (2020); An *et al.*, *Science* (2018); Werling *et al.*, *Nat. Genet.* (2018)
- 2015-2022** Structural Variation Group (Broad-SV), Broad Institute of MIT and Harvard
Collins *et al.*, *Cell* (2022); Collins *et al.*, *Genome Biol.* (2017)
- 2013-2022** Developmental Genome Anatomy Project / International Breakpoint Mapping Consortium
Lowther *et al.*, *medRxiv* (2022); Redin *et al.*, *Nat. Genet.* (2017)
- 2015-2019** Human Genome Structural Variation Consortium (1000 Genomes Project phase 4)
Zhao *et al.*, *Am. J. Hum. Genet.* (2021); Chaisson *et al.*, *Nat. Commun.* (2019)
- 2015-2017** Collaborative Center for X-Linked Dystonia Parkinsonism, Massachusetts General Hospital
Aneichyk *et al.*, *Cell.* (2018)

OTHER AFFILIATIONS

- 2017-Present** Sigma Xi Honors Society for Scientific Research
- 2014-Present** American Society of Human Genetics
- 2021-2022** European Society for Human Genetics

FUNDING

Totals below reflect direct costs (no indirect costs included). Pending grants not shown.

ACTIVE

- 2025-2028** \$419,661 NCI Pathway to Independence Award for Early-Stage Postdoctoral Researchers (PI; R00 CA286805)
Dissecting the role of germline genetics in RAS-driven cancers
- 2025-2027** \$112,500 Haroun Family Foundation (MPIs: Collins, Gillani)
Interrogating germline genome instability as a novel approach for prevention and clinical translation in Ewing Sarcoma
- 2026** \$55,000 American Cancer Society Institutional Research Grant (PI; IRG-24-1290526-01)
Secondary genetic effects on mismatch repair as a novel modifier of penetrance and expressivity in Lynch Syndrome
- 2023-2026** \$700,000 Dana-Farber Institute-Sponsored Research Award (MPIs: Collins, Van Allen, Gusev)
The Dana-Farber Germline Genomics of Cancer (G2C) Database

COMPLETED

- 2024-2025** \$303,200 NCI Pathway to Independence Award for Early-Stage Postdoctoral Researchers (PI; K99 CA286805)
Dissecting the Role of Germline Genetics in RAS-Driven Cancers
- 2023-2025** \$200,000 Trustee Science Committee Fellowship, Dana-Farber Cancer Institute
- 2017-2021** \$138,000 NSF Graduate Research Fellowship, National Science Foundation (GRFP #2017240332)

AWARDS & HONORS

MAJOR

- 2021** European Society for Human Genetics Early Career Award for Outstanding Science
€500 prize; one of five winners among 950 trainee applicants at the European Society of Human Genetics Annual Meeting 2021
- 2021** Massachusetts General Hospital Executive Committee on Research Celebration of Science Award
\$1,000 prize; given to MGH Martin Prize finalists; awarded for work on mapping structural variation in the human population
- 2021** European Society for Human Genetics Conference Fellowship of Excellence
Awarded to each of the five top-rated abstracts submitted by trainees to the European Society of Human Genetics Annual Meeting 2021
- 2018** Charles J. Epstein Trainee Award for Excellence in Human Genetics Research
\$2,000 prize; one of three predoctoral winners among 670 applicants at the American Society of Human Genetics Annual Meeting 2018
- 2017** Associate Member, Sigma Xi Honors Society for Scientific Research
- 2014** Clinical Research Team Award, Massachusetts General Hospital
\$2,500 team prize awarded to best entry among 386 teams for work on complex structural variation in neuropsychiatric disorders

MINOR

- 2023** Collaborative Impact Award, Broad Institute of M.I.T. and Harvard
Institute-wide honor awarded to the core team responsible for the production of the Genome Aggregation Database (gnomAD)
- 2022** Annual Most Valuable Member (Second Place), Talkowski Laboratory
- 2020** Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist)
\$750 prize; one of 30 predoctoral semifinalists among 433 applicants at the American Society of Human Genetics Annual Meeting 2020
- 2020** Annual Most Valuable Member, Talkowski Laboratory
- 2020** Reviewer's Choice Poster Award, American Society for Human Genetics
Distinction given to top 10% of best-scoring abstracts at the American Society of Human Genetics Annual Meeting 2020
- 2019** Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist)
\$750 prize; one of 30 predoctoral semifinalists among 511 applicants at the American Society of Human Genetics Annual Meeting 2019
- 2019** Trainee & Staff Abstract Award, Center for Genomic Medicine, Massachusetts General Hospital
\$750 travel award; one of three top submitted abstracts for a Center-wide talk competition
- 2019** Travel Award, Center for Genomic Medicine, Massachusetts General Hospital
\$1,500 travel award; one of three winners from a Center-wide competition based on submitted abstract, CV, and travel proposal
- 2018** Travel Award, Harvard University Graduate School of Arts and Sciences Professional Development Fund
\$600 travel award; awarded to present research at American Society of Human Genetics Annual Meeting 2018 in San Diego, CA
- 2018** Annual Most Valuable Member (Third Place), Talkowski Laboratory
- 2018** Selected for the American Society of Human Genetics (ASHG) Trainee Paper Spotlight
Described by ASHG: "The Trainee Paper Spotlight is a quarterly feature highlighting outstanding papers written by ASHG trainees"
Selected paper: Collins *et al.*, *Genome Biol.* (2017). Highlight [available online via ASHG](#).
- 2016** Annual Most Valuable Member (Third Place), Talkowski Laboratory
- 2015** Annual Most Valuable Member, Talkowski Laboratory
- 2014** Annual Most Valuable Member (Second Place), Talkowski Laboratory
- 2009** Robert Stone Memorial Award, Hanover High School
- 2009** Tom Hanlon Cup, Hanover High School

RESEARCH PRESENTATIONS

All presentations were intramural unless marked as [E], which designates an extramural presentation. **Bold** indicates highlighted presentations of note.

What can population genomics teach us about cancer predisposition and tumorigenesis?

[E]	March 2026	Invited Speaker	Boston University Genome Science Institute Seminar Series	Boston, MA, USA
	December 2025	Invited Speaker	DFCI Population Sciences Clinical Computational Oncology Group Seminar	Boston, MA, USA
	October 2025	Invited Speaker	Broad Institute Broad-SV Seminar Series	Cambridge, MA, USA
	September 2025	Invited Speaker	DFCI Cancer Genetics, Prevention, and Early Detection Seminar	Boston, MA, USA

Germline structural variants in pediatric and early-onset cancers

	May 2025	Invited Speaker	DFCI Center for Outcomes and Policy Research Seminar Series	Boston, MA, USA
[E]	April 2025	Invited Speaker	Harvard School of Public Health Quantitative Genomics Seminar Series	Boston, MA, USA
[E]	February 2025	Invited Speaker	Broad Institute Somatic Mutation (SMART) Journal Club	Cambridge, MA, USA
[E]	February 2025	Invited Speaker	International Society of Pediatric Oncology Seminar Series	International (Virtual)
	December 2024	Invited Platform Speaker	Broad Institute 20th Annual Retreat	Boston, MA, USA
	April 2024	Invited Speaker	Broad Institute Cancer Program Seminar	Cambridge, MA, USA
	April 2024	Invited Speaker	Broad Institute Broad-SV Seminar Series	Cambridge, MA, USA
	February 2024	Invited Speaker	DFCI Cancer Genetics, Prevention, and Early Detection Seminar	Boston, MA, USA
	January 2023	Invited Speaker	DFCI Program in Clinical Computational Oncology Seminar Series	Boston, MA, USA

Genomic structural variation as a lens for understanding human disease

[E]	January 2024	Invited Speaker	Feigin Rising Stars Research Symposium, Texas Children's Hospital	Houston, TX, USA
A cross-disorder dosage sensitivity map of the human genome				
[E]	December 2022	Invited Speaker	Pritchard Lab @ Stanford Journal Club	Palo Alto, CA, USA (Virtual)
[E]	November 2022	Invited Speaker	Boston Children's Hospital Digital Science Seminar Series (video recording here)	Boston, MA, USA (Virtual)
[E]	October 2022	Invited Speaker	ClinGen Dosage Sensitivity Curation Working Group	International (Virtual)
	November 2021	Invited Speaker	MGH Analytical and Translational Genetics Unit Seminar Series	Boston, MA, USA
	October 2021	Invited Speaker	Broad Institute Stanley Center for Psychiatric Research Seminar Series	Cambridge, MA, USA
[E]	June 2021	Selected Plenary Speaker	European Society for Human Genetics Annual Meeting	International (Virtual)
[E]	May 2021	Invited Panel Speaker	International Society for Autism Research Annual Meeting	International (Virtual)
	April 2021	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	Cambridge, MA, USA
	January 2021	Invited Speaker	Broad Institute Broad-SV Seminar Series	Cambridge, MA, USA
[E]	October 2020	Poster Presentation	American Society of Human Genetics Annual Meeting	International (Virtual)
[E]	March 2018	Poster Presentation	NHGRI Research Training and Career Development Annual Meeting	Los Angeles, CA, USA
	December 2017	Poster Presentation	Broad Institute Scientific Retreat	Boston, MA, USA
	September 2017	Poster Presentation	Massachusetts General Hospital Center for Genomic Medicine Annual Retreat	Cambridge, MA, USA
gnomAD-SV: an open resource of structural variation for medical and population genetics				
[E]	August 2021	Invited Speaker	Garvan Institute Computational Biology Seminar Series	Australia (Virtual)
[E]	May 2021	Invited Speaker	UK NHS Southwest Genomic Laboratory Hub Seminar Series	United Kingdom (Virtual)
	April 2021	Invited Speaker	Massachusetts General Hospital Executive Committee on Research Meeting	Boston, MA, USA
[E]	October 2020	Invited Speaker	Boston Children's Hospital Digital Science Seminar Series	Boston, MA, USA
[E]	September 2020	Invited Speaker	OpenBox Science Virtual Symposium	International (Conf. Call)
[E]	February 2020	Invited Speaker	ClinGen CNV Interpretation Webinar	International (Conf. Call)
	January 2020	Invited Speaker	Harvard Medical School BIG Ph.D. Recruitment Informational Session	Boston, MA, USA
	December 2019	Invited Plenary Speaker	Broad Institute 15 th Annual Retreat	Boston, MA, USA
[E]	December 2019	Invited Speaker	Yale Genetics Walter J. Burdette Trainee Symposium	New Haven, CT, USA
[E]	October 2019	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	Houston, TX, USA
[E]	July 2019	Selected Platform Speaker	Gordon Research Seminar: Human Genetics & Genomics	Waterville Valley, NH, USA
[E]	July 2019	Invited Speaker	ClinGen Biocurator Working Group Monthly Conference Call	International (Conf. Call)
[E]	May 2019	Invited Speaker	NHGRI Genome Sequencing Program Methods Group Conference Call	International (Conf. Call)
[E]	May 2019	Poster Presentation	Cold Spring Harbor Laboratory Biology of Genomes Annual Meeting	Cold Spring Harbor, NY, USA
	May 2019	Selected Platform Speaker	Massachusetts General Hospital CGM Annual Trainee & Staff Seminar	Boston, MA, USA
	March 2019	Invited Speaker	Massachusetts General Hospital Reproductive Endocrine Unit Seminar	Boston, MA, USA
	January 2019	Invited Speaker	Broad Institute Data Sciences Platform Methods & Applications Seminar	Cambridge, MA, USA
	November 2018	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	Cambridge, MA, USA
[E]	February 2018	Invited Speaker	gnomAD Consortium Monthly Analysis Conference Call	International (Conf. Call)
	January 2018	Invited Speaker	Harvard Medical School BIG Ph.D. Recruitment Informational Session	Boston, MA, USA
	August 2017	Selected Speaker	Talkowski Laboratory Retreat	Portsmouth, NH, USA
Methods for structural variation discovery from whole-genome sequencing				
	December 2018	Invited Speaker	Broad Institute Data Sciences Platform Methods Primer	Cambridge, MA, USA
	January 2018	Invited Speaker	Broad Institute Data Sciences Platform Methods & Applications Seminar	Cambridge, MA, USA
Convergence of copy-number variants and point mutations in neurodevelopmental disorders				
[E]	October 2018	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	San Diego, CA, USA
	October 2018	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	Cambridge, MA, USA
Complex structural variation in neurodevelopmental disorders				
[E]	July 2017	Poster Presentation	Gordon Research Conference: Human Genetics & Genomics	Stowe, VT, USA
[E]	July 2017	Poster Presentation	Gordon Research Seminar: Human Genetics & Genomics	Stowe, VT, USA
[E]	April 2017	Selected Platform Speaker	NHGRI Research Training and Career Development Annual Meeting	St. Louis, MO, USA
	December 2016	Poster Presentation	Broad Institute Scientific Retreat	Boston, MA, USA
[E]	May 2016	Poster Presentation	Cold Spring Harbor Laboratory Biology of Genomes Annual Meeting	Cold Spring Harbor, NY, USA
[E]	May 2016	Invited Speaker	Human Genome Structural Variation Consortium Semiannual Meeting	Cold Spring Harbor, NY, USA
[E]	October 2015	Poster Presentation	American Society of Human Genetics Annual Meeting	Baltimore, MD, USA
	April 2015	Poster Presentation	Massachusetts General Hospital Scientific Advisory Council Meeting	Boston, MA, USA
	April 2015	Poster Presentation	Massachusetts General Hospital Center for Human Genetics Research Retreat	Cambridge, MA, USA
Limited contribution of rare, noncoding variation to autism spectrum disorders				
	April 2017	Invited Speaker	Massachusetts General Hospital Analytical and Translational Genetics Unit	Boston, MA, USA
Understanding dosage biases in the analysis of whole-genome sequencing datasets				
	February 2016	Invited Speaker	Broad Institute Structural Variation and Assembly Group	Cambridge, MA, USA
	December 2016	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	Cambridge, MA, USA
	May 2015	Invited Speaker	Massachusetts General Hospital Molecular Neurogenetics Unit Seminar	Boston, MA, USA
Structural rearrangements in genome editing with CRISPR-Cas9 and TALENs				
	January 2015	Invited Speaker	Massachusetts General Hospital Center for Human Genetics Research Seminar	Boston, MA, USA
[E]	October 2014	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	San Diego, CA, USA
	April 2014	Poster Presentation	Massachusetts General Hospital Center for Human Genetics Research Retreat	Cambridge, MA, USA
	April 2014	Invited Speaker	Massachusetts General Hospital Molecular Neurogenetics Unit Seminar	Boston, MA, USA

Articles are grouped based on the extent of contributions to each (primary/major/minor/group) and ordered chronologically thereafter.

- **Primary contributions** include principal leadership roles in conducting and/or overseeing the overall study.
- **Major contributions** include core roles in conducting and/or managing one or more main components of the study.
- **Minor contributions** include supporting or secondary roles in conducting or otherwise facilitating the study.
- **Group authorship** indicates a general involvement as part of a larger consortium that facilitated the study.

Specific details of the contributions to each publication are enumerated below each citation (grey text)

Key: *^o/_† equal contributions

§ publication highlight

! highly cited (≥100 citations)

Citation statistics from [Google Scholar](#)

REFEREED SCIENTIFIC ARTICLES

Articles authored as a primary contributor

- 2025 § [57] **Collins RL**, Talkowski ME. The diversity and consequences of structural variation in the human genome. *Nature Reviews Genetics* (2025), 26: 443-462. PMID: [39838028](#). DOI: [10.1038/s41576-024-00808-9](#).
Performed all literature review; wrote manuscript; designed all figures.
§ **Featured on the cover of *Nature Reviews Genetics***
- § [56] Gillani R^o, **Collins RL**^{*}, Crowdis J, Garza A, Jones JK, Walker M, Sanchis-Juan A, Whelan C, Pierce-Hoffman E, Brookings T, Calamari N, Talkowski M, Brand H, Haigis K, LoPiccolo J, AlDubayan SH, Gusev A, Crompton BD, Janeway KA, Van Allen EM^o. Rare germline structural variants increase risk for pediatric solid tumors. *Science* (2025), 387: 6729. PMID: [39745975](#). DOI: [10.1126/science.adq0071](#).
Led or co-led all aspects of study; co-wrote manuscript; created Figures 1, 2, and 5.
§ **Highlighted by *Nature Genetics*, *Genome Web*, *Cancer Discovery*, *the National Cancer Institute*, and *Science***
- 2023 [55] Montanucci L^{*}, Lewis-Smith D^{*}, **Collins RL**^{*}, Niestroj L^{*}, Parthasarathy S, Xian J, Ganesan S, Macnee M, Brünger T, Thomas RH, Talkowski M, The Epi25 Collaborative, Helbig I, Leu C^o, Lal D^o. Genome-wide identification and phenotypic characterization of seizure-associated copy number variations in 741,075 individuals. *Nature Communications* (2023), 14(1): 4392. PMID: [37474567](#). DOI: [10.1038/s41467-023-39539-6](#).
Co-led rare CNV meta-analyses of seizure cohort with other neuropsychiatric disorders; contributed to manuscript drafting and figures
- 2022 § ! [54] **Collins RL**^{*}, Glessner JT, Porcu E, Lepamets M, Brandon R, Lauricella C, Han L, Morley T, Niestroj L, Ulirsch J, Everett S, Howrigan DP, Boone PM, Fu J, Karczewski K, Kellaris G, Lowther C, Lucente D, Mohajeri K, Nöukas M, Nuttle X, Samocha KE, Trinh M, Ullah F, Vösa U, Hurler ME, Aradhya S, Davis EE, Finucane H, Gusella JF, Janze A, Katsanis N, Matyakhina L, Neale BM, Sanders D, Warren S, Hodge JC, Lal D, Ruderfer DM, Meck J, Mägi R, Esko T, Reymond A, Kutalik Z, Hakonarson H, Sunyaev S, Brand H^{*}, Talkowski ME^{*}. A cross-disorder dosage sensitivity map of the human genome. *Cell* (2022), 185(16): 3041-3055. PMID: [35917817](#). DOI: [10.1016/j.cell.2022.06.036](#).
Conceived of study, curated all data, developed all methods, performed all analyses, and drafted manuscript and figures
§ **Winner of the 2021 ESHG Early Career Award for Outstanding Science (see Awards & Honors)**
§ **Highlighted by *Spectrum News*, *Genome Web*, *Nature Reviews Genetics*, and in a *Preview in Cell***
- 2020 § ! [53] **Collins RL**^{*}, Brand H^{*}, Karczewski KJ, Zhao X, Alföldi J, Khera AV, Francioli LC, Gauthier LD, Wang H, O'Donnell-Luria A, Solomonson M, Baumann A, Munshi R, Walker M, Whelan C, Huang Y, Brookings T, Sharpe T, Stone MR, Tiao G, Laricchia KM, Watts NA, Fu J, Valkanas E, Lowther C, Stevens C, Gupta N, Cusick C, Margolin L, The gnomAD Production Team, The gnomAD Consortium, Spertus JA, Taylor KD, Psaty BM, Rich SS, Post W, Chen YI, Rotter JI, Nusbaum C, Philippakis A, Lander E, Gabriel S, Neale BM, Kathiresan S, Daly MJ, Banks E, MacArthur DG, Talkowski ME. A structural variation reference for medical and population genetics. *Nature* (2020), 581: 444-451. PMID: [32461652](#). DOI: [10.1038/s41586-020-2287-8](#).
Led and conducted all analyses for duration of study (2016-2020); co-led all methods development; wrote manuscript & created all figures
§ **Featured by *Nature* in an editorial, a *News & Views* commentary, and on the cover of the May 28, 2020 issue**
§ **Highlighted in *Nature Reviews Genetics*, *GenomeWeb*, *The Broad Institute*, *Yahoo! News*, *Science Daily*, and others**
§ **Semifinalist for the 2019 ASHG Charles J. Epstein Trainee Award for Excellence in Human Genetics (see Awards & Honors)**
§ **Winner of a Travel Award and a Trainee & Staff Abstract Award from the Massachusetts General Hospital (see Awards & Honors)**
- 2017 § ! [52] **Collins RL**, Brand H, Redin CE, Hanscom C, Antolik C, Stone MR, Glessner JT, Mason T, Pregno G, Dorrani N, Mandrile G, Giachino D, Perrin D, Walsh C, Cipicchio M, Costello M, Stortchevoi A, An J, Currall BB, Seabra CM, Ragavendran A, Margolin L, Martinez-Agosto JA, Lucente D, Levy B, Sanders SJ, Wapner RJ, Quintero-Rivera F, Kloosterman W, Talkowski ME. Defining the diverse spectrum of inversions, complex structural variation, and chromothripsis in the morbid human genome. *Genome Biology* (2017), 18(36): 1-21. PMID: [28260531](#). DOI: [10.1186/s13059-017-1158-6](#).
Led study design, methods development, and analysis for duration of study (2014-2017); wrote manuscript & created all figures
§ **Selected for the *American Society of Human Genetics Trainee Paper Spotlight***
§ **Highlighted by *GenomeWeb*, *The Broad Institute*, *the 10X Genomics Blog***
§ **Selected by the editors of *Genome Biology* as one of their favorite articles from 2017**
- 2015 § [51] Brand H^{*}, **Collins RL**^{*}, Hanscom C, Rosenfeld JA, Pillalamarri V, Stone MR, Kelley F, Mason T, Margolin L, Eggert S, Mitchell E, Hodge J, Gusella JF, Sanders SJ, Talkowski ME. Paired duplications mark cryptic inversions and are a common signature of complex structural variation. *American Journal of Human Genetics* (2015), 97(1): 170-176. PMID: [26094575](#). DOI: [10.1016/j.ajhg.2015.05.012](#).
Co-designed study and co-led all analyses; created all figures; led manuscript writing
§ **Highlighted by *Spectrum News***

- 2013 [50] **Collins RL**, Hu T, Wejse C, Sirugo G, Williams SM, Moore JH. Multifactor dimensionality reduction reveals a three-locus epistatic interaction associated with susceptibility to pulmonary tuberculosis. *BioData Mining* (2013), 6(1): 4. PMID: [23418869](#). DOI: [10.1186/1756-0381-6-4](#).
Performed all analyses; interpreted all results; assisted in statistical methods development; wrote manuscript & created all figures

Articles authored as a **major contributor**

- 2022 §! [49] Fu JM*, Satterstrom FK*, Peng M*, Brand H*, **Collins RL**, Dong S, Klei L, Stevens CR, Cusick C, Babadi M, Banks E, Collins B, Dodge S, Gabriel SB, Gauthier L, Lee SK, Liang L, Ljungdahl A, Mahjani B, Sloofman L, Smirnov A, Barbosa M, Brusco A, Chung BH, Cuccaro ML, Domenici E, Ferrero GB, Gargus JJ, Herman GE, Hertz-Picciotto I, Maciel P, Manoach DS, Passos-Bueno MR, Persico AM, Renieri A, Tassone F, Trabetti E, Campos G, Chan MC, Fallerini C, Giorgio E, Girard AC, Hansen-Kiss E, Lee SL, Lintas C, Ludena Y, Nguyen R, Pavinato L, Pericak-Vance M, Pessah I, Riberi E, Schmidt R, Smith M, Souza CI, Trajkova S, Wang JY, Yu MH, The Autism Sequencing Consortium, Broad Center for Common Disease Genomics, iPSYCH-BROAD Consortium, Cutler DJ, Rubeis SD, Buxbaum JD°, Daly MJ°, Devlin B°, Roeder K°, Sanders SJ°, Talkowski ME°. Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. *Nat. Genet.* (2022), 54:1320-1331. PMID: [35982160](#). DOI: [10.1038/s41588-022-01104-0](#).
Contributed to methods development and CNV callset generation; contributed to manuscript writing & revising; co-created Figure 2
§ Highlighted by [Spectrum News](#) and in a [Research Briefing by Nature Genetics](#).
- 2021 §! [48] Zhao X, **Collins RL**, Lee W, Weber AM, Jun Y, Zhu Q, Weisburd B, Huang Y, Audano PA, Wang H, Walker M, Lowther C, Fu J, Gerstein MB, Devine SE, Marschall T, Korbel JO, Eichler EE, Chaisson MJ, Lee C, Mills RE, Brand H, Talkowski ME. Expectations and blind spots for structural variation detection from short-read alignment and long-read assembly. *American Journal of Human Genetics* (2021), 108(5): 919-928. PMID: [33789087](#). DOI: [10.1016/j.ajhg.2021.03.014](#).
Assisted in study design; contributed to analysis of short-read vs. long-read comparisons; assisted in manuscript & figure preparation
§ Highlighted by [GenomeWeb](#)
- 2020 §! [47] Karczewski KJ, Francioli LC, Tiao G, Cummings BB, Alföldi J, Wang Q, **Collins RL**, Laricchia KM, Ganna A, Birnbaum DP, Gauthier LD, Brand H, Solomonson M, Watts NA, Rhodes D, Singer-Berk M, Seaby EG, Kosmicki JA, Walters RK, Tashman K, Farjoun Y, Banks E, Poterba T, Wang A, Seed C, Whiffin N, Chong JX, Samocha KE, Pierce-Hoffman E, Zappala Z, O'Donnell-Luria AH, Minikel EV, Weisburd B, Lek M, Ware JS, Vittal C, Armean IM, Bergelson L, Cibulskis K, Connolly KM, Covarrubias M, Donnelly S, Ferriera S, Gabriel S, Gentry J, Gupta N, Jeandet T, Kaplan D, Llanwarne C, Munshi R, Novod S, Petrillo N, Roazen D, Ruano-Rubio V, Saltzman A, Schleicher M, Soto J, Tibbetts K, Tolonen C, Wade G, Talkowski ME, The gnomAD Consortium, Neale BM, Daly MJ, MacArthur DG. Variation across 141,456 human exomes and genomes reveals the spectrum of loss-of-function intolerance across human protein-coding genes. *Nature* (2020), 581. PMID: [32461654](#). DOI: [10.1038/s41586-020-2308-7](#).
Led generation of structural variation callset from whole-genome sequencing of 14,891 samples; designed Figure 3B
§ Winner of the 2018 ASHG Charles J. Epstein Trainee Award for Excellence in Human Genetics
§ Highlighted by Nature in an editorial, a News & Views commentary, and on the cover
§ Highlighted by [GenomeWeb](#), [The Broad Institute](#), [Yahoo! News](#), [Science Daily](#), and others
- §! [46] Satterstrom FK*, Kosmicki JA*, Wang J*, Breen M, Rubeis SD, An J, Peng M, **Collins RL**, Grove J, Klei L, Stevens C, Reichert J, Mulhern M, Artomov M, Gerges S, Sheppard B, Xu X, Bhaduri A, Norman U, Brand H, Schwartz G, Nguyen R, Guerrero E, Dias C, Aleksic B, Anney RJ, Barbosa M, Bishop S, Brusco A, Bybjerg-Grauholm J, Carracedo A, Chan MC, Chiochetti A, Chung B, Coon H, Cuccaro M, Curró A, Bernardina BD, Doan R, Domenici E, Dong S, Fallerini C, Fernández-Prieto M, Ferrero GB, Freitag CM, Fromer M, Gargus JJ, Geschwind D, Giorgio E, González-Peñas J, Guter S, Halpern D, Hassen-Kiss E, He X, Herman G, Hertz-Picciotto I, Hougaard DM, Hultman CM, Ionita-Laza I, Jacob S, Jamison J, Jugessur A, Kaartinen M, Knudsen GP, Kolvez'n A, Kushima I, Lee SL, Lehtimäki T, Lim ET, Lintas C, Lipkin WI, Lopergolo D, Lopes F, Ludena Y, Maciel P, Magnus P, Mahjani B, Maltman N, Manoach DS, Meiri G, Menashe I, Miller J, Minshew N, De Souza EM, Moreira D, Morrow E, Mors O, Mortensen PB, Mosconi M, Muglia P, Neale B, Nordentoft M, Ozaki N, Palotie A, Parellada M, Passos-Bueno MR, Pericak-Vance M, Persico A, Pessah I, Puura K, Reichenberg A, Renieri A, Riberi E, Robinson E, Samocha KE, Sandin S, Santangelo SL, Schellenberg G, Scherer S, Schlitt S, Schmidt R, Schmitt L, Silva IM, Singh T, Siper P, Smith M, Soares G, Stoltenberg C, Suren P, Susser E, Sweeney J, Szatmari P, Tang L, Tassone F, Teufel K, Trabetti E, Trelles MD, Walsh C, Weiss L, Werge T, Werling D, Wigdor EM, Wilkinson E, Willsey JA, Yu T, Yu MH, Yuen R, Zachi E, Betancur C, Cook EH, Gallagher L, Gill M, Lehner T, Senthil G, Sutcliffe JS, Thurm A, Zwick ME, Børglum AD, State MW, Cicek AE, Talkowski ME, Cutler DJ, Devlin B, Sanders SJ°, Roeder K°, Buxbaum JD°, Daly MJ°. Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism. *Cell* (2020), 80(3):568-584.e23. PMID: [31981491](#). DOI: [10.1016/j.cell.2019](#).
Led integration of *de novo* point mutations and autism risk genes with copy-number variation data; created Figure 2F-G
§ Winner of the 2018 ASHG Charles J. Epstein Trainee Award for Excellence in Human Genetics (see Awards & Honors)
- 2019 ! [45] Chaisson MJ*, Sanders AD*, Zhao X*, Malhotra A°, Porubsky D°, Rausch T°, Gardner EJ°, Rodriguez O°, Guo L°, **Collins RL**°, Fan X°, Wen J°, Handsaker RE°, Fairley S°, Kronenberg ZN°, Kong X°, Hormozdiari F°, Lee D°, Wenger AM°, Hastie A°, Antaki D°, Audano P, Brand H, Cantsilieris S, Cao H, Cerveira E, Chen C, Chen X, Chin C, Chong Z, Chuang NT, Church DM, Clarke L, Farrell A, Flores J, Galeev T, Gorkin D, Gujral M, Guryev V, Heaton WH, Korlach J, Kumar S, Kwon JY, Lee JE, Lee J, Lee W, Lee SP, Li S, Marks P, Viaud-Martinez K, Meiers S, Munson KM, Navarro F, Nelson BJ, Nodzak C, Noor A, Kyriazopoulou-Panagiotopoulou S, Pang A, Qiu Y, Rosanio G, Ryan M, Stütz A, Spierings DC, Ward A, Welch AE, Xiao M, Xu W, Zhang C, Zhu Q, Zheng-Bradley X, Yakneen S, McCarroll S, Jun G, Ding L, Koh CL, Ren B, Flicek P°, Chen K°, Gerstein MB°, Kwok P°, Lansdorp PM°, Marth G°, Sebat J°, Shi X°, Bashir A°, Ye K°, Devine SE°, Talkowski M°, Mills RE°, Marschall T°, Korbel J°, Eichler EE°, Lee C°. Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nature Communications* (2019), 10(1):1784. PMID: [30992455](#). DOI: [10.1038/s41467-018-08148-z](#).
Led analysis of long-insert jumping whole-genome libraries; assisted in cross-platform inversion discovery, analysis, and validation
- ! [44] Khera AV*, Chaffin M*, Zekavat SM, **Collins RL**, Roselli C, Natarajan P, Lichtman JH, D'Onofrio G, Mattered J, Dreyer R, Spertus JA, Taylor KD, Psaty BM, Rich SS, Post W, Gupta N, Gabriel S, Lander E, Chen YI, Talkowski ME, Rotter JI, Krumholz

HM^o, Kathiresan S^o. Whole genome sequencing to characterize monogenic and polygenic contributions in patients hospitalized with early-onset myocardial infarction. *Circulation* (2019), 139(13): 1593-1602. PMID: [30586733](https://pubmed.ncbi.nlm.nih.gov/30586733/). DOI: [10.1161/CIRCULATIONAHA.118.035658](https://doi.org/10.1161/CIRCULATIONAHA.118.035658).

Conducted all structural variation discovery and related disease association analyses; created Figure 2; assisted in revising manuscript

- 2018 §! [43] Werling DM*, Brand H*, An J*, Stone MR*, Zhu L*, Glessner JT, **Collins RL**, Dong S, Layer RM, Markenscoff-Papadimitriou E, Farrell A, Schwartz GB, Wang HZ, Currall BB, Zhao X, Dea J, Duhn C, Erdman C, Gilson M, Handsaker RE, Kashin S, Klei L, Mandell JD, Nowakowski TJ, Liu Y, Pochareddy S, Smith L, Walker MF, Waterman MJ, He X, Kriegstein AR, Rubenstein JL, Sestan N, McCarroll SA, Neale BM, Coon H, Willsey Ajeremy, Buxbaum JD, Daly MJ, State MW, Quinlan A, Marth GT, Roeder K, Devlin B^o, Talkowski ME^o, Sanders SJ^o. An analytical framework for whole genome sequence data and its implications for autism spectrum disorder. *Nature Genetics* (2018), 50(5): 727-736. PMID: [29700473](https://pubmed.ncbi.nlm.nih.gov/29700473/). DOI: [10.1038/s41588-018-0107-y](https://doi.org/10.1038/s41588-018-0107-y).

Assisted in study design; assisted in statistical & computational methods development; assisted in structural variant analyses & interpretation
Designed & created Figure 4; assisted in writing of manuscript

§ Highlighted in a News and Views commentary in *Nature Genetics*.

- §! [42] Anechik T*, Hendriks WT*, Yadav R*, Shin D*, Gao D*, Vaine CA, **Collins RL**, Domingo A, Currall B, Stortchevoi A, Multhaupt-Buell T, Penney EB, Cruz L, Dhakal J, Brand H, Hanscom C, Antolik C, Dy M, Ragavendran A, Underwood J, Cantsilieris S, Munson KM, Eichler EE, Acuña P, Go C, Jamora RDG, Rosales RL, Church DM, Williams SR, Garcia S, Klein C, Müller U, Wilhelmens KC, Timmers HTM, Ballouz S, Gillis J, Lyon GJ, Sapir Y, Wainger BJ, Henderson D, Ito N, Weisenfeld N, Jaffe D, Sharma N, Breakefield XO, Ozelius LJ, Bragg DC^o, Talkowski ME^o. Dissecting the causal mechanism of X-linked dystonia-parkinsonism by integrating genome and transcriptome assembly. *Cell* (2018), 172(5): 897-909.e21. PMID: [29474918](https://pubmed.ncbi.nlm.nih.gov/29474918/). DOI: [10.1016/j.cell.2018.02.011](https://doi.org/10.1016/j.cell.2018.02.011).

Led initial genotyping experiments; discovered initial recombination on disease haplotype; advised secondary studies & manuscript drafting

§ Highlighted in a Preview commentary in *Cell* and by *Massachusetts General Hospital*.

- 2017 ! [41] Redin C, Brand H, **Collins RL**, Kammin T, Mitchell E, Hodge JC, Hanscom C, Pillalamarri V, Seabra CM, Abbott M, Abdul-Rahman OA, Aberg E, Adley R, Alcaraz-Estrada SL, Alkuraya FS, An Y, Anderson M, Antolik C, Anyane-Yeboah K, Atkin JF, Bartell T, Bernstein JA, Beyer E, Bongers EMHF, Brilstra EH, Brown CW, Brüggewirth HT, Callewaert B, Corning K, Cox H, Cuppen E, Currall BB, Cushing T, David D, Deardorff MA, Dheedene A, D'hooghe M, de Vries BBA, Earl DL, Ferguson HL, Fisher H, FitzPatrick DR, Gerrol P, Giachino D, Glessner JT, Gliem T, Grady M, Graham BH, Griffis C, Gripp KW, Gropman AL, Hanson-Kahn A, Harris DJ, Hayden MA, Hochstenbach R, Hoffman JD, Hopkin RJ, Hubshman MW, Innes AM, Irons M, Irving M, Janssens S, Jewett T, Johnson JP, Jongmans MC, Kahler SG, Koolen DA, Korzelius J, Kroisel PM, Lacassie Y, Lawless W, Lemyre E, Leppig K, Levin AV, Li H, Li H, Liao EC, Lim C, Lose EJ, Lucente D, Macera MJ, Manavalan P, Mandrile G, Marcelis CL, Margolin L, Mason T, Masser-Frye D, McClellan MW, Mendoza CZ, Menten B, Middelkamp S, Mikami LR, Moe E, Mohammed S, Mononen T, Mortenson ME, Moya G, Nieuwint A, Ordule Z, Parkash S, Pauker SP, Pereira S, Perrin D, Phelan K, Piña-Aguilar RE, Poddighe PJ, Pregno G, Raskin S, Reis L, Rhead W, Rita D, Renkens I, Roelens F, Ruliera J, Rump P, Schilit SLP, Shaheen R, Sparkes R, Spiegel E, Stevens B, Stone MR, Tagoe J, Thakuria JV, van Bon BW, van de Kamp J, van der Burgt I, van Essen T, van Ravenswaaij-Arts CM, van Roosmalen MJ, Vergult S, Volker-Touw CML, Warburton DP, Waterman MJ, Wiley S, Wilson A, Vega M, Zori RT, Levy B, Brunner HG, de Leeuw N, Kloosterman WP, Thorland EC, Morton CC, Gusella JF, Talkowski ME. The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics* (2017), 49(1): 36-45. PMID: [27841880](https://pubmed.ncbi.nlm.nih.gov/27841880/). DOI: [10.1038/ng.3720](https://doi.org/10.1038/ng.3720).

Led development of structural variation discovery methods; analyzed ~10% of all samples; assisted in manuscript drafting & figure design

- 2014 §! [40] Mandal PK*, Ferreira LMR*, **Collins RL**, Meissner TB, Boutwell CL, Friesen M, Garrison BS, Stortchevoi A, Bryder D, Musunuru K, Brand H, Allen TM, Talkowski ME, Rossi DJ^o, Cowan CA^o. Efficient ablation of genes in human hematopoietic stem and effector cells using CRISPR/Cas9. *Cell Stem Cell* (2014), 15(5): 643-652. PMID: [25517468](https://pubmed.ncbi.nlm.nih.gov/25517468/). DOI: [10.1016/j.stem.2014.10.004](https://doi.org/10.1016/j.stem.2014.10.004).

Designed targeted capture sequencing experiments; executed all analyses associated with CRISPR on- and off-target efficiencies

Designed and created Figure 4; assisted in writing of manuscript

§ Featured by *TIME Magazine* and on the cover of *Cell Stem Cell*

- § [39] Brand H*, Pillalamarri V*, **Collins RL**, Eggert S, O'Dushlaine C, Braaten EB, Stone M, Chambert K, Doty ND, Hanscom C, Ditmars H, Blais J, Mills R, Lee C, Gusella JF, McCarroll S, Smoller JW, Talkowski ME^o, Doyle AE^o. Cryptic and complex chromosomal aberrations in early onset neuropsychiatric disorders. *American Journal of Human Genetics* (2014), 95:4, 454-461. PMID: [25279985](https://pubmed.ncbi.nlm.nih.gov/25279985/). DOI: [10.1016/j.ajhg.2014.09.005](https://doi.org/10.1016/j.ajhg.2014.09.005).

Led development of structural variation discovery methods; assisted in study design & writing of manuscript; led design for all figures

§ Recipient of 2014 Massachusetts General Hospital Clinical Research Team Award (see Awards & Honors)

- ! [38] Veres A, Gosis BS, Ding Q, **Collins RL**, Ragavendran A, Brand H, Erdin S, Talkowski ME, Musunuru K. Low incidence of off-target mutations in individual CRISPR-Cas9 and TALEN targeted human stem cell clones detected by whole-genome sequencing. *Cell Stem Cell* (2014), 15:1, 27-30. PMID: [24996167](https://pubmed.ncbi.nlm.nih.gov/24996167/). DOI: [10.1016/j.stem.2014.04.020](https://doi.org/10.1016/j.stem.2014.04.020).

Led structural variation discovery analyses; assisted in writing of manuscript

Articles authored as a minor contributor

- 2025 [37] Awasthi BW, Paulo JA, Burkhart DL, Smith IR, **Collins RL**, Harper JW, Gygi SP, Haigis KM. Integrative Analysis of Germline Rare Variants in Clear and Non-Clear Cell Renal Cell Carcinoma. The network response to Egf is tissue-specific. *iScience* (2025), 112146. PMID: TBD. DOI: <https://doi.org/10.1016/j.isci.2025.112146>.

Advised on statistical aspects of proteomic, phosphoproteomic, and transcriptomic analyses.

- 2024 [36] Han S, Camp SY, Chu H, Collins RL, Gillani R, Park J, Bakouny Z, Ricker CA, Reardon B, Moore N, Kofman E, Labaki C, Braun D, Choueiri TK, AlDubayan SH*, Van Allen EM*. Integrative Analysis of Germline Rare Variants in Clear and Non-Clear Cell Renal Cell Carcinoma. *European Urology Open Science* (2024), 62, 107-122. PMID: [38496821](https://pubmed.ncbi.nlm.nih.gov/38496821/). DOI: [10.1016/j.euro.2024.02.006](https://doi.org/10.1016/j.euro.2024.02.006).

Collaborated on integration of noncoding constraint and selection on SVs.

- ! [35] Chen S, Francioli L, Goodrich J, **Collins RL**, Wang Q, Alfoldi J, Watts N, Vittal C, Gauthier L, Poterba T, Wilson M, Tarasova Y, Phu W, Yohannes M, Koenig Z, Farjoun Y, Banks E, Donnelly S, Gabriel S, Gupta N, Ferreira S, Tolonen C, Novod S, Bergelson L, Roazen D, Ruano-Rubio V, Covarrubias M, Llanwarne C, Petrillo N, Wade G, Jeandet T, Munshi R, Tibbetts K, The gnomAD Project Consortium, O'Donnell-Luria A, Solomonson M, Seed C, Martin A, Talkowski M, Rehm H, Daly M, Tiao G, Neale B, MacArthur D, Karczewski K. A genomic mutational constraint map using variation in 76,156 human genomes. *Nature* (2023), 625:7993, 92-100. PMID: [38057664](#). DOI: [10.1038/s41586-023-06045-0](#). Collaborated on integration of noncoding constraint and selection on SVs.
- 2023 [34] Lowther C*, Valkanas E*, Giordano JL, Wang HZ, Currall BB, O'Keefe K, **Collins RL**, Zhao X, Aggarwal V, Lucente D, Margolin L, An JY, Werling DM, Dong S, Sanders SJ, Devlin B, Gilmore K, Powell B, Brandt A, O'Donnell-Luria AH, Lennon NJ, Goldstein DB, Rehm HL, Vora NL, MacArthur D, Levy B°, Brand H°, Wapner R°, Talkowski ME°. Systematic evaluation of genome sequencing for the diagnostic assessment of autism spectrum disorder and fetal structural anomalies. *American Journal of Human Genetics* (2023), 110:9, 1454-1469. PMID: [37595579](#). DOI: [10.1016/j.ajhg.2023.07.010](#). Assisted in detection and interpretation of variants from genome sequencing data; assisted in quality control; assisted in manuscript preparation
- ! [33] Babadi M*, Fu JM*, Lee SK*, Smirnov AN*, Gauthier LD, Walker M, Benjamin DI, Zhao X, Karczewski KJ, Wong I, **Collins RL**, Sanchis-Juan A, Brand H, Banks E, Talkowski ME. GATK-gCNV enables the discovery of rare copy number variants from exome sequencing data. *Nature Genetics* (2023), 55:9, 1589-1597. PMID: [37604963](#). DOI: [10.1038/s41588-023-01449-0](#). Contributed to application & quality control of gCNV methods; contributed to manuscript & figure design.
- [32] Macnee M, Pérez-Palma E, Brünger T, Klöckner C, Platzer K, Stefanski A, Montanucci L, Bayat A, Radtke M, **Collins RL**, Talkowski M, Blankenberg D, Moller R, Lemke J, Nothnagel M, May P, Lal D. CNV-ClinViewer: Enhancing the clinical interpretation of large copy-number variants online. *Bioinformatics* (2023), 39:5. PMID: [37104749](#). DOI: [10.1093/bioinformatics/btad290](#). Provided feedback on the design & implementation of CNV viewer interface.
- [31] Kaivola K, Chia R, Ding J, Rasheed M, Fujita M, Menon V, Walton RL, **Collins RL**, Billingsley K, Brand H, Talkowski M, Zhao X, Dewan R, Stark A, Ray A, Solaiman S, Jerez PA, Malik L, Dawson TM, Rosenthal LS, Albert MS, Pletnikova O, Troncoso JC, Masellis M, Keith J, Black SE, Ferrucci L, Resnick SM, Tanaka T, The American Genome Center, International LBD Genomics Consortium, International ALS/FTD Consortium, PROSPECT Consortium, Topol E, Torkamani A, Tienari P, Foroud TM, Ghetti B, Landers JE, Ryten M, Morris HR, Hardy JA, Mazzini L, D'Alfonso S, Moglia C, Calvo A, Serrano GE, Beach TG, Ferman T, Graff-Radford NR, Boeve BF, Wszolek ZK, Dickson DW, Chiò A, Bennett DA, De Jager PL, Ross OA, Dalgard CL, Gibbs JR, Traynor BJ, Scholz SW. *Cell Genomics* (2023), 3:100316. PMID: TBD. DOI: [10.1016/j.xgen.2023.100316](#). Advised on the generation, filtering, and quality control of SV calls from short-read genome sequencing.
- [30] Billingsley KJ, Ding J, Jerez PA, Illarionova A, Grenn FP, Makarios MB, Moore A, Vitale D, Reed X, Hernandez D, Torkamani A, Ryten M, Hardy J, Chia R, Scholz SW, Traynor BJ, Dalgard CL, Ehrlich DJ, Tanaka T, Ferrucci L, Beach T, Serrano GE, Quinn JP, Bubbs VJ, **Collins RL**, Zhao X, Walker M, Pierce-Hoffman E, Brand H, Talkowski M, Casey B, Cookson MR, Markham A, Nalls M, Mahmoud M, Sedlazeck FJ, Blauwendraat C, Gibbs JR, Singleton AB. Genome-wide analysis of structural variants in Parkinson's Disease using short-read sequencing data. *Annals of Neurology* (2023), online ahead of print. PMID: [36695634](#). DOI: [10.1002/ana.26608](#). Advised on the generation, filtering, and quality control of SV calls from short-read genome sequencing.
- 2022 [29] Tai DJC*, Razaz P*, Erdin S*, Gao D*, Wang J, Nuttle X, de Esch CE, Collins RL, Currall BB, O'Keefe K, Burt ND, Yadav R, Mohajeri K, Aneichyk T, Ragavendran A, Stortchevoi A, Morini E, Ma W, Lucente D, Hastie A, Kelleher RJ, Perlis RH, Talkowski ME°, Gusella JF°. Tissue and cell-type specific molecular and functional signatures of 16p11.2 reciprocal genomic disorder across mouse brain and human neuronal models. *American Journal of Human Genetics* (2022), 109:10, 1789-1813. PMID: [36152629](#). DOI: [10.1016/j.ajhg.2022.08.012](#). Conducted analyses of genomic properties of 16p11.2 CNV locus.
- 2021 ! [28] Nasser J*, Bergman DT*, Fulco CP*, Guckelberger P*, Doughty BR*, Patwardhan T, Jones TR, Nguyen TH, Ulirsch JC, Natri HM, Weeks EM, Munson G, Kane M, Kang HY, Cui A, Ray JP, Eisenhaure TM, Mualim K, **Collins RL**, Dey K, Price A, Epstein CB, Kundaje A, Xavier RJ, Daly MJ, Huang H, Finucane HK, Hacohen N, Lander ES°, Engreitz JM°. Genome-wide maps of enhancer regulation connect risk variants to disease genes. *Nature* (2021), 593: 238-243. PMID: [33828297](#). DOI: [10.1038/s41586-021-03446-x](#). Contributed to development of enhancer prediction model (ABC-Max) in 16 immune cell types
- [27] Gudmundsson S, Karczewski KJ, Francioli LC, Tiao G, Cummings BB, Alfoldi J, Wang Q, **Collins RL**, Laricchia KM, Ganna A, Birnbaum DP, Gauthier LD, Brand H, Solomonson M, Watts NA, Rhodes D, Singer-Berk M, England EM, Seaby EG, Kosmicki JA, Walters RK, Tashman K, Farjoun Y, Banks E, Poterba T, Wang A, Seed C, Whiffin N, Chong JX, Samocha KE, Pierce-Hoffman E, Zappala Z, O'Donnell-Luria AH, Vallabh-Minikel E, Weisburd B, Lek M, Ware JS, Vittal C, Armean IM, Bergelson L, Cibulskis K, Connolly KM, Covarrubias M, Donnelly S, Ferreira S, Gabriel S, Gentry J, Gupta N, Jeandet T, Kaplan D, Llanwarne C, Munshi R, Novod S, Petrillo N, Roazen D, Ruano-Rubio V, Saltzman A, Schleicher M, Soto J, Tibbetts K, Tolonen C, Wade G, Talkowski ME, Neale BM, Daly MJ, MacArthur DG. Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature* (2021), 597(7874): E3-E4. PMID: [34373650](#). DOI: [10.1038/s41586-021-03758-y](#). Contributed to the production of the Genome Aggregation Database (gnomAD)
- [26] Wahlster L*, Verboon JM*, Ludwig LS, Black SC, Luo W, Garg K, Voit RA, **Collins RL**, Garimella K, Costello M, Chao KR, Goodrich JK, DiTroia SP, O'Donnell-Luria A, Talkowski ME, Michelson AD, Cantor AB, Sankaran VG. Familial Thrombocytopenia Due to a WAC-ANKRD26 Gene Fusion. *Journal of Experimental Medicine* (2021), 218 (6): e20210444. PMID: [33857290](#). Assisted in sequencing data analysis to identify the causal complex inversion underlying this Mendelian blood disorder

- [25] Jung R, Lee Y, Barker D, Correia K, Shin B, Loupe J, **Collins RL**, Lucente D, Ruliera J, Gillis T, Mysore JS, Rodan L, Picker J, Lee J, Howland D, Lee R, Kwak S, MacDonald ME., Gusella JF., Seong IS. Mutations causing Lopes-Maciel-Rodan Syndrome are huntingtin hypomorphs. *Human Molecular Genetics* (2021), 30(3-4): 135-148. PMID: [33432339](https://pubmed.ncbi.nlm.nih.gov/33432339/). DOI: [10.1093/hmg/ddaa283](https://doi.org/10.1093/hmg/ddaa283).
Curated information on the presence of Huntington loss-of-function mutations in the general population
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Contributed to structural variation discovery analyses and statistical methods development; assisted in manuscript drafting & figure design
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GitHub <https://github.com/RCollins13>
Twitter <https://twitter.com/ryanlcollins13>