Ryan Lewis Collins

Ph.D. Candidate, Bioinformatics and Integrative Genomics Ph.D. Program, Harvard Medical School
NSF Graduate Research Fellow, Talkowski Laboratory, Massachusetts General Hospital
Team Leader, Population Genetics & Genome Biology, Broad-SV Group, Broad Institute of MIT and Harvard

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Professional status: sixth-year Ph.D. candidate at Harvard Medical School studying human genome structure & function in the lab of Dr. Michael Talkowski at Massachusetts General Hospital and The Broad Institute.

Career objective: to advance knowledge of the structure-function dynamics in the human genome and their implications for development, disease, population genetics, and precision medicine, ideally by leading an independent research group.

Scientific interests: genome biology, structural variation, chromatin, genome organization, sequencing technologies, interpretation of genetic variation, gene regulation, statistics, predictive modeling, bioinformatics, and medical genetics.

Education

Degrees

Ph.D. Harvard University, *Cambridge, MA, USA*
2016-Present 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.
Thesis Committee Shamil Sunyaev, Ph.D.; Mark Daly, Ph.D.; James Gusella, Ph.D.; Marcin Imielinski, M.D., 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.

Coursework

Harvard Medical School Principles of Molecular Biology; Principles of Genetics; Clinical Genetics; Computational and Functional Genomics; Advanced Topics in Gene Regulation; Conduct of Science; Analysis of Biological Literature

Massachusetts Institute of Technology (MIT) Quantitative Genomics; Machine Learning

Dartmouth College Molecular Genetics of Eukaryotes; Computational Molecular Biology; Experimental Genetic Analysis; Gene Expression & Inheritance; Biostatistics; Multivariate Calculus; Discrete Probability; Hazardous Data; General Physics; Cell Structure & Function; Physiology; Art of Science Writing; General Chemistry

Research Experience

Team Leader Population Genetics & Genome Biology Team, Broad-SV Group
2018-Present Broad Institute of MIT and Harvard | Cambridge, MA, USA

Ph.D. Candidate Laboratory of Michael E. Talkowski, Ph.D.
2017-Present 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

Teaching Assistant Clinical Genetics (GEN288; Instructor: David Sweetser, M.D.)
2020 Harvard Medical School | Boston, MA, USA

Invited Lecturer Human Genetics (GEN202; Instructor: Matthew Warman, M.D.)
2018 Harvard Medical School | Boston, MA, USA

Invited Lecturer Short Course on Rare Variant Interpretation in Mendelian Diseases
2017 Broad Institute of MIT and Harvard | Cambridge, MA, USA

Teaching Assistant Gene Expression and Inheritance (BIOL013; Instructor: Thomas Jack, Ph.D.)
2012-2013 Dartmouth College | Hanover, NH, USA

Volunteer Tutor Calculus and Advanced Mathematics
2010-2011 Hanover High School | Hanover, NH, USA

ACADEMIC SERVICE

Invited Referee 3 articles for 3 journals, including: *Cell Genomics; Human Genetics; npj Genomic Medicine*
2020-Present

***Ad Hoc* Referee** 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*
2013-Present All *ad hoc* referee activities conducted with Michael Talkowski, Ph.D.

Organizer Bioinformatics and Integrative Genomics Student Research Seminar Series
2019-2020 Harvard Medical School | Boston, MA, USA

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

Admissions Interviewer PhD Student Admissions, Program in Bioinformatics and Integrative Genomics
2018 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

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

2016-Present 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*
Including Collins *et al.*, *Nature* (2020)

2016-Present NIH Center for Mendelian Genomics, Broad Institute of MIT and Harvard
Wahlster *et al., J. Exp. Med.* (2021); Shaw *et al.*, *Nat. Genet.* (2017)

2015-Present Core Analysis Group, Autism Sequencing Consortium
Satterstrom, *et al., Cell* (2020); An *et al., Science* (2018); Werling *et al.*, *Nat. Genet.* (2018)

2015-Present Structural Variation Group (Broad-SV), Broad Institute of MIT and Harvard
Collins *et al., medRxiv* (2021); Collins *et al.*, *Genome Biol.* (2017)

2013-Present Developmental Genome Anatomy Project (DGAP) / International Breakpoint Mapping Consortium (IBMC)
Redin *et al.*, *Nat. Genet.* (2017)

2016-2020 NIH Center for Common Disease Genomics, Broad Institute of MIT and Harvard
Khera *et al., Circulation* (2018)

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)

AFFILIATIONS

2017-Present Sigma Xi Honors Society for Scientific Research

2015-Present Program in Population and Medical Genetics, Broad Institute of MIT and Harvard

2014-Present American Society of Human Genetics

FELLOWSHIPS & FUNDING

FELLOWSHIPS

2017-2021 $138,000 NSF Graduate Research Fellowship Program (GRFP; NSF #2017240332)

2016-2018 $92,086 NIH/NHGRI Institutional National Research Service Award (T32; 4T32HG002295-14; PI: Park)

AWARDS & HONORS

MAJOR

2021 European Society for Human Genetics 2021 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 2021 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

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

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

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

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 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.*

"A cross-disorder dosage sensitivity map of the human genome"

[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 Structural Variation Group	*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 Virtual 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 15th 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 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*

PUBLICATIONS

Peer-Reviewed Publications: 40 **Preprinted Papers:** 3 **Total Citations:** 6,550 **h-index:** 25 **i10-index:** 29

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: */°/† *equal contributions* § *publication highlight* ! *highly cited (≥100 citations)* Citation statistics from Google Scholar

REFEREED SCIENTIFIC ARTICLES

Articles authored as a primary contributor

2020 § ! [40] **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 § ! [39] **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 § [38] 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 [37] **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

2021 § [36] 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)*, online ahead of print.* PMID: 33789087. *bioRxiv* DOI: 10.1101/2020.07.03.168831.

Assisted in study design; contributed to analysis of short-read vs. long-read comparisons; assisted in manuscript & figure preparation

§ ***Highlighted by* GenomeWeb**

2020 § ! [35] 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***

§ ! [34] 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, Chiocchetti 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, Kolevzon 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 ! [33] 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

! [32] Khera AV*, Chaffin M*, Zekavat SM, **Collins RL**, Roselli C, Natarajan P, Lichtman JH, D'Onofrio G, Mattera 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°, Kathiresan S°. 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. DOI: 10.1161/CIRCULATIONAHA.118.035658.
Conducted all structural variation discovery and related disease association analyses; created Figure 2; assisted in revising manuscript

2018 § ! [31] 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°, Talkowski ME°, Sanders SJ°. An analytical framework for whole genome sequence data and its implications for autism spectrum disorder. ***Nature Genetics*** (2018), 50(5): 727-736. PMID: 29700473. DOI: 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.**

§ ! [30] Aneichyk 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, Wilhelmsen 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°, Talkowski ME°. Dissecting the causal mechanism of X-linked dystonia-parkinsonism by integrating genome and transcriptome assembly. ***Cell*** (2018), 172(5): 897-909.e21. PMID: 29474918. DOI: 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 ! [29] 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-Yeboa K, Atkin JF, Bartell T, Bernstein JA, Beyer E, Bongers EMHF, Brilstra EH, Brown CW, Brüggenwirth 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, Ordulu 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. DOI: 10.1038/ng.3720.
Led development of structural variation discovery methods; analyzed ~10% of all samples; assisted in manuscript drafting & figure design

2014 § ! [28] 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°, Cowan CA°. Efficient ablation of genes in human hematopoietic stem and effector cells using CRISPR/Cas9. ***Cell Stem Cell*** (2014), 15(5): 643-652. PMID: 25517468. DOI: 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**

§ [27] 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°, Doyle AE°. Cryptic and complex chromosomal aberrations in early onset neuropsychiatric disorders. ***American Journal of Human Genetics*** (2014). 95:4, 454-461. PMID: 25279985. DOI: 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)**

! [26] 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. DOI: 10.1016/j.stem.2014.04.020.
Led structural variation discovery analyses; assisted in writing of manuscript

Articles authored as a minor contributor

2021 [25] 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

[24] Gudmundsson S, 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, 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, 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, Neale BM, Daly MJ, MacArthur DG. Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. ***Nature*** (2021), online ahead of print. PMID: 34373650. DOI: 10.1038/s41586-021-03758-y.
Contributed to the production of the Genome Aggregation Database (gnomAD)

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Assisted in sequencing data analysis to identify the causal complex inversion underlying this Mendelian blood disorder

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Contributed to structural variation discovery analyses and statistical methods development; assisted in manuscript drafting & figure design

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Assisted in variant interpretation and manuscript editing

2019 [19] Toomer K*, Yu M*, Fulmer D, Guo L, Moore K, Moore R, Drayton K, Glover J, Peterson N, Ramos-Ortiz S, Drohan A, Catching BJ, Stairley R, Wessels A, Lipschutz JH, Delling FN, Jeunemaitre X, Dina C, **Collins RL**, Brand H, Talkowski ME, del Monte F, Mukherjee R, Awgulewitsch A, Body S, Hardiman G, Da Silveira W, Wang B, Leyne M, Durst R, Markwald R, Le Scouranec S, Hagege A, Le Tourneau T, Kohl P, Rog-Zielinska E, Schott J°, Levine RA°, Milan D°, Bouatia-Naji N°, Slaugenhaupt S°, Norris RA°. Primary cilia defects causing mitral valve prolapse. ***Science Translational Medicine*** (2019), 11(493). PMID: 31118289. DOI: 10.1126/scitranslmed.aax0290.
Conducted exome sequencing analysis that identified missense variants in *DZIP1* as a likely causal gene for mitral valve prolapse

2018 ! [18] An JY*, Lin K*, Zhu L*, Werling DM*, Dong S, Brand H, Wang HZ, Zhao X, Schwartz GB, **Collins RL**, Currall BB, Dastmalchi C, Dea J, Duhn C, Gilson MC, Klei L, Liang L, Markenscoff-Papadimitriou E, Pochareddy S, Ahituv N, Buxbaum JD, Coon H, Daly MJ, Kim YS, Marth GT, Neale BM, Quinlan AR, Rubenstein JL, Sestan N, State MW, Willsey AJ, Talkowski ME°, Devlin B°, Roeder K°, Sanders SJ°. Genome-wide *de novo* risk score implicates promoter variation in autism spectrum disorder. ***Science*** (2018), 362(6420). PMID: 30545857. DOI: 10.1126/science.aat8464.
Contributed to genome annotations, burden testing design, promoter-proximal analysis framework, and figure design

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Contributed to study design, analyses, and manuscript drafting

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Ferraro N, Verloes A, Rauch A, Steindl K, Zweier M, Scheer I, Sato D, Okamoto N, Jacobsen C, Tryggestad J, Chernausek S, Schimmenti LA, Brasseur B, Cesaretti C, García-Ortiz JE, Buitrago TP, Silva OP, Hoffman JD, Mühlbauer W, Ruprecht KW, Loeys BL, Shino M, Kaindl AM, Cho C, Morton CC, Meehan RR, van Heyningen V, Liao EC, Balasubramanian R, Hall JE, Seminara SB, Macarthur D, Moore SA, Yoshiura K, Gusella JF, Marsh JA, Graham Jr JM, Lin AE, Katsanis N, Jones PL, Crowley Jr WF, Davis EE°, FitzPatrick DR°, Talkowski ME°. *SMCHD1* mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. ***Nature Genetics*** (2017), 49(2): 238-248. PMID: 28067909. DOI: 10.1038/ng.3743.
Analyzed the initial set of 18/40 whole-exomes that primed discovery of *SMCHD1* as pleiotropic contributor to arhinia
§ *Highlighted in a* News and Views *commentary in* Nature Genetics *and reviewed in* Trends in Genetics.

§ [14] Maussion G*, Cruceanu C*, Rosenfeld JA, Bell SC, Jollant F, Szatkiewicz J, **Collins RL**, Hanscom C, Kolobova I, de Champfleur NM, Blumenthal I, Chiang C, Ota V, Hultman C, O'Dushlaine C, McCarroll S, Alda M, Jacquemont S, Ordulu Z, Marshall CR, Carter MT, Shaffer LG, Sklar P, Girirajan S, Morton CC, Gusella JF, Turecki G, Stavropoulos DJ, Sullivan PF, Scherer SW, Talkowski ME, Ernst C. Implication of LRRC4C and DPP6 in neurodevelopmental disorders. ***American Journal of Medical Genetics*** (2017), 173(2): 395-406. PMID: 27759917. DOI: 10.1002/ajmg.a.38021.
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§ *Featured on the cover of* The American Journal of Medical Genetics

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Performed structural variation analyses for 20% of samples presented in study

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Co-led analysis of whole-genome sequencing of patient in study

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Performed whole-genome analysis of chromosomal integrity in germline samples to validate genome structure of transgenic animal models

[10] Tai DJC, Ragavendran A, Manavalan P, Stortchevoi A, Seabra C, Erdin S, **Collins RL**, Blumenthal I, Chen X, Shen Y, Sahin M, Zheng C, Lee C, Gusella JF°, Talkowski ME°. Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. ***Nature Neuroscience*** (2016), 19(3): 517-522. PMID: 26829649. DOI: 10.1038/nn.4235.
Assisted in guide RNA design and selection to target highly homologous regions of 16p11.2 segmental duplications

2015 ! [9] Turner T, Sharma K, Oh EC, Liu YP, **Collins RL**, Sosa MX, Auer D, Brand H, Sanders SJ, Moreno-De-Luca D, Pihur V, Piona T, Pike K, Soppet D, Smith MW, Cheung SW, Martin CL, State M, Talkowski ME, Cook E, Huganir R, Katsanis N, Chakravarti A. Loss of delta catenin function in severe autism. ***Nature*** (2015), 519(7543): 51-56. PMID: 25807484. DOI: 10.1038/nature14186.
Analyzed *CTNND2* autism risk conferred by copy number variation in large cohort (N>50k); designed & created Extended Data Figure 6

2013 [8] Hu T, Chen Y, Kiralis JW, **Collins RL**, Wejse C, Sirugo G, Williams SM, Moore JH. An information-gain approach to detecting three-way epistatic interactions in genetic association studies. ***Journal of the American Medical Informatics Association*** (2013), 20(4): 630-636. PMID: 23396514. DOI: 10.1136/amiajnl-2012-001525.
Assisted in testing and application of statistical methods

Articles as a group or consortium author

2020 [7] Minikel EV, Karczewski KJ, Martin HC, Cummings BB, Whiffin N, Alfoldi J, Trembath RC, van Heel DA, Daly MJ, **The gnomAD Production Team**, The gnomAD Consortium, Schreiber SL, MacArthur DG. Evaluating potential drug targets through human loss-of-function genetic variation. ***Nature*** (2020), 581: 459-464. PMID: 32461653. DOI: 10.1038/s41586-020-2267-z.
Contributed to the production of the Genome Aggregation Database (gnomAD)

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Led integration of significant & pleiotropic GWAS loci with rare variants and genomic disorders

Books & Book Chapters

2019 [1] Antolik C, Hanscom C, Currall BB, **Collins RL**, Talkowski ME. Next-generation sequencing of prenatal structural chromosomal rearrangements. ***Methods in Molecular Biology****, Vol. 1885: Prenatal Diagnosis* (2019). Edited by Levy B.
Wrote subchapter on computational analysis of prenatal genome sequencing; assisted in drafting & editing of full chapter

Preprint Manuscripts

2021 § [3] **Collins RL**, Glessner JT, Porcu E, Niestroj L, Ulirsch J, Kellaris G, Howrigan DP, Everett S, Mohajeri K, Nuttle X, Lowther C, Fu J, Boone PM, Ullah F, Samocha KE, Karczewski K, Lucente D, Consortium E, Gusella JF, Finucane H, Matyakhina L, Aradhya S, Meck J, Lal D, Neale BM, Hodge JC, Reymond A, Kutalik Z, Katsanis N, Davis EE, Hakonarson H, Sunyaev S, Brand H, Talkowski ME. A cross-disorder dosage sensitivity map of the human genome. ***medRxiv*** (2021). DOI: 10.1101/2021.01.26.21250098.
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.***

2020 [2] 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 as a first-tier diagnostic test for prenatal and pediatric disorders. ***bioRxiv*** (2020). DOI: 10.1101/2020.08.12.248526.
Assisted in detection and interpretation of variants from genome sequencing data; assisted in quality control; assisted in manuscript preparation

2016 [1] **Collins RL**, Stone MR, Brand H, Glessner JT, Talkowski ME. CNView: a visualization and annotation tool for copy number variation from whole-genome sequencing. ***bioRxiv*** (2016). DOI: 10.1101/049536.
Developed all methods; wrote manuscript & created all figures

Other Scientific Works

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Public Profiles

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