

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

185 Cambridge St., Floor 5
Boston, MA, 02114
(603) 290-2200
www.RyanLCollins.com
RLCollins@g.harvard.edu

Professional status: fifth-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: structural variation, chromatin, genome topology, sequencing technologies, interpretation of genetic variation, gene regulation & regulatory elements, 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 Dr. 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

2018-Present NIH “All of Us” Genome Center, Broad Institute of MIT and Harvard

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
Shaw *et al.*, *Nat. Genet.* (2017)

2015-Present Core Analysis Group, Autism Sequencing Consortium
Kosmicki, *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.*, *bioRxiv* (2021); Collins *et al.*, *Genome Biol.* (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)
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

- 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 (Overall Winner)
\$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 (Overall Winner), 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 (Overall Winner), 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"

	January 2021	Invited Speaker	Broad Institute Structural Variation Group	Cambridge, MA, USA
[E]	October 2020	Poster Presentation	American Society of Human Genetics Annual Meeting	San Diego, CA, USA
[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]	October 2020	Invited Speaker	Boston Children's Hospital Virtual Seminar Series	Boston, MA
[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	<i>Boston, MA, USA</i>
[E]	December 2019	Invited Speaker	Yale Genetics Walter J. Burdette Trainee Symposium	<i>New Haven, CT, USA</i>
[E]	October 2019	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	<i>Houston, TX, USA</i>
[E]	July 2019	Selected Platform Speaker	Gordon Research Seminar: Human Genetics & Genomics	<i>Waterville Valley, NH, USA</i>
[E]	July 2019	Invited Speaker	ClinGen Biocurator Working Group Monthly Conference Call	<i>International (Conf. Call)</i>
[E]	May 2019	Invited Speaker	NHGRI Genome Sequencing Program Methods Group Conference Call	<i>International (Conf. Call)</i>
[E]	May 2019	Poster Presentation	Cold Spring Harbor Laboratory Biology of Genomes Annual Meeting	<i>Cold Spring Harbor, NY, USA</i>
	May 2019	Selected Platform Speaker	Massachusetts General Hospital CGM Annual Trainee & Staff Seminar	<i>Boston, MA, USA</i>
	March 2019	Invited Speaker	Massachusetts General Hospital Reproductive Endocrine Unit Seminar	<i>Boston, MA, USA</i>
	January 2019	Invited Speaker	Broad Institute Data Sciences Platform Methods & Applications Seminar	<i>Cambridge, MA, USA</i>
	November 2018	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	<i>Cambridge, MA, USA</i>
[E]	February 2018	Invited Speaker	gnomAD Consortium Monthly Analysis Conference Call	<i>International (Conf. Call)</i>
	January 2018	Invited Speaker	Harvard Medical School BIG Ph.D. Recruitment Informational Session	<i>Boston, MA, USA</i>
	August 2017	Selected Speaker	Talkowski Laboratory Retreat	<i>Portsmouth, NH, USA</i>

“Methods for structural variation discovery from whole-genome sequencing”

	December 2018	Invited Speaker	Broad Institute Data Sciences Platform Methods Primer	<i>Cambridge, MA, USA</i>
	January 2018	Invited Speaker	Broad Institute Data Sciences Platform Methods Seminar	<i>Cambridge, MA, USA</i>

“Convergence of copy-number variants and point mutations in neurodevelopmental disorders”

[E]	October 2018	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	<i>San Diego, CA, USA</i>
	October 2018	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	<i>Cambridge, MA, USA</i>
[E]	June 2018	Invited Speaker	Autism Sequencing Consortium Weekly Analysis Conference Call	<i>International (Conf. Call)</i>

“Complex structural variation in neurodevelopmental disorders”

[E]	July 2017	Poster Presentation	Gordon Research Conference: Human Genetics & Genomics	<i>Stowe, VT, USA</i>
[E]	July 2017	Poster Presentation	Gordon Research Seminar: Human Genetics & Genomics	<i>Stowe, VT, USA</i>
[E]	April 2017	Selected Platform Speaker	NHGRI Research Training and Career Development Annual Meeting	<i>St. Louis, MO, USA</i>
	December 2016	Poster Presentation	Broad Institute Scientific Retreat	<i>Boston, MA, USA</i>
	July 2016	Selected Speaker	Talkowski Laboratory Retreat	<i>Newport, RI, USA</i>
[E]	May 2016	Poster Presentation	Cold Spring Harbor Laboratory Biology of Genomes Annual Meeting	<i>Cold Spring Harbor, NY, USA</i>
[E]	May 2016	Invited Speaker	Human Genome Structural Variation Consortium Semiannual Meeting	<i>Cold Spring Harbor, NY, USA</i>
[E]	October 2015	Poster Presentation	American Society of Human Genetics Annual Meeting	<i>Baltimore, MD, USA</i>
	April 2015	Poster Presentation	Massachusetts General Hospital Scientific Advisory Council Meeting	<i>Boston, MA, USA</i>
	April 2015	Poster Presentation	Massachusetts General Hospital Center for Human Genetics Research Retreat	<i>Cambridge, MA, USA</i>

“Limited contribution of rare, noncoding variation to autism spectrum disorders”

	April 2017	Invited Speaker	Massachusetts General Hospital Analytical and Translational Genetics Unit	<i>Boston, MA, USA</i>
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“Understanding dosage biases in the analysis of whole-genome sequencing datasets”

	February 2016	Invited Speaker	Broad Institute Structural Variation and Assembly Group	<i>Cambridge, MA, USA</i>
	December 2016	Invited Speaker	Broad Institute Medical and Population Genetics Seminar	<i>Cambridge, MA, USA</i>
	May 2015	Invited Speaker	Massachusetts General Hospital Molecular Neurogenetics Unit Seminar	<i>Boston, MA, USA</i>

“Structural rearrangements in genome editing with CRISPR-Cas9 and TALENs”

	January 2015	Invited Speaker	Massachusetts General Hospital Center for Human Genetics Research Seminar	<i>Boston, MA, USA</i>
[E]	October 2014	Selected Platform Speaker	American Society of Human Genetics Annual Meeting	<i>San Diego, CA, USA</i>
	April 2014	Poster Presentation	Massachusetts General Hospital Center for Human Genetics Research Retreat	<i>Cambridge, MA, USA</i>
	April 2014	Invited Speaker	Massachusetts General Hospital Molecular Neurogenetics Unit Seminar	<i>Boston, MA, USA</i>

PUBLICATIONS

Peer-Reviewed Publications: 36 Preprinted Papers: 5 Total Citations: 4,386 h-index: 23 i10-index: 26

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 § ! [36] **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 YL, 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 §! [35] **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 § [34] 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 [33] **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

- 2020 §! [32] 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, 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, 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
- §! [31] 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, Kolvezon A, Kushima I, Lee SL, Lehtimäki T, Lim ET, Lintas C, Lipkin WI, Loperogo 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, Sussner E, Sweeney J, Szatmari P, Tang L, Tassone F, Teufel K, Trabetti E, Truelles 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 ! [30] 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
- 2018 §! [29] 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](#). 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](#).**

- § [28] Aneichyk T*, Hendriks WT*, Yadav R*, Shin D*, Gao D*, Vaine CA, **Collins RL**, Domingo A, Currall B, Stortchevoi A, Mulhaupt-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, Wilhelmensn 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](#). 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 ! [27] 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](#). DOI: [10.1038/ng.3720](#).

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

- 2014 § ! [26] 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](#). 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](#)**

- § [25] 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](#). 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)**

- ! [24] 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 [23] 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), online ahead of print. PMID: [33432339](#). DOI: [10.1093/hmg/ddaa283](#).

Curated information on the presence of Huntington loss-of-function mutations in the general population

- 2020 [22] Han L*, Zhao X*, Benton ML, Perumal T, **Collins RL**, Hoffman G, Johnson JS, Sloofman L, Consortium C, Brennand KJ, Brand H, Sieberts SK, Marengo S, Peters MA, Lipska BK, Roussos P, Capra JA, Talkowski M, Ruderfer DM. Functional annotation of rare structural variation in the human brain. *Nature Communications* (2020), 11:2990, 1-13. PMID: [32533064](#). DOI: [10.1038/s41467-020-16736-1](#).

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

- [21] Lilleväli H, Pajusalu S, Wojcik MH, Goodrich J, **Collins RL**, Murumets U, Tammur P, Blau N, Lilleväli K, Öunap K. Genome sequencing identifies a homozygous inversion disrupting *QDPR* gene as a cause for dihydropteridine reductase deficiency. *Molecular Genetics & Genomic Medicine* (2020), 5:e1154. PMID: [32022462](#). DOI: [10.1002/mgg3.1154](#).

Assisted in variant interpretation and manuscript editing

- 2019 [20] 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°. Mitral valve prolapse: a congenital disease of the primary cilia. *Science Translational Medicine* (2019), 11(493). PMID: [31118289](#). DOI: [10.1126/scitranslmed.aax0290](#).

Conducted exome sequencing analysis that identified missense variants in *DZ1P1* as a likely causal gene for mitral valve prolapse

- [19] Khera AV*, Chaffin M*, Zekavat SM, **Collins RL**, Roselli C, Natarajan P, Lichtman JH, D'Onofrio G, Maller 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 [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

- [17] Halgren C, Nielsen NM, Nazaryan-Petersen L, Silaharoglu A, **Collins RL**, Lowther C, Kjaergaard S, Frisch M, Kirchhoff M, Brøndum-Nielsen K, Lind-Thomsen A, Mang Y, El-Schich Z, Boring C, Mehriouy MM, Jensen PKA, Fagerberg C, Krogh LN, Hansen J, Bryndorf T, Hansen C, Talkowski ME, Bak M, Tommerup N*, Bache I*. Risk and recommendations in prenatally detected *de novo* balanced chromosomal rearrangements from assessment of long-term outcomes. *American Journal of Human Genetics* (2018). PMID: [29805044](#). DOI: [10.1016/j.ajhg.2018.04.005](#).

Led phenotype-blinded analysis of 27 prenatal WGS cases, including functional interpretation and phenotype prediction

- 2017 [16] Pedersen B, **Collins RL**, Talkowski ME, Quinlan A. Indexcov: fast whole-genome coverage quality-control from BAM or CRAM indexes. *GigaScience* (2017), 6(11), 1-6. PMID: [29048539](#). DOI: [10.1093/gigascience/gix090](#).

Contributed to study design, analyses, and manuscript drafting

- § [15] Shaw ND*, Brand H*, Kupchinsky ZA, Bengani H, Plummer L, Jones TI, Erdin S, Williamson KA, Rainger J, Stortchevoi A, Samochoa K, Currall BB, Dunican DS, **Collins RL**, Willer JR, Lek A, Lek M, Nassan M, Pereira S, Kammin T, Lucente D, Silva A, Seabra CM, Chiang C, An Y, Ansari M, Rainger JK, Joss S, Smith JC, Lippincott MF, Singh SS, Patel N, Jing JW, Law JR, Ferraro N, Verloes A, Rauch A, Steindl K, Zweier M, Scheer I, Sato D, Okamoto N, Jacobsen C, Tryggstad J, Chernauek S, Schimmenti LA, Brasseur B, Cesaretti C, Garcia-Ortiz JE, Buitrago TP, Silva OP, Hoffman JD, Mühlbauer W, Ruprecht KW, Loeyls 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 Champfleure 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](#).

Led copy number variation analyses to quantify neurodevelopmental disorder liability conferred by alterations to the netrin gene family

§ **Featured on the cover of [The American Journal of Medical Genetics](#)**

- 2016 [13] Ordulu Z, Kammin T, Brand H, Pillalamarri V, Redin CE, **Collins RL**, Blumenthal I, Hanscom C, Pereira S, Bradley I, Crandall BF, Gerrol P, Hayden MA, Hussain N, Kanengisser-Pines B, Kantarci S, Levy B, Macera MJ, Quintero-Rivera F, Spiegel E, Stevens B, Ulm JE, Warburton D, Wilkins-Haug LE, Yachevich N, Gusella JF, Talkowski ME, Morton CC. Structural chromosome rearrangements require nucleotide level resolution: lessons from next-generation sequencing in prenatal diagnosis. *American Journal of Human Genetics* (2016), 99(5): 1015-1033. PMID: [27745839](#). DOI: [10.1016/j.ajhg.2016.08.022](#).

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

- [12] Schilit S, Currall B, Yao R, Hanscom C, **Collins RL**, Pillalamarri V, Lee DYD, Kammin T, Zepeda-Mendoza CJ, Mononen T, Nolan LS, Gusella JF, Shen J, Talkowski M, Morton CC. Estrogen-related receptor gamma (*ESRRG*) implicated in a case of syndromic hearing loss and mild developmental delay. *European Journal of Human Genetics* (2016), 24(11): 1622-1626. PMID: [27381092](#). DOI: [10.1038/ejhg.2016.64](#).

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

- [11] Macakova M, Bohuslavova B, Vochozkova P, Pavlok A, Sedlackova M, Vidinska D, Vochoyanova K, Liskova I, Valekova I, Baxa M, Ellederova Z, Klima J, Juhas S, Juhasova J, Klouckova J, Haluzik M, Klempir J, Hansikova H, Spacilova J, **Collins RL**, Blumenthal I, Talkowski M, Gusella JF, Howland DS, DiFiglia M, Motlik J. Mutated Huntingtin causes testicular pathology in transgenic minipig boars. *Neurodegenerative Diseases* (2016), 16(3-4): 245-259. PMID: [26959244](#). DOI: [10.1159/000443665](#).

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, Haganir 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/amiainjnl-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)
- [6] Cummings BB, Karczewski KJ, Kosmicki JA, Seaby EG, Watts NA, Singer-Berk M, Mudge JM, Karjalainen J, Satterstrom KF, O'Donnell-Luria A, Poterba T, Seed C, Solomonson M, Alfoldi J, **The gnomAD Production Team**, The gnomAD Consortium, Daly MJ, MacArthur DG. Transcript expression-aware annotation improves rare variant discovery and interpretation. *Nature* (2020), 581: 452-457. PMID: [32461655](#). DOI: [10.1038/s41586-020-2329-2](#).
Contributed to the production of the Genome Aggregation Database (gnomAD)
- [5] Whiffin N*, Armean IM*, Kleinman A*, Marshall JL, Minikel EV, Karczewski KJ, Cummings BB, Francioli L, Laricchia K, Wang Q, Guan A, Alipanahi B, Morrison P, Baptista MAS, Merchant KM, **The gnomAD Production Team**, The gnomAD Consortium, Ware JS, Havulinna AS, Iliadou B, Lee J, Nadkarni GN, Whiteman C, The 23andMe Research Team, Daly M, Esko T, Hultman C, Loos RJF, Milani L, Palotie A, Pato C, Pato M, Saleheen D, Sullivan PF, Alfoldi J, Cannon P°, MacArthur DG°. The effect of *LRRK2* loss-of-function variants in humans. *Nature Medicine* (2020). PMID: [32461697](#). DOI: [10.1038/s41591-020-0893-5](#).
Contributed to the production of the Genome Aggregation Database (gnomAD)
- [4] Whiffin N, Karczewski KJ, Zhang X, Chothani S, Smith MJ, Evans DG, Roberts AM, Quaipe NM, Schafer S, Rackham O, Alfoldi J, O'Donnell-Luria AH, Francioli LC, **The gnomAD Production Team**, The gnomAD Consortium, Cook SA, Barton PJR, MacArthur DG*, Ware JS*. Characterising the loss-of-function impact of 5' untranslated region variants in whole genome sequence data from 15,708 individuals. *Nature Communications* (2020), 11(2523): 1-12. PMID: [32461616](#). DOI: [10.1038/s41467-019-10717-9](#).
Contributed to the production of the Genome Aggregation Database (gnomAD)
- [3] Wang Q, Pierce-Hoffman E, Cummings BB, Karczewski KJ, Alfoldi J, Francioli LC, Gauthier LD, Hill AJ, O'Donnell-Luria AH, **The gnomAD Production Team**, The gnomAD Consortium, MacArthur DG. Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. *Nature Communications* (2020), 11(2523): 1-13. PMID: [32461613](#). DOI: [10.1038/s41467-019-12438-5](#).
Contributed to the production of the Genome Aggregation Database (gnomAD)
- [2] Shanta O, Noor A, **The HGSV Consortium**, Sebat J. The effects of common structural variants on 3D chromatin structure. *BMC Genomics* (2020), 21(1):95. PMID: [32000688](#). DOI: [10.1186/s12864-020-6516-1](#).
Contributed to the creation of the HGSV structural variant callset
- 2019 ! [1] **Cross-Disorder Group of the Psychiatric Genomics Consortium**. Genome wide meta-analysis identifies genomic relationships, novel loci, and pleiotropic mechanisms across eight psychiatric disorders. *Cell* (2019), 179(7):1469-1482.e11. PMID: [31835028](#). DOI: [10.1016/j.cell.2019.11.020](#).
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 [5] **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, Samochoa 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
- 2020 [4] 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

- [3] 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. *bioRxiv* (2020). DOI: [10.1101/2020.07.03.168831](https://doi.org/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

- [2] 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^o, Engreitz JM^o. Genome-wide maps of enhancer regulation connect risk variants to disease genes. *bioRxiv* (2020). DOI: [10.1101/2020.09.01.278093](https://doi.org/10.1101/2020.09.01.278093).

Contributed to development of enhancer prediction model (ABC-Max) in 16 immune cell types

- 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](https://doi.org/10.1101/049536).

Developed all methods; wrote manuscript & created all figures

OTHER SCIENTIFIC WORKS

PUBLISHED & INDEXED CONFERENCE ABSTRACTS

- 2019 [4] Guo L, Toomer K, Fulmer D, Moore K, Yu M, 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, Naji NB, Slauchhaupt S, Norris RA. Mitral valve prolapse is associated with dysregulated β -catenin activities. *Structural Heart* (2019), 3(sup 1):184.

- 2017 [3] Shaw ND*, Brand H*, Kupchinsky ZA, Bengani H, Plummer L, Jones TI, Erdin S, Williamson KA, Rainger J, Stortchevoi A, Samocho K, Currall BB, Dunican DS, **Collins RL**, Willer JR, Lek A, Lek M, Nassan M, Pereira S, Kammin T, Lucente D, Silva A, Seabra CM, Chiang C, An Y, Ansari M, Rainger JK, Joss S, Smith JC, Lippincott MF, Singh SS, Patel N, Jing JW, Law JR, Ferraro N, Verloes A, Rauch A, Steindl K, Zweier M, Scheer I, Sato D, Okamoto N, Jacobsen C, Tryggstad J, Chernauek S, Schimmenti LA, Brasseur B, Cesaretti C, García-Ortiz JE, Buitrago TP, Silva OP, Hoffman JD, Mühlbauer W, Ruprecht KW, Loeyls 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^o, FitzPatrick DR^o, Talkowski ME^o. Identification of *SMCHD1* mutations in a severe form of Kallman Syndrome (KS) with absence of the nose (rhinia) attests to the power of extreme phenotypes in human reproductive gene discovery. *Hormone Res.* (2017), 88:5-7.

- 2016 [2] Redin C, Brand H, **Collins RL**, Hanscom C, Pillalamarri V, Kammin T, Mitchell E, Hodge JC, Schilit S, Curall BB, Pereira S, Seabra C, Stone M, Lawless W, Lucente D, Antolik C, Hochstenbach R, Renkens I, Brilstra E, Vergult S, Menten B, Janssens S, Callewaert B, D'heedene A, D'hooghe M, Roelens F, van de Kamp J, Nieuwint A, Poddighe PJ, van Ravenswaaij-Arts C, Rump P, vanEssen T, Freixo J, Dezso D, Liao EC, de Leew N, Brunner HG, Kloosterman W, Thorland EC, Morton CC, Gusella JF, Talkowski ME. Genomic landscape of balanced cytogenetic abnormalities in subjects with multiple congenital anomalies. *Eur. J. Hum. Genet.* (2016), 24(S1): 4. PL2.1.

- [1] Church DM, Jabara C, Kyriazopoulou-Panagiotopoulou S, **Collins RL**, Xu AW, Giorda K, Ordonez H, Heaton H, Pratt M, Marks P, Hardenbol P, Fehr A, Talkowski ME, Schnall-Levin M. Molecular cytogenetics using linked-reads. *Cancer Genetics* (2016), 209(6): 297. DOI: [10.1016/j.cancergen.2016.04.044](https://doi.org/10.1016/j.cancergen.2016.04.044).

POPULAR MEDIA & SCIENTIFIC COMMUNICATION

- 2019 [4] **Collins RL**, Brand H, Talkowski ME, MacArthur DG. Structural variants in gnomAD. *MacArthur Lab Blog* (March 2019). [Link](#).

- 2017 [3] **Collins RL**. Strength in numbers: genetic sequencing of large populations is shaping the future of medicine. *Science in the News* (June 2017). [Link](#).

- [2] **Collins RL**. Trainee leader interview: Beryl Cummings. *ASHG Nascent Transcript* (February 2017). [Link](#).

- 2016 [1] **Collins RL**. Separation anxiety: how balanced structural variation can sunder TADs and drive disease pathogenesis. *Structure Meets Function* (November 2016). [Link](#).

SCIENTIFIC ARTWORK & GRAPHICAL RESOURCES

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Creator & maintainer of database of vector art files for all 24 primary human chromosomes. Released under the open CC BY 4.0 license.

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Personal Website <http://www.ryanlcollins.com>

Google Scholar <https://scholar.google.com/citations?hl=en&user=9j9I-A8AAAAJ>

ResearchGate https://www.researchgate.net/profile/Ryan_Collins3

Mendeley <https://www.mendeley.com/profiles/ryan-collins7/>
LinkedIn <https://www.linkedin.com/pub/ryan-collins/64/2b9/27>
SCOPUS <https://www.scopus.com/authid/detail.uri?authorId=56504486900>
ORCID <http://orcid.org/0000-0003-1268-9995>
GitHub <https://github.com/RCollins13>
Twitter <https://twitter.com/ryancollins13>
Loop <http://loop.frontiersin.org/people/308215/overview>