CHELSEA LOWTHER

CURRICULUM VITAE

The Institute for Genomic Health, Icahn School of Medicine at Mount Sinai
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CURRENT APPOINTMENT

2024-Present Assistant Professor
The Institute for Genomic Health
Department of Genetics and Genomic Sciences
Icahn School of Medicine at Mount Sinai

EDUCATION AND TRAINING

2018-2024 Postdoctoral Research Fellowship
Center for Genomic Medicine, Massachusetts General Hospital
Department of Neurology, Harvard Medical School
Program in Medical and Population Genetics, Broad Institute of MIT and Harvard
Advised by Michael E. Talkowski

2012-2017 Ph.D., Medical Science
University of Toronto, Toronto, Canada
Advised by Anne S. Bassett

2007-2012 B.Sc., Psychology *(graduated first-class standing)*
University of Prince Edward Island, Prince Edward Island, Canada

AWARDS AND HONORS

2020 Finalist, Charles J. Epstein Trainee Award for Excellence in Human Genetics Research
2019 Winner, Charles J. Epstein Trainee Award for Excellence in Human Genetics Research
2019 Travel Award, Center for Genomic Medicine, Massachusetts General Hospital
2019 Finalist, European Society of Human Genetics Young Investigator Award
2018 Travel Award, Advances in Genome Biology and Technology Precision Health
2017 Finalist, Dr. Margaret Thompson Memorial Award
2015, 2016 Training Award, University of Toronto McLaughlin Centre
2015, 2017 Reviewer's Choice Abstract, American Society of Human Genetics Annual Meeting
2014, 2016 Travel Award, Canadian Institutes of Health Research
2013, 2014 Open Fellowship, University of Toronto
2012, 2013 Conference Grant, University of Toronto School of Graduate Studies
2011 Robert Haines Memorial Science Award of Merit, University of Prince Edward Island
2012 Convocation Award, Psychological Association of Prince Edward Island

SOCIETY MEMBERSHIP

2020-Present American College of Medical Genetics and Genomics (ACMG)
2016-Present European Society of Human Genetics (ESHG)
2013-Present American Society of Human Genetics (ASHG)

PREFESSIONAL SERVICE

2026-Present Member, Program Committee, American Society of Human Genetics
2025-Present Lead, SV Working Group, Mount Sinai Million Health Discoveries Program
2022-Present Member, ACMG/AMP/CAP/ClinGen CNV Technical Standards Working Group
2020-2022 Member, ACMG Cytogenetics and Molecular Lab Quality Assurance Subcommittee

RESEARCH SUPPORT

Current

NICHD R00HD108392 PI 2022-2027
Disruption of three-dimensional genome organization as a noncoding mechanism of disease in human developmental disorders
Total Costs: $1,019,028 USD

Completed

Research Fellowship Award PI 2021-2022
Fund for Medical Discovery (FMD), Massachusetts General Hospital Executive Committee on Research
Genome-wide identification of structural variants across population and disease cohorts
Total Costs: $81,216 USD

Postdoctoral Fellowship PI 2018-2021
Canadian Institutes of Health Research
Characterizing the genome-wide burden of rare variants in developmental disorders
Total Costs: $150,000 CAD

Accelerator Grant Co-I (PI: Bassett) 2018-2019
University of Toronto McLaughlin Centre
Optimized whole genome sequencing analysis of complex disease in a 22q11.2 deletion model
Total Costs: $50,000 CAD

Accelerator Grant Co-I (PI: Bassett) 2016-2017
University of Toronto McLaughlin Centre
Whole genome sequencing to uncover the genetic architecture of familial schizophrenia
Total Costs: $70,000 CAD

Doctoral Award PI 2015-2018
Canadian Institutes of Health Research Frederick Banting and Charles Best Canada Graduate Scholarship
Elucidating the genetic architecture of comorbid schizophrenia and intellectual disability
Total Costs: $105,000 CAD

MENTORSHIP AND TEACHING

Certificate programs

2026 Mentorship for Mutual Success: Strategies for Mentoring Excellence in Science, ISMMS
2024 Compass Scholar, Washington University School of Medicine, St. Louis, MO

Primary supervision

2025-Present Nandini Samanta (PhD student at ISMMS, Genetics and Genomic Sciences)
2025-Present Claire Coleman (PhD student at ISMMS, Genetics and Genomic Sciences)
2025-Present Saleh Sereshki (Postdoctoral Research Fellow)

Thesis advisory committee

2025-Present David D'Onofrio (PhD student at ISMMS, AI and Emerging Technologies)
2025-Present Ashley Richardson (PhD student at ISMMS, Immunology)

Secondary mentorship

2025 Nayaab Punjani (PhD student at the University of Toronto) as part of the Institute for Medical Science Career Mentorship Program
2020-2021 Selin Everett (4th year undergraduate student, Middlebury College); now a medical student at Stanford University
2018-2024 Elise Valkanas (PhD student, Harvard University); now Senior Manager at Serepta Therapeutics
2015-2016 Phil Fradkin (4th year undergraduate student, University of Toronto); now a PhD student in Computer Science at University of Toronto

Teaching

2025 Invited Lecture, 7101 Foundations IV: Biological Basis of Behavior, University of PEI
2013 Lecturer, Neuroscience Enrichment Program: A Lecture Series for High School Students, University of Toronto Collaborative Program in Neuroscience
2012 Teaching Assistant, Psychology 102: Introduction to Psychology, University of PEI
2009-2012 Teaching Assistant, Psychology 278 & 279: Statistics and Research Design I/II, University of PEI

ACADEMIC SERVICE

2026 Application screener, PhD Program in Biomedical Sciences, ISMMS

2025, 2026 Interviewer, PhD Program in Biomedical Sciences, ISMMS

2025 Panelist, *Pathways to Success in a Career Related to Human Genetics and Genomics*, Gordon Research Seminar in Human Genetics and Genomics, Portland, Maine

2023 Discussion Leader, *Defining the Landscape of Genomic Variation and Associated Phenotypes*, Gordon Research Seminar in Human Genetics and Genomics, Waterville, New Hampshire

2020, 2021 Invited grant reviewer, Canadian Institutes of Health Research Doctoral Research Awards

2018-Present Invited journal reviewer: *Nature*, *Biological Psychiatry*; *Psychiatric Genetics; npj Genomic Medicine, Prenatal Diagnosis, Translational Psychiatry, Genetics in Medicine*

2016 Invited abstract reviewer, University of Toronto Medical Student Research Day

2015 Poster judge, Summer Undergraduate Research Program (SURP), Institute of Medical Science, University of Toronto

2014, 2015 DNA Day Essay Judge, American Society of Human Genetics

2013-2015 Journalist, Institute of Medical Science Magazine, University of Toronto, Toronto, Ontario, Canada.

2013-2014 Alumni Lead, IMS Strategic Planning Initiative, University of Toronto

2012-2015 Member, University of Toronto Social Sciences, Humanities, and Education Research Ethics Board

2012-2013 Graduate Student Union Representative, IMS Student Association, University of Toronto

2010-2011 Student Representative, Department of Psychology, University of PEI

2010-2011 Treasurer, Psychology Arts & Sciences Society, University of PEI

INVITED TALKS AND ORAL PRESENTATIONS

2026 Digital Health Partnership Conference, ISMMS, New York City, NY
2023 Rising Stars in Genetics and Genomics Symposium, University of Utah
2022 European Society of Human Genetics, Vienna, Austria
2022 Biology of Genomes, Cold Spring Harbor Laboratory, Long Island, NY
2021 13th Annual European Cytogenetics Conference, virtual during COVID-19 pandemic
2021 Program in Medical and Population Genetics, Broad Institute of MIT and Harvard
2020 American Society of Human Genetics, virtual during the COVID-19 pandemic
2019 American Society of Human Genetics, Houston, TX
2018 European Society of Human Genetics, Gothenburg, Sweden
2016 Harvey Stancer Research Day, University of Toronto Department of Psychiatry
2015 Canadian College of Medical Geneticists Annual Conference, Ottawa, ON
2013 Canadian College of Medical Geneticists Annual Conference, Toronto, ON
2014 Humber River Regional Hospital Ground Rounds, Toronto, ON
2014 Brown Bag Lunch Ethics Series, Center for Addiction and Mental Health, Toronto, ON

PUBLICATIONS

Google scholar metrics: # of papers = 26 | h-index = 19 | i10-index = 23 | # of citations = 3,144

1. Gudmundsson S, Singer-Berk M, Stenton SL, Goodrich JK, Wilson MW, Einson J, et al. Exploring penetrance of clinically relevant variants in over 800,000 humans from the Genome Aggregation Database. Nat Commun. 2025 Oct 31;16(1):9623.

2. Epi25 Collaborative. Exome sequencing of 20,979 individuals with epilepsy reveals shared and distinct ultra-rare genetic risk across disorder subtypes. Nat Neurosci. 2024 Oct;27(10):1864–79.

3. Musgrave SM, Taylor J, Whitford W, Garton A, Poquérusse J, Hawkins V, et al. Genetic diagnostic outcomes from a 10-year research programme in autism in Aotearoa New Zealand. J R Soc N Z. 2024 Sept 18;1–17.

4. Liao C, Ye R, Ivankovic F, Fu JM, Walters R, Lowther C, et al. The landscape of gene loss and missense variation across the mammalian tree informs on gene essentiality [Internet]. bioRxiv. 2024. Available from: https://www.biorxiv.org/content/10.1101/2024.05.16.594531v1

5. Lowther C, Valkanas E, Giordano JL, Wang HZ, Currall BB, O'Keefe K, et al. Systematic evaluation of genome sequencing for the diagnostic assessment of autism spectrum disorder and fetal structural anomalies. Am J Hum Genet. 2023 Sept 7;110(9):1454–69.

6. Raca G, Astbury C, Behlmann A, De Castro MJ, Hickey SE, Karaca E, et al. Points to consider in the detection of germline structural variants using next-generation sequencing: A statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023;25(2):100316.

7. Mohajeri K, Yadav R, D'haene E, Boone PM, Erdin S, Gao D, et al. Transcriptional and functional consequences of alterations to MEF2C and its topological organization in neuronal models. Am J Hum Genet. 2022 Nov 3;109(11):2049–67.

8. Collins RL, Glessner JT, Porcu E, Lepamets M, Brandon R, Lauricella C, et al. A cross-disorder dosage sensitivity map of the human genome. Cell. 2022 Aug 4;185(16):3041-3055.e25.

9. Lowther C, Mehrjouy MM, Collins RL, Bak MC, Dudchenko O, Brand H, et al. Balanced chromosomal rearrangements offer insights into coding and noncoding genomic features associated with developmental disorders [Internet]. bioRxiv. 2022. Available from: http://medrxiv.org/lookup/doi/10.1101/2022.02.15.22270795

10. Cleynen I, Engchuan W, Hestand MS, Heung T, Holleman AM, Johnston HR, et al. Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Mol Psychiatry. 2021 Aug;26(8):4496–510.

11. Zhao X, Collins RL, Lee W-P, Weber AM, Jun Y, Zhu Q, et al. Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. Am J Hum Genet. 2021 May 6;108(5):919–28.

12. Collins RL, Brand H, Karczewski KJ, Zhao X, Alfoldi J, Francioli LC, et al. A structural variation reference for medical and population genetics. Nature. 2020;581(7809):444–51.

13. Reuter MS, Jobling R, Chaturvedi RR, Manshaei R, Costain G, Heung T, et al. Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot. Genet Med. 2019 Apr;21(4):1001–7.

14. Halgren C, Nielsen NM, Nazaryan-Petersen L, Silahtaroglu A, Collins RL, Lowther C, et al. Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. Am J Hum Genet. 2018;102(6):1090–103.

15. Lowther C, Merico D, Costain G, Waserman J, Boyd K, Noor A, et al. Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Med. 2017 Nov 30;9(1):105.

16. Lowther C, Costain G, Baribeau DA, Bassett AS. Genomic disorders in psychiatry-what does the clinician need to know? Curr Psychiatry Rep. 2017 Sept 20;19(11):82.

17. Bassett AS, Lowther C, Merico D, Costain G, Chow EWC, van Amelsvoort T, et al. Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. Am J Psychiatry. 2017 Nov 1;174(11):1054–63.

18. Mak CCY, Chow PC, Liu APY, Chan KYK, Chu YWY, Mok GTK, et al. De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. NPJ Genom Med. 2016 Sept 14;1:16033.

19. Loviglio MN, Leleu M, Männik K, Passeggeri M, Giannuzzi G, van der Werf I, et al. Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. Mol Psychiatry. 2017 June;22(6):836–49.

20. Lowther C, Speevak M, Armour CM, Goh ES, Graham GE, Li C, et al. Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression. Genet Med. 2017;19(1):53–61.

21. Isles AR, Ingason A, Lowther C, Walters J, Gawlick M, Stöber G, et al. Parental Origin of Interstitial Duplications at 15q11.2-q13.3 in Schizophrenia and Neurodevelopmental Disorders. PLoS Genet. 2016;12(5):e1005993.

22. Lowther C, Costain G, Bassett AS. Reproductive genetic testing and human genetic variation in the era of genomic medicine. Am J Bioeth. 2015;15(6):25–6.

23. Hashemi B, Bassett A, Chitayat D, Chong K, Feldman M, Flanagan J, et al. Deletion of 15q11.2(BP1-BP2) region: further evidence for lack of phenotypic specificity in a pediatric population. Am J Med Genet A. 2015;167A(9):2098–102.

24. Lowther C, Costain G, Stavropoulos DJ, Melvin R, Silversides CK, Andrade DM, et al. Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genet Med. 2015;17(2):149–57.

25. Lowther C, Costain G, Melvin R, Stavropoulos DJ, Lionel AC, Marshall CR, et al. Adult expression of a 3q13.31 microdeletion. Mol Cytogenet. 2014 Mar 20;7(1):23.

26. Costain G, Lionel AC, Merico D, Forsythe P, Russell K, Lowther C, et al. Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Hum Mol Genet. 2013 Nov 15;22(22):4485–501.

BOOK CHAPTERS

1. Lowther C, Boot E, Boyd K, Bassett AS (2015). Chapter 7: 22q11.2 Deletion Syndrome in S.L. Watson & D. Griffiths (2nd Edition.), New York. NADD Press.

MEDIA

August 2016 Researchers identify gene variations specifically associated with intellectual disability
Article written by the Brain and Behavior Research Foundation on the Lowther C et al. *Genet Med*. 2017 publication

August 2014 Large genetic deletion leads to autism, but not always
Article written by Spectrum on the Lowther C et al. *Genet Med*. 2015 publication