Phillip Andrew Richmond

BC Children's Hospital Research Institute
#3109, 950 West 28th Avenue
Vancouver, BC V5Z 4H4
Canada

Email (work): prichmond@bcchr.ca
Email (personal): phillip.a.richmond@gmail.com
GitHub: GitHub Profile
Twitter: @Phil_A_Richmond
LinkedIn: LinkedIn Profile
YouTube: YouTube Channel
URL: Personal Website

Current Position

Staff Scientist
Precision Health Initiative
BC Children's Hospital Research Institute
Provincial Health Services Authority

Areas of Specialization

Bioinformatics • Genomics • Rare Genetic Diseases • Molecular Biology • Gene Regulation

Appointments Held

2020-Present *Staff Scientist*, Precision Health Initiative, BC Children's Hospital Research Institute, Provincial Health Services Authority, Vancouver, BC.

2015-2020 *PhD Graduate Student*, Medical Genetics Lab of Dr Wyeth Wasserman, Centre for Molecular Medicine and Therapeutics, BC Children's Hospital Research Institute, University of British Columbia

2012/6-2015/7 *Professional Research Assistant*, Genomics Lab of Dr Robin Dowell, BioFrontier's Institute, University of Colorado–Boulder

2010/5-2012/5 *Undergraduate Research Assistant*, Genomics Lab of Dr Robin Dowell, University of Colorado–Boulder

2009/3-2009/8 *Undergraduate Research Assistant*, Behavioral Genetics Lab of Dr Chris Downing, University of Colorado–Boulder

Affiliations & Memberships

2017/8-Present Member; Global Organisation for Bioinformatics Learning, Education & Training (GOBLET)

2017/6-Present Member; European Society for Human Genetics (ESHG)

2017/5-Present Committee Member; BC Children's Hospital Research Institute Trainee Council

2017/1-Present Student Representative; NSERC CREATE Program Committee, University of British Columbia
2016/9-Present Committee Member & Teacher; Education, Outreach and Training (EOT) Compute Canada, WestGrid
2016/8-Present Member; American Society for Human Genetics (ASHG)
2016/5-Present Development Team Member; Vancouver Bioinformatics User Group (VanBUG)
2014-2015 Member; American Society for Brewing Chemists (ASBC)
2011-2013 Member; American Society for Cellular Biology (ASCB)

Education

2015-2020 PhD in Bioinformatics, University of British Columbia.
Supervised by Dr. Wyeth W. Wasserman (https://wassermanlab.github.io).
Expanding the utility of whole genome sequencing in the diagnosis of rare genetic disorders.
Available online at: https://open.library.ubc.ca/cIRcle/collections/ubctheses/24/items/1.0394775
2008-2012 B.A. in Molecular, Cellular, & Developmental Biology, University of Colorado–Boulder

Grants, Honors & Awards

2019 Advanced Research Computing Resource Allocation Competition, University of British Columbia
2018 Resources for Research Groups, Compute Canada
2017-2019 BC Children's Hospital Research Institute Graduate Studentship
2017 Resources for Research Groups, Compute Canada
2015-2017 NSERC CREATE Trainee Scholarship, NSERC
2012 *Summa Cum Laude* in Molecular, Cellular, & Developmental Biology

Publications, Talks, & Poster Presentations

Journal articles (Working)

2020 **Phillip A. Richmond***, Alice M. Kaye*, Godfrain Jacques Kounkou, Tamar V. Av-Shalom, Wyeth W Wasserman. "Demonstrating the utility of flexible sequence queries against indexed short reads with FlexTyper." *Accepted at PLoS Computational Biology*, (2020). (online at https://doi.org/10.1101/2020.03.02.973750). *Co-first author.

Journal articles (Published)

2021 Bhavi P Modi, Kate L Del Bel, Susan Lin, Mehul Sharma, **Phillip A Richmond**, Clara DM van Karnebeek, Edmond S Chan, Vishal Avinashi, Wingfield E Rehmus, Catherine M Biggs, Wyeth W Wasserman, Stuart E Turvey. Exome sequencing enables diagnosis of X-linked hpohidrotic ectodermal dysplasia in patient with eosinophilic esophagitis and severe atopy. Allergy, Asthma & Clinical Immunology, (2021). 17: 1-6.

2020 **Phillip A. Richmond***, Tamar V. Av-Shalom*, Oriol Fornes, Bhavi Modi, Alison Elliott, Wyeth W. Wasserman. "GeneBreaker: Variant simulation to improve the diagnosis of Mendelian rare genetic

diseases." *Human Mutation*, (2020). (online at https://doi.org/10.1002/humu.24163). *Co-first author.

2020 Xin (Cynthia) Ye, Nicole M Roslin, Andrew D. Paterson, Christopher Lyons, Victor Pegado, **Phillip Andrew Richmond**, Casper Shyr, Oriol Fornes, Xiaohua Han, Michelle Higginson, Colin Ross, Deborah Giaschi, Cheryl Y Gregory-Evans, Millan Patel, Wyeth W Wasserman. "Linkage analysis identifies an isolated strabismus locus at 14q12 overlapping with FOXG1 syndrome region." *Journal of Medical Genetics*, (2020). (online at https://doi.org/10.1101/2020.04.24.20077586).

2020 Clara van Karnebeek*, **Phillip A. Richmond***, Frans van der Kloet, Wyeth W. Wasserman, Marc Engelen, Stephan Kemp. "The variability conundrum in neurometabolic degenerative diseases." *Molecular Genetics and Metabolism*, (2020). (online at https://doi.org/10.1016/j.ymgme.2020.11.002). *Co-first author.

2020 Radcliffe RA, Dowell R, Odell AT, **Richmond PA**, Bennett B, Larson C, Kechris K, Saba LM, Rudra P, Wen S. Systems genetics analysis of the LXS recombinant inbred mouse strains:Genetic and molecular insights into acute ethanol tolerance. *PLoS One.* Oct 23;15(10):e0240253 (2020). (online at doi:10.1371/journal.pone.0240253).

2020 **Phillip A Richmond***, Frans van der Kloet*, Frederic M. Vaz, Antoine H.C. van Kampen, Anuli Uzozie, Philipp F. Lange, David Lin, Michael Kobor, Emma Graham, Sara Mostafavi, Perry Moerland, Wyeth W Wasserman***, Marc Engelen***, Stephan Kemp***, Clara van Karnebeek***. "Multi-omic approach to identify phenotypic modifiers undelrying cerebral demyelination in X-linked adrenoleukodystrophy." *Frontiers in Cellular and Developmental Biology*, 8, 520. (2020). (online at https://doi.org/10.1101/2020.03.19.20035063). *Co-first author, ***Co-last author.

2020 Emma J Graham, **Phillip Andrew Richmond**, Maja Tarailo-Graovac, Udo Engelke, Leo AJ Kluijtmans, Karlien LM Coene, Ron A Wevers, Wyeth W Wasserman, Clara DM van Karnebeek, Sara Mostafavi. "metPropagate: network-guided propagation of metabolomic information for prioritization of neurometabolic disease genes." *Genomic Medicine* 5, 25 (2020). (online at https://doi.org/10.1101/2020.01.12.20016691).

2020 Egor Dolzhenko*, Mark Benett*, **Phillip A Richmond***, Brett Trost, Sai Chen, Joke JFA van Vugt, Charlotte Nguyen, Giuseppe Narzisi, Vladimir G Gainullin, Andrew M Gross, Bryan R Lajoie, Ryan J Taft, Wyeth W Wasserman, Stephen W Shcerer, Jan H Veldink, David R Bentley, Ryan KC Yuen***, Melanie Bahlo***, Michael A Eberle***. "ExpansionHunter Denovo: A computational method for locating known and novel repeat expansions in short-read sequencing data", *Genome Biology*, 21, 102 (2020). (online at https://doi.org/0.1186/s13059-020-02017-z). *Co-first author, ***Co-last author.

2020 Oriol Fornes, Jaime A Castro-Mondragon, Aziz Khan, Robin van der Lee, Xi Zhang, **Phillip A Richmond**, Bhavi P Modi, Solenne Correard, Marius Gheorghe, Damir Baranašić, Walter Santana-Garcia, Ge Tan, Jeanne Chèneby, Benoit Ballester, François Parcy, Albin Sandelin, Boris Lenhard, Wyeth W Wasserman, Anthony Mathelier. "JASPAR 2020: update of the open-access database of transcription factor binding profiles", *Nucleic Acids Research* 42 (D1) 87-92 (2020). (online at https://doi.org/10.1093/nar/gkz1001).

2019 **Phillip A Richmond**, Wyeth Wasserman. "Introduction to Genomic Analysis Workshop: A catalyst for engaging life-science researchers in high throughput analysis", *F1000 Research*, (2019). (online at https://doi.org/10.12688/f1000research.19320.1).

2019 Andre BP van Kuilenburg*, Maja Tarailo-Graovac*, **Phillip A Richmond***, Britt I Drogemoller, Mahmoud A Pouladi, Rene Leen, Koroboshka Brand-Arzamendi, Doreen Dobritzsch, Egor Dolzhenko, Michael A Eberle, Bruce Hayward, Meaghan J Jones, Farhad Karbassi, Michael S Kobor, Janet Koster, Daman Kumari, Meng Li, Julia MacIsaac, Cassandra McDonald, Judith Meijer, Charlotte Nguyen, Indhu-Shree Rajan-Babu, Stephen W Scherer, Bernice Sim, Brett Trost, Laura A Tseng, Marjolein Turkenburg, Joke JFA van Vugt, Jan H Veldink, Jagdeep S Walia, Youdong Wang, Michel van Weeghel, Galen EB Wright, Xiaohong Xu, Ryan KC Yuen, Jinqiu Zhang, Colin J Ross, Wyeth W Wasserman, Michael T Geraghty, Saikat Santra, Ronald JA Wanders, Xiao-Yan Wen, Hans R Waterham, Karen Usdin, Clara DM van Karnebeek***. (2019), "Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in GLS", *New England Journal of Medicine* 380(15) 1433-1441 (2020). (online at https://doi.org/10.1056/NEJMoa1806627). *Co-first author, ***Co-last author.

2019 Timothy H. Webster, Madeline Couse, Bruno M. Grande, Eric Karlins, Tanya Phung, **Phillip Richmond**, Whitney Whitford, Melissa A. Wilson Sayres. "XYalign: Inferring sex chromosome content and correcting for technical biases in next-generation sequencing data", *GigaScience* 8 (7) giz074 (2019). (online at https://doi.org/10.1093/gigascience/giz074).

2018 Oriol Fornes, Marius Gheorghe, **Phillip A Richmond**, David Arenillas, Wyeth Wasserman and Anthony Mathelier. "MANTA2, update of the Mongo database for the analysis of transcription factor binding site alterations", *Scientific Data*, (2018). (online at https://doi.org/10.1038/sdata.2018.141).

2017 Gilson Sanchez, **Phillip Richmond**, Eric Bunker, Joseph Azofeifa, Aaron Garnett, Qinghong Zhang, Robin Dowell, and Xuedong Liu. (2017), "Dose-dependent Inhibition of Histone Deacetylases Reprograms Gene Expression Through Global Remodeling of the Enhancer Landscape", *Nucleic Acids Research*, 2017. (online at https://doi.org/10.1093/nar/gkx1225).

2017 Amber L Scott, **Phillip A Richmond**, Robin D Dowell, Anna M Selmecki. (2017), "The influence of polyploidy on the evolution of yeast grown in a sub-optimal carbon source", *Molecular Biology and Evolution* 34(10) 2690–2703, (2017). (online at https://doi.org/10.1093/molbev/msx205).

2016 Bennett B, Larson C, **Richmond PA**, Odell AT, Saba LM, Tabakoff B, Dowell R, Radcliffe RA. (2016), "Quantitative trait locus mapping of acute functional tolerance in the LXS recombinant inbred strains", *Alcoholism: Clinical and Experimental Research* 39(4): 611-620, (2016). (online at https://doi.org/10.1111/acer.12678).

2016 Kamens HM, Corley RP, **Richmond PA**, Darlington TM, Dowell R, Hopfer CJ, Stallings MC, Hewitt JK, Brown SA, Ehringer MA. "Evidence for Association Between Low Frequency Variants in CHRNA6/ CHRNB3 and Antisocial Drug Dependence", *Behavior Genetics* 46(5): 693-704 (2016). (online at https://doi.org/10.1007/s10519-016-9792-4).

2016 Timothy J Read, **Phillip A Richmond**, Robin D Dowell. (2016), "A trans-acting variant within the transcription factor RIM101 interacts with genetic background to determine its regulatory capacity", *PLoS Genetics* 12(1): e1005746, (2016). (online at https://doi.org/10.1371/journal.pgen.1005746).

2016 Robin Dowell, Aaron Odell, **Phillip Richmond**, Daniel Malmer, Eitan Halper-Stromberg, Beth Bennett, Colin Larson, Sonia Leach, Richard A Radcliffe. "Genome Characterization of the Selected Long and Short Sleep Mouse Lines", *Mammalian Genome*: 27(11): 574-586, (2016). (online at https://doi.org/10.1007/s00335-016-9663-6).

2015 Emily K Pugach, **Phillip A Richmond** Joseph G Azofeifa, Robin D Dowell, Leslie A Leinwand. (2015), "Prolonged Cre expression driven by the alpha-myosin heavy chain promoter can be cardiotoxic", *Journal of Molecular and Cellular Cardiology* 86: 54-61 (2015). (online at https://doi.org/10.1016/j.yjmcc.2015.06.019).

2015 Anna M. Selmecki, Yosef E. Maruvka, **Phillip A. Richmond**, Marie Guillet, Noam Shoresh, Amber L. Sorenson, Subhajyoti De, Roy Kishony, Franziska Michor, Robin Dowell *&* David Pellman. "Polyploidy can drive rapid adaptation in yeast", *Nature* (519): 349-352, (2015). (online at https://doi.org/10.1038/nature14187).

Conference Publications

2016 RA Radcliffe, RD Dowell, A Odell, **P Richmond**, B Bennett, C Larson, K Kechris, P Rudra, WJ Shi. (2016) "Ethanol-specific effects on the genetic regulation of gene expression: potential relationship to acute ethanol sensitivity", *Alcoholism-clinical and experimental research*

2016 Daniel Malmer, **Phillip A Richmond**, Aaron Odell, Robin D Dowell. (2016), "Inferring Ancestry In Mouse Genomes Using A Hidden Markov Model", *The 5th ACM Conference*

2013 MA Ehringer, HM Kamens, RP Corley, M Simonson, A Poole, **P Richmond**, JA Stitzel, R Dowell, K Krauter, MB Mcqueen, MC Stallings, C Hopfer, T Crowley, JK Hewitt. (2013), "Behavioral Disinhibition: Sequencing Chrn Genes In A Selected Sample To Identify Novel Variants", *Alcoholism: Clinical & Experimental Research*

Poster Presentations

2019 Bioinformatics, Integrative Oncology, Genome Sciences *&* Technology Research Day (March 2019), "Short tandem repeats in undiagnosed rare genetic disease", Vancouver, BC.

2018 American Society of Human Genetics (October 2018), "Short tandem repeat expansions in undiagnosed rare genetic disease", Vancouver, BC.

2018 Bioinformatics, Integrative Oncology, Genome Sciences *&* Technology Research Day (March 2018), "Noncoding variant interpretation in rare genetic disease", Vancouver, BC.

2017 BC Children's Hospital Research Day (June 2017), "Clinical Grade CNV Calling For Rare Genetic Disorders", Vancouver, BC.

2017 Bioinformatics, Integrative Oncology, Genome Sciences *&* Technology Research Day (March 2017), "Clinical Grade CNV Calling", *2nd Place*, Vancouver, BC.

2014 Rocky Mountain Brewing Symposium (October 2014) "Leveraging Next Generation Sequencing in Brewing QC", Colorado Springs, Colorado.

2011 American Society for Cellular Biology Conference (December 2011), "The Genotypic Impact of Polyploidy on Directed Evolution", Denver, Colorado.

Talks

2020 "ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data." (2020), UBC ARC Summer School Session, Vancouver, Canada.

2020 "Multi-omic approach to identify markers of cerebral demyelination in X-linked adrenoleukodystrophy" (2020), Trainee Omics Group Seminar Series, BC Children's Hospital Research Institute, Vancouver, Canada.

2019 "Introduction to Short Read Mapping: The foundation of next generation sequencing analysis" (2019), GrasPods Seminar Series, BC Cancer Research Centre, Vancouver, Canada.

2018 ”Clinical Genomics: The Next Generation of Medicine for Rare Genetic Disorders” (2018), Vancouver Summer Program in Clinical Research and Medicine, Canada.

2018 ”A STRange inborn error of metabolism” (2018), VanBUG Student Presenter, BC Cancer Agency, Vancouver, Canada.

2018 ”Noncoding Variants in Genetic Disease” (2018), VanBUG Student Presenter, BC Cancer Agency, Vancouver, Canada.

2017 ”Clinical Genomics: The Next Generation of Medicine for Rare Genetic Disorders” (2017), Vancouver Summer Program in Medicine, BC Children’s Hospital, Vancouver, Canada.

2017 ”Clinical Grade CNV Calling from WGS Data” (2017), BC Children’s Hospital Research Institute TGIF Seminar, Vancouver, Canada.

2016 ”The Next Generation of the Fight Against Rare Genetic Disorders” (2016), BC Children’s Hospital Foundation, Vancouver, Canada.

2014 ”Leveraging Next Generation Sequencing in Brewing Quality Control” (2014), American Society for Brewing Chemists Annual Conference, Chicago, USA.

2013 ”Impact of Ploidy on Directed Evolution” (2013), MCDB Departmental talk, University of Colorado–Boulder, USA.

Leadership

2018-2020 Co-Chair and Co-Founder, Trainee Omics Group, BC Children’s Hospital Research Institute. http://bcchr.ca/tog

2017-2020 Chair, Trainee Council, BC Children’s Hospital Research Institute.

Mentorship

2018-2020 Supervisor of Co-op student, followed by summer research assistant Tamar Av’Shalom in the Wasserman Lab (2018-2020)

Teaching

High School

2019 GeneSkool Content Creation: Mini Medical Genetics Case Study for Genome BC, Vancouver, BC. (online at https://www.genomebc.ca/education-resource/rare-genetic-diseases)

2019 Richmond High School #38 Medical Genetics Case Study, Richmond, BC

2018 SHAD student workshop at BC Children’s Hospital, Vancouver, BC

2018 Career panel for Undergraduate Summer Research Program at BC Children’s Hospital Research institute, Vancouver, BC

2018 Mini Med School for BC Children’s Hospital Research Institute, Fort Saint John, BC

2018 City-School BC Children’s Hospital Research Institute Visit, BC Children’s Hospital

2017-Present Genome BC’s GeneSkool, Volunteer High School Teaching Program.

2016-Present Gairdiner Symposium, BC Children’s Hospital

2016-Present Research Open House, BC Children’s Hospital

Online Curriculum (Inverted Classroom Format)

2016 Bioinformatics Introductory Analysis Course.
URL: http://phillip-a-richmond.github.io/Bioinformatics-Introductory-Analysis-Course/

2014 Introduction to Python for Biologists,
URL: http://dowell.colorado.edu/education-python.html

Hybrid Online/In-person Curriculum

2020 *In-progress* UBC Master's in Genetic Counselling Training Program, "Genome Analysis Module": November 4th-18th, 2020.

2020 UBC Advanced Research Computing Summer School: "Introduction to Short Read Mapping: The foundation of next generation sequencing analysis": June 26th, 2019.
Attendees: 110
Sponsors: Compute Canada, WestGrid, Advanced Research Computing (UBC).
Slide Deck: Google Slides

2019 EOT Tutorials: Introduction to Short Read Mapping: April 3, 2019.
Attendees:21
Sponsors: Compute Canada, WestGrid, Advanced Research Computing (UBC).
Slide Deck: Google Slides
Recording: YouTube

2018 Advanced Research Computing Summer School: Introduction to Short Read Mapping: The foundation of next generation sequencing analysis, June 12, 2018.
Attendees: 25
Sponsors: University of British Columbia, Compute Canada, WestGrid, Advanced Research Computing.
slide deck: Google Slides
URL: https://westgrid.github.io/ubcSummerSchool2018/4-materials.html

2017 Introduction to Linux: Command Line Basics, September 23, 2017.
Attendees: 30
Sponsors: University of British Columbia, Compute Canada, WestGrid, Advanced Research Computing.
slide deck: Google Slides
recording: YouTube
URL: https://phillip-a-richmond.github.io/ComputeCanada_EOT/

2017 Introduction to Genomic Analysis Workshop Series, June 7-15, 2017.
Attendees: 91
Sponsors: University of British Columbia, BC Children's Hospital, Evidence2Innovation, Compute Canada, WestGrid, Advanced Research Computing.
URL: https://phillip-a-richmond.github.io/Introduction-to-Genomic-Analysis/

2016 Introduction to Next Generation Sequencing Analysis, November 24, 2016.
Attendees: 60
Sponsors: University of British Columbia, Compute Canada, WestGrid, Advanced Research Computing.
Slide Deck: Google Slides
Recording: YouTube

Invited Guest Lectures

2021

"Problem Based Learning in Bioinformatics" (2021). Designed and taught 2-session module on diagnosing rare genetic disorders using whole genome sequencing". Bioinformatics 520, University of British Columbia, Vancouver, Canada.

2021 "Medical Genetics 421: Bioinformatics in Cancer Genomics" (2021). Guest Lecture for Medical Genetics 421: Genetics and Cell Biology of Cancer, University of British Columbia, Vancouver, Canada.

2020 "Medical Genetics 421: Bioinformatics in Cancer Genomics" (2020). Guest Lecture for Medical Genetics 421: Genetics and Cell Biology of Cancer, University of British Columbia, Vancouver, Canada.

2019 "Problem Based Learning in Bioinformatics" (2019). Designed and taught 2-session module on diagnosing rare genetic disorders using whole genome sequencing". Bioinformatics 520, University of British Columbia, Vancouver, Canada.

2019 "Medical Genetics 421: Bioinformatics in Cancer Genomics" (2019). Guest Lecture for Medical Genetics 421: Genetics and Cell Biology of Cancer, University of British Columbia, Vancouver, Canada.

2018 "Medical Genetics 421: Bioinformatics in Cancer Genomics" (2018). Guest Lecture for Medical Genetics 421: Genetics and Cell Biology of Cancer, University of British Columbia, Vancouver, Canada.

2017 "Medical Genetics 421: NGS Bioinformatics" (2017). Guest Lecture for Medical Genetics 421: Genetics and Cell Biology of Cancer, University of British Columbia, Vancouver, Canada.

Miscellaneous

International Collaborations

2017-Present xALD twin cohort collaboration with researchers at Amsterdam University Medical Centre.

Peer Review

2018 Nucleic Acids Research: "Performance evaluation of pathogenicity-computation methods for missense variants" (Assisted Review)

Programming Languages

Proficient: Python, Bash, Perl, R, sed
Novice: C++, Java, MySQL

Bioinformatics Software Experience

DNA-sequencing: Bowtie, Bowtie2, BWA, BWAmem, Samtools, HTSLib, GATK, FreeBayes, Platypus, Picard, CNVnator, LUMPY, Pindel, Breakdancer, CANVAS, ERDS, MetaSV, GangSTR, ExpansionHunter, ExpansionHunter Denovo, STRetch, TREDPARSE, LobSTR, GEMINI, VCFAnno, VCF2DB, AnnotSV, ANNOVAR, SNPeff, VEP, RUFUS, VarSim, ART, wgsim, Peddy, Intervene

RNA-sequencing: Tophat, Tophat2, Stringtie, Cufflinks, HTSeq, DESeq, DESeq2, EdgeR, DEXSeq, HISAT2, GSNAP

ChIP-sequencing: MACS2, HOMER

Generic Bioinformatics: BedTools, InterVene

Software Management: Conda, Docker, Brew, GitHub, Singularity

Linux Schedulers: SLURM, Torque/Moab, SGE, PBS Pro