

Konrad J Karczewski, Ph.D.

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Analytic and Translational Genetics Unit
Boston, MA 02111

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Educational Background:

- 2009 - 2013 Stanford University (Stanford, CA)
 Degree(s): M.S. (2011), Ph.D. (2013) in Biomedical Informatics
 Advisor: Michael Snyder (Department of Genetics)
 Co-advisor: Stephen Montgomery
- 2005 - 2009 Princeton University (Princeton, NJ)
 Degree(s): B.A. in Molecular Biology, Certificate in Quantitative and
 Computational Biology (Integrated Science Curriculum)
 Advisor: Amy Caudy

Research Experience:

- 2014 - Present Research Fellow (Massachusetts General Hospital; Boston, MA)
 Topics: Loss-of-function mutations in humans
 Advisor(s): Dr. Daniel MacArthur (Analytic and Translational Genetics Unit)
- 2009 - 2013 Graduate Student (Stanford University; Stanford, CA)
 Topics: Variation of transcriptional regulation in humans, Integrative analysis of
 personal genomes
 Advisor(s): Dr. Michael Snyder (Department of Genetics), Dr. Stephen
 Montgomery (Department of Pathology)
- 2008 - 2009 Undergraduate Thesis Student (Princeton University; Princeton, NJ)
 Topics: Mitochondrial RNA and its regulation in mammals, Characterization of
 the transcriptome of *Saccharomyces bayanus* and comparisons of regulation of
 gene expression to *S. cerevisiae*
 Advisor: Dr. Amy Caudy (Lewis-Sigler Institute for Integrative Genomics)
- 2007 Visiting Researcher (L'École Polytechnique Fédérale de Lausanne; Lausanne, Switzerland)
 Topics: Physical behavior of biological polymers (DNA), using atomic force
 microscopy (AFM)
 Advisor: Dr. Giovanni Dietler (Laboratory of Physics of Living Matter)
- 2006 Lab Assistant (University of Pennsylvania; Philadelphia, PA)
 Topics: Study of structure and function of *E. coli* aspartate receptor and variants
 (Cloning, protein expression, crystallography, biochemistry)
 Advisor: Dr. William DeGrado (Department of Biochemistry and Biophysics)
- 2005 - 2006 Lab Assistant (Princeton University; Princeton, NJ)
 Topics: Preparation of expression constructs, Responsibilities included
 preparation of all applicable reagents, solutions, and general lab maintenance
 Advisor: Dr. Jean Schwarzbauer (Department of Molecular Biology)
- 2005 Lab Assistant (University of Pennsylvania; Philadelphia, PA)

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Topics: Study of protein-protein interactions of a junction adhesion molecule in platelets

Advisor: Dr. Lawrence Brass (Department of Medicine)

2004 - 2005

Lab Assistant (University of Pennsylvania; Philadelphia, PA)

Topics: Growing and manipulating mouse embryonic stem cells

Advisor: Dr. Mark Kahn (Department of Molecular Cardiology)

2003

Lab Assistant (Children's Hospital of Philadelphia; Philadelphia, PA)

Topic: Cloning of mammalian chemokine receptor

Advisor: Dr. Mortimer Poncz (Department of Pediatrics)

Books:

Dudley JT and Karczewski KJ. *Exploring Personal Genomics*. January 2013. Oxford University Press.

Publications:

Lanktree MB, Elbers CC, Li Y, Zhang G, Duan Q, Karczewski KJ, Guo Y, Tragante V, North KE, Cushman M, Asselbergs FW, Wilson JG, Lange LA, Drenos F, Reiner AP, Barnes MR, Keating BJ. "Genetic meta-analysis of 15,901 African Americans identifies variation in EXOC3L1 is associated with HDL concentration." *J Lipid Res*. 2015 Jul 21. pii: jlr.P059477.

Rivas MA, Pirinen M, Conrad DF, Lek M, Tsang EK, Karczewski KJ, Maller JB, Kukurba KR, DeLuca DS, Fromer M, Ferreira PG, Smith KS, Zhang R, Zhao F, Banks E, Poplin R, Ruderfer DM, Purcell SM, Tukiainen T, Minikel EV, Stenson PD, Cooper DN, Huang KH, Sullivan TJ, Nedzel J; GTEX Consortium; Geuvadis Consortium, Bustamante CD, Li JB, Daly MJ, Guigo R, Donnelly P, Ardlie K, Sammeth M, Dermitzakis ET, McCarthy MI, Montgomery SB, Lappalainen T, MacArthur DG. "Effect of predicted protein-truncating genetic variants on the human transcriptome." *Science*. 2015 May 8;348(6235):666-9. doi: 10.1126/science.1261877.

Layer RM, Kindlon N, Karczewski KJ, Exome Aggregation Consortium, Quinlan AR. "Efficient compression and analysis of large genetic variation datasets." *bioRxiv*. doi: 10.1101/018259. 2015 Apr 20.

Li X, Battle A, Karczewski KJ, Zappala Z, Knowles DA, Smith KS, Kukurba KR, Wu E, Simon N, Montgomery SB. "Transcriptome sequencing of a large human family identifies the impact of rare noncoding variants." *Am J Hum Genet*. 2014 Sep 4;95(3):245-56.

Daneshjou R, Gamazon ER, Burkley B, Cavallari LH, Johnson JA, Klein TE, Limdi N, Hillenmeyer S, Percha B, Karczewski KJ, Langae T, Patel SR, Bustamante CD, Altman RB, Perera MA. "Genetic Variant in Folate Homeostasis Associated with Lower Warfarin Dose in African Americans." *Blood*. doi: 10.1182/blood-2014-04-568436

Tragante et al., "Gene-centric Meta-analysis in 87,736 Individuals of European Ancestry Identifies Multiple Blood-Pressure-Related Loci." *Am J Hum Genet*. 2014 Feb 19. pii: S0002-9297(13)00613-7. doi: 10.1016/j.ajhg.2013.12.016.

Karczewski KJ, Snyder M, Altman RB, Tatonetti NP. "Coherent functional modules improve transcription factor target identification, cooperativity prediction, and disease association." *PLoS Genetics*. 10(2): e1004122. doi: 10.1371/journal.pgen.1004122.s012

Holmes MV et al., "Mendelian randomization of blood lipids for coronary heart disease." *Eur Heart J*. Epub 2014 Jan 27.

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- Karczewski KJ, Fernald GH, Martin AR, Snyder M, Tatonetti NP, Dudley JT. "STORM-Seq: An Open, Community-Based Pipeline for Processing Personal Genomics Data in the Cloud." *PLOS ONE*. 9(1): e84860. doi:10.1371/journal.pone.0084860
- Karczewski KJ. "Progress in genomics according to bingo: 2013 edition." *Genome Biology*. 2013 Dec 24;14(12): 143.
- Karczewski KJ, Montgomery SB. "Performance of genomic medicine". *Genome Biology*. 2013 Dec 23;14(12):316.
- Yoneyama S, Guo Y, Lanktree MB, Barnes MR, Elbers CC, Karczewski KJ, et al., "Gene-centric meta-analyses for central adiposity traits using up to 57,412 individuals of European descent confirm known loci and reveal several novel associations." *Hum Mol Genet*. 2014 May 1;23(9):2498-510. doi: 10.1093/hmg/ddt626. Epub 2013 Dec 17.
- Franceschini N et al. "Genome-wide Association Analysis of Blood-Pressure Traits in African-Ancestry Individuals Reveals Common Associated Genes in African and Non-African Populations." *Am J Hum Genet*. 2013 Sep 5;93(3):545-54. doi: 10.1016/j.ajhg.2013.07.010. Epub 2013 Aug 22.
- Salari K, Karczewski KJ, Hudgins L, Ormond KE. "Evidence that personal genome testing enhances student learning in a course on genomics and personalized medicine." *PLOS ONE*. 8(7): e68853. doi:10.1371/journal.pone.0068853
- Caudy AA et al. "A New System for Comparative Functional Genomics of Saccharomyces Yeasts." *Genetics*. Epub 2013 Jul 12.
- Daneshjou R, Tatonetti NP, Karczewski KJ, Sagreiya H, Bourgeois S, Drozda K, Burmestor JK, Tsunoda T, Nakamura Y, Kubo M, Tector M, Limdi NA, Cavallari LH, Perera M, Johnson JA, Klein TE, Altman RB. "Pathway Analysis of Genome-Wide Data Improves Warfarin Dose Prediction." *BMC Genomics*. 2013, 14(Suppl 3):S11 doi:10.1186/1471-2164-14-S3-S11. Epub 2013 May 28.
- Karczewski KJ*, Dudley JT*, Kukurba KR, Chen R, Butte AJ, Montgomery SB, Snyder M. "Systematic functional regulatory assessment of disease-associated variants." *Proc Natl Acad Sci U S A*. Epub 2013 May 20. doi: 10.1073/pnas.1219099110.
- Karczewski KJ. "The future of genomic medicine is here." *Genome Biol*. 2013 Mar 27;14(3):304. doi:10.1186/gb-2013-14-3-304
- Montgomery SB, Goode D, Kvikstad E, Albers CA, Zhang Z, Mu XJ, Ananda G, Howie B, The 1000 Genomes Pilot Project Consortium, MacArthur DG, Karczewski KJ, Smith KS, Anaya V, Richardson R, Sidow A, Duret A, Gerstein M, Makova K, Marchini J, McVean G, Lunter G. "The origin, evolutionary dynamics and functional impact of short insertion-deletion variants identified in 179 human genomes." *Genome Res*. 2013 May;23(5):749-61. doi: 10.1101/gr.148718.112. Epub 2013 Mar 11.
- Karczewski KJ, Daneshjou R, Altman RB. "Chapter 7: Pharmacogenomics." *PLoS Comput Biol*. 2012;8(12):e1002817. doi: 10.1371/journal.pcbi.1002817. Epub 2012 Dec 27.
- O'Huallachain M, Karczewski KJ, Weissman S, Urban AE, Snyder M. "Extensive Genetic Variation in Somatic Human Tissues." *Proc Natl Acad Sci U S A*. 2012 Oct 30; 109(44): 18018-18023. doi: 10.1073/pnas.1213736109. Epub 2012 Oct 5
- The ENCODE Project Consortium. "An Integrated Encyclopedia of DNA Elements in the Human Genome." *Nature*. 2012 Sep 6;489(7414):57-74. doi: 10.1038/nature11247.

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- Boyle AP, Hong EL, Hariharan M, Chen Y, Schaub MA, Kasowski M, **Karczewski KJ**, Park J, Hitz BC, Weng S, Cherry JM, Snyder M. "Annotation of Functional Variation in Personal Genomes Using RegulomeDB." *Genome Res.* 2012 Sep;22(9):1790-7. doi: 10.1101/gr.137323.112.
- Adamcik J, Jeon J, **Karczewski KJ**, Metzler R, Dietler G. "Quantifying supercoiling-induced denaturation bubbles in DNA." *Soft Matter*. Epub 2012 Jul 09. DOI: 10.1039/c2sm26089a
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- Karczewski KJ***, Tirrell RP*, Tatonetti NP, Dudley JT, Cordero P, Salari K, Altman RB, Snyder M, and Kim SK. "Interpretome: A Freely Available, Modular, and Secure Personal Genome Interpretation Engine." *Pac Symp Biocomput.* Epub 2011 Oct 25. 17:339-350(2012).
- Clark MJ*, Chen R*, Lam HY, **Karczewski KJ**, Chen R, Euskirchen G, Butte AJ, Snyder M. "Performance comparison of exome DNA sequencing technologies" *Nat Biotechnol.* Epub 2011 Sep 25. doi:10.1038/nbt.1975.
- Dewey FE, Chen R, Cordero SP, Ormond KE, Caleshu C, **Karczewski KJ**, Whirl-Carrillo M, Wheeler MT, Dudley JT, Byrnes JK, Cornejo OE, Knowles JK, Woon M, Sangkuhl K, Gong L, Thorn CF, Hebert JM, Capriotti E, David SP, Pavlovic A, West A, Thakuria JV, Ball MP, Zaranek AW, Rehm HL, Church GM, West JS, Bustamante CD, Snyder M, Altman RB, Klein TE, Butte AJ, Ashley EA. "Phased whole genome genetic risk in a family quartet using a major allele reference sequence." *PLoS Genet.* 2011 Sep;7(9):e1002280. doi: 10.1371/journal.pgen.1002280. Epub 2011 Sep 15.
- Karczewski KJ**, Tatonetti NP, Landt SG, Yang X, Slifer T, Altman RB, Snyder M. "Cooperative Transcription Factor Associations Discovered using Regulatory Variation." *Proc Natl Acad Sci U S A.* 2011 Aug 9;108(32):13353-8. doi: 10.1073/pnas.1103105108. Epub 2011 Jul 26.
- Zheng W, Gianoulis TA, **Karczewski KJ**, Zhao H, Snyder M. "Regulatory Variation Within and Between Species: Implications for Diversity and Evolution." *Annu Rev Genomics Hum Genet.* 2011 Sep 22;12:327-46. doi: 10.1146/annurev-genom-082908-150139. Review
- Fernald GH, Capriotti E, Daneshjou R, **Karczewski KJ**, Altman RB. "Bioinformatics Challenges for Personalized Medicine." *Bioinformatics.* 2011 Jul 1;27(13):1741-8. Epub 2011 May 19.
- Kasowski M, Grubert F, Heffelfinger C, Hariharan M, Asabere A, Waszak SM, Habegger L, Rozowsky J, Shi M, Urban AE, Hong MY, **Karczewski KJ**, Huber W, Weissman SM, Gerstein MB, Korbel JO, Snyder M. "Variation in Transcription Factor Binding Among Humans." *Science.* 2010 Apr 9;328(5975):232-5. Epub 2010 Mar 18.

Public Projects:

STORMSeq: Scalable Tools for Open-Source Read Mapping.

<http://www.stormseq.org>

Interpretome: A Freely Available, Modular, and Secure Personal Genome Interpretation Engine.

<http://www.interpretome.com>

Konrad J Karczewski, Ph.D.

Talks:

Karczewski KJ, MacArthur DG. “The Human Knockout Project: Systematic discovery of loss-of-function variants in humans” *Talk*.

-2014 Genome Informatics Conference

-2014 American Society of Human Genetics (Plenary Presentation)

Karczewski KJ, Hammerbacher J, Dudley JT. “Exploring Personal Genomics.” *Workshop*.

-2014 South by Southwest Interactive Festival

-2015 South by Southwest Interactive Festival

Karczewski KJ, Dudley JT. “Software for Clinical Genomics.” *Talk*.

-2013 The Clinical Genomics Conference

Karczewski KJ, Fernald GH, Martin AR, Snyder M, Tatonetti NP, and Dudley JT. “STORM-Seq: An Open, Community-Based Pipeline for Processing Personal Genomics Data in the Cloud.” *Talk*.

-2013 Pacific Symposium on Biocomputing (PSB)

Karczewski KJ, Tirrell RP, Tatonetti NP, Dudley JT, Cordero P, Salari K, Altman RB, Snyder M, and Kim SK. “Interpretome: A Freely Available, Modular, and Secure Personal Genome Interpretation Engine.” *Talk*.

-2012 Pacific Symposium on Biocomputing (PSB)

Karczewski KJ, Dudley JT, Tatonetti NP, Chen R, Butte AJ, Snyder M. “Assessing Functional and Clinical Significance of Regulatory Variants.” *Talk*.

-2011 SNP-SIG Meeting, Int’l Conference on Intelligent Systems for Molecular Biology (ISMB)

-2011 Biomedical Computation at Stanford (BCATS) Conference

Karczewski KJ, Tatonetti NP, Landt SG, Snyder M. “The ABC test and the ALPHABIT pipeline”. *Talk*.

-2010 Centers of Excellence in Genomic Science Meeting

Teaching Experience:

Instructor, *Pharmacogenomics*, San Francisco State University (Spring Semester, 2012)

Instructor, *Principles of Pharmacogenomics*, Stanford University (Winter 2012)

Head Teaching Assistant & Lecturer, *Genetics 210: Genomics and Personalized Medicine*, Stanford University (Summer 2010 and Spring 2011)

Head Teaching Assistant & Lecturer (Comparative Genomics), *Biomedical Informatics 214: Representations and Algorithms for Computational Molecular Biology*, Stanford University (Fall 2010 and Fall 2011)

Invited Lectures:

Personal Genomics 101, March 2013, Biology 109A (Stanford University)

Advanced Personal Genomics Workshop (with Dudley JT), multiple sessions (Singularity University, July 2011; BioCurious, October 2011, July 2012; GET Conference, April 2013)

Personalized Medicine and Genomics, March 2011, March 2012, Med School 101 (high school outreach program, Stanford University School of Medicine)

Exome Sequencing in Disease Gene Identification, November 2010, Genetics 202 (Stanford University)

Academic Honors/Funding/Fellowships:

2015 NIGMS F32 Postdoctoral Fellowship

Konrad J Karczewski, Ph.D.

2014 ASHG/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research
-Finalist

2011 National Science Foundation Graduate Research Fellowship Program
-Award Winner

2010 National Science Foundation Graduate Research Fellowship Program
-Honorable Mention

2009 “Omics Meets Cell Biology” Keystone Symposia Conference
-Keystone Symposia Scholarship Winner

Meeting Organizer:

2010 Biomedical Computation at Stanford (BCATS) Conference, Chair

Skills:

Computational analysis tools: Python, MySQL, R, Perl, PHP, Bash, Matlab, and Java programming; Web development (Javascript, JQuery, HTML5, Django, D3); Cloud computing (Amazon AWS); Various computational biology applications

Basic laboratory and Molecular Biology procedures: PCR, DNA and Protein purification, Western blots, Transformation, Cloning, Tissue Culture, Microarrays, High-throughput sequencing (RNA-Seq, CHIP-Seq)

Languages: Fluent in English, Polish, and French, Conversational in Spanish

References available upon request