Curriculum Vitae

Christine R. Beck, Ph.D.

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Faculty Position

2017-Present Assistant Professor, Tenure Track
University of Connecticut Heath and the Jackson Laboratory for Genomic Medicine
Department of Genetics and Genome Sciences
Department of Molecular Biology and Biophysics

Education

1998-2002 B.S., Biochemistry, Iowa State University, Ames, Iowa

2004 Graduate Non-Degree Student, University of Iowa, Iowa City, Iowa

2005-2012 Ph.D., Human Genetics, Laboratory of Dr. John V. Moran, Ph.D., University of Michigan, Ann Arbor, Michigan

Postdoctoral Training

2012-2017 Research Fellow, Laboratory of Dr. James R. Lupski, M.D., Ph.D., Department of Molecular & Human Genetics, Baylor College of Medicine, Houston TX

Other Professional Experience and Activities

2002-2005 Research Assistant, Laboratory of Beverly L. Davidson, Ph.D., University of Iowa, Iowa City, Iowa

2012-Present Ad hoc manuscript reviewer: *Genome Research*, *Genetics in Medicine*, *Brain*, *Neurogenetics*, *Human Mutation,* and *PLOS One*.

2008-Present	Society Memberships: American Society of Human Genetics, Parents and Researchers Interested in Smith-Magenis Syndrome.
2016	American Society of Human Genetics, Annual Meeting Vancouver, Canada Session co-Organizer and Chair, "The Role of DNA Repair in Genomic Variation, Instability, and Human Disease."
2017	American Society of Human Genetics, Annual Meeting Orlando, Florida Abstract Reviewer and Session Chair, "Genome Structure and Function: The Contribution of Mutations to Human Genetic Diversity, Disease, and Evolution"
2018	American Society of Human Genetics, Annual Meeting San Diego, California Abstract Reviewer and Session Chair, "Emerging Omics Technologies"

Honors and Awards

2007	Anita and Howard Cramer Scholarship Award University of Michigan Department of Human Genetics, Ann Arbor, Michigan
2006-2008	NIH Genetics Training Grant (Pre-doctoral) University of Michigan, Ann Arbor, Michigan
2008-2011	NIH Genome Science Training Program (Pre-doctoral) University of Michigan, Ann Arbor, Michigan
2009	ASHG Trainee Award Semi-Finalist American Society of Human Genetics Annual Meeting, Honolulu, Hawaii
2010	Neel Fellowship University of Michigan Department of Human Genetics, Ann Arbor, Michigan
2012-2013	NIH Brain Disorders and Development Training Program (Post-doctoral) Baylor College of Medicine, Houston, Texas
2013-2016	HHMI Fellow of the Damon Runyon Cancer Research Foundation Baylor College of Medicine, Houston, Texas

Teaching Activity

2006	Teaching Assistant, Human Genetics 541 University of Michigan, Ann Arbor, Michigan
2006-2008	Volunteer Instructor, Summer Science Academy University of Michigan, Ann Arbor, Michigan
2018-present	Organizer, Structural Variation Journal Club The Jackson Laboratory, Farmington, Connecticut
2018-present	Lecturer, MEDS 6448; Foundations of Biomedical Sciences University of Connecticut Heath Center, Farmington, Connecticut

Mentoring Activities

2013-2014	Janson White, Molecular and Human Genetics PhD Candidate, BCM
2015-2017	Xiaofei Song, Molecular and Human Genetics PhD Candidate, BCM
2016-2017	Vahid Bharambeigi, Master's Student, MD Anderson
2017-present	Kirby Madden-Hennessey, PhD Candidate; Thesis Committee Member Molecular Biology and Biochemistry program, UCHC
2018	Jacob Dayton, Summer Science Program Mentor, The Jackson Laboratory
2018-present	Alex Nesta, PhD Candidate; Thesis Mentor Genetics and Developmental Biology program, UCHC
2018	Nicholas Jannetty, PhD Candidate; Rotation Mentor, Fall Semester

Committee Membership (Department and University)

2018-	Scientific Advisory Committee The Jackson Laboratory of Genomic Medicine Farmington, CT
2018-	Faculty Retreat Planning Committee The Jackson Laboratory of Genomic Medicine Farmington, CT

Competitive Funding Secured

2013-2016	**HHMI Fellow of the Damon Runyon Cancer Research Foundation** Baylor College of Medicine, Houston, Texas PI: Christine R. Beck Title: Mechanisms underlying copy number gain in cancer and genomic disorder-associated complex rearrangements Duration: 3.5 years
2016-2017	**K99 GM120453 Pathway to Independence Award** NIH / National Institute of General Medical Sciences (NIGMS) Baylor College of Medicine, Houston, Texas PI: Christine R. Beck Title: Mechanisms of Repetitive Element Mediated Genomic Rearrangements
2018	**R00 GM120453 Pathway to Independence Award** NIH / National Institute of General Medical Sciences (NIGMS) UCONN Health/Jackson Laboratory for Genomic Medicine PI: Christine R. Beck Title: Mechanisms of Repetitive Element Mediated Genomic Rearrangements

Publications

1. Harper SQ, Staber PD, **Beck CR**, Fineberg SK, Stein C, Ochoa D, Davidson BL. (2006). Optimization of feline immunodeficiency virus vectors for RNA interference. *J Virol*. *80,* 9371-80.

2. **Beck CR**, Collier P, Macfarlane C, Malig M, Kidd JM, Eichler EE, Badge RM, Moran JV. (2010) LINE-1 Activity in Human Genomes. *Cell. 14,* 1159-1170.

3. **Beck CR**, Garcia-Perez JL, Badge RM, Moran, JV. (2011). LINE-1 Elements in Structural Variation and Disease. *Annu Rev Genomics Hum Genet*. *12*, 187-215.

4. Bank LM, Bianchi LM, Ebisu F, Lerman-Sinkoff D, Smiley EC, Shen YC, Ramamurthy P, Thompson DL, Roth TM, **Beck CR**, Flynn M, Teller RS, Feng L, Llewellyn GN, Holmes B, Sharples C, Coutinho-Budd J, Linn SA, Chervenak AP, Dolan DF, Benson J, Kanicki A, Martin CA, Altschuler R, Koch AE, Jewett EM, Germiller JA, Barald KF. (2012). Macrophage Migration Inhibitory Factor Acts as a Neurotrophin in the Developing Inner Ear. *Development*. *139*, 4666-74.

5. Macfarlane CM, Collier P, Rahbari R, **Beck CR**, Igoe S, Moran JV, Badge RM. (2013). Transduction-Specific ATLAS Reveals a Cohort of Highly Active L1 Retrotransposons in Human Populations. *Hum Mutat*. *34*, 974-85.

6. Shuvarikov A, Campbell IM, Dittwald P, Neill NJ, Bialer MG, Moore C, Wheeler PG, Wallace SE, Hannibal MC, Murray MF*,* Giovanni MA, Terespolski D, Sodhi S, Cassina M, Viskochil D, Moghaddam B, Herman K, Brown CW, **Beck CR**, Gambin A, Cheung SW, Patel A, Lamb AN, Shaffer LG, Ellison JW, Ravnan JB, Stankiewicz P, Rosenfeld JA (2013). Recurrent HERV-H-mediated 3q13.2-q13.31 deletions cause a syndrome of hypotonia and motor, language, and cognitive delays. *Hum Mutat 34*, 1415-1423.

7. Okamoto Y, Pehlivan D, Wiszniewski W, **Beck CR**, Snipes GJ, Lupski JR, and Khajavi M (2013). Curcumin facilitates a transitory cellular stress response in Trembler-J mice. *Hum Mol Genet. 22*, 4698-4705.

8. Okamoto Y, Goksungur MT, Pehlivan D, **Beck CR**, Gonzaga-Jauregui C, Muzny DM, Atik MM, Carvalho CM, Matur Z, Bayraktar S*, et al.* (2013). Exonic duplication CNV of *NDRG1* associated with autosomal-recessive HMSN-Lom/CMT4D. *Genet Med*. *16*, 386-94.

9. Pehlivan D, Karaca E, Aydin H, **Beck CR**, Gambin T, Muzny DM, Bilge Geckinli B, Karaman A, Jhangiani SN, Centers for Mendelian Genomics*, et al.* (2014). Whole-exome sequencing links *TMCO1* defect syndrome with cerebro-facio-thoracic dysplasia. *Eur J Hum Genet*. *22*, 1145-8.

10. Stray-Pedersen A, Backe PH, Sorte HS, Mørkrid L, Chokshi NY, Erichsen HC, Gambin T, Elgstøen KB, Bjørås M, Wlodarski MW, Krüger M, Jhangiani SN, Muzny DM, Patel A, Raymond KM, Sasa GS, Krance RA, Martinez CA, Abraham SM, Speckmann C, Ehl S, Hall P, Forbes LR, Merckoll E, Westvik J, Nishimura G, Rustad CF, Abrahamsen TG, Rønnestad A, Osnes LT, Egeland T, Rødningen OK, **Beck CR**, *et al.* (2014). *PGM3* mutations cause a congenital disorder of glycosylation with severe immunodeficiency and skeletal dysplasia. *Am J Hum Genet. 95*, 96-107.

11. Boone PM, Yuan B, Campbell IM, Scull JC, Withers MA, Baggett BC, **Beck CR**, Shaw CJ, Stankiewicz P, Moretti P, Goodwin WE, Hein N, Fink JK, Seong MW, Seo SH, Park SS, Karbassi ID, Batish SD, Ordóñez-Ugalde A, Quintáns B, Sobrido MJ, Stemmler S, and Lupski JR. (2014). The *Alu*-rich genomic architecture of *SPAST* predisposes to diverse and functionally distinct disease-associated CNV alleles. *Am J Hum Genet. 95*, 143-61.

12. Campbell IM, Gambin T, Dittwald P, **Beck CR**, Shuvarikov A, Hixson P, Patel A, Gambin A, Shaw CA, Rosenfeld JA, and Stankiewicz P. (2014). Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. *BMC Biol*. *12*, 74.

13. Yuan B, Pehlivan D, Karaca E, Patel N, Charng WL, Gambin T, Gonzaga-Jauregui C, Sutton VR, Yesil G, Bozdogan ST, Tos T, Koparir A, Koparir E, **Beck CR**, Gu S, Aslan H, Yuregir OO, Al Rubeaan K, Alnaqeb D, Alshammari MJ, Bayram Y, Atik MM, Aydin H, Geckinli BB, Seven M, Ulucan H, Fenercioglu E, Ozen M, Jhangiani S, Muzny DM, Boerwinkle E, Tuysuz B, Alkuraya FS, Gibbs RA, and Lupski JR. (2014). Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. *J Clin Invest*. *125*, 636-51.

14. Beck CR, Carvalho CMB, Banser L, Gambin T, Stubbolo D, Yuan B, Sperle K, McCahan SM, Henneke M, Seeman P, Garbern JY, Hobson GM, and Lupski JR. (2015). Complex genomic rearrangements at the *PLP1* locus include triplication and quadruplication. *PLoS Genet*. *11,* e1005050.

15. English AC, Salerno WJ, Hampton OA, Gozaga-Jauregui C, Ambreth S, Ritter, DI, **Beck CR**, Davis CF, Dahdouli M, Ma S, Carroll A, Veeraraghavan N, Bruestle J, Drees B, Hastie A, Lam ET, White S, Mishra P, Wang M, Han Y, Zhang F, Stankiewicz P, Wheeler DA, Reid JG, Muzny DM, Rogers J, Sabo A, Worley KC, Lupski JR, Boerwinkle E, and Gibbs RA. (2015). Assesing structural variation in a personal genome-towards a human reference diploid genome. *BMC Genomics*. *11*, 286.

16. Wang M[*], **Beck CR**[*], English AC, Meng Q, Buhay C, Han Y, Doddapaneni HV, Yu F, Boerwinkle E, Lupski JR, Muzny DM, and Gibbs RA. (2015). PacBio-LITS: A large-insert targeted sequencing method for characterization of human disease-associated chromosomal structural variations. *BMC Genomics*. *16*, 214. ([*]co-first author).

17. Gu S, Yuan B, Campbell IM, **Beck CR**, Carvalho CM, Nagamani SC, Erez A, Patea A, Bacino CA, Shaw CA, Stankiewicz P, Cheung SW, Bi W, Lupski JR. (2015). Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. *Hum Mol Genet*. *24*, 4061-77.

18. Mayle R, Campbell IM, **Beck CR**, Yu Y, Wilson M, Shaw CA, Bjergbaek L, Lupski JR, Ira G. (2015). Mus81 and converging forks limit the mutagenicity of replication fork breakage. *Science. 349*, 742-7.

19. Pehlivan D[*], **Beck CR**[*], Okamoto Y, Harel T, Akdemir ZHC, Jhangiani SN, Withers MA, Goksungur MT, Carvalho CMB, Czesnik D, Gonzaga-Jauregui C, Wiszniewski W, Muzny DM, Baylor-Hopkins Center for Mendelian Genomics, Gibbs RA, Rautenstrauss B, Sereda MW, Lupski JR. (2015). The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. *Genet In Med. 18,* 443-51. ([*]co-first author)

20. Yuan B, Harel T, Gu S, Liu P, Burglen L, Chantot-Bastaraud S, Gelowani V, **Beck CR**, Carvalho CM, Cheung SW*, et al.* (2015). Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The *PMP22-RAI1* Contiguous Gene Duplication Syndrome. Am J Hum Genet *97*, 691-707.

21. Yuan B, Liu P, Gupta A, **Beck CR**, Tejomurtula A, Campbell IM, Gambin T, Simmons AD, Withers MA, Harris RA*, et al.* (2015). Comparative Genomic Analyses of the Human *NPHP1* Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. *PLoS Genet 11*, e1005686.

22. White J[*], **Beck CR**[*], Harel T, Posey JE, Jhangiani SN, Tang S, Farwell KD, Powis Z, Mendelsohn NJ, Baker JA*, et al.* (2016). *POGZ* truncating alleles cause syndromic intellectual disability. *Genome Med 8*, 3. (*co-first author).

23. Stray-Pedersen A, Sorte HS, Samarakoon P, Gambin T, Chinn IK, Coban-Akdemir ZH, Erichsen HC, Forbes LR, Gu S, Yuan B, Jhangiani SN, Muzny DM, Rodningen OK, Sheng Y, Nicholas SK, Noroski LM, Seeborg FO, Davis CM, Canter DL, Mace EM, Vece TJ, Allen CE, Abhyankar HA, Boone PM, **Beck CR**, *et al.* (2016). Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. *J Allergy Clin Immunol*. *139,* 232-245.

24. Loviglio MN*, **Beck CR***, White JJ, Leleu M, Harel T, Guex N, Niknejad A, Bi W, Chen ES, Crespo I, Yan J, Charng WL, Gu S, Fang P, Coban-Akdemir Z, Shaw CS, Jhangiani SN, Muzny DM, Gibbs RA, Rougemont J, Xenarios I, Lupski JR, Reymond A. (2016). Identification of a *RAI1*-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. *Genome Med 8*, 105. (*co-first author)

25. Larson P, Moldovan JB, Jasti N, Kidd JM, Beck CR, Moran JV. (2018) Spliced integrated retrotransposed element (SpIRE) formation in the human genome. *PLoS Biology. 16,* e2003067.

26. Song, X., Beck, C.R., Du, R., Campbell, I.M., Coban-Akdemir, Z., Gu, S., Breman, A.M., Stankiewicz, P., Ira, G., Shaw, C.A.*, et al.* (2018). Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Res.

Selected Oral Presentations at Scientific Meetings and Invited Speaking Engagements

October 2009- Beck CR, Collier P, Macfarlane C, Malig M, Kidd JM, Eichler EE, Badge RM, Moran JV. LINE-1 variation in human genomes. American Society of Human Genetics, Honolulu, Hawaii.

June 2012- Beck CR, Liu P, Carvalho CMB, Stray-Pedersen A, Lacaria M, Lupski JR. The Molecular Basis of Phenotypic Variation in Smith-Magenis and Potocki-Lupski Syndromes. Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS) Research Symposium, Denver, Colorado.

November 2013- Wang M, English A, **Beck CR,** Buhay C, Han Y, Kovar C, Santibanez J, Doddapanen HV, Yu F, Reid J, Boerwinkle E, Lupski JR, Muzny DM, Gibbs RA. PacBio Long-Read-Length Sequencing for Structural Variation Analysis of the Human Genome. Personal Genomes Meeting, Cold Spring Harbor Labs, New York. (co-first author)

August 2014- Beck CR, Liu P, Lupski JR. Smith-Magenis Syndrome and Complex Genomic Rearrangements. Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS) Research Symposium, St. Louis, Missouri.

November 2014- Beck CR, Carvalho CMB, Banser L, Gambin T, Stubbolo D, Yuan B, Sperle K, McCahan SM, Henneke M, Seeman P, Hobson GM, and Lupski JR. Complex Genomic Rearrangements at the *PLP1* Locus Include Triplication and Quadruplication. Personal Genomes: Discovery, Treatment & Outcomes Meeting, Cold Spring Harbor Labs, New York.

February 2016- Beck CR, Loviglio, MN, White JJ, Harel T, Mikhaleva A, Yalcin B, Bi W, Coban-Akdemir Z, Yan J, Charng W, Chen E, Shaw CA, Reymond A, Lupski JR. Integration of exome sequencing and genomic analyses reveals potential Smith-Magenis-like causative genes. Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS) Research Symposium, Houston, Texas.

May 2016- Beck CR, Carvalho CMB, Wang M, Meng Q, English AC, Bainbridge MN, Salerno WJ, Muzny DM, Lupski JR, Gibbs RA. The use of capture and long-read PacBio sequencing for clinically relevant loci. **Invited Speaker**. European Society of Human Genetics PacBio Satellite Meeting, Barcelona, Spain.

September 2016- Beck CR, Carvalho CMB, Wang M, Meng Q, Chen ES, Chong Z, English AC, Salerno WJ, Muzny DM, Shaw CA, Chen K, Gibbs RA, Lupski JR. Targeted Capture and SMRT sequencing for Complex Genomic Loci. **Invited Speaker**. PacBio Users Group Meeting, Stanford, California.

October 2016- Beck CR. Mechanisms for DNA Rearrangements in Genomic Disorders- From the Bedside to the Bench Side. **Invited Speaker**. American Society of Human Genetics Meeting; Vancouver, Canada.

April 2018- Beck CR. Repetitive sequences and genomic plasticity. **Invited Speaker**. The University of Connecticut, MCB Spring Seminar Series; Storrs, Connecticut.

May 2018- Beck CR. Repetitive sequences and genomic plasticity. **Invited Speaker**. Annual New England Genome Instability and Cancer Conference; Worcester, Massachusetts.

May 2018- Beck CR. Repetitive elements and genomic change. **Keynote Speaker**. Big Data for Genomics Professors Course; Farmington, Connecticut.