Curriculum Vitae, Gregory M. Cooper, Ph.D.

Contact Information

HudsonAlpha Institute for Biotechnology
601 Genome Way
Hunstville, AL 35806-2908
E-mail: gcooper@hudsonalpha.org
Phone: (256) 327-9490
Web: http://research.hudsonalpha.org/CooperG/

Education

2001 – 2006	Ph.D. Genetics; Stanford University; Stanford, CA
1997 – 2001	B.S. Mathematics and Statistics; B.A. Microbiology; *summa cum laude*; Miami University; Oxford, OH

Research Positions

2010 –	Faculty Investigator; HudsonAlpha Institute for Biotechnology; Huntsville, AL
2010 –	Adjunct Instructor; Department of Genetics; University of Alabama-Birmingham; Birmingham, AL
2009 – 2010	Acting Assistant Professor; Department of Genome Sciences; University of Washington; Seattle, WA
2006 – 2009	Senior Fellow; Department of Genome Sciences; University of Washington; Seattle, WA; Advisors: Evan E. Eichler and Deborah A. Nickerson
2001 – 2006	Ph.D. Student; Department of Genetics; Stanford University; Stanford, CA; Advisor: Arend Sidow; Thesis: Evolutionary constraints on the human genome

Honors and Awards

2007 – 2010	Merck, Jane Coffin Childs Memorial Fund Postdoctoral Fellowship
2007	UW Genome Training Grant Postdoctoral Fellowship
2001 – 2006	Howard Hughes Medical Institute Doctoral Fellowship
1999 – 2001	Goldwater Scholar
1997 – 2001	Harrison Scholar, Miami University

Teaching and Reviewer Activity

2014 –	Course Master and Lecturer for "Genomics" offered by HudsonAlpha, the University of Alabama-Birmingham, and the University of Alabama-Huntsville
2011 –	Guest Lecturer for 'Genomics' offered by the University of Alabama-Birmingham
2010 –	Editorial Board Member; *Genome Research*

2002 –	Reviewer for *Genome Research*, *PLoS Genetics, Nature Genetics, Nature, American Journal of Human Genetics, Nature Reviews Genetics, Nature Methods, PNAS, Science, Cell Stem Cell, Molecular Biology and Evolution, PLoS Computational Biology, BMC Bioinformatics, Genomics, FEBS, Diabetes, Physiological Genomics, Pacific Symposium on Biocomputing, Trends in Genetics*, and *CSHL Press*
2008 – 2010	Guest Lecturer for 'Pharmacogenetics and Toxicogenomics', 'Statistical Genetics', and 'Genetic Epidemiology'; University of Washington
2004 – 2005	Teaching assistant for 'Genomics'; Stanford University
2001	Teaching assistant for 'Introduction to Statistics'; Miami University

Publications

1. Kircher, M.*, Witten, D.M.*, Jain, P., O'Roak, B.J., **Cooper, G.M.**†, and Shendure, J.† 2014. A general framework for estimating the relative pathogenicity of human genetic variants. *Nature Genetics* 46:310-315. PMID: 24487276.
2. Gertz, J., Savic, D., Varley, K.E., Partridge, E.C., Safi, A., Jain, P., **Cooper, G.M**., Reddy, T.E, Crawford, G.E., and Myers, R.M.† 2013. Distinct properties of cell-type-specific and shared transcription factor binding sites. *Molecular Cell* 52(1):25-36. PMID: 24076218. PMCID: 3811135.
3. Savic, D., Gertz, J., Jain, P., **Cooper, G.M.**, and Myers, R.M.† Mapping genome-wide transcription factor binding in frozen tissue. 2013. *Epigenetics & Chromatin* 6:30. PMID: 24279905. PMCID: 3848595.
4. Lorente-Galdos, B., Bleyhl, J., Vives, L., **Cooper, G.M.**, Navarro, A.†, Eichler, E.E.†, and Marques-Bonet, T.† Accelerated exon evolution in duplicated regions in hominids. 2013. *Genome Biology* 14(1):R9. PMID: 23360670. PMCID: 3906575.
5. Kaelin, C.B.*, Xu, X.*, Hong, L.Z., David, V.A., McGowan, K.A., Schmidt-Kuntzel, A., Roelke, M.E., Pontius, J., **Cooper, G.M.**, Manuel, H., Kraus, L.M., Harper, C.K., van Dyk, A., Yue, B., Mullikin, J.C., Warren, W.C., Eizirik, E., O'Brien, S.J., Barsh, G.S.†, and Menotti-Raymond, M. 2012. Specifying and sustaining pigmentation patterns in domestic and wild cats. *Science* 337 (6101):1536-1541. PMID: 22997338. PMCID: 3709578.
6. Patwardhan, R.P., Hiatt, J.B., Witten, D.M., Kim, M.J., Smith, R.P., May, D., Lee, C., Andrie, J.M., Lee, S.I., **Cooper, G.M.**, Ahituv, N.†, Pennacchio, L.A.†, and Shendure, J.† 2012. Massively parallel functional dissection of mammalian enhancers *in vivo*. *Nature Biotechnology* 30 (3): 265-270. PMID: 22371081. PMCID: 3402344.
7. **Cooper, G.M.***, Coe, B.P.*, Girirajan, S.J.*, Rosenfeld, J.A., Vu, T., Baker, C., Williams, C., Stalker, H., Hamid, R., Hannig, V., Abdel-Hamid, H., Bader, P., McCracken, E., Niyazov, D., Leppig, K., Thiese, H., Hummel, M., Alexander, N., Gorski, J., Kussmann, J., Shashi, V., Johnson, K., Rehder, C., Bejjani, B., Shaffer, L., and Eichler, E.E.† 2011. A copy number variation morbidity map of developmental delay. *Nature Genetics* 43 (9): 838-846. PMID: 21841781. PMCID: 3171215.
8. Innocenti, F.I.*, **Cooper, G.M.***, Stanaway, I.B., Gamazon, E.R., Smith, J.D., Mirkov, S., Ramirez, J., Liu, W., Lin, Y.S., Maloney, C., Aldred, S.F., Trinklein, N.D., Shuetz, E., Nickerson, D.A., Cox, N.J., Thummel, K.E., Rieder, M.J., Rettie, A.E., Ratain, M.J., and Brown, C.D.† 2011. Identification, replication, and functional fine-mapping of expression quantitative trait loci in primary human liver tissue. *PLoS Genetics* 7 (5): e1002078. PMID: 21637794. PMCID: 3102751.

9. Davydov, E., Goode, D.L., Sirota, M., **Cooper, G.M.**, Sidow, A.†, and Batzoglou, S. 2010. Identifying a high fraction of the human genome to be under selective constraint using GERP++. *PLoS Computational Biology* 6 (12): e1001025. PMID: 21152010. PMCID: 2996323.

10. Ng, S.B.*, Bigham, A.W.*, Buckingham, K.J., Hannibal, M., McMillin, M., Gildersleeve, H., Beck, A.E., Tabor, H., **Cooper, G.M.**, Mefford, H., Lee, C., Turner, E.H., Smith, J.D., Rieder, M.J., Yoshiura, K., Matsumoto, N., Ohta, T., Niikawa, N., Nickerson, D.A., Bamshad, M.J.†, and Shendure, J.† 2010. Exome sequencing reveals that mutations in *MLL2* cause Kabuki syndrome. *Nature Genetics* 42 (9): 7990-7993. PMID: 20711175. PMCID: 2930028.

11. **Cooper, G.M.**†, Goode, D.L., Ng, S.B., Sidow, A., Bamshad, M., Shendure, J., and Nickerson, D.A. 2010. Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. *Nature Methods* 7 (4): 250-251. PMID: 20354513. PMCID: 3145250.

12. Goode, D.L., **Cooper, G.M.,** Schmutz, J., Dickson, M., Gonzales, E., Tsai, M., Davydov, E., Batzoglou, S., Myers, R.M., and Sidow, A.† 2010. Evolutionary constraint facilitates interpretation of genetic variation in resequenced human genomes. *Genome Research* 20 (3): 301-310. PMID: 20067941. PMCID: 2840986.

13. Girirajan, S.*, Rosenfeld, J.A.*, **Cooper, G.M,** Antonacci, F., Kidd, J.M., Siswara, P., Itsara, A., Vives, L., Walsh, T., McCarthy, S.E., Baker, C., Mefford, H.C., Kidd, J.M., Browning, S.R., Browning, B.L., Dickel, D.E., Levy, D.L., Ballif, B.C., Platky, K., Farber, D.M, Gowans, G.C, Wetherbee, J.J., Asamoah, A., Weaver, D.D., Mark, P.R., Dickerson, J., Garg, B.P, Ellingwood, S.A., Smith, R., Banks, V.C., Smith, W., McDonald, M.T., Hoo, J.J., French, B.N., Hudson, C., Johnson, J.P., Ozmore, J.R., Moeschler, J.B., Surti, U., Escobar, L.F., El-Khechen, D., Gorski, J.L, Kussmann, J., Salbert, B., Lacassie, Y., Biser, A., McDonald-McGinn, D.M., Zackai, E.H., Deardorff, M.A., Shaikh, T.H., Haan, E., Friend, K.L., Fichera, M., Romano, C., Gecz, J., DeLisi, L.E., Sebat, J., King, M.C., Shaffer, L.G., and Eichler, E.E.† 2010. A recurrent 16p12.1 microdeletion suggests a two-hit model for severe developmental delay. *Nature Genetics* 42 (3): 203-209. PMID: 20154674. PMCID: 2847896.

14. Zerr, T., **Cooper, G.M.**, Eichler, E.E., and Nickerson, D.A.† Targeted interrogation of copy number variation using SCIMMkit. 2010. *Bioinformatics* 26 (1): 120-122. PMID: 19846438. PMCID: 2796813.

15. Mefford, H.C.*, **Cooper, G.M.***, Zerr, T.*, Smith, J.D., Baker, C., Shafer, N., Thorland, E.C., Skinner, C., Schwartz, C.E., Nickerson, D.A., and Eichler, E.E.† 2009. A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. *Genome Research* 19 (9): 1579-1585. PMID: 19506092. PMCID: 2752120.

16. Itsara, A.*, **Cooper, G.M.*,** Baker, C., Girirajan, S., Li, J., Absher, D., Krauss, R.M., Myers, R.M., Ridker, P.M., Chasman, D.I., Mefford, H., Ying, P., Nickerson, D.A., and Eichler, E.E.† 2009. Population analysis of large copy number variants and hotspots of human genetic disease. *American Journal of Human Genetics* 84 (2): 148-161. PMID: 19166990. PMCID: 2668011.

17. **Cooper, G.M.***†, Zerr, T.R.*, Kidd, J.M., Eichler, E.E., and Nickerson, D.A. 2008. Systematic assessment of copy-number variant detection via genome-wide SNP genotyping. *Nature Genetics* 40 (10): 1199-1203. PMID: 18776910. PMCID: 2759751.

18. **Cooper, G.M.,** Johnson, J.A., Langaee, T.Y., Feng, H., Stanaway, I.B., Schwarz, U., Ritchie, M.D., Stein, C.M., Roden, D.M., Smith, J.D., Veenstra, D.L., Rettie, A.E., and Rieder, M.J.† 2008. A genome-wide scan for common genetic variants with a large influence on warfarin maintenance dose. *Blood* 112 (4): 1022-1027. PMID: 18535201. PMCID: 2515139.

19. Hannes, F.D., Sharp, A.J., Mefford, H.C., de Ravel, T., Ruivenkamp, C.A., Breuning, M.H., Fryns, J.P., Devriendt, K., Van Buggenhout, G., Vogels, A., Stewart, H.H, Hennekam, R.C., **Cooper, G.M.,** Regan, R., Knight, S.J.L, Eichler, E.E., and Vermeesch, J.R.† 2008. Recurrent reciprocal deletions and duplications of 16p13.11: The deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. *Journal of Medical Genetics* 46 (4): 223-232. PMID: 18550696. PMCID: 2658752.

20. Kidd, J.M., **Cooper, G.M.**, Donahue, W.F., Hayden, H.S., Sampas, N., Graves, T., Hansen, N., Teague, B., Alkan, C., Antonacci, F., Haugen, E., Zerr, T., Yamada, NA., Tsang, P., Newman, T.L., Tüzün, E., Cheng, Z., Ebling, H.M., Tusneem, N., David, R., Gillett, W., Phelps, K.A., Weaver, M., Saranga, D., Brand, A., Tao, W., Gustafson, E., McKernan, K., Chen, L., Malig, M., Smith, J.D., Korn, J.M., McCarroll, S.A., Altshuler, D.A., Peiffer, D.A., Dorschner, M., Stamatoyannopoulos, J., Schwartz, D., Nickerson, D.A., Mullikin, J.C., Wilson, R.K., Bruhn, L., Olson, M.V., Kaul, R., Smith, D.R., and Eichler, E.E.† 2008. Mapping and sequencing of structural variation from eight human genomes. *Nature* 453:56-64. PMID: 18451855. PMCID: 2424287.

21. Reiner, A.P.†, Barber, M.J., Guan, Y., Ridker, P.M., Lange, L.A., Chasman, D.I., Walston, J.D., **Cooper, G.M.,** Jenny, N.S., Rieder, M.J., Durda, J.P., Smith, J.D., Novembre, J., Tracy, R.P., Rotter, J.I., Stephens, M., Nickerson, D.A., and Krauss, R.M. 2008. Polymorphisms of the *HNF1A* gene encoding Hepatocyte Nuclear Factor-1α are associated with C-Reactive Protein. *American Journal of Human Genetics* 82:1-9. PMID: 18439552. PMCID: 2427318.

22. Walsh T., McClellan J.M.†, McCarthy, S.E., Pierce, S.B., **Cooper, G.M.**, Nord, A.S., Kusenda, M., Malhotra, D., Bhandari, A., Stray, S.M., Rippey, C.F., Roccanova, P., Makarov, V., Lakshmi, B., Findling, R.L., Sikich, L., Stromberg, T., Merriman, B., Gogtay, N., Butler, P., Eckstrand, K., Noory, L., Gochman, P., Long, R., Chen, Z., Davis, S., Baker, C., Eichler, E.E., Meltzer, P.S., Nelson, S.F., Singleton, A.B., Lee, M.K., Rapoport, J.L., King, M.C, and Sebat, J. 2008. Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science* 320 (5875): 539-543. PMID: 18369103. PMCID: None.

23. Sharp, A.J., Mefford, H., Li, K., Baker, C., Skinner, C., Stevenson, R.E., Schroer, R.J., Novara, F., De Gregori, M., Ciccone, R., Broomer, A., Casuga, I., Wang, Y., Xiao, C., Barbacioru, C., Gimelli, G., Bernardina, B.D., Torniero, C., Giorda, R., Regan, R., Murday, V., Mansour, S., Fichera, M., Castiglia, L., Failla, P., Ventura, M., Jiang, Z., **Cooper, G.M.**, Knight, S.J., Romano, C., Zuffardi, O., Chen, C., Schwartz, C.E., and Eichler, E.E.† 2008. A recurrent 15p13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genetics* 40 (3): 322-328. PMID: 18278044. PMCID: 2365467

24. Kathiresan, S.†, Melander, O., Guiducci, C., Surti, A., Burtt, N.P., Rieder, M.J., **Cooper, G.M.**, Roos, C., Voight, B.F., Havulinna, A.S., Wahlstrand, B., Hedner, T., Corella, D., Tai, E.S., Ordovas, J.M., Berglund, G., Vartiainen, E., Jousilahti, P., Hedblad, B., Taskinen, M.R., Newton-Cheh, C., Salomaa, V., Peltonen, L., Groop, L., Altshuler, D.M., and Orho-Melander, M.† 2008. Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol, or triglycerides in humans. *Nature Genetics* 40 (2): 189-197. PMID: 18193044. PMCID: 2682493.

25. Bovee, D., Zhou, Y., Haugen, E., Wu, Z., Hayden, H.S., Gillett, W., Tuzun, E., **Cooper, G.M.,** Sampas, N., Phelps, K., Levy, R., Morrison, V.A., Sprague, J., Jewett, D., Buckley, D., Subramaniam, S., Chang, J., Smith, D.R, Olson, M.V., Eichler, E.E, and Kaul, R.† 2008. Closing gaps in the human genome with fosmid resources generated from multiple individuals. *Nature Genetics* 40 (1): 96-101. PMID: 18157130

26. Mefford, H.C., Clauin, S., Sharp, A.J., Moller, R.S., Ullmann, R., Kapur, R., Pinkel, D., **Cooper, G.M.**, Ventura, M., Ropers, H.H., Tommerup, N., Eichler, E.E.†, and Bellanne-Chantelot, C. 2007. Recurrent

reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. *American Journal of Human Genetics* 81 (5): 1057-1069. PMID: 17924346. PMCID: 2265663.

27. Clark, T.G.† Andrew, T., **Cooper, G.M.,** Margulies, E.H., Mullikin, J.C., and Balding, D.J. 2007. Functional constraint and small insertions and deletions in the ENCODE regions of the human genome. *Genome Biology* 8 (9): R180. PMID: 17784950. PMCID: 2375018.

28. Kaelin, C.B., **Cooper, G.M.,** Sidow, A., and Barsh, G.S.† 2007. Mammalian comparative sequence analysis of the *Agrp* locus. *PLoS ONE* 2 (8): e702. PMID: 17684549. PMCID: 1931611.

29. The ENCODE Project Consortium (including **Cooper, G.M.**). 2007. Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature* 447 (7146): 799-816. PMID: 17571346. PMCID: 2212820.

30. Margulies, E.H.*†, **Cooper, G.M.***, Asimenos G.*, Thomas D.J.*, Dewey C.N.*, Siepel, A., Birney, E., Keefe, D., Schwartz, A.S., Hou, M., Taylor, J., Nikolaev, S., Montoya-Burgos, J.I., Löytynoja, A., Whelan, S., Pardi, F., Massingham, T., Brown, J.B., Bickel, P., Holmes, I., Mullikin, J.C., Ureta-Vidal, A., Paten, B., Stone, E.A., Rosenbloom, K.R., Kent, W.J., Bouffard, G.G., Guan, X., Hansen, N.F., Idol, J.R., Maduro, V.V., Maskeri, B., McDowell, J.C., Park, M., Thomas, P.J., Young, A.C., Blakesley, R.W., Muzny, D.M., Sodergren, E., Wheeler, D.A., Worley, K.C., Jiang, H., Weinstock, G.M., Gibbs, R.A., Graves, T., Fulton, R., Mardis, E.R., Wilson, R.K., Clamp, M., Cuff, J., Gnerre, S., Jaffe, D.B., Chang, J.L., Lindblad-Toh, K., Lander, E.S., Hinrichs, A., Trumbower, H., Clawson, H., Zweig, A., Kuhn, R.M., Barber, G., Harte, R., Karolchik, D., Field, M.A., Moore, R.A., Matthewson, C.A., Schein, J.E., Marra, M.A., Antonarakis, S.E., Batzoglou, S., Goldman, N., Hardison, R., Haussler, D., Miller, W., Pachter, L., Green, E.D, and Sidow, A. 2007. Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. *Genome Research* 17 (6): 760-774. PMID: 17567995. PMCID: 1891336.

31. **Cooper, G.M.**, Stone, E.A., Asimenos, G., NISC Comparative Sequencing Program, Green, E.D., Batzoglou, S., and Sidow, A.† 2005. Distribution and intensity of constraint in mammalian genomic sequence. *Genome Research* 15 (7): 978-986. PMID: 15965027. PMCID: 1172034.

32. **Cooper, G.M**., Singaravelu, S.A.G., and Sidow, A.† 2004. ABC: Software for interactive browsing of genomic multiple sequence alignment data. *BMC Bioinformatics* 5: 192. PMID: 15588288. PMCID: 539296.

33. Okun, A.†, **Cooper, G.M.**, Bailer, A.J., Bena, J., and Stayner, L. 2004. Trends in occupational lead exposure since the 1978 OSHA lead standard. *American Journal of Industrial Medicine* 45 (6): 558-572. PMID: 15164400

34. Rat Genome Sequencing Project Consortium, including **Cooper, G.M. (Co-Leader, Evolutionary Analysis Group)**. 2004. Genome sequence of the Brown Norway Rat yields insights into mammalian evolution. *Nature* 428 (6982): 493-521. PMID: 15057822

35. **Cooper, G.M.**, Brudno, M., Stone, E.A., Dubchak, I., Batzoglou, S., and Sidow, A.† 2004. Characterization of evolutionary rates and constraints in three mammalian genomes. *Genome Research* 14 (4): 539-548. PMID: 15059994. PMCID: 383297.

36. Brudno, M., Poliakov, A., Salamov, A., **Cooper, G.M.**, Sidow, A., Rubin, E.M., Solovyev, V., Batzoglou, S., and Dubchak, I.† 2004. Automated whole-genome multiple alignment of rat, mouse, and human. *Genome Research* 14 (4): 685-692. PMID: 15060011. PMCID: 383314.

37. **Cooper, G.M.**, Brudno, M., NISC Comparative Sequencing Program, Green, E.D., Batzoglou, S., and Sidow, A.† 2003. Quantitative estimates of sequence divergence for comparative analyses of mammalian genomes. *Genome Research* 13 (5): 813-820. PMID: 12727901. PMCID: 430923.

38. Brudno, M., Do, C.B., **Cooper, G.M.**, Kim, M.F., Davydov, E., NISC Comparative Sequencing Program, Green, E.D., Sidow, A., and Batzoglou, S.† 2003. LAGAN and Multi-LAGAN: efficient tools for large-scale multiple alignment of genomic DNA. *Genome Research* 13 (4): 721-731. PMID: 12654723. PMCID: 430158.

Reviews and Perspectives

39. **Cooper, G.M.**† and Shendure, J.† 2011. Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. *Nature Reviews Genetics* 12 (9): 628-640. PMID: 21850043.

40. **Cooper, G.M.**† and Mefford, H.C.† 2011. Detection of copy number variation using SNP genotyping. *Methods in Molecular Biology* 767: 243-252. PMID: 21822880.

41. **Cooper, G.M.***† and Brown, C.D.* 2008. Qualifying the relationship between sequence conservation and molecular function. *Genome Research* 18 (2): 201-205. PMID: 18245453.

42. **Cooper, G.M.**†, Nickerson, D.A., and Eichler, E.E.† 2007. Mutational and selective effects on copy-number variants in the human genome. *Nature Genetics* 39 (7 Suppl): S22-S29. PMID: 17597777.

43. Stone, E.A., **Cooper, G.M.**, and Sidow, A.† 2005. Trade-offs in detecting evolutionarily constrained sequence by comparative genomics. *Annual Review of Genomics and Human Genetics* 6: 143-164. PMID: 16124857.

44. The ENCODE Project Consortium (including **Cooper, G.M.**). 2004. The ENCODE (ENCyclopedia Of DNA Elements) Project. *Science* 306 (5696): 636-640. PMID: 15499007.

45. **Cooper, G.M.** and Sidow, A.† 2003. Genomic regulatory regions: insights from comparative sequence analysis. *Current Opinion in Genetics and Development* 13 (6): 604-610. PMID: 14638322.

* - equal contributions
† - corresponding author