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

Contact Information

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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
2017 –	Associate Professor Adjunct; Department of Genetics; University of Alabama-Birmingham; Birmingham, AL
2010 – 2017	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: Dr. Evan E. Eichler and Dr. Deborah A. Nickerson
2001 – 2006	Ph.D. Student; Department of Genetics; Stanford University; Stanford, CA; Advisor: Dr. Arend Sidow; Thesis: Evolutionary constraints on the human genome

Honors and Awards

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

Teaching and Reviewer Activity

2018 – 2022	Member of NIH Genomics, Computational Biology, and Technology study section
2015 –	Ad hoc reviewer for 10 NIH study sections
2014 –	Editorial Board Member, *PLoS Genetics*
2002 –	Peer reviewer for numerous manuscripts submitted to journals including *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, *Nature Communications,* and *CSHL Press*

2010 – 2019 Editorial Board Member, *Genome Research*

2014 – 2016 Course Director for "Genomics" offered by HudsonAlpha, the University of Alabama-Birmingham, and the University of Alabama-Huntsville

2001 – 2010 Teaching assistant and guest lecturer for several classes at Miami University, Stanford University, the University of Washington, and the University of Alabama-Birmingham

Active Grant Support (PI/Co-PI only)

Long-read genome sequencing for the molecular diagnosis of neuromuscular disorders

Muscular Dystrophy Association

09/01/22 – 08/31/2025

This project seeks to test the ability for long-read genome sequencing to diagnose probands with neuromuscular disorders that are refractory to standard clinical testing, including short-read genome sequencing. Sequencing will be performed on 50 probands, using Pacific Biosciences "HiFi", that were previously recruited into a HudsonAlpha translational genomics study but remain undiagnosed.

Utility of genome sequencing in community care contexts

NIH (NHGRI) - 1R01HG011598

Role: Co-Principal Investigator

This project seeks to assess the long-term impacts of genomic diagnostic testing on children with neurodevelopmental disorders. Recruitment includes undiagnosed probands who will be prospectively sequenced probands and probands sequenced within the previous five years. Interviews and surveys will be conducted with parents of diagnosed probands and the various professionals that treat, educate, evaluate, or otherwise work with the diagnosed probands.

09/22/2021 – 06/30/2025

Genome sequencing and diagnosis for children in our community

Alpha Foundation

12/17/2018 – 12/16/2025

Role: Principal Investigator

This project seeks to enroll children with developmental disorders in or around Huntsville and conduct genome sequencing to identify, and return to families, medically relevant genetic variation.

Genomic diagnosis of children with developmental delay or other undiagnosed phenotypes

State of Alabama and the HudsonAlpha Foundation

01/01/2015 - 12/31/2025

Role: Principal Investigator

The goal of this project is to explore the ability for whole genome sequence analysis to identify the genetic causes of rare diseases, including intellectual disability, developmental delay, and related congenital problems, in children for which there is a significant suspicion of a genetic cause for disease. Recruiting will be performed at clinics in underserved parts of southern Alabama.

Alabama Genomic Health Initiative

State of Alabama

01/01/2017 – 09/30/2023

Role: Subrecipient Principal Investigator

The goal of the Alabama Genomic Health Initiative (AGHI) is to accelerate the discovery of genomic variants with health implications and to bring these discoveries to the citizens of Alabama. Participants will be enrolled into a research study and their genome will be sequenced or genotyped by microarray.

Transcription factor function in healthy and psychiatric disorder-affected brains

NIH (NIMH) - R01MH110472

08/01/2017 – 05/31/2023

Role: Co-Principal Investigator

This project will generate a deep resource of knowledge about the locations and properties of transcription factor activities, which are crucial to controlling gene expression, in both healthy and disorder-affected brain tissue. This work will improve our knowledge of the genes, pathways, and molecular mechanisms that contribute to the risk and manifestation of psychiatric disorders.

Completed Grant Support (PI/Co-PI only)

South-seq: DNA sequencing for newborn nurseries in the south

NIH (NHGRI) - UM1HG007301

06/14/2017 - 11/30/2022

Role: Co-Principal Investigator

This project aims to apply whole genome sequencing (WGS) to diagnose neonates with rare disorders, increase participation of individuals from underrepresented racial/ethnic groups in genomics clinical trials, provide educational materials appropriate to diverse audiences, equip non-genetics healthcare providers to return WGS results, assess the impact of WGS testing and results, and engage a broad community to implement safer, more effective, and more equitably distributed genomic medicine.

Building a HudsonAlpha variant catalog from tens of thousands of human genomes

UAB-HudsonAlpha Center for Genomic Medicine and Hugh Kaul Precision Medicine Institute

10/01/2018 – 09/30/2020

Role: Principal Investigator

The goal of this project is to generate a useful catalog of genetic variation found in individuals sequenced at HudsonAlpha so as to better understand human genetic diversity and to improve clinical and research interpretation of genetic variants.

Integrative interpretation of the organismal consequences of non-coding variation

NIH (NCI) - R01CA197139

03/01/2015 - 02/28/2018

Role: Co-Principal Investigator

The goal of this proposal is to dramatically improve our ability to identify and interpret non-coding variants that causally contribute to a variety of human phenotypes. A recently developed computational approach will be substantially improved and evaluated in a variety of genetic studies, and predictions will be experimentally evaluated with several approaches.

Discovery of regulatory variants underlying pediatric neurological disease

SFARI - 385330

1/01/2015 - 10/31/2017

Role: Principal Investigator

In this project we will perform systematic analyses of the Simons Foundation Autism Research Initiative (SFARI) whole genome sequence (WGS) data from 500 children with autism, their parents, and their siblings to identify regulatory variant contributors to autism and related phenotypes like intellectual disability (ID). We will use our own clinical sequencing data from children with similar, overlapping conditions as well as our unique data resources and innovative methods that we have developed to analyze WGS data.

Genomic diagnosis in children with developmental delay

NIH (NHGRI) - UM1HG007301

06/14/2013 - 05/31/2017

Role: Co-Principal Investigator

The goal of this project is to address technological, analytical, and ethical challenges that prevent optimal use of DNA sequencing to improve treatment of diseases and life planning for patients and their families. We are applying next-generation DNA sequencing to meet the diagnostic needs of children with developmental delay, intellectual disability and related health problems.

Publications

1. Hiatt, S. M., Trajkova, S., Sebastiano, M. R., Partridge, E. C., Abidi, F. E., Anderson, A., Ansar, M., Antonarakis, S. E., Azadi, A., Bachmann-Gagescu, R., Bartuli, A., Benech, C., Berkowitz, J. L., Betti, M. J., Brusco, A., Cannon, A., Caron, G., Chen, Y., Cochran, M. E., Coleman, T. F., …[63 additional authors]…, and **Cooper, G.M.**† 2023. Deleterious, protein-altering variants in the transcriptional coregulator ZMYM3 in 27 individuals with a neurodevelopmental delay phenotype. *American Journal of Human Genetics* S0002-9297(22)00541-9. PMID: 36586412.

2. Bowling, K.M., Thompson, M.L., Kelly, M.A., Scollon, S., Slavotinek, A.M., Powell, B.C., Kirmse, B.M., Hendon, L.G., Brothers, K.B., Korf, B.R., **Cooper, G.M**., Greally, J.M., and Hurst, A.C.E.† 2022. Return of non-ACMG recommended incidental genetic findings to pediatric patients: considerations and opportunities from experiences in genomic sequencing. *Genome Medicine* 4(1):131. PMID: 36414972. PMCID: PMC9682742.

3. Bowling, K.M., Thompson, M.L., Finnila, C.R., Hiatt, S.M., Latner, D.R., Amaral, M.D., Lawlor, J.M.J., East, K.M., Cochran, M.E., Greve, V., Kelley, W.V., Gray, D.E., Felker, S.A., Meddaugh, H., Cannon, A., Luedecke, A., Jackson, K.E., Herndon, L.G., Janani, H.M., Johnston, M., Merin, L.A., Deans, S.L., Tuura, C., Williams, H., Laborde, K., Neu, M.B., Patrick-Esteve, J., Hurst, A.C.E., Kandasamy, J., Carlo, W., Brothers, K.B., Kirmse, B.M., Savich, R., Superneau, D., Spedale, S.B., Knight, S.J., Barsh, G.S., Korf, B.R., and **Cooper, G.M.**† 2022. Genome sequencing as a first-line diagnostic test for hospitalized infants. *Genetics in Medicine* 24(4):851-861. PMID: 34930662. PMCID: 8995345.

4. Voisin, N., Schnur, R.E., Douzgou, S., Hiatt, S.M., Rustad, C.F., Brown, N.J., Earl, D.L., Keren, B., Levchenko, O., Geuer, S., Verheyen, S., Johnson, D., Zarate, Y.A., Hančárová, M., Amor, D.J., Bebin, E.M., Blatterer, J., Brusco, A., Cappuccio, G., Charrow, J., Chatron, N., **Cooper, G.M**., Courtin, T., Dadali, E., Delafontaine, J., Del Giudice, E., Doco, M., Douglas, G., Eisenkölbl, A., Funari, T., Giannuzzi, G., Gruber-Sedlmayr, U., Guex, N., Heron, D., Holla, Ø.L., Hurst, A.C.E., Juusola, J., Kronn, D., Lavrov, A., Lee, C., Lorrain, S., Merckoll, E., Mikhaleva, A., Norman, J., Pradervand, S., Prchalová, D., Rhodes, L., Sanders, V.R., Sedláček, Z., Seebacher, H.A., Sellars, E.A., Sirchia, F., Takenouchi, T., Tanaka, A.J., Taska-Tench, H., Tønne, E., Tveten, K., Vitiello, G., Vlčková, M., Uehara, T., Nava, C., Yalcin, B., Kosaki, K., Donnai, D., Mundlos, S., Brunetti-Pierri, N., Chung, W.K., Reymond, A.†, 2021. Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. *American Journal of Human Genetics* 108(5): 857–873. PMID: 33961779. PMCID: 8206167.

5. Hiatt, S.M., Lawlor, J.M.J., Handley, L.H., Ramaker, R.C., Rogers, B.B., Partridge, E.C., Boston, L.B., Williams, M., Plott, C.B., Jenkins, J.., Gray, D.E., Holt, J.M., Bowling, K.M., Bebin, E.M., Grimwood, J., Schmutz, J., and **Cooper, G.M.**† 2021. Long-read genome sequencing for the molecular diagnosis of

neurodevelopmental disorders. *Human Genetics and Genomics Advances* 2(2):100023. PMID: 33937879. PMCID: 8087252.

6. Roberts, B.S., Partridge, E.C., Moyers, B.A., Agarwal, V., Newberry, K.M., Martin, B.K., Shendure, J., Myers, R.M.†, and **Cooper, G.M.†,** 2021. Genome-wide strand asymmetry in massively parallel reporter activity favors genic strands. *Genome Research* 31(5): 866–876. PMID: 33879525. PMCID: 80920006.

7. East, K.M.†, Kelley, W.V., Cannon, A., Cochran, M.E., Moss, I.P., Nakano-Okuno, M., Sodeke, S.O., Edberg, J.C., Cimino, J.J., Fouad, M., Curry, W.A., Hurst, A.C.E., Bowling, K.M., Thompson, M.L., Bebin, E.M., Johnson, R.D., AGHI Consortium, **Cooper, G.M.,** Might, M., Barsh, G.S., and Korf, B.R.† 2021. A state-based approach to genomics for rare disease and population screening. *Genetics in Medicine* 23(4):777-781. PMID: 33244164.

8. Childerhose, J.E., Rich, C., East, K.M., Kelley, W.V., Simmons, S., Finnila, C.R., Bowling, K.M., Amaral, M., Hiatt, S.M., Thompson, M., Gray, D.E., Lawlor, J.M.J, Myers, R.M., Barsh, G.S., Lose, E.J., Bebin, M.E., **Cooper, G.M.**, Brothers, K.B.† 2021. The Therapeutic Odyssey: Positioning Genomic Sequencing in the Search for a Child's Best Possible Life. *AJOB Empir Bioeth*. 12(3):179-189. PMID: 33843487.

9. Bowling, K.M., Thompson, M.L., Gray, D.E., Lawlor, J.M.J., Williams, K., East, K.M., Kelley, W.V., Moss, I.P., Absher, D.M., Partridge, E.C., Hurst, A.C.E., Edberg, J.C., Barsh, G.S., Korf, B.R., and **Cooper, G.M.†** 2021. Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. *Genetics in Medicine* 23(2):280-288. PMID: 32989269.

10. Voskobiynyk, Y.*, Battu, G.*, Felker, S.A., Cochran, J.N., Newton, M.P., Lambert, L.J., Kesterston, R.A., Myers, R.M., **Cooper, G.M.**, Roberson, E.D.†, and Barsh, G.S.† 2021. Aberrant regulation of a poison exon caused by a non-coding variant in a mouse model of *Scn1a*-associated epileptic encephalopathy. *PLoS Genetics* 17(1):e1009195. PMID: 33411788. PMCID: 7790302.

11. Kazamel, M., Lopez, M.A., Bebin, E.M., Bowling, K., Korf, B.R., Barsh, G.S., **Cooper, G.M.,** Hurst, A.C.E., and Ubogu, E.E.† 2020. *Fibulin-5* mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity and scoliosis. *Neurology Genetics* 6(4):e476. PMID: 32802946. PMCID: 7413605.

12. Trivisano, M.*, Ferretti, A.*, Bebin, E.M., Huh, L., Lesca, G., Siekierska, A., Takeguchi, R., Carneiro, M., De Palma, L., Guella, I., Haginoya, K., Ming Shi, R., Kikuchi, A., Kobayashi, T., Jung, J., Lage, L., Milh, M., Mathieu, M., Minassian, B.A., Novelli, A., Pietrafusa, N., Takeshita, E., Tartaglia, M., Terracciano, A., Thompson, M.L., **Cooper, G.M.,** Vigevano, F., Villard, L., Villeneuve, N., Busye, G.M., Demos, M., Scheffer, I.E., and Specchio, N. 2020. Defining the phenotype of *FHF1* developmental and epileptic encephalopathy. *Epilepsia* 7:e71-e78. PMID: 32645220.

13. Drivas, T.G., Li, D., Nair, D., Alaimo, J.T., Alders, M., Altmuller, J., Barakat, T.S., Bebin, E.M., Bertsch, N.L., Blackburn, P.R., Blesson, A., Bouman, A.M., Brockmann, K., Brunelle, P., Burmeister, M., **Cooper, G.M.,** Deneck, J., Dieux-Coeslier, A., Dubbs, H., Ferrer, A., Gal, D., Bartik, L.E., Gunderson, L.B., Hasadri, L., Jain, M., Karimov, C., Keena, B., Klee, E.W., Kloth, K., Lace, B., Macchiaiolo, M., Marcadier, J.L., Milunksy, J.M., Napier, M.P.,Ortiz-Gonzalez, X.R., Pichurin, P.N., Pinner, J., Powis, Z., Prasad, C., Clementina Radio, F., Rasmussen, K.J., Renaud, D.L., Rush, E.T., Saunders, C., Selcen, D., Seman, A.R., Shinde, D.N., Smith, E.D., Smol, T., Snijders Blok, L., Stoler, J.M., Tang, S., Tartaglia, M., Thompson, M.L., van de Kamp, J.M., Wang, J., Weise, D., Weiss, K., Woitschach, R., Wonik, B., Yan, H., Zackai, E.H., Zampino, G., Campeua, P.†, and Bhoj, E.† A second cohort of *CHD3* patients expands the molecular mechanisms known to cause Snijders Block-Campeau syndrome. *European Journal of Human Genetics*. 28(10): 1422-1431. PMID: 32483341. PMCID: 7608102.

14. Cochran, J.N., Geier, E.G., Bonham, L.W., Newberry, J.S., Amaral, M.D., Thompson, M.L., Lasseigne, B.N., Karydas, A.M., Roberson, E.D., **Cooper, G.M.**, Rabinovici, G.D., Miller, B.L., Myers, R.M.,

Yokoyama, J.S.†, Alzheimer's Disease Neuroimaging Initiative. 2020. Non-coding and loss-of-function coding variants in *TET2* are associated with multiple neurodegenerative diseases. *American Journal of Human Genetics* 106(5):632-645. PMID: 32330418.

15. Nambot, S., Faivre, L., Mirzaa, G., Thevenon, J., Bruel, A.L., Mosca-Boidron, A.L., Masurel-Paulet, A., Goldenberg, A., Le Meur, N., Charallais, A., Mignot, C., Petif, F., Rossi, M., Metreau, J., Layet, V., Amram, D., Boute-Benejean, O., Bhoj, E., Cousin, M.A., Kruisselbrink, T.M., Lanpher, B.C., Klee, E.W., Fiala, E., Grange, D.K., Meschino, W.S., Hiatt, S.M. **Cooper, G.M.**, Olivie, H., Smith, W.E., Dumas, M., Lehman, A., CAUSES Study, Inglese, C., Nizon, M., Guerrini, R., Vetro, A., Kaplan, E.S., Miramar, D., Van Gils, J., Fergelot, P., Bodamer, O., Herkert, J.C., Pajusalu, S., Ounap, K., Filiano, J.J., Smol, T., Piton, A., Gerard, B., Chantot-Bastaraud, S., Bienvenu, T., Li, D., Juusola, J., Devriendt, K., Bilan, F., Poe, C., Chevarin, M., Jouan, T., Tisserant, E., Riviere, J.B., Tran Mau-Them, F., Philippe, C., Duffourd, Y., Dobyns, W.B., Hevner, R., and Thauvin-Robinet, C.† 2020. De novo *TBR1* variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *European Journal of Human Genetics*. PMID: 32005960.

16. Scott, T.M., Guo, H., Eichler, E.E., Rosenfeld, J.A., Pang, K., Liu, Z., Lalani, S., Bi, W., Yang, Y., Bacino, C.A., Streff, H., Lewis, A.M., Koenig, M.K., Thiffault, I., Bellomo, A., Everman, D.B., Jones, J.R., Stevenson, R.E., Bernier, R., Gilissen, C., Pfundt, R., Hiatt, S.M., **Cooper, G.M.**, Holder, J.L., and Scott, D.A.† 2020. *BAZ2B* haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. *Human Mutation* 41(5): 921-925. PMID: 31999386.

17. Cochran, J.N.†, McKinley, E.C., Cochran, M., Amaral, M.D., Moyers, B.A., Lasseigne, B.N., Gray, D.E., Lawlor, J.M.J, Prokop, J.W., Geier, E.G., Holt, J.M., Thompson, M.L., Newberry, J.S., Yokoyama, J.S., Worthey, E.A., Geldmacher, D.S., Love, M.N., **Cooper, G.M.**, Myers, R.M., and Roberson, E.D. 2019. Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. *Cold Spring Harbor Molecular Case Studies* 13:5(6):pii:a003491. PMID: 31836585. PMCID: 6913143.

18. Dean, M.†, Messiaen, L., **Cooper, G.M.**, Amaral, M.D., Rashid, S., Korf, B.R., and Standaert, D.G. 2019. Child Neurology: Spastic parapesis and dystonia with a novel *ADCY5* mutation. *Neurology* 93(11): 510-514. PMID: 31501304. PMCID: 64746208.

19. LeDuc, D., Giulivi, C., Hiatt, S.M., Napoli, E., Panoutsopoulos, A., Harlan De Crescenzo, A., Kotzaeridou, U., Syrbe, S., Anagnostou, E., Azage, M., Bend, R., Begtrup, A., Brown, N.J., Buttner, B., Cho, M.T., **Cooper, G.M.,** Doering, J.H., Dubourg, C., Everman, D.B., Hildebrand, M.S., Santos, F.J.R., Kellam, B., Keller-Ramey, J., Lemke, J.R., Liu, S., Niyazov, D., Payne, K., Person, R., Quelin, C., Schnur, R.E., Smith, B.T., Strober, J., Walker, S., Wallis, M., Walsh, L., Yang, S., Yuen, R.K.C., Zielger, A., Sticht, H., Pride, M.C., Orsoco, L., Martinez-Cerdeno, V., Silverman, J.L., Crawley, J.N., Scherer, S.W., Zarbalis, K.S.†, and Jamra, R.† 2019. Pathogenic *WDFY3* variants cause neurodevelopmental disorders and opposing effects on brain size. *Brain* 142(9): 2617-2630. PMID: 31327001. PMCID: 6736092.

20. Bell, S.*, Roussau, J.*, Peng, H., Aouabed, Z., Priam, P., Theroux, J.F., Jefri, M., Tanti, A., Wu, H., Kolobova, I., Silviera, H., Manzano-Vargas, K., Ehresmann, S., Hadman, F.F., Hettige, N., Zhang, X., Nassif, C., Gonzalez, L.G., Sebastian, J., Vockley, J., Begrup, A.G., Wentzensen, I.M., Crunk, A., Nicholls, R.D., Herman, K.C., Deignan, J., Al-Hetani, W., Efthymiou, S., Salpietro, V., Miyake, N., Makita, Y., Matsumoto, N., Au, M., Ostern, R., Houge, G., Hafstrom, M., Fassi, E., Houlden, H., Wassink-Ruiter, J.S.K., Nelson, D., Goldstein, A., Dabir, T., van Gils , J., Bourgeron, T., Delorme, R., **Cooper, G.M.,** Martinez, J.E., Finnila, C.R., Carmant, L., Lortie, A., Oegema, R., van de Gassen, K., Mehta, S.G., Huhle, D., Jamra, R.A., Martin, S., Brunner, H., Lindhout, D., Au, M., Graham, J.M., Turecki, G., Gravel, S., Mechawar, N., Rossignol, E., Michaud, J.L., Lessard, J., Ernst, C.†, and Campeau, P.M.† 2019. Mutations in *ACTL6B* cause neurodevelopmental deficits and epilepsy and lead to loss of dendrites in human neurons. *American Journal of Human Genetics* 104(5): 815-834. PMID: 31031012. PMCID: 6507050.

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23. Popov, I.K., Hiatt, S.M.†, Whalen, S., Keren, B., Ruivenkamp, C., van Haeringen, A., Chen, M., **Cooper, G.M.,** Korf, B.R., and Chang, C.† 2019. A YWHAZ variant associated with cardiofaciocutaneous syndrome actives the RAF-ERK pathway. *Frontiers in Physiology* 10:388. PMID: 31024343. PMCID: 6465419.

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