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.

21. Zweier, M., Begemann, A., McWalter, K., Cho, M.T., Abela, L., Banka, S., Behring, B., Brown, C., Chen, J., **Cooper, G.M.,** DDD Study, Finnila, C.R., Sacoto, M.J.G., Henderson, A., Huffmeier, U., Kerr, B., McDermott, J.H., Meltzer, M.R., Monaghan, K.G., Mostafavi, R., Ounap, K., Plecko, B., Powis, Z., Purcarin, G., Reimand, T., Riedhammer, K.M., Schreiber, J.M., Sirsi, D., Wierenga, K.J., Wojcik, M.H., Papuc, S.M., Steindl, K., Sticht, H., and Rauch, A.† 2019. Spatially clustering de novo variants in *CYFIP2*, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. *European Journal of Human Genetics* 27(5): 747-759. PMID: 30664714. PMCID: 6461771.

22. Hiatt, S.M., Thompson, M.L., Prokop, J.W., Lawlor, J.M.J., Gray, D.E., Bebin, E.M., Rinne, T., Kempers, M., Mignot, C., Nava, C., Depienne, C., Pfundt, R., van Bon, B., Kalsner, L., Rauch, A., Joset, P., Bachmann-Gagescu, R., Wentzensen, I.M., McWalter, K., and **Cooper, G.M.†** 2019. Deleterious variation in *BRSK2* associates with a neurodevelopmental disorder. *American Journal of Human Genetics* 104(4): 701-708. PMID: 30879638. PMCID: 6451696.

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.

24. Carapito, R., Ivanova, E.L., Morlon, A., Meng, L., Molitor, A., Erdmann, E., Kieffer, B., Pichot, A., Naegely, L., Kolmer, A., Paul, N., Hanauer, A., Dutreux, F., Tran-Mau-Them, F., Jean-Marcais, N., Hiatt, S.M., **Cooper, G.M.,** Tvrdik, T., Garde, A., Thauvin-Robinet, C., Wang, X., Phillips, M., Crawford, H.P., Kukolich, M.K., Hunt, D., Harrison, V., Smigiel, R., Gold, N., Hung, C.Y., Viskochil, D.H., Dugan, S.L., Bayrak-Toydemir, P., Kharbanda, M., Guerrot, A.M., Schluth-Bolard, C., Cho, M., Wentzensen, I.M., McWalter, K., Schnur, R.E., Liu, S., Lewis, A.M., Lalani, S., Mensah-Bonsu, N., Ceraline, J., Sun, Z., Ploski, R., Bacino, C.A., Faivre, L., Bodamer, O., Chelly, J., Isidor, B., and Bahram, S.† 2019. ZMIZ1 variants cause a syndromic neurodevelopmental disorder. *American Journal of Human Genetics* 104(2): 319-330. PMID: 30639322. PMCID: 6369415.

25. Rentzsch, P., Witten, D., **Cooper, G.M.,** Shendure, J.†, and Kircher, M.† 2019. CADD: Predicting the deleteriousness of variants throughout the human genome. *Nucleic Acids Research* 47(D1): D886-D894. PMID: 30371827. PMCID: 6323892.

26. Carvill, G.L.*, Engel, K.L.*, Ramamurthy, A., Cochran, J.N., Roovers, J., Stamberger, H., Lim, N., Schneider, A.L., Hollingsworth, G., Holder, D.H., Regan, B.M., Lagae, L., Ceulemans, B., Bebin, E.M., Nguyen, J., EuroEPINOMICS Rare Epilepsy Syndrome Myclonic-Astatic Epilepsy and Dravet Working Group, Barsh, G.S., Weckhuysen, S., Meisler, M., Berkovic, S.F., De Jonghe, P., Scheffer, I.E., Myers, R.M., **Cooper, G.M.†,** and Mefford, H.C.† 2018. Aberrant inclusion of a poison exon causes Dravet syndrome and related *SCN1A*-associated genetic epilepsies. *American Journal of Human Genetics* 103(6): 1022-1029. PMID: 30526861. PMCID: 6288405.

27. Thompson, M.L*, Finnila, C.R.*, Bowling, K.M., Brothers, K.B., Neu, M.B., Amaral, M.D., Hiatt, S.M., East, K.M., Gray, D.E., Lawlor, J., Kelley, W.V., Lose, E.J., Rich, C.A., Simmons, S., Myers, R.M., Barsh, G.S., Bebin, E.M., and **Cooper, G.M.†** 2018. Genomic sequencing identifies secondary findings in a cohort of parent study participants. *Genetics in Medicine* 20(12): 1635-1643. PMID: 29790872. PMCID: 6185813.

28. Claasen, D., Boals, M., Bowling, K.M., **Cooper, G.M.,** Cox, J., Hershfield, M., Lewis, S., Wlodarski, M., Weiss, M.J., and Estepp, J.H. 2018. Complexities of genetic diagnosis illustrated by an atypical case of congenital hypoplastic anemia. *Molecular Case Studies* 4(6): a003384. PMID: 30559313. PMCID: 6318771.

29. Tripathy, R., Leca, I., van Dijk, T., Weiss, J., van Bon, B., Gstrein, T., Breuss, M., Guoling, T., Bahi-Buisson, N., Paciorkowski, A.R., Pagnamenta, A., Lise, S., Taylor, J., Terrone, G., Vitiello, G., Del Giudice, E., Brunetti-Pierri, N., D'Amico, A., Reymond, A., Voisin, N., Bernstein, J.A., Farrelly, E., Kini, U., Leonard, T.A., Armstrong, L., Hiatt, S.M., **Cooper, G.M**., Mirzaa, G., Pierson, T.M., Baas, F., Chelly,

J., Cowan, N.J., and Keays, D.A.† 2018. Mutations in *MAST1* cause mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations. *Neuron* 100(6): 1354-1368. PMID: 30449657. PMCID: 6436622.

30. Hiatt, S.M.*, Neu, M.B.*, Ramaker, R.C., Hardigan, A.A.,Prokop, J.W., Hancarova, M., Prchalova, D., Havlocvicova, M., Prchal, J., Stranecky, V., Yim, D.K.C., Powis, Z., Keren, B., Nava, C., Mignot, C., Rio, M., Revah-Politi, A., Hemati, P., Stong, N., Iglesias, A.D., Suchy, S.F., Willaert, R., Wentzensen, I.M., Wheeler, P.G., Brick, L., Kozenko, M., Hurst, A.C.E., Wheless, J.W., Lacassie, Y., Myers R.M., Barsh, G.S., Sedlacek Z., and **Cooper, G.M.†** 2018. De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. *PLoS Genetics* 14(11): e1007671. PMID: 30500825. PMCID: 6291162.

31. Porter, K.M.†, Kauffman, T.L., Koenig, B.A., Lewis, K.L., Rehm, H.L., Richards, C.S., Strande, N.T., Tabor, H.K., Wolf, S.M., Yang, Y., Amendola, L.M., Azzariti, D.R., Berg, J.S., Bergstrom, K., Biesecker, L.G., Biswas, S., Bowling, K.M., Chung W.K., Clayton, E.W., Conlin, L.K., **Cooper, G.M.**, Dulik, M.C., Garraway, L.A., Ghazani, A.A., Green, R.C., Hiatt, S.M., Jamal, S.M., Jarvik, G.P., Goddard, K.A.B., Wilfond, B.S., and members of the CSER Actionability and Return of Results Working Group. 2018. Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. *Molecular Genetics & Genomic Medicine* 6(6): 898-909. PMID: 30133189. PMCID: 6305639.

32. Mulhern, M., Stumpel, C., Stong, N., Brunner, H., Bier, L., Lippa, N., Riviello, J., Rouhl, R., Kempers, M., Pfundt, R., Stegmann, S., Kukolich, M., Telegrafi, A., Lehman, A., Lopez-Rangel, E., Houcinat, N., Barth, M., den Hollander, N., Hoffer, M.J.V., Weckhuysen, S., Roovers, J., Djemie, T., Barca, D., Ceulemans, B., Craiu, D., Lemke, J., Korff, C., Mefford, H., Myers, C., Siegler, Z., Hiatt, S.M., **Cooper, G.M.,** Bebin, E.M., Snijders-Blok, L., Veenstra-Knol, H., Baugh, E., Brilstra, E., Volker-Touw, C., van Binsbergen, E., Revah-Politi, A., Pereira, E., McBrian, D., Pacault, M., Isidor, B., Le Caignec, C., Gilbert-Dussardier, Br., Bilan, F., Heinzen, E., Goldstein, D., Stevens, S., and Sands, T. *NBEA*: developmental disease gene with myoclonic-astatic epilepsy-like phenotypes. 2018. *Annals of Neurology* 84(5): 788-795. PMID: 30269351. PMCID: 6249120.

33. Sanghvi, R.V., Buhay, C.J., Powell, B.C., Tsai, E.A., Dorschner, M.O., Hong, C.S., Lebo, M.S., Sasson, A., Hanna, D.S., McGee, S., Bowling, K.M., **Cooper, G.M.**, Gray, D.E., Lonigro, R.J., Dunford, A., Brennan, C.A., Cibulskis, C., Walker, D., Carneiro, M.O., Salisbery, J., Hindorff, L.A., Robinson, D.A., Santani, A., Sarmady, M., Rehm, H.L., Biesecker, L.G., Nickerson, D.A., Hutter, C.M., Garraway, L., Muzny, D.M.†, and Wagle, N.† 2018. Characterizing reduced coverage regions through comparisons of exome and genome sequencing data across 10 centers. *Genetics in Medicine* 20(8): 855-866. PMID: 29144510.

34. Hiatt, S.M.*, Amaral, M.D.*, Bowling, K.M., Finnila, C.R., Thompson, M.L, Gray, D.E., Lawlor, J., Cochran, M., Bebin, E.M., Brothers, K.B., East, K.M., Kelley, W.V., Lamb, N.E., Levy, S.E., Lose, E.J., Neu, M.B., Rich, C.A., Simmons, S., Myers, R.M., Barsh, G.S., and **Cooper, G.M.†** Systematic reanalysis of genomic data improves quality of variant interpretation. 2018. *Clinical Genetics* 94(1): 174-178. PMID: 29652076. PMCID: 5995667.

35. Hart, M.R.†, Biesecker, B.B., Blout, C.L., Christensen, K.D., Amendola, L.M., Bergstrom, K.L., Biswas, S., Bowling, K.M., Brothers, K.B., Conlin, L.K., **Cooper, G.M.,** Dulik, M.C., East, K.M., Everett, J.N., Finnila, C.R., Ghazani, A.A., Gilmore, M.J., Goddard, K.A.B., Jarvik, G.P., Johnston, J.J., Kauffman, T.L., Kelley, W.V., Krier, J.B., Lewis, K.L., McGuire, A.L., McMullen, C., Ou, J., Plon, S.E., Rehm, H., Richards, C.S., Romasko, E.J., Sagardia, A.M., Spinner, N.B., Thompson, M.L., Turbitt, E., Vassy, J.L., Wilfond, B.S., Veenstra, D.L.*, Berg, J.S.*, Green, R.C.*, Biesecker, L.G.*, and Hindorff, L.A.*† Secondary findings from clinical genomic sequencing: Prevalence, patient perspectives, family history assessment, and healthcare costs from a multi-site study. *Genetics in Medicine* 21(5): 1100-1110. PMID: 30287922. PMCID: 6450774.

36. Snijders-Blok, L.*, Hiatt, S.M.*, Bowling, K.M., Prokop, J.W., Engel, K.L., Cochran, J.N., Bebin, M.E., Bijlsma, E.K., Ruivenkamp, C.A., Terhal, P., Simon, M.E.H., Smith, R., Hurst, J.A., The DDD Study, McLaughlin, H., Person, R., Crunk, A., Wangler, M.F., Streff, H., Symonds, J.D., Zuberi, S.M., Elliott, K.S., Sanders, V.R., Masunga, A., Hopkin, R.J., Dubbs, H.A., Ortiz-Gonzalez, X.R., Pfundt, R., Brunner, H.G., Fisher, S.E., Kleefstra, T.†, and **Cooper, G.M.**† De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. 2018. *Human Genetics* 137(5): 375-388. PMID: 29740699. PMCID: 5973976.

37. Cheng, H.*, Dharmadhikari, A.V.*, Kleyner, R., Varland, S., Rope, A.F., Yoon, M., Stray-Pederson, A., Posey, J.E., Crews, S., Eldomery, M., Akdemir, Z.C., Lewis, A.M., Sutton, V.R., Conboy, E., Agre, K., Xia, F., Walkiewicz, M., Longoni, M., High, F.A., van Slegtenhorst, M.A., Mancini, G.M.S., Finnila, C.R., van Haeringen, A., den Hollander, N., Ruivenkamp, C., Naidu, S., Desai, S., Palmer, E.E., Murray, L., Lim, D., Jayakar, P., Parker, M.J., Giusto, S., Stracuzzi, E., Romano, C., Beighley, J.S., Bernier, R.A., Kury, S., Mathilde, N., Corbett, M.A., Barnett, C., Shaw, M., Armstrong, R., Kassahn, K.S., Van Dijck, A., Vandeweyer, G., Rojas, S.K., Boycott, K.M., Person, R., Willaert, R., Eichler, E.E., Kooy, R.R., Yang, Y., Lupski, J.R., Arnesen, T., **Cooper, G.M.,** Chung, W.K., Gecz, J., Stessman, H.F.†, Meng, L.†, and Lyon, G.J.† 2018. Truncating variants in *NAA15* are associated with variable levels of intellectual disability, autism spectrum disorder, and congenital anomalies. *American Journal of Human Genetics* 102(5): 985-994. PMID: 29656860. PMCID: 5932113.

38. Roberts, B.S., Hardigan, A.A., Moore, D.E., Ramaker, R.C., Jones, A.L., Fitz-Gerald, M.B., **Cooper, G.M.,** Wilcox, C.M., Kimberly, R.P., and Myers, R.M.† 2018. Discovery and validation of circulating biomarkers of colorectal adenoma by high-depth small RNA sequencing. *Clinical Cancer Research* 24(9): 2092-2099. PMID: 29490987.

39. Ramaker, R.C., Savic, D., Hardigan, A.A., Newberry, K., **Cooper, G.M**., Myers, R.M.†, and Cooper, S.J.† 2017. A genome-wide interactome of DNA-associated proteins in the human liver. *Genome Research* 27(11): 1950-1960. PMID: 29021291. PMCID: 5668951.

40. Bowling, K.M., Thompson, M.L., Amaral, M.D., Finnila, C.R., Hiatt, S.M., Engel, K.L., Cochran, N.J., Brothers, K.B., East, K.M., Gray, D.E., Kelley, W.V., Lamb, N.E., Lose, E.J., Rich, C.A., Simmons, S., Whittle, J.S., Weaver, B.T., Nesmith, A.S., Myers, R.M., Barsh, G.S., and **Cooper, G.M.**† 2017. Genomic diagnosis for children with intellectual disability and/or developmental delay. *Genome Medicine* 9(1): 43. PMID: 28554332. PMCID: 5448144.

41. Blanchet, P., Bebin, M., Bruet, S., **Cooper, G.M.**, Thompson, M.L., Duban-Bedu, B., Gerard, B., Piton, A., Suckno, S., Desphande, C., Clowes, V., Vogt, J., Turnpenny, P., Wiliamson, M.P., Alembik, Y., Clinical Sequencing Exploratory Research Consortium, Deciphering Developmental Disorders Consortium, Glasgow, E.†, and McNeill, A.† 2017. MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. *PLoS Genetics* 13(8): e1006957. PMID: 28859103. PMCID: 5597252.

42. O'Daniel, J.M., McLaughlin, H.M., Amendola, L.M., Bale, S.J., Berg, J.S., Bick, D., Bowling, K.M., Chung, W.K., Conlin, L.K., **Cooper, G.M**., Das, S., Deignan, J.L., Dorschner, M.O., Evans, J.P., Ghazani, A.A., Goddard, K.A., Gornick, M., Farwell Hagman, K.D., Hambuch, T., Hegde, M., Knight Johnson, A., Mighion, L., Morra, M., Plon, S.E., Punj, S., Richards, C.S., Santani, A., Shirts, B.H., Spinner, N.B., Tang, S., Weck, K.E., Wolf, S.M., Yang, Y., and Rehm, H.L.† 2017. A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. *Genetics in Medicine* 19(5): 575-582. PMID: 27811861. PMCID: 5415437.

43. Harms, F.L.*, Girish, K.M.*, Hardigan, A.A.*, Kortüm, F., Shukla, A., Alawi, M., Dala, A., Brady, L., Tarnopolsky, M., Bird, L.M., Ceulemans, S., Bebin, E.M., Bowling, K.M., Hiatt, S.M., Lose, E.J., Primiano, M., Chung, W.K., Juusola, J., Akdemir, Z.C., Bainbridge, M., Charng, W., Drummond-Borg, M., Eldomery, M.K., El-Hattab, A.W., Saleh, M.A.M, Bézieau, S., Cogné, B., Isidor, B., Küry, S., Lupski, J.R., Myers, R.M., **Cooper, G.M.†,** and Kutsche, K.† 2017. Mutations in *EBF3* disturb transcriptional

profiles and cause intellectual disability, ataxia, and facial dysmorphism. *American Journal of Human Genetics* 100(1): 117-127. PMID: 28017373. PMCID: 5223027.

44. Brothers, K.B.†, East, K.M., Kelley, W.V., Wright, F., Westbrook, M.J., Rich, C.A., Bowling, K.M., Lose, E.J., Bebin, E.M., Simmons, S., Myers, J.A., Barsh, G.S., Myers, R.M., **Cooper, G.M**., Pulley, J.M., Rothstein, M.A., and Wright, E. 2017. Eliciting preference on secondary findings: the Preferences Instrument for Genomic Secondary Results. *Genetics in Medicine* 19(3): 337-344. PMID: 27561086. PMCID: 5326612.

45. Inoue, F., Kircher, M., Martin, B., **Cooper, G.M.**, Witten, D.M., McManus, M.T., Ahituv, N., and Shendure, J.† 2017. A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. *Genome Research* 27(1): 38-52. PMID: 27831498. PMCID: 5204343.

46. Guella, I., Huh, L., McKenzie, M.B., Toyota, E.B., Bebin, E.M., Thompson, M.L., **Cooper, G.M**., Evans, D.M., Buerki, S.E., Adam, S., Van Allen, M.I., Nelson, T.N., Connolly, M.B., Farrer, M.J., and Demos, M.† 2016. De-novo FGF12 mutation in two patients with neonatal onset epilepsy. *Neurology: Genetics* 2(6): e120. PMID: 27872899. PMCID: 5113095.

47. Møller, R.S., Weckhuysen, S., Chipaux, M., Marsan, E., Taly, V., Bebin, E.M., Hiatt, S., Prokop, J.W., Bowling, K.M., Mei, D., Conti, V., de la Grange, P., Ferrand-Sorbets, S., Dorfmüller, G., Lambrecq, V., Larsen, L., Leguern, E., Guerrini, R., Rubboli, G., **Cooper, G.M**., and Baulac, S.† 2016. Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. *Neurology: Genetics* 2(6): e118. PMID: 27830187. PMCID: 5089441.

48. Ng, B.G., Shiryaev, S.A., Rymen, D., Eklund, E.A., Raymond, K., Kircher, M., Abdenur, J.E., Alehan, F., Midro, A.T., Bamshad, M.J., Barone, R., Berry, G.T., Brumbaugh, J.E., Buckingham, K.J., Clarkson, K., Cole, F.S., O'Connor, S., **Cooper, G.M.**, Van Coster, R., Demmer, L.A., Diogo, L., Fay, A.J., Ficicioglu, C., Fiumara, A., Gahl, W.A., Ganetzky, R., Goel, H., Harshman, L.A., He, M., Jaeken, J., James, P.M., Katz, D., Keldermans, L., Kibaek, M., Kornberg, A.J., Lachlan, K., Lam, C., Yaplito-Lee, J., Nickerson, D.A., Peters, H.L., Race, V., Régal, L., Rush, J.S., Rutledge, S.L., Shendure, J., Souche, E., Sparks, S.E., Trapane, P., Sanchez-Valle, A., Vilain, E., Vøllo, A., Waechter, C.J., Wang, R.Y., Wolfe, L.A., Wong, D.A., Wood, T., Yang, A.C., University of Washington Center for Mendelian Genomics, Matthijs, G., and Freeze, H.H.† 2016. ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Human Mutation* 37(7): 653-60. PMID: 26931382. PMCID:4907823.

49. Amendola, L.M., Jarvik, G.P., Leo, M.C., McLaughlin, H.M., Akkari, Y., Amaral, M.D., Berg, J.S., Biswas, S., Bowling, K.M., Conlin, L.K., **Cooper, G.M**., Dorschner, M.O., Dulik, M.C., Ghazani, A.A., Ghosh, R., Green, R.C., Hart, R., Horton, C., Johnston, J.J., Lebo, M.S., Milosavljevic, A., Ou, J., Pak, C.M., Patel, R.Y., Punj, S., Richards, C.S., Salama, J., Strande, N.T., Yang, Y., Plon, S.E., Biesecker, L.G., and Rehm, H.L.† 2016. Performance of ACMG-AMP variant-interpretation guidelines among nine laboratories in the Clinical Sequencing Exploratory Research Consortium. *American Journal of Human Genetics* 98(6): 1067-1076. PMID: 27181684. PMCID: 4908185.

50. Savic D., Roberts B.S., Carleton J.B., Partridge E.C., White M.A., Cohen B.A., **Cooper G.M.**, Gertz J., and Myers R.M.† 2015. Promoter-distal RNA polymerase II binding discriminates active from inactive CCAAT/enhancer-binding protein beta binding sites. *Genome Research* 25(12): 1791-1800. PMID: 26486725. PMCID: 4665001.

51. Cirulli, E.T.*, Lasseigne, B.N.*, Petrovski, S., Sapp, P.C., Dion, P.A., Leblond, C.S., Couthouis, J., Lu, Y.F., Wang, Q., Krueger, B.J., Ren, Z., Keebler, J., Han, Y., Levy, S.E., Boone, B.E., Wimbish, J.R., Waite, L.L., Jones, A.L., Carulli, J.P., Day-Williams, A.G., Staropoli, J.F., Xin, W.W., Chesi, A., Raphael, A.R., McKenna-Yasek, D., Cady, J., Vianney de Jon, J.M.B., Kenna, K.P., Smith, B.N., Topp, S., Miller, J., Gkazi, A., FALS Sequencing Consortium, Al-Chalabi, A., van den Berg, L.H., Veldink, J., Silani, V., Ticozzi, N., Shaw, C.E., Baloh, R.H., Appel, S., Simpson, E., Lagier-Tourenne, C., Pulst, S.M., Gibson, S., Trojanowski, J.Q., Elman, L., McCluskey, L., Grossman, M., Shneider, N.A., Chung,

W.K., Ravits, J.M., Glass, J.D., Sims, K.B., Van Deerlin, V.M., Maniatis, T., Hayes, S.D., Ordureau, A., Swarup, S., Landers, J., Baas, F., Allen, A.S., Bedlack, R.S., Harper, J.W., Gitler, A.D., Rouleau, G.A., Brown, R., Harms, M.B., **Cooper, G.M.**, Harris, T.†, Myers, R.M.†, and Goldstein, D.B.†. 2015. Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. *Science* 347(6229): 1436-1441. PMID: 25700176. PMCID: 4437632.

52. Amendola, L.M., Dorschner, M.O., Robertson, P.D., Salama, J.S., Hart, R., Shirts, B.H., Murray, M.L., Tokita, M.J., Gallego, C.J., Kim, D.S., Bennett, J.T., Crosslin, D.R., Ranchalis, J., Jones, K.L., Rosenthal, E.A., Jarvik, E.R., Itsara, A., Turner, E.H., Herman, D.S., Schleit, J., Burt, A., Jamal, S.M., Abrudan, J.L., Johnson, A.D., Conlin, L.K., Dulik, M.C., Santani, A., Metterville, D.R., Kelly, M., Foreman, A.K., Lee, K., Taylor, K.D., Guo, X., Crooks, K., Kiedrowski, L.A., Raffel, L.J., Gordon, O., Machini, K., Desnick, R.J., Biesecker, L.G., Lubitz, S.A., Mulchandani, S., **Cooper, G.M.**, Joffe, S., Richards, C.S., Yang, Y., Rotter, J.I., Rich, S.S., O'Donnell, C.J., Berg, J.S., Spinner, N.B., Evans, J.P., Fullerton, S.M., Leppig, K.A., Bennett, R.L., Bird, T., Sybert, V.P., Grady, W.M., Tabor, H.K., Kim, J.H., Bamshad, M.J., Wilfond, B., Motulsky, A.G., Scott, C.R., Pritchard, C.C., Walsh, T.D., Burke, W., Raskind, W.H., Byers, P., Hisama, F.M., Rehm, H., Nickerson, D.A., and Jarvik, G.P. 2015. Actionable exomic incidental findings in 6503 participants: challenges of variant classification. *Genome Research* 25(3): 305-315. PMID: 25637381. PMCID: 4352885.

53. 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(3): 310-315. PMID: 24487276. PMCID: 3992975.

54. 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.

55. 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(1): 30. PMID: 24279905. PMCID: 3848595.

56. 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.

57. 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.

58. 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.

59. **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.

60. 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.

61. 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.

62. 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.

63. **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. 2010. *Nature Methods* 7(4): 250-251. PMID: 20354513. PMCID: 3145250.

64. 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.

65. 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.

66. 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.

67. 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.

68. 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.

69. **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.

70. **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.

71. 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.

72. 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(7191): 56-64. PMID: 18451855. PMCID: 2424287.

73. 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(5): 1-9. PMID: 18439552. PMCID: 2427318.

74. 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.

75. 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.

76. 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.

77. 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.

78. 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.

79. 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.

80. 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.

81. 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.

82. 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.

83. **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.

84. **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.

85. 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.

86. 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.

87. **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.

88. 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.

89. **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.

90. 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, Opinions, and Book Chapters

91. Bowling, K.M., Thompson, M.L., and **Cooper, G.M.**† 2020. How secondary findings are made. *Secondary Findings in Genomic Research* p59. Academic Press.

92. Neu, M.B., Bowling, K.M., and **Cooper, G.M.**[†] 2019. Clinical utility of genomic sequencing. *Current Opinion in Pediatrics* 31(6): 732-738. PMID: 31693580.

93. Barsh, G.S.[†], **Cooper, G.M.**, Copenhaver, G.P.[†], Sirugo, G., Tang, H., and Williams, S.M. 2019. Evaluating the strength of genetic results: Risks and responsibilities. *PLoS Genetics* 15(10): e1008437. PMID: 31603891. PMCID: 6788678.

94. Amendola, L., Berg, J.S., Horowitz, C.R., Angelo, F., Bensen, J.T., Biesecker, B.B., Biesecker, L.G., **Cooper, G.M.,** East, K.M., Filipski, K., Fullerton, S.M., Gelb, B.D., Goddard, K.A.B., Hailu, B., Hart, R., Lich, K.H., Joseph, G., Kenny, E.E., Koenig, B.A., Knight, S.J., Kwok, P.Y., Lewis, K.L., McGuire, A.L., Norton, M.E., Ou, J., Parsons, D.W., Powell, B.C., Risch, N., Robinson, M., Rini, C., Scollon, S., Slavotinek, A.M., Veenstra, D.L., Wasserstein, M.P., Wilfond, B.S., Hindorff, L.A., Plon, S.E., and Jarvik, G.P. [†] 2018. The Clinical Sequencing Evidence-Generating Research (CSER) Consortium: Integrating genomic sequencing in diverse and medically underserved populations. *American Journal of Human Genetics* 103(3): 319-327. PMID: 30193136. PMCID: 6128306.

95. Manolio, T.A.[†], Fowler, D.M., Starita, L.M., Haendel, M.A., MacArthur, D.G., Biesecker, L.G., Worthy, E., Chisholm, R.L., Green, E.D., Jacob, H.J., McLeod, H.L., Roden, D., Rodriguez, L.L., Williams, M.S., **Cooper, G.M**., Cox, N.J., Herman, G.E., Kingsmore, S., Lo, C., Lutz, C., MacRae, C.A., Nussbaum, R.L., Ordova, J.M., Ramos, E.M., Robinson, P.N., Rubinstein, W.S., Seidman, C., Stranger, B.E., Wang, H., Westerfield, M., and Bult, C. 2017. Bedside back to bench: Building bridges between basic and clinical genomic research. *Cell* 169(1): 6-12. PMID: 28340351. PMCID: 5511379.

96. Green, R.C. [†], Goddard, K.A., Jarvik, G.P., Amendola, L.M., Appelbaum, P.S., Berg, J.S., Bernhardt, B.A., Biesecker, L.G., Biswas, S., Blout, C.L., Bowling, K.M., Brothers, K.B., Burke, W., Caga-Anan, C.F., Chinnaiyan, A.M., Chung, W.K., Clayton, E.W., **Cooper, G.M**., East, K., Evans, J.P., Fullerton, S.M., Garraway, L.A., Garrett, J.R., Gray, S.W., Henderson, G.E., Hindorff, L.A., Holm, I.A., Lewis, M.H., Hutter, C.M., Janne, P.A., Joffe, Ss, Kaufman, D., Knoppers, B.M., Koenig, B.A., Krantz, I.D., Manolio, T.A., McCullough, L., McEwen, J., McGuire, A., Muzny, D., Myers, R.M., Nickerson, D.A., Ou ,J., Parsons, D.W., Petersen, G.M., Plon, S.E., Rehm, H.L., Roberts, J.S., Robinson, D., Salama, J.S., Scollon, S., Sharp, R.R., Shirts, B., Spinner, N.B., Tabor, H.K., Tarczy-Hornoch, P., Veenstra, D.L., Wagle, N., Weck, K., Wilfond, B.S., Wilhelmsen, K., Wolf, S.M., Wynn, J., Yu, J.H., and CSER Consortium. 2016. Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics* 98(6): 1051-1066. PMID: 27181682. PMCID: 4908179.

97. Barsh, G.S.[†], **Cooper, G.M.,** Copenhaver, G.P., Gibson, G., McCarthy, M.I., Tang, H., and Williams, S. 2015. *PLoS Genetics* Data Sharing Policy: In pursuit of functional utility. *PLoS Genetics* 11(12): e1005716. PMID: 26655768. PMCID: 4678900.

98. **Cooper, G.M.**[†] 2015. Parlez-vous VUS? *Genome Research* 25(10): 1423-1426. PMID: 26430151. PMCID: 4579326.

99. MacArthur, D.G., Manolio, T.A., Dimmock, D.P., Rehm, H.L., Shendure, J., Abecasis, G.R., Adams, D.R., Altman, R.B., Antonarakis, S.E., Ashley, E.A., Barrett, J.C., Biesecker, L.G., Conrad, D.F., **Cooper, G.M**., Cox, N.J., Daly, M.J., Gerstein, M.B., Goldstein, D.B., Hirschorn, J.N., Leal, L.A., Pennacchio, L.A., Stamatoyannopoulos, J.A., Sunyaev, S.R., Valle, D., Voight, B.F, Winckler, W., and Gunter, C. 2014. Guidelines for investigating causality of sequence variants in human disease. *Nature* 508(7497): 469-476. PMID: 24759409. PMCID: 4180223.

100. **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.

101. **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.

102. **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.

103. **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.

104. 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.

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

106. **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.

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