

CURRICULUM VITAE

Name: Douglas Michael Ruderfer
Work Address: 515B Light Hall
Division of Genetic Medicine
Department of Medicine
Vanderbilt University Medical Center
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EDUCATION

2000-2004 Johns Hopkins University, Baltimore, MD. B.S. in Computer Engineering
2002-2004 Johns Hopkins University, Baltimore, MD. M.S. in Computer Science
2010-2013 Cardiff University, Cardiff, UK. Ph.D. in Genetics
Thesis: Inferring Schizophrenia Biology from Genome-wide Data

ACADEMIC APPOINTMENTS

2014-2016 Instructor, Department of Psychiatry, Icahn School of Medicine at Mount Sinai, New York, NY
2016-present Assistant Professor, Departments of Medicine, Biomedical Informatics and Psychiatry, Vanderbilt University Medical Center

EMPLOYMENT

2005-2007 Bioinformatics Specialist, Lewis-Sigler Institute for Integrative Genomics, Princeton University
2007-2011 Research Affiliate, Stanley Center for Psychiatric Research, Broad Institute
2007-2011 Bioinformatics Specialist, Center for Human Genetic Research, Massachusetts General Hospital
2011-2016 Bioinformatician III, Department of Psychiatry, Icahn Institute of Genomics and Multiscale, Icahn School of Medicine at Mount Sinai

PROFESSIONAL ORGANIZATIONS

American College of Neuropsychopharmacology (Associate Member since 2017)
International Society of Psychiatric Genetics
American Society of Human Genetics
Molecular Psychiatry Association

PROFESSIONAL ACTIVITIES

Intramural service:

2017 Chair, Vanderbilt Genomics Institute (VGI) Annual Symposium
2017-2019 Interdisciplinary Graduate Program (IGP) Admissions Committee
2017-2019 Co-organizer, Vanderbilt Genetics Institute (VGI) Weekly Seminar Series
2018-2019 Faculty Search Committee, Vanderbilt Genomics Institute (VGI)
2019-2019 Human Genetics Direct Admit Exploration Committee
2019-2019 BioVU Genetic Taskforce

Extramural service:

2017 Co-Chair, Genes in the Context of the Medical phenotype: The challenges, opportunities, and Pitfalls of Psychiatric Genetics Using Biobanks and Registries, World Congress of Psychiatric Genetics (WCPG)
2018-2020 Program Committee, American College of Neuropsychopharmacology (ACNP)
2018 Grant Review Panel, Simons Foundation Autism Research Initiative (SFARI)
2018 Co-Chair, Somatic Mosaicism in the Brain: Diverse Neuronal Genomics and Neuropsychiatric Disorders, Molecular Psychiatry Meeting

Peer reviewer:

Cell, Nature Genetics, Nature Neuroscience, JAMA Neurology, Molecular Psychiatry, JAMA Psychiatry, PLOS Genetics, Biological Psychiatry, Bioinformatics, Pharmacogenomics, Bipolar Disorders, Journal of Affected Disorders

HONORS AND AWARDS

- 2018 Leading Research Achievements by Foundation Grantees, Brain & Behavior Research Foundation
- 2018 Vanderbilt University Medical Center Biomedical Science Impact Award
- 2015 American College of Neuropsychopharmacology Travel Award
- 2015 UK Alumni Award Finalist, Professional Achievement
- 2014 Early Career Investigator Program, World Congress of Psychiatric Genetics, Copenhagen, Denmark
- 2014 Best Oral Presentation Award, World Congress of Psychiatric Genetics, Copenhagen, Denmark

TEACHING**Graduate Students:**

- 2017- Course Director and Primary Lecturer, Introduction to Behavioral Genetics, Interdisciplinary Graduate Program
- 2017 Guest Lecturer, Human Genetics Module II, Interdisciplinary Graduate Program

Psychiatry Residents:

- 2017- Guest Lecturer, Psychiatry Residents, Department of Psychiatry

Extramural:

- 2016 Faculty, Summer School for Computation Genomics, Icahn School of Medicine at Mount Sinai, New York, NY
- 2015 Faculty, International Workshop on Statistical Genetic Methods for Human Complex Traits, Institute for Behavioral Genetics, University of Colorado Boulder, Boulder, CO
- 2014 Teaching Assistant, Mount Sinai Institute of Technology, Icahn School of Medicine at Mount Sinai, New York, NY. Design, Technology and Entrepreneurship Makers Studio II: Pharmacogenetics

Research Supervision:**Thesis students:**

- Alexander Charney (Ph.D. Genetics graduated 2017)
- Jooeun “Jay” Kang (MSTP, started 2018)
- Tess Vessels (Human Genetics, started 2019)

Research Staff:

- Elizabeth Webster (Junior Bioinformatics Specialist, 2012-2014)
- Tymor Hamamsy (Junior Bioinformatics Specialist, 2014-2015)
- Lide Han (Senior Staff Scientist, 2017-)
- Theodore Morley (Junior Bioinformatics Specialist, 2018-)

Thesis Committees:

- Mary Lauren Benton (Biomedical Informatics)
- Ying Ji (Human Genetics)
- Annika Foucan (Human Genetics)
- Nick Strayer (Biostatistics)
- Annah Moore (Pharmacology)
- Julia Sealock (Human Genetics)

RESEARCH PROGRAM

Completed:

2016-2018 NARSAD Young Investigator Grant, Brain and Behavior Research Foundation
Intersecting Genetics and Therapeutic Information to Improve Treatment of Schizophrenia
Role: Principal Investigator
\$35,000/yr

Active:

2016-2020 R01MH111776
Transcriptional consequences of structural variation in brains of schizophrenia patients
Role: Principal Investigator
\$397,407/yr
38% effort

2018-2022 R01MH116269
Leveraging electronic health records for pharmacogenomics of psychiatric disorders
Role: Co-Principal Investigator
\$397,407/yr
12% effort

2017-2022 R01MH113362
Discovering Biology for Neuropsychiatric Diseases Through Omics Studies on Comorbidities
Role: Co-Investigator (PI: Knapik)
\$472,225/yr
4% effort

2018-2019 5U54HD083211
Eunice Kennedy Shriver Intellectual and Developmental Disabilities Research Center at
Vanderbilt University
Role: Co-Investigator (PI: Neul)
\$401,145/yr
4% effort

2018-2023 R01AG059716
Genetic Drivers of Resilience to Alzheimer's Disease
Role: Co-Investigator (PI: Hohman)
\$631,409/yr
10% effort

2018-2022 NIMH R01
PsycheMERGE: Leveraging electronic health records and genomics for mental health research
Role: Co-Investigator (PI: Davis)
\$425,000/yr
15% effort

2018-2023 R01AG061518
Neuroprotective Effects of Vascular Endothelial Growth Factor in Alzheimer's Disease
Role: Co-Investigator (PI: Hohman)
\$696,254/yr
10% effort

2019-2024 R01MH120736

Mental health and chronic disease: A PsychEmerge investigation into the shared biology underlying psychiatric disorders and their physical comorbidities
Role: Co-Investigator
\$597,185/yr
5% effort

Pending:

2019-2024 R01MH121455
Distinguishing Clinical and Genetic Risk of Suicidal Ideation from Attempts to Inform Prevention
Role: Co-Principal Investigator
\$567,688/yr

2019-2024 R01MH119822
Convergence of Diverse Neurodevelopmental Loci on Synaptic Function
Role: Co-Principal Investigator
\$847,886/yr

PUBLICATIONS AND PRESENTATIONS

Peer reviewed research articles: (*corresponding author)

1. Gresham D, **Ruderfer DM**, Pratt SC, Schacherer J, Dunham MJ, Botstein D, Kruglyak L. Genome-wide detection of polymorphisms at nucleotide resolution with a single DNA microarray. *Science*. 2006 Mar 31;311(5769):1932-6. Epub 2006 Mar 9.
2. Gatbonton T, Imbesi M, Nelson M, Akey JM, **Ruderfer DM**, Kruglyak L, Simon JA, Bedalov A. Telomere length as a quantitative trait: genome-wide survey and genetic mapping of telomere length-control genes in yeast. *PLoS Genet*. 2006 Mar;2(3):e35. Epub 2006 Mar 17. Erratum in: *PLoS Genet*. 2006 Jun;2(6):e104.
3. Perlstein EO, **Ruderfer DM**, Ramachandran G, Haggarty SJ, Kruglyak L, Schreiber SL. Revealing complex traits with small molecules and naturally recombinant yeast strains. *Chem Biol*. 2006 Mar;13(3):319-27.
4. **Ruderfer DM**, Pratt SC, Seidel HS, Kruglyak L. Population genomic analysis of outcrossing and recombination in yeast. *Nat Genet*. 2006 Sep;38(9):1077-81. Epub 2006 Aug 6.
5. Perlstein EO, **Ruderfer DM**, Roberts DC, Schreiber SL, Kruglyak L. Genetic basis of individual differences in the response to small-molecule drugs in yeast. *Nat Genet*. 2007 Apr;39(4):496-502. Epub 2007 Mar 4.
6. Schacherer J, **Ruderfer DM**, Gresham D, Dolinski K, Botstein D, Kruglyak L. Genome-wide analysis of nucleotide-level variation in commonly used *Saccharomyces cerevisiae* strains. *PLoS One*. 2007 Mar 28;2(3):e322.
7. Foss EJ, Radulovic D, Shaffer SA, **Ruderfer DM**, Bedalov A, Goodlett DR, Kruglyak L. Genetic basis of proteome variation in yeast. *Nat Genet*. 2007 Nov;39(11):1369-75. Epub 2007 Oct 21.
8. Weiss LA, Shen Y, Korn JM, Arking DE, Miller DT, Fossdal R, Saemundsen E, Stefansson H, Ferreira MA, Green T, Platt OS, **Ruderfer DM**, Walsh CA, Altshuler D, Chakravarti A, Tanzi RE, Stefansson K, Santangelo SL, Gusella JF, Sklar P, Wu BL, Daly MJ; Autism Consortium. Association between microdeletion and microduplication at 16p11.2 and autism. *N Engl J Med*. 2008 Feb 14;358(7):667-75. doi: 10.1056/NEJMoa075974. Epub 2008 Jan 9.
9. International Schizophrenia Consortium. Rare chromosomal deletions and duplications increase risk of schizophrenia. *Nature*. 2008 Sep 11;455(7210):237-41. doi: 10.1038/nature07239. Epub 2008 Jul 30.

10. Ferreira MA, O'Donovan MC, Meng YA, Jones IR, **Ruderfer DM**, Jones L, Fan J, Kirov G, Perlis RH, Green EK, Smoller JW, Grozeva D, Stone J, Nikolov I, Chambert K, Hamshere ML, Nimgaonkar VL, Moskvina V, Thase ME, Caesar S, Sachs GS, Franklin J, Gordon-Smith K, Ardlie KG, Gabriel SB, Fraser C, Blumenstiel B, Defelice M, Breen G, Gill M, Morris DW, Elkin A, Muir WJ, McGhee KA, Williamson R, MacIntyre DJ, MacLean AW, St CD, Robinson M, Van Beck M, Pereira AC, Kandaswamy R, McQuillin A, Collier DA, Bass NJ, Young AH, Lawrence J, Ferrier IN, Anjorin A, Farmer A, Curtis D, Scolnick EM, McGuffin P, Daly MJ, Corvin AP, Holmans PA, Blackwood DH, Gurling HM, Owen MJ, Purcell SM, Sklar P, Craddock N; Wellcome Trust Case Control Consortium. Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. *Nat Genet.* 2008 Sep;40(9):1056-8. doi: 10.1038/ng.209.
11. Schacherer J, Shapiro JA, **Ruderfer DM**, Kruglyak L. Comprehensive polymorphism survey elucidates population structure of *Saccharomyces cerevisiae*. *Nature.* 2009 Mar 19;458(7236):342-5. doi: 10.1038/nature07670. Epub 2009 Feb 11.
12. International Schizophrenia Consortium, Purcell SM, Wray NR, Stone JL, Visscher PM, O'Donovan MC, Sullivan PF, Sklar P. Common polygenic variation contributes to risk of schizophrenia and bipolar disorder. *Nature.* 2009 Aug 6;460(7256):748-52. doi: 10.1038/nature08185. Epub 2009 Jul 1.
13. **Ruderfer DM**, Roberts DC, Schreiber SL, Perlstein EO, Kruglyak L. Using expression and genotype to predict drug response in yeast. *PLoS One.* 2009 Sep 4;4(9):e6907. doi: 10.1371/journal.pone.0006907.
14. O'Dushlaine C, Kenny E, Heron E, Donohoe G, Gill M, Morris D; International Schizophrenia Consortium, Corvin A. Molecular pathways involved in neuronal cell adhesion and membrane scaffolding contribute to schizophrenia and bipolar disorder susceptibility. *Mol Psychiatry.* 2011 Mar;16(3):286-92. doi: 10.1038/mp.2010.7. Epub 2010 Feb 16.
15. **Ruderfer DM**, Korn J, Purcell SM. Family-based genetic risk prediction of multifactorial disease. *Genome Med.* 2010 Jan 15;2(1):2. doi: 10.1186/gm123.
16. Chen X, Lee G, Maher BS, Fanous AH, Chen J, Zhao Z, Guo A, van den Oord E, Sullivan PF, Shi J, Levinson DF, Gejman PV, Sanders A, Duan J, Owen MJ, Craddock NJ, O'Donovan MC, Blackman J, Lewis D, Kirov GK, Qin W, Schwab S, Wildenauer D, Chowdari K, Nimgaonkar V, Straub RE, Weinberger DR, O'Neill FA, Walsh D, Bronstein M, Darvasi A, Lencz T, Malhotra AK, Rujescu D, Giegling I, Werge T, Hansen T, Ingason A, Nöthen MM, Rietschel M, Cichon S, Djurovic S, Andreassen OA, Cantor RM, Ophoff R, Corvin A, Morris DW, Gill M, Pato CN, Pato MT, Macedo A, Gurling HM, McQuillin A, Pimm J, Hultman C, Lichtenstein P, Sklar P, Purcell SM, Scolnick E, St Clair D, Blackwood DH, Kendler KS; GROUP investigators; International Schizophrenia Consortium. GWA study data mining and independent replication identify cardiomyopathy-associated 5 (CMYA5) as a risk gene for schizophrenia. *Mol Psychiatry.* 2011 Nov;16(11):1117-29. doi: 10.1038/mp.2010.96. Epub 2010 Sep 14.
17. Raychaudhuri S, Korn JM, McCarroll SA; International Schizophrenia Consortium, Altshuler D, Sklar P, Purcell S, Daly MJ. Accurately assessing the risk of schizophrenia conferred by rare copy-number variation affecting genes with brain function. *PLoS Genet.* 2010 Sep 9;6(9):e1001097. doi: 10.1371/journal.pgen.1001097.
18. Talkowski ME, McCann KL, Chen M, McClain L, Bamne M, Wood J, Chowdari KV, Watson A, Prasad KM, Kirov G, Georgieva L, Toncheva D, Mansour H, Lewis DA, Owen M, O'Donovan M, Papasaikas P, Sullivan P, **Ruderfer D**, Yao JK, Leonard S, Thomas P, Miyajima F, Quinn J, Lopez AJ, Nimgaonkar VL. Fine-mapping reveals novel alternative splicing of the dopamine transporter. *Am J Med Genet B Neuropsychiatr Genet.* 2010 Dec 5;153B(8):1434-47. doi: 10.1002/ajmg.b.31125. Epub 2010 Oct 18.

19. Richards AL, Jones L, Moskvina V, Kirov G, Gejman PV, Levinson DF, Sanders AR; Molecular Genetics of Schizophrenia Collaboration (MGS); International Schizophrenia Consortium (ISC), Purcell S, Visscher PM, Craddock N, Owen MJ, Holmans P, O'Donovan MC. Schizophrenia susceptibility alleles are enriched for alleles that affect gene expression in adult human brain. *Mol Psychiatry*. 2012 Feb;17(2):193-201. doi: 10.1038/mp.2011.11. Epub 2011 Feb 22.
20. **Ruderfer DM**, Kirov G, Chambert K, Moran JL, Owen MJ, O'Donovan MC, Sklar P, Purcell SM. A family-based study of common polygenic variation and risk of schizophrenia. *Mol Psychiatry*. 2011 Sep;16(9):887-8. doi: 10.1038/mp.2011.34. Epub 2011 Apr 12.
21. Bridges M, Heron EA, O'Dushlaine C, Segurado R; International Schizophrenia Consortium (ISC), Morris D, Corvin A, Gill M, Pinto C. Genetic classification of populations using supervised learning. *PLoS One*. 2011 May 12;6(5):e14802. doi: 10.1371/journal.pone.0014802.
22. Chen J, Lee G, Fanous AH, Zhao Z, Jia P, O'Neill A, Walsh D, Kendler KS, Chen X; International Schizophrenia Consortium. Two non-synonymous markers in PTPN21, identified by genome-wide association study data-mining and replication, are associated with schizophrenia. *Schizophr Res*. 2011 Sep;131(1-3):43-51. doi: 10.1016/j.schres.2011.06.023. Epub 2011 Jul 14.
23. Psychiatric GWAS Consortium Bipolar Disorder Working Group. Large-scale genome-wide association analysis of bipolar disorder identifies a new susceptibility locus near ODZ4. *Nat Genet*. 2011 Sep 18;43(10):977-83. doi: 10.1038/ng.943.
24. Schizophrenia Psychiatric Genome-Wide Association Study (GWAS) Consortium. Genome-wide association study identifies five new schizophrenia loci. *Nat Genet*. 2011 Sep 18;43(10):969-76. doi: 10.1038/ng.940.
25. Talkowski ME, Mullegama SV, Rosenfeld JA, van Bon BW, Shen Y, Repnikova EA, Gastier-Foster J, Thrush DL, Kathiresan S, **Ruderfer DM**, Chiang C, Hanscom C, Ernst C, Lindgren AM, Morton CC, An Y, Astbury C, Brueton LA, Lichtenbelt KD, Ades LC, Fichera M, Romano C, Innis JW, Williams CA, Bartholomew D, Van Allen MI, Parikh A, Zhang L, Wu BL, Pyatt RE, Schwartz S, Shaffer LG, de Vries BB, Gusella JF, Elsea SH. Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. *Am J Hum Genet*. 2011 Oct 7;89(4):551-63. doi: 10.1016/j.ajhg.2011.09.011.
26. Kirov G, Pocklington AJ, Holmans P, Ivanov D, Ikeda M, **Ruderfer D**, Moran J, Chambert K, Toncheva D, Georgieva L, Grozeva D, Fjodorova M, Wollerton R, Rees E, Nikolov I, van de Lagemaat LN, Bayés A, Fernandez E, Olason PI, Böttcher Y, Komiyama NH, Collins MO, Choudhary J, Stefansson K, Stefansson H, Grant SG, Purcell S, Sklar P, O'Donovan MC, Owen MJ. De novo CNV analysis implicates specific abnormalities of postsynaptic signalling complexes in the pathogenesis of schizophrenia. *Mol Psychiatry*. 2012 Feb;17(2):142-53. doi: 10.1038/mp.2011.154. Epub 2011 Nov 15.
27. Jia P, Wang L, Fanous AH, Chen X, Kendler KS; International Schizophrenia Consortium, Zhao Z. A bias-reducing pathway enrichment analysis of genome-wide association data confirmed association of the MHC region with schizophrenia. *J Med Genet*. 2012 Feb;49(2):96-103. doi: 0.1136/jmedgenet-2011-100397. Epub 2011 Dec 20.
28. Talkowski ME, Rosenfeld JA, Blumenthal I, Pillalamarri V, Chiang C, Heilbut A, Ernst C, Hanscom C, Rossin E, Lindgren AM, Pereira S, **Ruderfer D**, Kirby A, Ripke S, Harris DJ, Lee JH, Ha K, Kim HG, Solomon BD, Gropman AL, Lucente D, Sims K, Ohsumi TK, Borowsky ML, Loranger S, Quade B, Lage K, Miles J, Wu BL, Shen Y, Neale B, Shaffer LG, Daly MJ, Morton CC, Gusella JF. Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*. 2012 Apr 27;149(3):525-37. doi: 10.1016/j.cell.2012.03.028. Epub 2012 Apr 19.

29. Bergen SE, O'Dushlaine CT, Ripke S, Lee PH, **Ruderfer DM**, Akterin S, Moran JL, Chambert KD, Handsaker RE, Backlund L, Ösby U, McCarroll S, Landen M, Scolnick EM, Magnusson PK, Lichtenstein P, Hultman CM, Purcell SM, Sklar P, Sullivan PF. Genome-wide association study in a Swedish population yields support for greater CNV and MHC involvement in schizophrenia compared with bipolar disorder. *Mol Psychiatry*. 2012 Sep;17(9):880-6. doi: 10.1038/mp.2012.73. Epub 2012 Jun 12.
30. Derks EM, Vorstman JA, Ripke S, Kahn RS; Schizophrenia Psychiatric Genomic Consortium, Ophoff RA. Investigation of the genetic association between quantitative measures of psychosis and schizophrenia: a polygenic risk score analysis. *PLoS One*. 2012;7(6):e37852. doi: 10.1371/journal.pone.0037852. Epub 2012 Jun 22. Erratum in: *PLoS One*. 2013;8(3). doi: 10.1371/annotation/6ff0353a-cc91-4d12-896a-d1de0dcb0fe0.
31. Kim HG, Kim HT, Leach NT, Lan F, Ullmann R, Silahatoglu A, Kurth I, Nowka A, Seong IS, Shen Y, Talkowski ME, **Ruderfer D**, Lee JH, Glotzbach C, Ha K, Kjaergaard S, Levin AV, Romeike BF, Kleefstra T, Bartsch O, Elsea SH, Jabs EW, MacDonald ME, Harris DJ, Quade BJ, Ropers HH, Shaffer LG, Kutsche K, Layman LC, Tommerup N, Kalscheuer VM, Shi Y, Morton CC, Kim CH, Gusella JF. Translocations disrupting PHF21A in the Potocki-Shaffer-syndrome region are associated with intellectual disability and craniofacial anomalies. *Am J Hum Genet*. 2012 Jul 13;91(1):56-72. doi: 10.1016/j.ajhg.2012.05.005. Epub 2012 Jul 5.
32. Jia P, Wang L, Fanous AH, Pato CN, Edwards TL; International Schizophrenia Consortium, Zhao Z. Network-assisted investigation of combined causal signals from genome-wide association studies in schizophrenia. *PLoS Comput Biol*. 2012;8(7):e1002587. doi: 10.1371/journal.pcbi.1002587. Epub 2012 Jul 5.
33. Rueckert EH, Barker D, **Ruderfer D**, Bergen SE, O'Dushlaine C, Luce CJ, Sheridan SD, Theriault KM, Chambert K, Moran J, Purcell SM, Madison JM, Haggarty SJ, Sklar P. Cis-acting regulation of brain-specific ANK3 gene expression by a genetic variant associated with bipolar disorder. *Mol Psychiatry*. 2013 Aug;18(8):922-9. doi: 10.1038/mp.2012.104. Epub 2012 Jul 31.
34. Perlis RH, **Ruderfer D**, Hamilton SP, Ernst C. Copy number variation in subjects with major depressive disorder who attempted suicide. *PLoS One*. 2012;7(9):e46315. doi: 10.1371/journal.pone.0046315. Epub 2012 Sep 27.
35. Terwisscha van Scheltinga AF, Bakker SC, van Haren NE, Derks EM, Buizer-Voskamp JE, Boos HB, Cahn W, Hulshoff Pol HE, Ripke S, Ophoff RA, Kahn RS; Psychiatric Genome-wide Association Study Consortium. Genetic schizophrenia risk variants jointly modulate total brain and white matter volume. *Biol Psychiatry*. 2013 Mar 15;73(6):525-31. doi: 10.1016/j.biopsych.2012.08.017. Epub 2012 Oct 3.
36. Fromer M, Moran JL, Chambert K, Banks E, Bergen SE, **Ruderfer DM**, Handsaker RE, McCarroll SA, O'Donovan MC, Owen MJ, Kirov G, Sullivan PF, Hultman CM, Sklar P, Purcell SM. Discovery and statistical genotyping of copy-number variation from whole-exome sequencing depth. *Am J Hum Genet*. 2012 Oct 5;91(4):597-607. doi: 10.1016/j.ajhg.2012.08.005.
37. Ernst C, Marshall CR, Shen Y, Metcalfe K, Rosenfeld J, Hodge JC, Torres A, Blumenthal I, Chiang C, Pillalamarri V, Crapper L, Diallo AB, **Ruderfer D**, Pereira S, Sklar P, Purcell S, Wildin RS, Spencer AC, Quade BF, Harris DJ, Lemyre E, Wu BL, Stavropoulos DJ, Geraghty MT, Shaffer LG, Morton CC, Scherer SW, Gusella JF, Talkowski ME. Highly penetrant alterations of a critical region including BDNF in human psychopathology and obesity. *Arch Gen Psychiatry*. 2012 Dec;69(12):1238-46. doi: 10.1001/archgenpsychiatry.2012.660.
38. Perlis RH, **Ruderfer D**, Maussion G, Chambert K, Gallagher P, Turecki G, Ernst C. Bipolar disorder and a history of suicide attempts with a duplication in 5HTR1A. *Am J Psychiatry*. 2012 Nov;169(11):1213-4. doi: 10.1176/appi.ajp.2012.12050592.
39. Talkowski ME, Maussion G, Crapper L, Rosenfeld JA, Blumenthal I, Hanscom C, Chiang C, Lindgren A, Pereira S, **Ruderfer D**, Diallo AB, Lopez JP, Turecki G, Chen ES, Gigeck C, Harris DJ, Lip V, An Y, Biagioli M, Macdonald

- ME, Lin M, Haggarty SJ, Sklar P, Purcell S, Kellis M, Schwartz S, Shaffer LG, Natowicz MR, Shen Y, Morton CC, Gusella JF, Ernst C. Disruption of a large intergenic noncoding RNA in subjects with neurodevelopmental disabilities. *Am J Hum Genet.* 2012 Dec 7;91(6):1128-34. doi: 10.1016/j.ajhg.2012.10.016.
40. **Ruderfer DM**, Chambert K, Moran J, Talkowski M, Chen ES, Gigeck C, Gusella JF, Blackwood DH, Corvin A, Gurling HM, Hultman CM, Kirov G, Magnusson P, O'Donovan MC, Owen MJ, Pato C, St Clair D, Sullivan PF, Purcell SM, Sklar P, Ernst C. Mosaic copy number variation in schizophrenia. *Eur J Hum Genet.* 2013 Sep;21(9):1007-11. doi: 10.1038/ejhg.2012.287. Epub 2013 Jan 16.
41. Guha S, Rees E, Darvasi A, Ivanov D, Ikeda M, Bergen SE, Magnusson PK, Cormican P, Morris D, Gill M, Cichon S, Rosenfeld JA, Lee A, Gregersen PK, Kane JM, Malhotra AK, Rietschel M, Nöthen MM, Degenhardt F, Priebe L, Breuer R, Strohmaier J, **Ruderfer DM**, Moran JL, Chambert KD, Sanders AR, Shi J, Kendler K, Riley B, O'Neill T, Walsh D, Malhotra D, Corvin A, Purcell S, Sklar P, Iwata N, Hultman CM, Sullivan PF, Sebat J, McCarthy S, Gejman PV, Levinson DF, Owen MJ, O'Donovan MC, Lencz T, Kirov G; Molecular Genetics of Schizophrenia Consortium; Wellcome Trust Case Control Consortium 2. Implication of a rare deletion at distal 16p11.2 in schizophrenia. *JAMA Psychiatry.* 2013 Mar;70(3):253-60. doi: 10.1001/2013.jamapsychiatry.71.
42. Lim ET, Raychaudhuri S, Sanders SJ, Stevens C, Sabo A, MacArthur DG, Neale BM, Kirby A, **Ruderfer DM**, Fromer M, Lek M, Liu L, Flannick J, Ripke S, Nagaswamy U, Muzny D, Reid JG, Hawes A, Newsham I, Wu Y, Lewis L, Dinh H, Gross S, Wang LS, Lin CF, Valladares O, Gabriel SB, dePristo M, Altshuler DM, Purcell SM; NHLBI Exome Sequencing Project, State MW, Boerwinkle E, Buxbaum JD, Cook EH, Gibbs RA, Schellenberg GD, Sutcliffe JS, Devlin B, Roeder K, Daly MJ. Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron.* 2013 Jan 23;77(2):235-42. doi: 10.1016/j.neuron.2012.12.029.
43. Andreassen OA, Djurovic S, Thompson WK, Schork AJ, Kendler KS, O'Donovan MC, Rujescu D, Werge T, van de Bunt M, Morris AP, McCarthy MI; International Consortium for Blood Pressure GWAS; Diabetes Genetics Replication and Meta-analysis Consortium; Psychiatric Genomics Consortium Schizophrenia Working Group, Roddey JC, McEvoy LK, Desikan RS, Dale AM. Improved detection of common variants associated with schizophrenia by leveraging pleiotropy with cardiovascular-disease risk factors. *Am J Hum Genet.* 2013 Feb 7;92(2):197-209. doi: 10.1016/j.ajhg.2013.01.001. Epub 2013 Jan 31.
44. van Scheltinga AF, Bakker SC, van Haren NE, Derks EM, Buizer-Voskamp JE, Cahn W, Ripke S; Psychiatric Genome-Wide Association Study (GWAS) Consortium, Ophoff RA, Kahn RS. Schizophrenia genetic variants are not associated with intelligence. *Psychol Med.* 2013 Dec;43(12):2563-70. doi:10.1017/S0033291713000196. Epub 2013 Feb 15.
45. Chen Z, Tang H, Qayyum R, Schick UM, Nalls MA, Handsaker R, Li J, Lu Y, Yanek LR, Keating B, Meng Y, van Rooij FJ, Okada Y, Kubo M, Rasmussen-Torvik L, Keller MF, Lange L, Evans M, Bottinger EP, Linderman MD, **Ruderfer DM**, Hakonarson H, Papanicolaou G, Zonderman AB, Gottesman O; BioBank Japan Project; CHARGE Consortium, Thomson C, Ziv E, Singleton AB, Loos RJ, Sleiman PM, Ganesh S, McCarroll S, Becker DM, Wilson JG, Lettre G, Reiner AP. Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network. *Hum Mol Genet.* 2013 Jun 15;22(12):2529-38. doi: 10.1093/hmg/ddt087. Epub 2013 Feb 26.
46. Cross-Disorder Group of the Psychiatric Genomics Consortium. Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis. *Lancet.* 2013 Apr 20;381(9875):1371-9. doi: 10.1016/S0140-6736(12)62129-1. Epub 2013 Feb 28. Erratum in: *Lancet.* 2013 Apr 20;381(9875):1360.
47. Schork AJ, Thompson WK, Pham P, Torkamani A, Roddey JC, Sullivan PF, Kelsoe JR, O'Donovan MC, Furberg H; Tobacco and Genetics Consortium; Bipolar Disorder Psychiatric Genomics Consortium; Schizophrenia Psychiatric Genomics Consortium, Schork NJ, Andreassen OA, Dale AM. All SNPs are not created equal: genome-wide association studies reveal a consistent pattern of enrichment among functionally annotated SNPs. *PLoS Genet.* 2013 Apr;9(4):e1003449. doi: 10.1371/journal.pgen.1003449. Epub 2013 Apr 25.

48. Cross-Disorder Group of the Psychiatric Genomics Consortium. Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nat Genet.* 2013 Sep;45(9):984-94. doi: 10.1038/ng.2711. Epub 2013 Aug 11.
49. Ripke S, O'Dushlaine C, Chambert K, Moran JL, Kähler AK, Akterin S, Bergen SE, Collins AL, Crowley JJ, Fromer M, Kim Y, Lee SH, Magnusson PK, Sanchez N, Stahl EA, Williams S, Wray NR, Xia K, Bettella F, Borglum AD, Bulik-Sullivan BK, Cormican P, Craddock N, de Leeuw C, Durmishi N, Gill M, Golimbet V, Hamshere ML, Holmans P, Hougaard DM, Kendler KS, Lin K, Morris DW, Mors O, Mortensen PB, Neale BM, O'Neill FA, Owen MJ, Milovancevic MP, Posthuma D, Powell J, Richards AL, Riley BP, **Ruderfer D**, Rujescu D, Sigurdsson E, Silagadze T, Smit AB, Stefansson H, Steinberg S, Suvisaari J, Tosato S, Verhage M, Walters JT; Multicenter Genetic Studies of Schizophrenia Consortium, Levinson DF, Gejman PV, Kendler KS, Laurent C, Mowry BJ, O'Donovan MC, Owen MJ, Pulver AE, Riley BP, Schwab SG, Wildenauer DB, Dudbridge F, Holmans P, Shi J, Albus M, Alexander M, Champion D, Cohen D, Dikeos D, Duan J, Eichhammer P, Godard S, Hansen M, Lerer FB, Liang KY, Maier W, Mallet J, Nertney DA, Nestadt G, Norton N, O'Neill FA, Papadimitriou GN, Ribble R, Sanders AR, Silverman JM, Walsh D, Williams NM, Wormley B; Psychosis Endophenotypes International Consortium, Arranz MJ, Bakker S, Bender S, Bramon E, Collier D, Crespo-Facorro B, Hall J, Iyegbe C, Jablensky A, Kahn RS, Kalaydjieva L, Lawrie S, Lewis CM, Lin K, Linszen DH, Mata I, McIntosh A, Murray RM, Ophoff RA, Powell J, Rujescu D, Van Os J, Walshe M, Weisbrod M, Wiersma D; Wellcome Trust Case Control Consortium 2, Donnelly P, Barroso I, Blackwell JM, Bramon E, Brown MA, Casas JP, Corvin AP, Deloukas P, Duncanson A, Jankowski J, Markus HS, Mathew CG, Palmer CN, Plomin R, Rautanen A, Sawcer SJ, Trembath RC, Viswanathan AC, Wood NW, Spencer CC, Band G, Bellenguez C, Freeman C, Hellenthal G, Giannoulatou E, Pirinen M, Pearson RD, Strange A, Su Z, Vukcevic D, Donnelly P, Langford C, Hunt SE, Edkins S, Gwilliam R, Blackburn H, Bumpstead SJ, Dronov S, Gillman M, Gray E, Hammond N, Jayakumar A, McCann OT, Liddle J, Potter SC, Ravindrarajah R, Ricketts M, Tashakkori-Ghanbaria A, Waller MJ, Weston P, Widaa S, Whittaker P, Barroso I, Deloukas P, Mathew CG, Blackwell JM, Brown MA, Corvin AP, McCarthy MI, Spencer CC, Bramon E, Corvin AP, O'Donovan MC, Stefansson K, Scolnick E, Purcell S, McCarroll SA, Sklar P, Hultman CM, Sullivan PF. Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nat Genet.* 2013 Oct;45(10):1150-9. doi: 10.1038/ng.2742. Epub 2013 Aug 25.
50. **Ruderfer DM**, Fanous AH, Ripke S, McQuillin A, Amdur RL; Schizophrenia Working Group of Psychiatric Genomics Consortium; Bipolar Disorder Working Group of Psychiatric Genomics Consortium; Cross-Disorder Working Group of Psychiatric Genomics Consortium, Gejman PV, O'Donovan MC, Andreassen OA, Djurovic S, Hultman CM, Kelsoe JR, Jamain S, Landén M, Leboyer M, Nimgaonkar V, Nurnberger J, Smoller JW, Craddock N, Corvin A, Sullivan PF, Holmans P, Sklar P, Kendler KS. Polygenic dissection of diagnosis and clinical dimensions of bipolar disorder and schizophrenia. *Mol Psychiatry.* 2014 Sep;19(9):1017-24. doi: 10.1038/mp.2013.138. Epub 2013 Nov 26.
51. Fromer M, Pocklington AJ, Kavanagh DH, Williams HJ, Dwyer S, Gormley P, Georgieva L, Rees E, Palta P, **Ruderfer DM**, Carrera N, Humphreys I, Johnson JS, Roussos P, Barker DD, Banks E, Milanova V, Grant SG, Hannon E, Rose SA, Chambert K, Mahajan M, Scolnick EM, Moran JL, Kirov G, Palotie A, McCarroll SA, Holmans P, Sklar P, Owen MJ, Purcell SM, O'Donovan MC. De novo mutations in schizophrenia implicate synaptic networks. *Nature.* 2014 Feb 13;506(7487):179-84. doi: 10.1038/nature12929. Epub 2014 Jan 22.
52. Purcell SM, Moran JL, Fromer M, **Ruderfer D**, Solovieff N, Roussos P, O'Dushlaine C, Chambert K, Bergen SE, Kähler A, Duncan L, Stahl E, Genovese G, Fernández E, Collins MO, Komiyama NH, Choudhary JS, Magnusson PK, Banks E, Shakir K, Garimella K, Fennell T, DePristo M, Grant SG, Haggarty SJ, Gabriel S, Scolnick EM, Lander ES, Hultman CM, Sullivan PF, McCarroll SA, Sklar P. A polygenic burden of rare disruptive mutations in schizophrenia. *Nature.* 2014 Feb 13;506(7487):185-90. doi: 10.1038/nature12975. Epub 2014 Jan 22.
53. Lange LA, Hu Y, Zhang H, Xue C, Schmidt EM, Tang ZZ, Bizon C, Lange EM, Smith JD, Turner EH, Jun G, Kang HM, Peloso G, Auer P, Li KP, Flannick J, Zhang J, Fuchsberger C, Gaulton K, Lindgren C, Locke A,

Manning A, Sim X, Rivas MA, Holmen OL, Gottesman O, Lu Y, **Ruderfer D**, Stahl EA, Duan Q, Li Y, Durda P, Jiao S, Isaacs A, Hofman A, Bis JC, Correa A, Griswold ME, Jakobsdottir J, Smith AV, Schreiner PJ, Feitosa MF, Zhang Q, Huffman JE, Crosby J, Wassel CL, Do R, Franceschini N, Martin LW, Robinson JG, Assimes TL, Crosslin DR, Rosenthal EA, Tsai M, Rieder MJ, Farlow DN, Folsom AR, Lumley T, Fox ER, Carlson CS, Peters U, Jackson RD, van Duijn CM, Uitterlinden AG, Levy D, Rotter JI, Taylor HA, Gudnason V Jr, Siscovick DS, Fornage M, Borecki IB, Hayward C, Rudan I, Chen YE, Bottinger EP, Loos RJ, Sætrum P, Hveem K, Boehnke M, Groop L, McCarthy M, Meitinger T, Ballantyne CM, Gabriel SB, O'Donnell CJ, Post WS, North KE, Reiner AP, Boerwinkle E, Psaty BM, Altshuler D, Kathiresan S, Lin DY, Jarvik GP, Cupples LA, Kooperberg C, Wilson JG, Nickerson DA, Abecasis GR, Rich SS, Tracy RP, Willer CJ; NHLBI Grand Opportunity Exome Sequencing Project. Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. *Am J Hum Genet.* 2014 Feb 6;94(2):233-45. doi: 10.1016/j.ajhg.2014.01.010.

54. O'Dushlaine C, Ripke S, **Ruderfer DM**, Hamilton SP, Fava M, Iosifescu DV, Kohane IS, Churchill SE, Castro VM, Clements CC, Blumenthal SR, Murphy SN, Smoller JW, Perlis RH. Rare copy number variation in treatment-resistant major depressive disorder. *Biol Psychiatry.* 2014 Oct 1;76(7):536-41. doi: 10.1016/j.biopsych.2013.10.028. Epub 2014 Jan 19.
55. Bergen SE, O'Dushlaine CT, Lee PH, Fanous AH, **Ruderfer DM**, Ripke S; International Schizophrenia Consortium, Swedish Schizophrenia Consortium, Sullivan PF, Smoller JW, Purcell SM, Corvin A. Genetic modifiers and subtypes in schizophrenia: investigations of age at onset, severity, sex and family history. *Schizophr Res.* 2014 Apr;154(1-3):48-53. doi: 10.1016/j.schres.2014.01.030. Epub 2014 Feb 26.
56. Nurnberger JI Jr, Koller DL, Jung J, Edenberg HJ, Foroud T, Guella I, Vawter MP, Kelsoe JR; Psychiatric Genomics Consortium Bipolar Group. Identification of pathways for bipolar disorder: a meta-analysis. *JAMA Psychiatry.* 2014 Jun;71(6):657-64. doi: 10.1001/jamapsychiatry.2014.176.
57. Szatkiewicz JP, O'Dushlaine C, Chen G, Chambert K, Moran JL, Neale BM, Fromer M, **Ruderfer D**, Akterin S, Bergen SE, Kähler A, Magnusson PK, Kim Y, Crowley JJ, Rees E, Kirov G, O'Donovan MC, Owen MJ, Walters J, Scolnick E, Sklar P, Purcell S, Hultman CM, McCarroll SA, Sullivan PF. Copy number variation in schizophrenia in Sweden. *Mol Psychiatry.* 2014 Jul;19(7):762-73. doi: 10.1038/mp.2014.40. Epub 2014 Apr 29.
58. Nicodemus KK, Hargreaves A, Morris D, Anney R, Gill M, Corvin A, Donohoe G; Schizophrenia Psychiatric Genome-wide Association Study (GWAS) Consortium; Wellcome Trust Case Control Consortium 2. Variability in working memory performance explained by epistasis vs polygenic scores in the ZNF804A pathway. *JAMA Psychiatry.* 2014 Jul 1;71(7):778-85. doi: 10.1001/jamapsychiatry.2014.528.
59. Schizophrenia Working Group of the Psychiatric Genomics Consortium. Biological insights from 108 schizophrenia-associated genetic loci. *Nature.* 2014 Jul 24;511(7510):421-7. doi: 10.1038/nature13595. Epub 2014 Jul 22.
60. **Ruderfer DM**, Lim ET, Genovese G, Moran JL, Hultman CM, Sullivan PF, McCarroll SA, Holmans P, Sklar P, Purcell SM. No evidence for rare recessive and compound heterozygous disruptive variants in schizophrenia. *Eur J Hum Genet.* 2015 Apr;23(4):555-7. doi: 10.1038/ejhg.2014.228. Epub 2014 Nov 5.
61. Gusev A, Lee SH, Trynka G, Finucane H, Vilhjálmsón BJ, Xu H, Zang C, Ripke S, Bulik-Sullivan B, Stahl E; Schizophrenia Working Group of the Psychiatric Genomics Consortium; SWE-SCZ Consortium, Kähler AK, Hultman CM, Purcell SM, McCarroll SA, Daly M, Pasaniuc B, Sullivan PF, Neale BM, Wray NR, Raychaudhuri S, Price AL; Schizophrenia Working Group of the Psychiatric Genomics Consortium; SWE-SCZ Consortium. Partitioning heritability of regulatory and cell-type-specific variants across 11 common diseases. *Am J Hum Genet.* 2014 Nov 6;95(5):535-52. doi: 10.1016/j.ajhg.2014.10.004. Epub 2014 Nov 6.

62. Roussos P, Mitchell AC, Voloudakis G, Fullard JF, Pothula VM, Tsang J, Stahl EA, Georgakopoulos A, **Ruderfer DM**, Charney A, Okada Y, Siminovitch KA, Worthington J, Padyukov L, Klareskog L, Gregersen PK, Plenge RM, Raychaudhuri S, Fromer M, Purcell SM, Brennand KJ, Robakis NK, Schadt EE, Akbarian S, Sklar P. A role for noncoding variation in schizophrenia. *Cell Rep*. 2014 Nov 20;9(4):1417-29. doi: 10.1016/j.celrep.2014.10.015. Epub 2014 Nov 6.
63. Network and Pathway Analysis Subgroup of Psychiatric Genomics Consortium. Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. *Nat Neurosci*. 2015 Feb;18(2):199-209. doi: 10.1038/nn.3922. Epub 2015 Jan 19. Erratum in: *Nat Neurosci*. 2015 Jun;18(6):926.
64. Maier R, Moser G, Chen GB, Ripke S; Cross-Disorder Working Group of the Psychiatric Genomics Consortium, Coryell W, Potash JB, Scheftner WA, Shi J, Weissman MM, Hultman CM, Landén M, Levinson DF, Kendler KS, Smoller JW, Wray NR, Lee SH. Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. *Am J Hum Genet*. 2015 Feb 5;96(2):283-94. doi: 10.1016/j.ajhg.2014.12.006. Epub 2015 Jan 29.
65. Bulik-Sullivan BK, Loh PR, Finucane HK, Ripke S, Yang J; Schizophrenia Working Group of the Psychiatric Genomics Consortium, Patterson N, Daly MJ, Price AL, Neale BM. LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. *Nat Genet*. 2015 Mar;47(3):291-5. doi: 10.1038/ng.3211. Epub 2015 Feb 2.
66. Castro VM, Minnier J, Murphy SN, Kohane I, Churchill SE, Gainer V, Cai T, Hoffnagle AG, Dai Y, Block S, Weill SR, Nadal-Vicens M, Pollastri AR, Rosenquist JN, Goryachev S, Ongur D, Sklar P, Perlis RH, Smoller JW; International Cohort Collection for Bipolar Disorder Consortium. Validation of electronic health record phenotyping of bipolar disorder cases and controls. *Am J Psychiatry*. 2015 Apr;172(4):363-72. doi: 10.1176/appi.ajp.2014.14030423. Epub 2014 Dec 12.
67. Rivas MA, Pirinen M, Conrad DF, Lek M, Tsang EK, Karczewski KJ, Maller JB, Kukurba KR, DeLuca DS, Fromer M, Ferreira PG, Smith KS, Zhang R, Zhao F, Banks E, Poplin R, **Ruderfer DM**, Purcell SM, Tukiainen T, Minikel EV, Stenson PD, Cooper DN, Huang KH, Sullivan TJ, Nedzel J; GTEx Consortium; Geuvadis Consortium, Bustamante CD, Li JB, Daly MJ, Guigo R, Donnelly P, Ardlie K, Sammeth M, Dermitzakis ET, McCarthy MI, Montgomery SB, Lappalainen T, MacArthur DG. Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*. 2015 May 8;348(6235):666-9. doi: 10.1126/science.1261877.
68. Rees E, Kirov G, Walters JT, Richards AL, Howrigan D, Kavanagh DH, Pocklington AJ, Fromer M, **Ruderfer DM**, Georgieva L, Carrera N, Gormley P, Palta P, Williams H, Dwyer S, Johnson JS, Roussos P, Barker DD, Banks E, Milanova V, Rose SA, Chambert K, Mahajan M, Scolnick EM, Moran JL, Tsuang MT, Glatt SJ, Chen WJ, Hwu HG; Taiwanese Trios Exome Sequencing Consortium, Neale BM, Palotie A, Sklar P, Purcell SM, McCarroll SA, Holmans P, Owen MJ, O'Donovan MC. Analysis of exome sequence in 604 trios for recessive genotypes in schizophrenia. *Transl Psychiatry*. 2015 Jul 21;5:e607. doi: 10.1038/tp.2015.99.
69. Eduati F, Mangravite LM, Wang T, Tang H, Bare JC, Huang R, Norman T, Kellen M, Menden MP, Yang J, Zhan X, Zhong R, Xiao G, Xia M, Abdo N, Kosyk O; NIEHS-NCATS-UNC DREAM Toxicogenetics Collaboration. Prediction of human population responses to toxic compounds by a collaborative competition. *Nat Biotechnol*. 2015 Aug 10. doi: 10.1038/nbt.3299. [Epub ahead of print]
70. Lee SH, Byrne EM, Hultman CM, Kähler A, Vinkhuyzen AA, Ripke S, Andreassen OA, Frisell T, Gusev A, Hu X, Karlsson R, Mantzioris VX, McGrath JJ, Mehta D, Stahl EA, Zhao Q, Kendler KS, Sullivan PF, Price AL, O'Donovan M, Okada Y, Mowry BJ, Raychaudhuri S, Wray NR; Schizophrenia Working Group of the Psychiatric Genomics Consortium and Rheumatoid Arthritis Consortium International; Schizophrenia Working Group of the Psychiatric Genomics Consortium, Rheumatoid Arthritis Consortium International. New data and an old puzzle:

the negative association between schizophrenia and rheumatoid arthritis. *Int J Epidemiol.* 2015 Aug 18. pii: dyv136. [Epub ahead of print]

71. **Ruderfer DM***, Charney AW, Readhead B, Kidd BA, Kahler AK, Kenny PJ, Keiser MJ, Moran JL, Hultman CM, Scott SA, Sullivan PF, Purcell SM, Dudley JT, Sklar P. Polygenic overlap between schizophrenia risk and antipsychotic response: a genomic medicine approach. *Lancet Psychiatry.* 2016 Feb 22. pii: S2215-0366(15)00553-2. doi: 10.1016/S2215-0366(15)00553-2. ***Corresponding author**
72. Topol A, Zhu S, Hartley BJ, English J, Hauberg ME, Tran N, Rittenhouse CA, Simone A, **Ruderfer DM**, Johnson J, Readhead B, Hadas Y, Gochman PA, Wang YC, Shah H, Cagney G, Rapoport J, Gage FH, Dudley JT, Sklar P, Mattheisen M, Cotter D, Fang G, Brennand KJ. Dysregulation of miRNA-9 in a Subset of Schizophrenia Patient-Derived Neural Progenitor Cells. *Cell Rep.* 2016 May 3;15(5):1024-36. doi: 10.1016/j.celrep.2016.03.090. Epub 2016 Apr 21.
73. Lek M, Karczewski KJ, Minikel EV, Samocha KE, Banks E, Fennell T, O'Donnell-Luria AH, Ware JS, Hill AJ, Cummings BB, Tukiainen T, Birnbaum DP, Kosmicki JA, Duncan LE, Estrada K, Zhao F, Zou J, Pierce-Hoffman E, Berghout J, Cooper DN, Deflaux N, DePristo M, Do R, Flannick J, Fromer M, Gauthier L, Goldstein J, Gupta N, Howrigan D, Kiezun A, Kurki MI, Moonshine AL, Natarajan P, Orozco L, Peloso GM, Poplin R, Rivas MA, Ruano-Rubio V, Rose SA, **Ruderfer DM**, Shakir K, Stenson PD, Stevens C, Thomas BP, Tiao G, Tusie-Luna MT, Weisburd B, Won HH, Yu D, Altshuler DM, Ardissino D, Boehnke M, Danesh J, Donnelly S, Elosua R, Florez JC, Gabriel SB, Getz G, Glatt SJ, Hultman CM, Kathiresan S, Laakso M, McCarroll S, McCarthy MI, McGovern D, McPherson R, Neale BM, Palotie A, Purcell SM, Saleheen D, Scharf JM, Sklar P, Sullivan PF, Tuomilehto J, Tsuang MT, Watkins HC, Wilson JG, Daly MJ, MacArthur DG; Exome Aggregation Consortium. Analysis of protein-coding genetic variation in 60,706 humans. *Nature.* 2016 Aug 17;536(7616):285-91. doi: 10.1038/nature19057.
74. **Ruderfer DM***, Hamamsy T, Lek M, Karczewski KJ, Kavanaugh D, Samocha K, Exome Aggregation Consortium, Daly MJ, MacArthur DG, Fromer M, Purcell SM. Patterns of genic intolerance of rare copy number variations in 59,898 human exomes. *Nat Genet.* 2016 Aug 17. doi: 10.1038/ng.3638.
75. Fromer M, Roussos P, Sieberts SK, Johnson JS, Kavanagh DH, Perumal TM, **Ruderfer DM**, Oh EC, Topol A, Shah HR, Klei LL, Kramer R, Pinto D, Gümüş ZH, Cicek AE, Dang KK, Browne A, Lu C, Xie L, Readhead B, Stahl EA, Xiao J, Parvizi M, Hamamsy T, Fullard JF, Wang YC, Mahajan MC, Derry JM, Dudley JT, Hemby SE, Logsdon BA, Talbot K, Raj T, Bennett DA, De Jager PL, Zhu J, Zhang B, Sullivan PF, Chess A, Purcell SM, Shinobu LA, Mangravite LM, Toyoshiba H, Gur RE, Hahn CG, Lewis DA, Haroutunian V, Peters MA, Lipska BK, Buxbaum JD, Schadt EE, Hirai K, Roeder K, Brennand KJ, Katsanis N, Domenici E, Devlin B, Sklar P. Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nat Neurosci.* 2016 Nov;19(11):1442-1453. doi: 10.1038/nn.4399. Epub 2016 Sep 26.
76. Genovese G, Fromer M, Stahl EA, **Ruderfer DM**, Chambert K, Landén M, Moran JL, Purcell SM, Sklar P, Sullivan PF, Hultman CM, McCarroll SA. Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. *Nat Neurosci.* 2016 Nov;19(11):1433-1441. doi: 10.1038/nn.4402. Epub 2016 Oct 3.
77. Johnson EC, Bjelland DW, Howrigan DP, Abdellaoui A, Breen G, Borglum A, Cichon S, Degenhardt F, Forstner AJ, Frank J, Genovese G, Heilmann-Heimbach S, Herms S, Hoffman P, Maier W, Mattheisen M, Morris D, Mowry B, Müller-Mhysok B, Neale B, Nenadic I, Nöthen MM, O'Dushlaine C, Rietschel M, **Ruderfer DM**, Rujescu D, Schulze TG, Simonson MA, Stahl E, Strohmaier J, Witt SH; Schizophrenia Working Group of the Psychiatric Genomics Consortium., Sullivan PF, Keller MC. No Reliable Association between Runs of Homozygosity and Schizophrenia in a Well-Powered Replication Study. *PLoS Genet.* 2016 Oct 28;12(10):e1006343. doi: 10.1371/journal.pgen.1006343. eCollection 2016 Oct.

78. CNV and Schizophrenia Working Groups of the Psychiatric Genomics Consortium.; Psychosis Endophenotypes International Consortium. Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. *Nat Genet.* 2017 Jan;49(1):27-35. doi: 10.1038/ng.3725. Epub 2016 Nov 21.
79. Karczewski KJ, Weisburd B, Thomas B, Solomonson M, **Ruderfer DM**, Kavanagh D, Hamamsy T, Lek M, Samocha KE, Cummings BB, Birnbaum D; The Exome Aggregation Consortium., Daly MJ, MacArthur DG. The ExAC browser: displaying reference data information from over 60 000 exomes. *Nucleic Acids Res.* 2017 Jan 4;45(D1):D840-D845. doi: 10.1093/nar/gkw971. Epub 2016 Nov 28.
80. Charney AW, **Ruderfer DM**, Stahl EA, Moran JL, Chambert K, Belliveau RA, Forty L, Gordon-Smith K, Di Florio A, Lee PH, Bromet EJ, Buckley PF, Escamilla MA, Fanous AH, Fochtmann LJ, Lehrer DS, Malaspina D, Marder SR, Morley CP, Nicolini H, Perkins DO, Rakofsky JJ, Rapaport MH, Medeiros H, Sobell JL, Green EK, Backlund L, Bergen SE, Juréus A, Schalling M, Lichtenstein P, Roussos P, Knowles JA, Jones I, Jones LA, Hultman CM, Perlis RH, Purcell SM, McCarroll SA, Pato CN, Pato MT, Craddock N, Landén M, Smoller JW, Sklar P. Evidence for genetic heterogeneity between clinical subtypes of bipolar disorder. *Transl Psychiatry.* 2017 Jan 10;7(1):e993. doi: 10.1038/tp.2016.242.
81. Song J, Bergen SE, Di Florio A, Karlsson R, Charney A, **Ruderfer DM**, Stahl EA; Members of the International Cohort Collection for Bipolar Disorder (ICCBD)., Chambert KD, Moran JL, Gordon-Smith K, Forty L, Green EK, Jones I, Jones L, Scolnick EM, Sklar P, Smoller JW, Lichtenstein P, Hultman C, Craddock N, Landén M. Genome-wide association study identifies SESTD1 as a novel risk gene for lithium-responsive bipolar disorder. *Mol Psychiatry.* 2017 Feb 14. doi: 10.1038/mp.2016.246. [Epub ahead of print]
82. Gorski M, van der Most PJ, Teumer A, Chu AY, Li M, Mijatovic V, Nolte IM, Cocca M, Taliun D, Gomez F, Li Y, Tayo B, Tin A, Feitosa MF, Aspelund T, Attia J, Biffar R, Bochud M, Boerwinkle E, Borecki I, Bottinger EP, Chen MH, Chouraki V, Ciullo M, Coresh J, Cornelis MC, Curhan GC, d'Adamo AP, Dehghan A, Dengler L, Ding J, Eiriksdottir G, Endlich K, Enroth S, Esko T, Franco OH, Gasparini P, Gieger C, Girotto G, Gottesman O, Gudnason V, Gyllenstein U, Hancock SJ, Harris TB, Helmer C, Höllerer S, Hofer E, Hofman A, Holliday EG, Homuth G, Hu FB, Huth C, Hutri-Kähönen N, Hwang SJ, Imboden M, Johansson Å, Kähönen M, König W, Kramer H, Krämer BK, Kumar A, Kutalik Z, Lambert JC, Launer LJ, Lehtimäki T, de Borst M, Navis G, Swertz M, Liu Y, Lohman K, Loos RJF, Lu Y, Lyytikäinen LP, McEvoy MA, Meisinger C, Meitinger T, Metspalu A, Metzger M, Mihailov E, Mitchell P, Nauck M, Oldehinkel AJ, Olden M, Wjh Penninx B, Pistis G, Pramstaller PP, Probst-Hensch N, Raitakari O'T, Rettig R, Ridker PM, Rivadeneira F, Robino A, Rosas SE, **Ruderfer D**, Ruggiero D, Saba Y, Sala C, Schmidt H, Schmidt R, Scott RJ, Sedaghat S, Smith AV, Sorice R, Stengel B, Stracke S, Strauch K, Toniolo D, Uitterlinden AG, Ulivi S, Viikari JS, Völker U, Vollenweider P, Völzke H, Vuckovic D, Waldenberger M, Jin Wang J, Yang Q, Chasman DI, Tromp G, Snieder H, Heid IM, Fox CS, Köttgen A, Pattaro C, Böger CA, Fuchsberger C. 1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Sci Rep.* 2017 Apr 28;7:45040. doi: 10.1038/srep45040. Erratum in: *Sci Rep.* 2017 May 26;7:46835. de Borst, Martin H [corrected to de Borst, Martin].
83. Scott RA, Scott LJ, Mägi R, Marullo L, Gaulton KJ, Kaakinen M, Pervjakova N, Pers TH, Johnson AD, Eicher JD, Jackson AU, Ferreira T, Lee Y, Ma C, Steinthorsdottir V, Thorleifsson G, Qi L, Van Zuydam NR, Mahajan A, Chen H, Almgren P, Voight BF, Grallert H, Müller-Nurasyid M, Ried JS, Rayner NW, Robertson N, Karssen LC, van Leeuwen EM, Willems SM, Fuchsberger C, Kwan P, Teslovich TM, Chanda P, Li M, Lu Y, Dina C, Thuillier D, Yengo L, Jiang L, Sparso T, Kestler HA, Chheda H, Eisele L, Gustafsson S, Fränberg M, Strawbridge RJ, Benediktsson R, Hreidarsson AB, Kong A, Sigurðsson G, Kerrison ND, Luan J, Liang L, Meitinger T, Roden M, Thorand B, Esko T, Mihailov E, Fox C, Liu CT, Rybin D, Isomaa B, Lyssenko V, Tuomi T, Couper DJ, Pankow JS, Grarup N, Have CT, Jørgensen ME, Jørgensen T, Linneberg A, Cornelis MC, van Dam RM, Hunter DJ, Kraft P, Sun Q, Edkins S, Owen KR, Perry JRB, Wood AR, Zeggini E, Tajes-Fernandes J, Abecasis GR, Bonnycastle LL, Chines PS, Stringham HM, Koistinen HA, Kinnunen L, Sennblad B, Mühleisen TW, Nöthen MM, Pechlivanis S, Baldassarre D, Gertow K, Humphries SE, Tremoli E, Klopp N, Meyer J, Steinbach G, Wennauer R, Eriksson JG, Männistö S, Peltonen L, Tikkanen E, Charpentier G, Eury E, Lobbens S, Gigante B, Leander K, McLeod O,

- Bottinger EP, Gottesman O, **Ruderfer D**, Blüher M, Kovacs P, Tonjes A, Maruthur NM, Scapoli C, Erbel R, Jöckel KH, Moebus S, de Faire U, Hamsten A, Stumvoll M, Deloukas P, Donnelly PJ, Frayling TM, Hattersley AT, Ripatti S, Salomaa V, Pedersen NL, Boehm BO, Bergman RN, Collins FS, Mohlke KL, Tuomilehto J, Hansen T, Pedersen O, Barroso I, Lannfelt L, Ingelsson E, Lind L, Lindgren CM, Cauchi S, Froguel P, Loos RJF, Balkau B, Boeing H, Franks PW, Barricarte Gurrea A, Palli D, van der Schouw YT, Altshuler D, Groop LC, Langenberg C, Wareham NJ, Sijbrands E, van Duijn CM, Florez JC, Meigs JB, Boerwinkle E, Gieger C, Strauch K, Metspalu A, Morris AD, Palmer CNA, Hu FB, Thorsteinsdottir U, Stefansson K, Dupuis J, Morris AP, Boehnke M, McCarthy MI, Prokopenko I; DIABetes Genetics Replication And Meta-analysis (DIAGRAM) Consortium. An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*. 2017 Nov;66(11):2888-2902. doi: 10.2337/db16-1253. Epub 2017 May 31.
84. Topol A, Zhu S, Hartley BJ, English J, Hauberg ME, Tran N, Rittenhouse CA, Simone A, **Ruderfer DM**, Johnson J, Readhead B, Hadas Y, Gochman PA, Wang YC, Shah H, Cagney G, Rapoport J, Gage FH, Dudley JT, Sklar P, Mattheisen M, Cotter D, Fang G, Brennand KJ. Dysregulation of miRNA-9 in a Subset of Schizophrenia Patient-Derived Neural Progenitor Cells. *Cell Rep*. 2017 Sep 5;20(10):2525. doi: 10.1016/j.celrep.2017.08.073. No abstract available.
85. Belbin GM, Odgis J, Sorokin EP, Yee MC, Kohli S, Glicksberg BS, Gignoux CR, Wojcik GL, Van Vleck T, Jeff JM, Linderman M, Schurmann C, **Ruderfer D**, Cai X, Merkelson A, Justice AE, Young KL, Graff M, North KE, Peters U, James R, Hindorff L, Kornreich R, Edelmann L, Gottesman O, Stahl EE, Cho JH, Loos RJ, Bottinger EP, Nadkarni GN, Abul-Husn NS, Kenny EE. Genetic identification of a common collagen disease in puerto ricans via identity-by-descent mapping in a health system. *Elife*. 2017 Sep 12;6. pii: e25060. doi: 10.7554/eLife.25060.
86. Nguyen HT, Bryois J, Kim A, Dobbyn A, Huckins LM, Munoz-Manchado AB, **Ruderfer DM**, Genovese G, Fromer M, Xu X, Pinto D, Linnarsson S, Verhage M, Smit AB, Hjerling-Leffler J, Buxbaum JD, Hultman C, Sklar P, Purcell SM, Lage K, He X, Sullivan PF, Stahl EA. Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. *Genome Med*. 2017 Dec 20;9(1):114. doi: 10.1186/s13073-017-0497-y.
87. Hoffman GE, Hartley BJ, Flaherty E, Ladran I, Gochman P, **Ruderfer DM**, Stahl EA, Rapoport J, Sklar P, Brennand KJ. Transcriptional signatures of schizophrenia in hiPSC-derived NPCs and neurons are concordant with post-mortem adult brains. *Nat Commun*. 2017 Dec 20;8(1):2225. doi: 10.1038/s41467-017-02330-5.
88. Pardiñas AF, Holmans P, Pocklington AJ, Escott-Price V, Ripke S, Carrera N, Legge SE, Bishop S, Cameron D, Hamshere ML, Han J, Hubbard L, Lynham A, Mantripragada K, Rees E, MacCabe JH, McCarroll SA, Baune BT, Breen G, Byrne EM, Dannlowski U, Eley TC, Hayward C, Martin NG, McIntosh AM, Plomin R, Porteous DJ, Wray NR, Caballero A, Geschwind DH, Huckins LM, **Ruderfer DM**, Santiago E, Sklar P, Stahl EA, Won H, Agerbo E, Als TD, Andreassen OA, Bækvad-Hansen M, Mortensen PB, Pedersen CB, Børglum AD, Bybjerg-Grauholm J, Djurovic S, Durmishi N, Pedersen MG, Golimbet V, Grove J, Hougaard DM, Mattheisen M, Molden E, Mors O, Nordentoft M, Pejovic-Milovancevic M, Sigurdsson E, Silagadze T, Hansen CS, Stefansson K, Stefansson H, Steinberg S, Tosato S, Werge T; GERAD1 Consortium; CRESTAR Consortium; Collier DA, Rujescu D, Kirov G, Owen MJ, O'Donovan MC, Walters JTR; GERAD1 Consortium; CRESTAR Consortium; GERAD1 Consortium; CRESTAR Consortium. Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. *Nat Genet*. 2018 Mar;50(3):381-389. doi: 10.1038/s41588-018-0059-2. Epub 2018 Feb 26.
89. Maier RM, Zhu Z, Lee SH, Trzaskowski M, **Ruderfer DM**, Stahl EA, Ripke S, Wray NR, Yang J, Visscher PM, Robinson MR. Improving genetic prediction by leveraging genetic correlations among human diseases and traits. *Nat Commun*. 2018 Mar 7;9(1):989. doi: 10.1038/s41467-017-02769-6.
90. Bastarache L, Hughey JJ, Hebring S, Marlo J, Zhao W, Ho WT, Van Driest SL, McGregor TL, Mosley JD, Wells QS, Temple M, Ramirez AH, Carroll R, Osterman T, Edwards T, **Ruderfer D**, Velez Edwards DR, Hamid R,

Cogan J, Glazer A, Wei WQ, Feng Q, Brilliant M, Zhao ZJ, Cox NJ, Roden DM, Denny JC. Phenotype risk scores identify patients with unrecognized Mendelian disease patterns. *Science*. 2018 Mar 16;359(6381):1233-1239. doi: 10.1126/science.aal4043.

91. Chen CY, Lee PH, Castro VM, Minnier J, Charney AW, Stahl EA, **Ruderfer DM**, Murphy SN, Gainer V, Cai T, Jones I, Pato CN, Pato MT, Landén M, Sklar P, Perlis RH, Smoller JW. Genetic validation of bipolar disorder identified by automated phenotyping using electronic health records. *Transl Psychiatry*. 2018 Apr 18;8(1):86. doi: 10.1038/s41398-018-0133-7.
92. Bipolar Disorder and Schizophrenia Working Group of the Psychiatric Genetics Consortium. Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Bipolar Disorder and Schizophrenia Working Group of the Psychiatric Genomics Consortium. *Cell*. 2018 Jun 14;173(7):1705-1715.e16. doi: 10.1016/j.cell.2018.05.046. ***Lead and corresponding author**
93. Gulyás-Kovács A, Keydar I, Xia E, Fromer M, Hoffman G, **Ruderfer D**, Sachidanandam R, Chess A. Unperturbed expression bias of imprinted genes in schizophrenia. *Nat Commun*. 2018 Jul 25;9(1):2914. doi: 10.1038/s41467-018-04960-9.
94. Velthorst E, Froudust-Walsh S, Stahl E, **Ruderfer D**, Ivanov I, Buxbaum J; iPSYCH-Broad ASD Group, the IMAGEN consortium, Banaschewski T, Bokde ALW, Dipl-Psych UB, Büchel C, Quinlan EB, Desrivières S, Flor H, Frouin V, Garavan H, Gowland P, Heinz A, Ittermann B, Martinot MP, Artiges E, Nees F, Orfanos DP, Paus T, Poustka L, Hohmann S, Fröhner JH, Smolka MN, Walter H, Whelan R, Schumann G, Reichenberg A. Genetic risk for schizophrenia and autism, social impairment and developmental pathways to psychosis. *Transl Psychiatry*. 2018 Sep 26;8(1):204. doi: 10.1038/s41398-018-0229-0.
95. **Ruderfer DM***, Walsh CG, Aguirre MW, Ribeiro JD, Franklin JC, Rivas MA. Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide. *Mol Psychiatry*. 2019 Jan 4. doi: 10.1038/s41380-018-0326-8.
96. Charney AW, Stahl EA, Green EK, Chen CY, Moran JL, Chambert K, Belliveau Jr. RA, Forty L, Gordon-Smith K, Lee PH, Bromet EJ, Buckley PF, Escamilla MA, Fanous AH, Fochtmann LJ, Lehrer DS, Malaspina D, Marder SR, Morley CP, Nicolini H, Perkins DO, Rakofsky JJ, Rapaport MH, Medeiros H, Sobell JL, Backlund L, Bergen SE, Juréus A, Schalling M, Lichtenstein P, Knowles JA, Burdick KE, Jones I, Jones LA, Hultman CA, Perlis R, Purcell SM, McCarroll SA, Pato CN, Pato MT, Di Florio A, Craddock N, Landén M, Smoller JW, **Ruderfer DM***, Sklar P. Contribution of rare copy number variants to bipolar disorder risk is limited to schizoaffective cases. *Biol Psychiatry*. 2018 Dec 20. pii: S0006-3223(18)32087-0. doi: 10.1016/j.biopsych.2018.12.009. [Epub ahead of print] ***co-senior and corresponding author**
97. Huckins LM, Dobbyn A, **Ruderfer DM**, Hoffman G, Wang W, Pardiñas AF, Rajagopal VM, Als TD, T Nguyen H, Girdhar K, Boocock J, Roussos P, Fromer M, Kramer R, Domenici E, Gamazon ER, Purcell S; CommonMind Consortium; Schizophrenia Working Group of the Psychiatric Genomics Consortium; iPSYCH-GEMS Schizophrenia Working Group, Demontis D, Børglum AD, Walters JTR, O'Donovan MC, Sullivan P, Owen MJ, Devlin B, Sieberts SK, Cox NJ, Im HK, Sklar P, Stahl EA. Gene expression imputation across multiple brain regions provides insights into schizophrenia risk. *Nat Genet*. 2019 Apr;51(4):659-674. doi: 10.1038/s41588-019-0364-4. Epub 2019 Mar 25.
98. Stahl EA, Breen G, Forstner AJ, McQuillin A, Ripke S, Trubetskoy V, Mattheisen M, Wang Y, Coleman JRI, Gaspar HA, de Leeuw CA, Steinberg S, Pavlides JMW, Trzaskowski M, Byrne EM, Pers TH, Holmans PA, Richards AL, Abbott L, Agerbo E, Akil H, Albani D, Alliey-Rodriguez N, Als TD, Anjorin A, Antilla V, Awasthi S, Badner JA, Bækvad-Hansen M, Barchas JD, Bass N, Bauer M, Belliveau R, Bergen SE, Pedersen CB, Bøen E, Boks MP, Boocock J, Budde M, Bunney W, Burmeister M, Bybjerg-Grauholm J, Byerley W, Casas M, Cerrato F, Cervantes P, Chambert K, Charney AW, Chen D, Churchhouse C, Clarke TK, Coryell W, Craig DW, Cruceanu C,

Curtis D, Czerski PM, Dale AM, de Jong S, Degenhardt F, Del-Favero J, DePaulo JR, Djurovic S, Dobbyn AL, Dumont A, Elvsåshagen T, Escott-Price V, Fan CC, Fischer SB, Flickinger M, Foroud TM, Forty L, Frank J, Fraser C, Freimer NB, Frisén L, Gade K, Gage D, Garnham J, Giambartolomei C, Pedersen MG, Goldstein J, Gordon SD, Gordon-Smith K, Green EK, Green MJ, Greenwood TA, Grove J, Guan W, Guzman-Parra J, Hamshere ML, Hautzinger M, Heilbronner U, Herms S, Hipolito M, Hoffmann P, Holland D, Huckins L, Jamain S, Johnson JS, Juréus A, Kandaswamy R, Karlsson R, Kennedy JL, Kittel-Schneider S, Knowles JA, Kogevinas M, Koller AC, Kupka R, Lavebratt C, Lawrence J, Lawson WB, Leber M, Lee PH, Levy SE, Li JZ, Liu C, Lucae S, Maaser A, MacIntyre DJ, Mahon PB, Maier W, Martinsson L, McCarroll S, McGuffin P, McInnis MG, McKay JD, Medeiros H, Medland SE, Meng F, Milani L, Montgomery GW, Morris DW, Mühleisen TW, Mullins N, Nguyen H, Nievergelt CM, Adolfsson AN, Nwulia EA, O'Donovan C, Loohuis LMO, Ori APS, Oruc L, Ösby U, Perlis RH, Perry A, Pfennig A, Potash JB, Purcell SM, Regeer EJ, Reif A, Reinbold CS, Rice JP, Rivas F, Rivera M, Roussos P, **Ruderfer DM**, Ryu E, Sánchez-Mora C, Schatzberg AF, Scheftner WA, Schork NJ, Shannon Weickert C, Shekhtman T, Shilling PD, Sigurdsson E, Slaney C, Smeland OB, Sobell JL, Söholm Hansen C, Spijker AT, St Clair D, Steffens M, Strauss JS, Streit F, Strohmaier J, Szlinger S, Thompson RC, Thorgeirsson TE, Treutlein J, Vedder H, Wang W, Watson SJ, Weickert TW, Witt SH, Xi S, Xu W, Young AH, Zandi P, Zhang P, Zöllner S; eQTLGen Consortium; BIOS Consortium, Adolfsson R, Agartz I, Alda M, Backlund L, Baune BT, Bellivier F, Berrettini WH, Biernacka JM, Blackwood DHR, Boehnke M, Børglum AD, Corvin A, Craddock N, Daly MJ, Dannlowski U, Esko T, Etain B, Frye M, Fullerton JM, Gershon ES, Gill M, Goes F, Grigoriu-Serbanescu M, Hauser J, Hougaard DM, Hultman CM, Jones I, Jones LA, Kahn RS, Kirov G, Landén M, Leboyer M, Lewis CM, Li QS, Lissowska J, Martin NG, Mayoral F, McElroy SL, McIntosh AM, McMahon FJ, Melle I, Metspalu A, Mitchell PB, Morken G, Mors O, Mortensen PB, Müller-Myhsok B, Myers RM, Neale BM, Nimgaonkar V, Nordentoft M, Nöthen MM, O'Donovan MC, Oedegaard KJ, Owen MJ, Paciga SA, Pato C, Pato MT, Posthuma D, Ramos-Quiroga JA, Ribasés M, Rietschel M, Rouleau GA, Schalling M, Schofield PR, Schulze TG, Serretti A, Smoller JW, Stefansson H, Stefansson K, Stordal E, Sullivan PF, Turecki G, Vaaler AE, Vieta E, Vincent JB, Werge T, Nurnberger JI, Wray NR, Di Florio A, Edenberg HJ, Cichon S, Ophoff RA, Scott LJ, Andreassen OA, Kelsoe J, Sklar P; Bipolar Disorder Working Group of the Psychiatric Genomics Consortium. Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nat Genet.* 2019 May;51(5):793-803. doi: 10.1038/s41588-019-0397-8.

99. Zheutlin AB, Dennis J, Karlsson Linnér R, Moscati A, Restrepo N, Straub P, **Ruderfer D**, Castro VM, Chen CY, Ge T, Huckins LM, Charney A, Kirchner HL, Stahl EA, Chabris CF, Davis LK, Smoller JW. Penetrance and Pleiotropy of Polygenic Risk Scores for Schizophrenia in 106,160 Patients Across Four Health Care Systems. *Am J Psychiatry.* 2019 Aug 16;appiajp201918091085. doi: 10.1176/appi.ajp.2019.18091085.

Significant manuscripts in review:

Szatkiewicz JP, Fromer M, Nonneman RJ, Ancalade N, Johnson JS, Stahl EA, Rees E, Bergen S, Hultman C, Kirov G, O'Donovan M, Owen M, Holmans P, Sklar P, Sullivan PF, Purcell SM, Crowley JJ, Douglas M. **Ruderfer DM***. Characterization of single gene copy number variants in schizophrenia. *BioRxiv* doi: <https://doi.org/10.1101/550863>

Han L, Zhao X, Benton ML, Perumal T, Collins RL, Hoffman, GE, Johnson JS, Sloofman L, CommonMind Consortium, Brennand KJ, Brand H, Sieberts SK, Marengo S, Peters MA, Lipska BK, Roussos P, Capra JA, Talkowski M, **Ruderfer DM***. Functional annotation of rare structural variation in the human brain. *BioRxiv* doi: <https://doi.org/10.1101/711754>

Book Chapters:

AW Charney, JR Scarpa, **DM Ruderfer**, DS Charney. Drug Repositioning Opportunities in Psychiatry, Drug Repositioning: Approaches and Applications for Neurotherapeutics. CRC Press 2017

Invited Commentary:

Ruderfer DM, Dudley JT. Deep phenotyping predicts Huntington's genotype. *Nat Biotechnol.* 2016 Aug 9;34(8):823-4. doi: 10.1038/nbt.3648. No abstract available.

Invited scientific lectures (Internal/Local):

- 2015 From Genomics to Medicine: Genetic Discovery to Treatment Response in Schizophrenia. Vanderbilt Genetics Institute
- 2017 Genetic Dissection of Bipolar Disorder and Schizophrenia. Genetics Interest Group
- 2017 Opportunities for Precision Medicine in Behavioral Health. Center for Precision Medicine Retreat
- 2017 Enabling advances in neuropsych research through 'omic and informatic approaches. Vanderbilt Memory and Alzheimer's Center
- 2017 Leveraging Informatics for Precision Medicine in Behavioral Health. Department of Biomedical Informatics Research Colloquium
- 2018 Leveraging Machine Learning and EHR Data to Improve the Power of Genetic Studies in Behavioral Health, Seminars in Precision Medicine, Vanderbilt University Medical Center
- 2019 Quantifying Psychiatry through Genomics, EHR and Machine Learning, Department of Biomedical Informatics Seminar, Vanderbilt University Medical Center
- 2019 Genomic dissection of bipolar disorder and schizophrenia, including 28 subphenotypes. Executive Faculty Meeting, Vanderbilt University Medical Center

Invited scientific lectures and conference presentations (National):

- 2006 Population genomic analysis of outcrossing and recombination in yeast. (Poster). Yeast Genetics and Molecular Biology Meeting, Princeton, NJ
- 2009 A genome-wide study of rare copy-number variation in bipolar disorder. Center for Human Genetics Research Meeting, Massachusetts General Hospital, Boston, MA
- 2009 A combined genome-wide association analysis of schizophrenia and bipolar disorder in 20,000 individuals. (Poster). World Congress of Psychiatric Genetics, San Diego, CA
- 2011 Characterizing short exonic insertions and deletions (indels) and understanding their role in schizophrenia. Center for Human Genetics Research Meeting, Massachusetts General Hospital, Boston, MA
- 2013 Shared burden of ultra-rare disruptive mutations between bipolar disorder and schizophrenia further implicates calcium channel function. World Congress of Psychiatric Genetics, Boston, MA
- 2014 Integrating large-scale genetic studies with drug target information to inform drug design and repurposing (Poster). American College of Neuropsychopharmacology, Phoenix, AZ
- 2015 Complex genetic overlap between schizophrenia risk and antipsychotic response. Mood and Anxiety Program Meeting, Icahn School of Medicine at Mount Sinai, New York, NY
- 2015 Complex genetic overlap between schizophrenia risk and antipsychotic response. Psychiatric Genomics Club Meeting, Icahn School of Medicine at Mount Sinai, New York, NY
- 2015 From Genomics to Medicine: Genetic Discovery to Treatment Response in Schizophrenia. The Jackson Laboratory for Genomic Medicine, Farmington, CT
- 2015 Genic intolerance to copy number variation in 60,000 individuals and applications to identifying risk genes in schizophrenia (Poster). American Society of Human Genetics, Baltimore, MD
- 2015 Complex genetic overlap between schizophrenia risk and antipsychotic response (Poster). American College of Neuropsychopharmacology, Hollywood, FL
- 2016 From Genomics to Medicine: Genetic Discovery to Treatment Response in Schizophrenia. Institute for Genomic Medicine, Columbia University, New York, NY
- 2016 From Genomics to Medicine: Genetic Discovery to Treatment Response in Schizophrenia. Department of Psychiatry, Yale University, New Haven, CT
- 2016 From Genomics to Medicine: Genetic Discovery to Treatment Response in Schizophrenia. Vertex Pharmaceuticals, Boston, MA
- 2016 Complex genetic overlap between schizophrenia risk and antipsychotic response. Biology of Genomes, Cold Spring Harbor, NY
- 2017 Genomic dissection of Bipolar disorder and schizophrenia. World Congress of Psychiatric Genetics, Orlando, FL
- 2017 Genomic dissection of Bipolar disorder and schizophrenia (Poster). American College of Neuropsychopharmacology, Hollywood, FL

- 2018 Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide (Poster). Biology of Genomes, Cold Spring Harbor, NY
- 2018 Quantifying Psychiatry through Genomics, EHR and Machine Learning, Integrative Center for Neurogenetics, The University of California, Los Angeles, CA
- 2018 Quantifying Psychiatry through Genomics, EHR and Machine Learning, Genetic and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY
- 2018 Transcriptional consequences of Structural variation in post-mortem schizophrenia brains. Molecular Psychiatry Meeting, Kauai, HI
- 2018 Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide (Poster). American College of Neuropsychopharmacology, Hollywood, FL
- 2019 Quantifying Psychiatry through Genomics, EHR and Machine Learning, Department of Psychiatry, Johns Hopkins University, Baltimore, MD
- 2019 A personal perspective on polygenic risk scores: Applications and limitations, European Bioinformatics Institute workshop: Pharmacogenetics in Drug Discovery. Boston, MA

Invited scientific lectures and conference presentations (International):

- 2011 Identifying relevant pathways from common polygenic SNP burden in schizophrenia. MRC Centre for Neuropsychiatric Genetics & Genomics Meeting, Cardiff University, Cardiff, UK
- 2010 Identification and characterization of small insertion and deletion events in an exome sequenced sample of 50 schizophrenia patients and 58 controls. (Poster). World Congress of Psychiatric Genetics. Athens, Greece
- 2012 Analysis of recessive and compound heterozygous variants in a schizophrenia exome sequencing sample of 5,000 individuals. World Congress of Psychiatric Genetics, Hamburg, Germany
- 2014 Genic copy number variants in an exome-sequencing study of 4,978 schizophrenia cases and 6,256 controls. World Congress of Psychiatric Genetics, Copenhagen, Denmark
- 2014 Integrating large-scale genetic studies with drug target information to inform drug design and repurposing (poster). World Congress of Psychiatric Genetics, Copenhagen, Denmark
- 2015 Genic intolerance to copy number variation in 60,000 individuals and applications to identifying risk genes in schizophrenia. World Congress of Psychiatric Genetics, Toronto, Canada
- 2015 Complex genetic overlap between schizophrenia risk and antipsychotic response. Pharmacogenetics in Psychiatry, Toronto, Canada
- 2018 The Application of Stem Cell Models to Validate Rare and Common Variants Contributing to Schizophrenia (Discussant). Schizophrenia International Research Society, Florence, Italy
- 2018 Significant shared heritability underlies suicide attempt and clinically predicted probability of attempting suicide. World Congress of Psychiatric Genetics, Glasgow, Scotland

PATENTS

Genome-wide mapping of polymorphisms at nucleotide resolution with a single DNA microarray. PCT Patent Application PCT/US2007/000987.