RSF Summer Institute in Social Science Genomics 2021 Reading List

Note: Readings in brackets are optional.

Topic: Using Genetic Data in Sociology

- Conley, D. and Rauscher, E. (2012). "Genetic Interactions with Prenatal Social Environment: Effects on Academic and Behavioral Outcomes," *Journal of Health and Social Behavior* 54(1).
- Conley, D., Rauscher, E., Dawes, C., Magnusson P.K.E. and Siegel, M.L. (2013). "Heritability and the Equal Environments Assumption: Evidence from Multiple Samples of Misclassified Twins," *Behavior Genetics* 43(4).
- Guo, G., Fu Y., Lee, H., Cai, T. et al. (2014). "Genetic Bio-Ancestry and Social Construction of Racial Classification in Social Surveys in the Contemporary United States," *Demography* 51(1).
- Laidley, T., Domingue, B., Sinsub, P., Harris, K.M. and Conley, D., 2019. New evidence of skin color bias and health outcomes using sibling difference models: a research note. *Demography*, *56*(2), pp.753-762.
- Sotoudeh, R., Harris, K. M., & Conley, D. (2019). Effects of the peer metagenomic environment on smoking behavior. *Proceedings of the National Academy of Sciences*, *116*(33), 16302-16307.

Topic: Heritability

- Falconer, D, and T Mackay. (1996). "Continuous Variation." *Introduction to Quantitative Genetics*, 4th ed. Harlow, Essex: Longman Group Ltd, 101-107.
- Goldberger, A.S. (2005). "Structural Equation Models in Human Behavior Genetics." *Identification and Inference for Econometric Models Essays in Honor of Thomas Rothenberg*, eds. DW Andrews and JS Stock. Cambridge: Cambridge University Press, 11–26.
- Polderman, T.J.C., Benyamin, B, de Leeuw, C.A, Sullivan, P.F. et al. (2015). "Meta-analysis of the heritability of human traits based on fifty years of twin studies," *Nature Genetics* 47 (7).
- Sacerdote, B. (2011). "Nature and Nurture Effects On Children's Outcomes: What Have We Learned From Studies of Twins And Adoptees?" *Handbook of Social Economics*, Chapter 1.

[Goldberger, A.S. (1979). "Heritability," *Economica* 46 (184).]

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- [Jencks, C. (1980). "Heredity, Environment, and Public Policy Reconsidered." *American Sociological Review* 45 (5).]
- [Kwan, JS, S Purcell, and PC Sham. (2007). "Introduction to Biometrical Genetics." *Statistical Genetics: Gene Mapping through Linkage and Association*, eds. BN Neale, MA Ferreira, SE Medland, and D Posthuma. Taylor & Francis Group, 17–42.]

Topic: Using Genetic Data in Psychology

Harden, K. P. (2021). "Reports of my death were greatly exaggerated": Behavior genetics in the postgenomic era. *Annual Review of Psychology*, *72*, 37-60

Topic: Molecular Genetics

[Strachan, T. and Read, A. (2011). *Human Molecular Genetics*, 4th Ed.] *Note: Reference book – not available in list of pdfs, but will be available to borrow at the Summer Institute*

Topic: Social and Ethical Implications of Social-Science Genomics

- Martschenko, Daphne, Sam Trejo, and Benjamin W. Domingue (2019). "Genetics and Education: Recent Developments in the Context of an Ugly History and an Uncertain Future." *AERA Open*, 5(1), 1–15.
- Martschenko, Daphne (2020). "DNA Dreams': Teacher Perspectives on the Role and Relevance of Genetics for Education." *Research in Education*, 107(1), 33–54.
- Martschenko, Daphne, and Markia Smith (2021). "Genes do not operate in a vacuum, and neither should our research." *Nature Genetics*, 53, 255–256.

Topic: Gene Discovery I: Power, Candidate Genes, GWAS

Chabris, C. F., et al. (2012). "Most Reported Genetic Associations with General Intelligence Are Probably False Positives." *Psychological Science*, 23(11), 1314–1323. doi:10.1177/0956797611435528

- Rietveld, C. A., Conley, D., Eriksson, N, Esko, T. et al. (2014). "Replicability and Robustness of Genome-Wide Association Studies for Behavioral Traits," *Psychological Science* 25 (11).
- Rietveld, C. A., Conley, D., Eriksson, N, Esko, T. et al. (2014). "Replicability and Robustness of Genome-Wide Association Studies for Behavioral Traits, Supplemental Material" *Psychological Science* 25 (11).
- Bayarri, M. J., Benjamin, D., Berger, J. O., and Sellke, T. M. (2016). "Rejection Odds and Rejection Ratios: A Proposal for Statistical Practice in Testing Hypotheses," *Journal of Mathematical Psychology*.
- Lee, J. J., Wedow, R., Okbay, A. et al. (2018). "Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals, "*Nature Genetics*, 50, 1112-1121.
- Lee, J. J., Wedow, R., Okbay, A. et al. (2018). "Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals (Supplementary Information)," *Nature Genetics*, 50, 1112-1121.
- Mills, M. C., Barban, N., & Tropf, F. C. (2020). An introduction to statistical genetic data analysis. MIT Press Note: Reference book – not available in list of pdfs.
- [Okbay, A et al. (2016). "Genome-wide association study identifies 74 loci associated with educational attainment," *Nature*. Published online May 11.doi:10.1038/nature17671.]
- [Okbay, A et al (2016). "Genetic variants associated with subjective well-being, depressive symptoms and neuroticism identified through genome-wide analyses." *Nature Genetics*. Published online April 18.]
- [Evangelou, E. and Ioannidis, J. P. A. (2013). "Meta-analysis methods for genome-wide association studies and beyond," *Nature Reviews Genetics* 14.]
- [Price, A.L, Zaitlen, N.A., Reich, D. and Patterson, N. (2010). "New approaches to population stratification in genome-wide association studies," *Nature Reviews Genetics* 11.]
- [Sham, P. C. and Purcell, S. M. (2014). "Statistical power and significance testing in largescale genetics studies," *Nature Reviews Genetics* 15.]

- [Visscher, P. M., Wray, N.R., Zhang, Q. et al. (2017). "10 Years of GWAS Discovery: Biology, Function and Translation," *American Journal of Human Genetics*, 101:5-22. <u>https://doi.org/10.1016/j.ajhg.2017.06.005</u>.]
- [Mills, M.C. and Rahal, C. (2019). "A scientometric review of genome-wide association studies," *Communications Biology*, 2(9).]

Topic: Gene Discovery II: Winner's Curse and Stratification

- Gelman, A., and Carlin, J. (2014). "Beyond Power Calculations: Assessing Type S (Sign) and Type M (Magnitude) Errors," *Perspectives on Psychological Science*, 9(6).
- Lee, J. J., Wedow, R., Okbay, A. et al. (2018). "Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals (Supplementary Information, **Section 2.4-2.6**)," *Nature Genetics*, 50, 1112-1121.
- [Rietveld, C. A., Esko, T., Davies, G., Pers, et al. (2014). "Common genetic variants associated with cognitive performance identified using the proxy-phenotype method," *PNAS*, 111(38). Supplemental Section 8 ("Correction of Effect Sizes for Winner's Curse")]

Topic: Quality Control of Genomic Data

- Okbay, A., Beauchamp, J. P., Fontana, M. A., Lee, et al. (2016). "Genome-wide association study identifies 74 loci associated with educational attainment," *Nature*. Supplemental Section 1.5 ("Quality Control").
- Winkler, T. W., et al. (2014). "Quality control and conduct of genome-wide association meta-analyses." *Nature Protocols*, 9(5): 1192–1212.

Topic: LD Score Regression and Stratification

- Bulik-Sullivan, B., Loh, P, Finucane H.K., Ripke, S. et al. (2015). "LD Score regression distinguishes confounding from polygenicity in genome-wide association studies," *Nature Genetics* 47(3).
- Lee, J. J., Wedow, R., Okbay, A. et al. (2018). "Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals," *Nature Genetics*, 50, 1112-1121. Supplementary Information, Section 2
- Evans, L. M., Tahmasbi, R., Vrieze, S. I. et al. (2018). "Comparison of methods that use whole genome data to estimate the heritability and genetic architecture of complex traits," *Nature Genetics*, 50, 737-745. **Table 1**

- [Evans, L. M., Tahmasbi, R., Vrieze, S. I. et al. (2018). "Comparison of methods that use whole genome data to estimate the heritability and genetic architecture of complex traits," *Nature Genetics*, 50, 737-745.]
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- [Loh, P., Tucker, G., Bulik-Sullivan, B., VIIjalmas, et al. (2015). "Efficient Bayesian mixedmodel analysis increases association power in large cohorts," *Nature Genetics* 47(3).]
- [Novembre, J., Johnson, T., Bryc, K., Kutalik, Z et al. (2008). "Genes mirror geography within Europe," *Nature* 456(7218).]
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- [Yang, J., Zaitlen, N. A., Goddard, M. E., Visscher, P. M., & Price, A. L. (2014). "Advantages and pitfalls in the application of mixed-model association methods," *Nature Genetics* 46(2).]
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- [Lee, J. J., McGue, M., Iacono, W. G., and Chow, C. C. (2018). "The accuracy of LD Score regression as an estimator of confounding and genetic correlations in genome-wide association studies," *Genetic Epidemiology*, 42(8).]
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Topic: Genetic Correlation and Partitioning

- Bulik-Sullivan, B., Finucane, H. K., Anttila, Y., Gusev, A., et al. (2015). "An atlas of genetic correlations across human diseases and traits," *Nature Genetics* 47(11).
- Finucane, H. K., Bulik-Sullivan, B, Gusev, A., Trynka, G. et al. (2015). "Partitioning heritability by functional annotation using genome-wide association summary statistics," *Nature Genetics* 47(11).
- Okbay, A., Beauchamp, J. P., Fontana, M. A., Lee, et al. (2016). "Genome-wide association study identifies 74 loci associated with educational attainment," *Nature*. Supplemental Section 3 ("Genetic Overlap").
- Neale Lab blog posts: <u>http://www.nealelab.is/blog</u>

- [Lee, S. H., Yang, J., Goddard, M. E., VIsscher, P. M. and Wray N.R. (2012). "Estimation of pleiotropy between complex diseases using SNP-derived genomic relationships and restricted maximum likelihood," *Bioinformatics* 28(19).]
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- [Gazal, S., Marquez-Luna, C., Finucane, H.K. and Price, A.L. (2018). "Reconciling S-LDSC and LDAK models and functional enrichment estimates," bioRxiv, <u>https://doi.org/10.1101/256412</u>]
- [Brown, B. C., Asian Genetic Epidemiology Network Type 2 Diabetes Consortium, Ye, C. J., Price, A. L. and Zaitlen, N. (2016). "Transethnic Genetic-Correlation Estimates from Summary Statistics", *American Journal of Human Genetics*, 99(1).]
- [van Reenan, W., Peyrot, W.J., et al. (2019). "Genetic correlations of polygenic disease traitsfrom theory to practice", *Nature Reviews*.]

Topic: Responsibly Communicating Social-Science Genomics

FAQs for: Becker et al. (2021), "Resource profile and user guide of the Polygenic Index Repository," *Nature Human Behavior* (in press), available at <u>https://www.biorxiv.org/content/biorxiv/early/2021/05/10/2021.05.08.443158/DC5/e</u> <u>mbed/media-5.pdf?download=true</u>

→ Skim the whole thing to get a sense of the content and structure, but carefully read FAQs 1.4–1.6 & 3.1–3.7

FAQs for: Turley et al. (2021), "Multi-ancestry meta-analysis yields novel genetic discoveries and ancestry-specific associations," bioRxiv, available at <u>https://www.biorxiv.org/content/biorxiv/early/2021/04/24/2021.04.23.441003/DC1/e mbed/media-1.pdf?download=true</u>

→ Skim the whole thing to get a sense of the content and structure, but carefully read FAQs 2.6–2.9, 4.3 & 5.1–5.7

Broad Communications, "Perspectives on the complex genetics of same-sex sexual behavior," a Broad Institute website devoted to discussion of Ganna et al. (2019), "Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior,"

Science, <u>https://www.broadinstitute.org/news/perspectives-complex-genetics-same-sex-sexual-behavior</u>.

- → Read the webpage and at least a few of the critical "Broadminded blog" commentaries on the study linked at the bottom (and also in the right-hand side bar)
- → In the right-hand side bar, view at least a few of the 5 brief videos by senior author Ben Neale
- → In the right-hand side bar, click the link to a separate website (https://geneticsexbehavior.info) devoted to the study; browse the FAQs there and watch the animated video

Browse the Hastings Center repository of FAQs for a variety of social-science genomics studies and read at least one of relevance to you (e.g., a study of a phenotype you're interested in): https://www.thehastingscenter.org/genomics-research-index/

Turley et al. (2021), "Challenges with embryo selection using polygenic scores," *New England Journal of Medicine* (in press)

[Nuffield Council on Bioethics, Genetics and Human Behavior: The Ethical Context (2002)

- Summary and Recommendations, pp. xix-xxxiii
- Ch. 2, Historical Context, pp. 13-22
- Ch. 12: Determinism, pp. 121-130
- Ch. 13: Selecting & Changing Behavioural Traits, pp. 133-156
- Ch. 14: Legal Responsibility, pp. 149-171
- Ch. 15: Testing & Selection in Employment, Education, & Insurance, pp. 175-188]

[Nature editorial, "Dangerous Work," *Nature* 502: 5-6 (Oct. 3, 2013), <u>http://www.nature.com/polopoly_fs/1.13861!/menu/main/topColumns/topLeftColumn/pdf/502005b.pdf]</u>

[Hayden, E. C. (2013). Ethics: Taboo genetics. *Nature News*, *502*(7469), 26. Available at <u>http://www.nature.com/news/ethics-taboo-genetics-1.13858]</u>

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Topic: Polygenic Scores

- Daetwyler, H.D., Villaneuva, B., and Woolliams, J.A. (2008). "Accuracy of Predicting the Genetic Risk of Disease Using a Genome-Wide Approach," *PLoS One* 3(10).
- Vilhjálmsson, B.J., Yang J., Finucane, H.K., Gusev, A. et al. (2015). "Modeling linkage disequilibrium increases accuracy of polygenic risk scores." *The American Journal of Human Genetics* 97(4).
- Martin, A.R., Kanai, M. et al. (2019). "Clinical use of current polygenic risk scores may exacerbate health disparities." *Nature Genetics* (51), pp. 584-591.
- Martin, A.R., Daly, M.J. et al. (2019). "Predicting polygenic risk of psychiatric disorders." *Biological Psychiatry.*
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- de Vlaming, R., Okbay, A. Rietveld, C.A. et al. (2017). "Meta-GWAS Accuracy and power calculator shows that hiding heritability is partially due to imperfect genetic correlations across studies," *PLOS Genetics*, 13(1): e1006495.
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- [Ge, T., Chen, C., Ni, Y. et al. (2019). "Polygenic prediction via Bayesian regression and continuous shrinkage priors," *Nature Communications* 10(1776).]

Topic: Population Genetics

- Lee, J.J. (2019). "Psychology 5135: Class Notes on Genetics." Sections 1 and 4.
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- [Sella, Guy, and Nicholas H. Barton (2019). "Thinking About the Evolution of Complex Traits in the Era of Genome-Wide Association Studies." *Annual Review of Genomics and Human Genetics*, 20, 461–93.]

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Diverse Ancestry, Admixed Populations, and Eurocentric Bias

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Topic: Assortative Mating

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Topic: Gene-Environment Interactions

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Topic: Gene-Environment Correlation and Genetic Nurture

- Young, A. "Heritability estimation: what is heritability?" <u>https://geneticvariance.wordpress.com/2017/09/28/heritability-estimation-what-is-heritability-i/</u>
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Topic: Mendelian Randomization

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- [Verbanck M., Chen C. Y., Neale B. and Do R. (2018). "Detection of widespread horizontal pleiotropy in causal relationships inferred from Mendelian randomization between complex traits and diseases," *Nature Genetics* 50(5), 693-698.]

Topic: Epigenetics and Social Science

- Zimmer, Carl. Growing Pains for Field of Epigenetics as Some Call for Overhaul. NY Times. July 1, 2016. <u>https://www.nytimes.com/2016/07/02/science/epigenetic-marks-dna-genes.html?action=click&module=RelatedLinks&pgtype=Article</u>
- Carey, Benedict. Can We Really Inherit Trauma? NY Times. December 10, 2018. https://www.nytimes.com/2018/12/10/health/mind-epigenetics-genes.html
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- Horvath, S., and Raj, K. (2018). "DNA methylation-based biomarkers and the epigenetic clock theory of ageing," *Nature Reviews Genetics*, 19: 371-384. **[students: please read to the top of page 376]
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Topic: Genomic Structural Equation Modeling

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Topic: Applications in Social Science

TBD

Topic: Computing in Social Science Genomics

"Introduction to the Linux Command Line." https://cscar.research.umich.edu/wp-content/uploads/sites/5/2016/09/Intro-to-Command-Line.pdf.

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