

Human Genomics: Science, Ethics, and Society

Placing Recent Advances in Human Genetics and Genomics in Social Context

PHG 551 / B H 551 / GENOME 573 (3 credits)
Autumn 2014 | Thursdays 1:30-4:20 pm | Location T-663

<https://canvas.uw.edu/courses/913526>

Stephanie Malia Fullerton, *DPhil*
Associate Professor
Department of Bioethics & Humanities

Office: 616-1864
Email: smflrtn@uw.edu
Office hours: By Appointment

“Disappointed love, not hatred or aversion, is the strongest motivation behind the urge to make an argument sing, rather than shout. And that is why the screamers on every side leave me so cold. They argue without love— and they are not true polemicists.” -- Lee Siegel

*“In science it often happens that scientists say, ‘You know that’s a really good argument; my position is mistaken,’ and then they would actually change their minds and you never hear that old view from them again. They really do it. It doesn’t happen as often as it should, because scientists are human and change is sometimes painful. But it happens every day. I cannot recall the last time something like that happened in politics or religion.”
— Carl Sagan*

COURSE DESCRIPTION

This course aims to provide an overview of recent topics in human genetics and genomics while simultaneously placing those topics in a broader social and ethical context. The approach to learning is unstructured and collaborative. Students will explore, in a sustained manner, the science involved in recent research advances, ranging from next gen sequencing to mitochondrial gene transfer. They will also use a range of ethical arguments to assess the implications of such advances for scientists, clinicians, research participants, and society at large.

COURSE OBJECTIVES

At the end of this course, students should be able to:

1. Describe and discuss key genetic and genomic approaches to the identification of genes and genetic variants relevant to health and disease;
2. Identify the ways in which human genomic investigation both determines, and is determined by, prevailing social norms and values;
3. Apply scientific insights to the critical assessment of current ethical debates in human genetics and genomics.

COURSE PREREQUISITES

LAW H 504/B H 514/PHG 512 or permission of instructor.

COURSE REQUIREMENTS

1. **CLASS PARTICIPATION** (25%): including careful reading and engaged discussion
2. **COMMENTARY OUTLINES** (30%): three one-page bulleted outlines of a genomics research ethics-oriented argument, suitable for use as the basis of a full length commentary. Outlines will be due by 5 PM on the **SUNDAY AFTER** the class for which an outline has been assigned. Assignments will be determined by random draw on the first day of class.
3. **FINAL COMMENTARY** (40%): one 1500-2000 word essay, written in the form of a commentary appropriate for a general scientific audience. Please follow standard conventions for peer-reviewed publication, i.e. double-space throughout, adequately cite all sources, proof read your work, etc. This essay, which must be based on one of the three outlines submitted earlier in the quarter, is due **MONDAY, DECEMBER 8th at 5 PM**.
4. **SELF EVALUATION** (5%): write a 1-page statement of self-evaluation with respect to your course participation (while I welcome feedback on my performance, this is really supposed to be about YOU). What are you taking away from this experience? What helped your understanding the most? Due by **WEDNESDAY, DECEMBER 10th at 5 PM**.

COURSE TEXTS

Links to all readings (outlined in Course Schedule below) will be posted to the Syllabus page of the course website:

➤ <https://canvas.uw.edu/courses/913526>

COURSE POLICIES

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| ATTENDANCE | Regular attendance is expected. If you must miss more than one class meeting you will be required to make up work through additional writing assignments. Advance notice of anticipated absence(s) is appreciated. |
| ELECTRONIC SUBMISSION | I prefer to receive writing assignments electronically (via Assignments page). If access to, or regular engagement with, the website is likely to be a problem for you, please come see me so we can discuss alternative mechanisms for participation. |
| LATE ASSIGNMENTS | In the interest of fairness, late assignments (received after due dates/times noted above) will lose 5% of the final grade per late day. |

WEEK 1 (9/25): Introduction to Course, Art of Argument

- Siegel, L. (2010). "The lost art of argument." *The Wall Street Journal*. November 13.
- deLaplante, K. (2013). "How to write a good argumentative essay: logical structure." <http://www.youtube.com/watch?v=tAmgEa1B1vI&index=2&list=PLDF3C877967DFDEC6>
- deLaplante, K. (2013). "What is a good argument? (Parts I & II)" <http://www.youtube.com/watch?v=AlRYrBFoQqA>
<http://www.youtube.com/watch?v=B2WWeWUBhKU>
- deLaplante, K. (2013). "Identifying premises and conclusions." <http://www.youtube.com/watch?v=07mehbgE5jc>
- Vaughn, L. (2010). "Evaluating moral arguments." *Doing ethics: moral reasoning and contemporary issues*, 2nd edn. New York, W. W. Norton & Company. Focus on pp. 51-56.

Optional

- Northwest Association for Biomedical Research (2008). "Ethics Background" Excerpt from *an Ethics Primer: Lesson Ideas and Ethics Background*, pp. 15-25.
- Lee, L. M. (2012). "Public health ethics theory: review and path to convergence." *The Journal of Law, Medicine & Ethics* 40(1): 85-98.

WEEK 2 (10/2): Genomic (Big) Data Sharing and Implications for Informed Consent

- Paltoo, D. N., et al. (2014). "Data use under the NIH GWAS data sharing policy and future directions." *Nature Genetics*, 9: 934-938.
- NIH Genomic Data Sharing Policy, <http://grants.nih.gov/grants/guide/notice-files/NOT-OD-14-124.html>, Release Date: August 27 2014.
- Trinidad, S. B., et al. (2011). "Research ethics. Research practice and participant preferences: the growing gulf." *Science* 331(6015): 287-288.
- Allen, A. (2014). "Can Big Data and patient informed consent coexist?" *Politico*, September 3.
- Callier, S., Husain, R., & Simpson, R. (2014) "Genomic data-sharing: what will be our legacy?" *Frontiers in Genetics*, 5 (34): 1-3.
- Pereira, S., et al. (2014). "Open access data sharing in genomic research." *Genes* 5(3): 739-747.

Optional

- Wilson, A. (2014). "Sequenced in the U.S.A.: a desperate town hands over its DNA." *Pacific Standard*, July 21.
- Holt, N. (2014). "Bump tracker: nine months of Big Data." *The Atlantic*, August 12.

WEEK 3 (10/9): Return of “Incidental” Findings from Genomic Testing (J. Kocarnik)

- Christenhusz, G. M., et al. (2013). “To tell or not to tell? A systematic review of ethical reflections on incidental findings arising in genetics contexts.” *European Journal of Human Genetics*, 21(3): 248-255.
- Jarvik, G.P., et al. (2014). “Return of genomic results to research participants: the floor, the ceiling, and the choices in between.” *American Journal of Human Genetics*, 94(6):818-26.
- Green, R. C., et al. (2013). “ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing.” *Genetics in Medicine*, 15(7): 565-574.
- Kocarnik, J.M. & Fullerton, S.M. (2014). “Returning pleiotropic genetic results to patients and research participants.” *Journal of the American Medical Association*, 311(8):795-6.

Optional

- Burke, W., et al. (2013). “Recommendations for returning genomic incidental findings? We need to talk!” *Genetics in Medicine* 15(11): 854-859.
- Yu, J. H., et al. (2013). “Self-guided management of exome and whole-genome sequencing results: changing the results return model.” *Genetics in Medicine*, 15(9): 684-690.
- Dewey, F. D., et al. (2014). “Clinical interpretation and implications of whole-genome sequencing.” *Journal of the American Medical Association*, 311(10):1035-45.

WEEK 4 (10/16): Protecting Genomic Privacy in Research and Public Spaces

- Rodriguez, L. L., et al. (2013). “Research ethics. The complexities of genomic identifiability.” *Science*, 339(6117): 275-276.
- Erlich, Y. & Narayanan, A. (2014). “Routes for breaching and protecting genetic privacy.” *Nature Reviews Genetics* 15(6): 409-421.
- Belluz, J. (2014). “Genetic testing brings families together: and sometimes tears them apart.” *Vox*, September 9.
- Belluz, J. (2014). “23andMe reverses its decision to move to more lax privacy settings.” *Vox*, September 16.
- Sulmasy, D. P. (2014). "Naked bodies, naked genomes: the special (but not exceptional) nature of genomic information." *Genetics in Medicine*.
- Lee, S. S. J. & Borgelt, E. (in press) “Protecting posted genes: social networking and the limits of GINA.” *American Journal of Bioethics*.

Optional

- Brenner, S. E. (2013). “Be prepared for the big genome leak.” *Nature*, 498(7453): 139.
- Wagner, J. K., et al. (2014). “‘Use it or lose it’ as an alternative approach to protect genetic privacy in personalized medicine.” *Urologic Oncology* 32(2): 198-201.
- “Current issues in research ethics: privacy and confidentiality.” Columbia University Center for Bioethics. <http://ccnmtl.columbia.edu/projects/cire/pac/foundation/>

WEEK 5 (10/23): Addressing Kidney Disease Disparities with Genetic Testing

- Pollak, M. R., et al. (2012). “*APOL1* and kidney disease.” *Current Opinion in Nephrology and Hypertension* 21(2): 179-182.
- Freedman, B. I. (2013). “*APOL1* and nephropathy progression in populations of African ancestry.” *Seminars in Nephrology* 33(5): 425-432.
- Cohen, D. M., et al. (2011). “African American living-kidney donors should be screened for *APOL1* risk alleles.” *Transplantation* 92(7): 722-725.
- Fullerton, S. M., et al. (2012). “Finding a place for genomics in health disparities research.” *Public Health Genomics* 15(3-4): 156-163.
- Wailoo, K. (2006). “Stigma, race, and disease in 20th century America.” *Lancet* 367(9509): 531-533.

Optional

- Long, K. A., et al. (2011). “Attitudes and beliefs of African-Americans toward genetics, genetic testing, and sickle cell disease education and awareness.” *Journal of Genetic Counseling* 20(6): 572-592.
- Yu, J. H., et al. (2013). “Attitudes of African Americans toward return of results from exome and whole genome sequencing.” *American Journal of Medical Genetics. Part A*, 161A(5): 1064-1072.

WEEK 6 (10/30): Epigenetics and Geneticists’ “Obsession” with Parents and Parenthood

- Lane, M., et al. (2014). “Parenting from before conception.” *Science* 345(6198): 756-760.
- Richardson, S. S., et al. (2014). “Don’t blame the mothers.” *Nature*, 512: 131-132.
- Shulevitz, J. (2014). “Geneticists’ new obsession with pregnancy isn’t bad for women: how epigenetics can bolster feminism.” *New Republic*, August 17.
- Juengst, E. T., et al. (2014). “Serving epigenetics before its time.” *Trends in Genetics*, 30(10): 427-429.
- Schmidt, C. W. (2013). “Uncertain inheritance: transgenerational effects of environmental exposures.” *Environmental Health Perspectives* 121(10): A298-303.

Optional

- Russell, A. (2014). “Epigenetics, you, and your kids.” *Early Life Nutrition*, <http://www.kidspot.com.au/epigenetics-you-and-your-kids/>
- “Feminist Bioethics.” *Stanford Encyclopedia of Bioethics*, <http://plato.stanford.edu/entries/feminist-bioethics/>.

WEEK 7 (11/6): RNA-guided “Gene Drives”: Applications and Concerns

- Oye, K. A., et al. (2014). “Biotechnology. Regulating gene drives.” *Science* 345(6197): 626-628.
- Gurwitz, D. (2014). “Gene drives raise dual-use concerns.” *Science* 345(6200): 1010 with Oye, K. A. & Esvelt, K. M. “Response.” 1010-1011.
- Esvelt, K. M., A. L. Smidler, et al. (2014). “Concerning RNA-guided gene drives for the alteration of wild populations.” *eLife*: e03401.
- Sample, I. (2014). “Muscular dystrophy prevented in mice.” *The Guardian*, August 14.

Optional

- Grens, K. (2014). “CRISPR corrects blood disorder gene.” *The Scientist*, August 5.
- Grens, K. (2014). “Genome editing cuts out HIV.” *The Scientist*, July 21.
- Selgelid, M. J. (2009). “Governance of dual-use research: an ethical dilemma.” *Bulletin of the World Health Organization*, 87: 720-723.

WEEK 8 (11/13): The Perfect 46 (K. Edwards)

- ThePerfect46.com
- Manaster, J. (2014). “The Perfect 46: the future is near.” Scientific American Blog, <http://blogs.scientificamerican.com/psi-vid/2014/04/21/the-perfect-46-the-future-is-near/>
- De Lange, C. (2014). “Meet your unborn child – before its even conceived.” *New Scientist*, April 9.
- Tabor, H. K., et al. (2014). “Pathogenic variants for Mendelian and complex traits in exomes of 6,517 European and African Americans: implications for the return of incidental results.” *American Journal of Human Genetics* 95(2): 183-193.

Optional

- Bianchi, D. W., et al. (2014). “DNA sequencing versus standard prenatal aneuploidy screening.” *The New England Journal of Medicine* 370(9): 799-808.

WEEK 9 (11/20): Population-Based Screening for BRCA Mutations (W. Burke)

- King, M. C., et al. (2014). “Population-based screening for *BRCA1* and *BRCA2*: 2014 Lasker Award.” *Journal of the American Medical Association* 312(11): 1091-1092.
- Gabai-Kapara, E., et al. (in press). “Population-based screening for breast and ovarian cancer risk due to *BRCA1* and *BRCA2*.” *Proceedings of the National Academy of Sciences of the United States of America*.
- Burke, W., et al. (2011). “Genetic screening.” *Epidemiologic Reviews* 33(1): 148-164.

- Caskey, C. T., et al. (2014). “Adult genetic risk screening.” *Annual Review of Medicine* 65: 1-17.

Optional

- Altman, L. K. & Rabin, R. C. (2014). “Lasker winner calls for more genetic testing for cancer.” *The New York Times*, September 8.
- Burke, W., S. B. Trinidad, et al. (2013). “Seeking Genomic Knowledge: The Case for Clinical Restraint.” *The Hastings Law Journal* 64(6): 1650-1664.

11/27: THANKSGIVING DAY HOLIDAY – NO CLASS

WEEK 10 (12/4): Genomic Surveillance in the 2014 Ebola Outbreak in West Africa

- Gatherer, D. (2014). “The 2014 Ebola virus disease outbreak in West Africa.” *Journal of General Virology* 95(Pt 8): 1619-1624.
- Gire, S. K., et al. (2014). “Genomic surveillance elucidates Ebola virus origin and transmission during the 2014 outbreak.” *Science* 345(6202): 1369-1372.
- Vogel, G. (2014). “Infectious Disease. Genomes reveal start of Ebola outbreak.” *Science* 345(6200): 989-990.
- Jack, C. & Berlinger, N. (2014). “Responding to Ebola: organizational ethics, frontline perspectives.” *Bioethics Forum - The Hastings Center*.

Optional

- Hayden, E. C. (2014). “Ebola virus mutating rapidly as it spreads.” *Nature News*, August 28.
- Vogel, G. (2014). “Ebola’s heavy toll on study authors.” *Science News*, August 28.
- Sabeti, P. (2014). “Studying Ebola, then dying from it.” *New York Times*, September 5.

SELF-EVALUATIONS DUE 12/10 BY 5 PM

EQUAL OPPORTUNITY STATEMENT

The University of Washington reaffirms its policy of equal opportunity regardless of race, color, creed, religion, national origin, sex, sexual orientation, age, marital status, disability, or status as a disabled veteran or Vietnam-era veteran in accordance with University of Washington policy and applicable federal and state statutes and regulations.

DISABILITY ACCOMMODATION

The University of Washington is committed to providing access, equal opportunity and reasonable accommodation in its services, programs, activities, education and employment for individuals with disabilities. For information or to request disability accommodation contact: Disabled Students Services (Seattle campus) at (206) 543-8924/V, (206) 543-8925/TTY, (206) 616-8379/Fax, or e-mail at uwdss@u.washington.edu.

ACADEMIC INTEGRITY

Matters of academic integrity are handled by the Vice Provost for Student Relations and the Committee for Academic Misconduct. The Office of the Vice Provost for Student Relations assumes responsibility for investigating and resolving allegations of scientific and scholarly misconduct by faculty, students, and staff of the University of Washington. It coordinates, in consultation and cooperation with the Schools and Colleges, inquiries and investigations into allegations of scientific and scholarly misconduct. The Office of the Vice Provost for Student Relations is responsible for compliance with reporting requirements established by various Federal and other funding agencies in matters of scientific or scholarly misconduct. The Vice Provost maintains all records resulting from inquiries and investigations of such allegations. University rules (Handbook, Vol. II, Section 25-51, Executive Order #61) define scientific and scholarly misconduct to include the following forms of inappropriate activities: intentional misrepresentation of credentials; falsification of data; plagiarism; abuse of confidentiality; deliberate violation of regulations applicable to research. Students can report cases of scientific or scholarly misconduct either to the Office of the Vice Provost for Student Relations, to their faculty adviser, or the department Chair. The student should report such problems to whomever he or she feels most comfortable.