Genomics Salon: *Health Disparities and the Limits of Genomics*

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Discussion Questions

- What does it meant to diversify genomic research? And why is it important?
- Why might some consider the APOL1 case to be a success story of genomics for health disparities?
- What assumptions must be made in order to call this a success for health disparities? What information is still needed?
- Given the limitations, what role, if any, might genomics play in reducing health disparities?

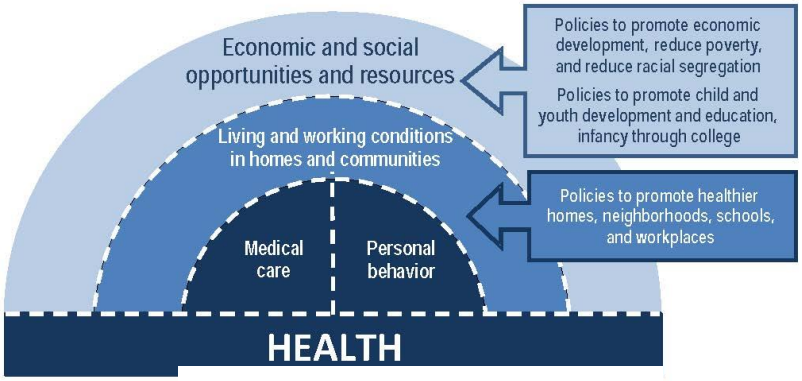
Key Definitions

Health disparities: "A particular type of health difference that is closely linked with social, economic, and/or environmental disadvantage. Health disparities adversely affect groups of people who have systematically experienced greater obstacles to health based on their racial or ethnic group; religion; socioeconomic status; gender; age; mental health; cognitive, sensory, or physical disability; sexual orientation or gender identity; geographic location; or other characteristics historically linked to discrimination or exclusion," (Secretary's Advisory Committee on Health Promotion and Disease Prevention Objectives for 2020, 2008, p. 28).

Social determinants of health (SDOH): social (including economic) factors with important direct or indirect effects on health. (See figure.)

Population health differences: Differences in health outcomes among population groups.

Health care disparities: Differences in access to and quality of healthcare among population groups.



SDOH: Braveman, et al. 2011. Am J Prev Med

APOL1: Apolipoprotein 1 gene; 2 variants found only in Sub-Saharan African ancestry. 13% of African Americans have the high-risk genotype; confers 7-10 fold increased risk of End-Stage Renal disease (ESRD).

Conclusions

- Concept of health disparities is unique, and about population health differences (e.g. PGx) vs. consensus in other scientific fields.
- Language of urgency: Emphasizes benefit to African Americans from APOL1 research, but benefit is speculative and undoubtedly a long way off; may prioritize genomics over SDOH.
- This research is potentially important for medical care, but not likely to reduce a health disparity: a) it does not address the root social causes, b) it operates on individuals not communities, and c) addresses a subtype only.
- Cultivating trustworthiness within genomics regarding minority communities includes developing clarity regarding benefits of participation and expected timelines, plus learning from the communities about what they value, to guide communication.

Selected Additional Resources

Commentary describing issues raised here: West, Blacksher & Burke. "Genomics, health disparities, and missed opportunities for the nation's research agenda." *JAMA*. 2017; 317(18): 1831-1832.

Health disparities and SDOH key references:
Braveman (2014). What are health disparities and health equity? We need to be clear. *PHR*, *129 Suppl 2*, 5–8.
Phelan, Link, & Tehranifar. (2010). Social conditions as fundamental causes of health inequalities: Theory, evidence, and policy implications. *Journal of Health and Social Behavior*, *51*(1 Suppl), S28–S40.
Woolf SH, et al. (2007). Giving everyone the health of the educated: an examination of whether social change would save more lives than medical advances. *American Journal of Public Health.* 97(4):679-683.
WHO Commission on Social Determinants of Health, 2008

Social gradient in health: Marmot, Rose, & Shipley. (1984). Inequalities in death—Specific explanations of a general pattern? *The Lancet*, *323*(8384), 1003–1006.

Early warnings about claims about genetics and health disparities: Sankar, P. et al. (2004). Genetic research and health disparities. *JAMA-Journal of the American Medical Association*, *291*(24), 2985–2989

Examples of harm and distrust: Gray, F. D. (2013). *The Tuskegee Syphilis Study: An insiders' account of the shocking medical experiment conducted by government doctors against African American men*. Montgomery, AL: New South Books.
Kennedy, B. R., Mathis, C. C., & Woods, A. K. (2007). African Americans and their distrust of the health care system: Healthcare for diverse populations. *Journal of Cultural Diversity*, *14*(2), 56–60.

Kidney disease statistics: US Renal Data System: *Annual data report: Epidemiology of kidney disease in the United States*. Bethesda, MD: National Institutes of Health. National Institute of Diabetes and Digestive and Kidney Disease. Retrieved from https://www.usrds.org/adr.aspx

APOL1 risk variants and epidemiology key references:
Freedman, et al. (2015). APOL1 and kidney disease: New insights leading to novel therapies. *American Journal of Kidney Diseases*, *66*(1), 9–11.
Genovese, et al. (2010). Association of trypanolytic ApoL1 variants with kidney disease in African Americans. *Science*, *329*(5993), 841–845.
Limou et al. (2014). APOL1 kidney risk alleles: population genetics and disease associations. *Advances in Chronic Kidney Disease*, *21*(5), 426–433.
Parsa et al. (2013). APOL1 risk variants, race, and progression of CKD. *NEJM*, *369*(23), 2183–2196.
Peralta, C. A., et al. (2015). APOL1 genotype and race differences in incident albuminuria and renal function decline. *JASN*. https://doi.org/10.1681/ASN.2015020124

Diversity in genomics: Popejoy & Fullerton. (2016). Genomics is failing on diversity. *Nature*, *538*(7624), 161–164.

Sample claims for genomics to reduce health disparities:
Rotimi, C., et al. (2013). Genome science and health disparities: a growing success story? *Gen Med*, *5*(7), 61.
Bustamente, et al. (2011). Genomics for the world. *Nature*, *475*(7355), 163–165.
Smith C. E., et al. (2016). Using genetic technologies to reduce, rather than widen, health disparities. *Health Affairs*, *35*(8), 1367–1373.
Williams & Pollack (2013). Health disparities in kidney disease--emerging data from the human genome. *NEJM*, *369*(23), 2260–2261.

Critical Discourse Analysis methods: Fairclough (1985). Critical and descriptive goals in discourse analysis. *Journal of Pragmatics*, *9*(6), 739–763.