

Suggested References for JHS 2017 ROR Workshop:

Recommendations and Considerations

- **Ethical and practical guidelines for reporting genetic research results to study participants: updated guidelines from a National Heart, Lung, and Blood Institute working group.** National Heart, Lung, and Blood Institute working group, Fabsitz RR, McGuire A, Sharp RR, Puggal M, Beskow LM, Biesecker LG, Bookman E, Burke W, Burchard EG, Church G, Clayton EW, Eckfeldt JH, Fernandez CV, Fisher R, Fullerton SM, Gabriel S, Gachupin F, James C, Jarvik GP, Kittles R, Leib JR, O'Donnell C, O'Rourke PP, Rodriguez LL, Schully SD, Shuldiner AR, Sze RK, Thakuria JV, Wolf SM, Burke GL. *Circ Cardiovasc Genet*. 2010 Dec;3(6):574-80. doi: 10.1161/CIRCGENETICS.110.958827. <https://www.ncbi.nlm.nih.gov/pubmed/21156933>
- **Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between.** Gail P. Jarvik, Laura M. Amendola, Jonathan S. Berg, Kyle Brothers, Ellen W. Clayton, Wendy Chung, Barbara J. Evans, James P. Evans, Stephanie M. Fullerton, Carlos J. Gallego, Nanibaa' A. Garrison, Stacy W. Gray, Ingrid A. Holm, Iftikhar J. Kullo, Lisa Soleymani Lehmann, Cathy McCarty, Cynthia A. Prows, Heidi L. Rehm, Richard R. Sharp, Joseph Salama, Saskia Sanderson, Sara L. Van Driest, Marc S. Williams, Susan M. Wolf, Wendy A. Wolf, eMERGE Act-ROR Committee and CERC Committee, CSER Act-ROR Working Group, and Wylie. *Am J Hum Genet*. 2014 Jun 5; 94(6): 818–826. doi: [10.1016/j.ajhg.2014.04.009](https://doi.org/10.1016/j.ajhg.2014.04.009) PMID: PMC4121476. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4121476/>
- **Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics.** Sarah S. Kalia ScM, Kathy Adelman, Sherri J. Bale PhD, Wendy K. Chung MD, PhD, Christine Eng MD, James P. Evans MD, PhD, Gail E. Herman MD, PhD, Sophia B. Hufnagel MD, Teri E. Klein PhD, Bruce R. Korf MD, PhD, Kent D. McKelvey MD, Kelly E. Ormond MS, C. Sue Richards PhD, Christopher N. Vlangos PhD, Michael Watson PhD, Christa L. Martin PhD & David T. Miller MD, PhD; on behalf of the ACMG Secondary Findings Maintenance Working Group. *Genetics in Medicine*. (2017). 19, 249–255. doi:10.1038/gim.2016.190. <http://www.nature.com/gim/journal/v19/n2/full/gim2016190a.html>
- **Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology.** Sue Richards PhD, Nazneen Aziz PhD, Sherri Bale PhD, David Bick MD, Soma Das PhD, Julie Gastier-Foster PhD, Wayne W. Grody MD, PhD, Madhuri Hegde PhD, Elaine Lyon PhD, Elaine Spector PhD, Karl Voelkerding MD & Heidi L. Rehm PhD; on behalf of the ACMG Laboratory Quality Assurance Committee. *Genetics in Medicine* (2015) 17, 405–423 doi:10.1038/gim.2015.30 <http://www.nature.com/gim/journal/v17/n5/full/gim201530a.html>
- **Regulatory changes raise troubling questions for genomic testing.** Barbara J. Evans, PhD, JD, Michael O. Dorschner, PhD, Wylie Burke, MD, PhD, and Gail P. Jarvik, MD, PhD. *Genet Med*. 2014 Nov; 16(11): 799–803. doi:10.1038/gim.2014.127 PMID: PMC4308037 NIHMSID: NIHMS656321 <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4308037/>
- **ANTICIPATE and COMMUNICATE. Ethical Management of Incidental and Secondary Findings in the Clinical, Research, and Direct-to-Consumer Contexts.** Washington, D.C., December 2013. Presidential Commission for the Study of Bioethics. <http://www.bioethics.gov> http://bioethics.gov/sites/default/files/FINALAnticipateCommunicate_PCSBI_0.pdf

Procedures and Resources

- **Returning a Research Participant's Genomic Results to Relatives: Analysis and Recommendations.** Wolf SM, Branum R, Koenig BA, Petersen GM, Berry SA, Beskow LM, Daly MB, Fernandez CV, Green RC, LeRoy BS, Lindor NM, O'Rourke PP, Breitkopf CR, Rothstein MA, Van Ness B, Wilfond BS. *J Law Med Ethics*. 2015 Fall;43(3):440-63. doi: 10.1111/jlme.12288. PMID: 26479555 PMCID: PMC4617203. <https://www.ncbi.nlm.nih.gov/pubmed/26479555>
- **Aggregate penetrance of genomic variants for actionable disorders in European and African Americans.** Natarajan P, Gold NB, Bick AG, McLaughlin H, Kraft P, Rehm HL, Peloso GM, Wilson JG, Correa A, Seidman JG, Seidman CE, Kathiresan S, Green RC. *Sci Transl Med*. 2016 Nov 9;8(364):364ra151. DOI: 10.1126/scitranslmed.aag2367 PMID: 27831900 <https://www.ncbi.nlm.nih.gov/pubmed/27831900>
- **Management of Hemoglobin Variants Detected Incidentally in HbA1c Testing: A Common Problem Currently Lacking a Standard Approach.** Lewis MR, Macauley RC, Sheehan PR, Staten MA, Phillips LS, Rasouli N, Pittas AG; D2d Research Group. *Diabetes Care*. 2017 Feb;40(2):e8-e9. doi: 10.2337/dc16-1667. Epub 2016 Nov 29. PMID: 27899488 PMCID: [PMC5250694](https://www.ncbi.nlm.nih.gov/pubmed/27899488) [Available on 2018-02-01 <https://www.ncbi.nlm.nih.gov/pubmed/27899488>

Stakeholders' Perspectives

- **Can I be sued for that? Liability risk and the disclosure of clinically significant genetic research findings.** McGuire AL, Knoppers BM, Zawati MH, Clayton EW. *Genome Res*. 2014 May;24(5):719-23. doi: 10.1101/gr.170514.113. Epub 2014 Mar 27. PMID: 24676095; PMCID: [PMC4009601](https://www.ncbi.nlm.nih.gov/pubmed/24676095) <https://www.ncbi.nlm.nih.gov/pubmed/24676095>
- **Attitudes of African Americans toward return of results from exome and whole genome sequencing.** Yu JH, Crouch J, Jamal SM, Tabor HK, Bamshad MJ. *Am J Med Genet A*. 2013 May;161A(5):1064-72. doi: 10.1002/ajmg.a.35914. PMID: 23610051 PMCID: [PMC3635144](https://www.ncbi.nlm.nih.gov/pubmed/23610051) <https://www.ncbi.nlm.nih.gov/pubmed/23610051>
- **Research Participants' Preferences for Hypothetical Secondary Results from Genomic Research.** Wynn J, Martinez J, Duong J, Chiuzan C, Phelan JC, Fyer A, Klitzman RL, Appelbaum PS, Chung WK. *J Genet Couns*. 2016 Dec 29. doi: 10.1007/s10897-016-0059-2. PMID: 28035592 <https://www.ncbi.nlm.nih.gov/pubmed/28035592>
- **The First Amendment Right to Speak about the Human Genome.** Barbara J. Evans. *University of Pennsylvania Journal of Constitutional Law* (2014), Vol. 16(3):549-636. U of Houston Law Center No. 2013-A-4. https://papers.ssrn.com/sol3/papers.cfm?abstract_id=2219522
- **Genetic research and health disparities.** Sankar P¹, Cho MK, Condit CM, Hunt LM, Koenig B, Marshall P, Lee SS, Spicer P. *JAMA*. 2004 Jun 23;291(24):2985-9. DOI: 10.1001/jama.291.24.2985 PMID: 15213210 PMCID: [PMC2271142](https://www.ncbi.nlm.nih.gov/pubmed/15213210). <https://www.ncbi.nlm.nih.gov/pubmed/15213210>
- **Research participants' opinions on genetic research and reasons for participation: a Jackson Heart Study focus group analysis.** Walker ER, Nelson CR, Antoine-LaVigne D, Thigpen DT, Puggal MA, Sarpong DE, Smith AM. *Ethn Dis*. 2014 Summer;24(3):290-7. PMID: 25065069 <https://www.ncbi.nlm.nih.gov/pubmed/25065069>