

Unit 9 References

- 100,000 Genomes Project Pilot Investigators, Smedley, D., Smith, K.R., Martin, A., Thomas, E.A., McDonagh, E.M., Cipriani, V., Ellingford, J.M., Arno, G., Tucci, A., Vandrovčová, J., Chan, G., Williams, H.J., Ratnaike, T., Wei, W., Stirrups, K., Ibanez, K., Moutsianas, L., Wielscher, M., Need, A., Barnes, M.R., Vestito, L., Buchanan, J., Wordsworth, S., Ashford, S., Rehmström, K., Li, E., Fuller, G., Twiss, P., Spasic-Boskovic, O., Halsall, S., Floto, R.A., Poole, K., Wagner, A., Mehta, S.G., Gurnell, M., Burrows, N., James, R., Penkett, C., Dewhurst, E., Gräf, S., Mapeta, R., Kasanicki, M., Haworth, A., Savage, H., Babcock, M., Reese, M.G., Bale, M., Baple, E., Boustred, C., Brittain, H., de Burca, A., Bleda, M., Devereau, A., Halai, D., Haraldsdóttir, E., Hyder, Z., Kasperaviciute, D., Patch, C., Polychronopoulos, D., Matchan, A., Sultana, R., Ryten, M., Tavares, A.L.T., Tregidgo, C., Turnbull, C., Welland, M., Wood, S., Snow, C., Williams, E., Leigh, S., Foulger, R.E., Daugherty, L.C., Niblock, O., Leong, I.U.S., Wright, C.F., Davies, J., Crichton, C., Welch, J., Woods, K., Abulhoul, L., Aurora, P., Bockenhauer, D., Broomfield, A., Cleary, M.A., Lam, T., Dattani, M., Footitt, E., Ganesan, V., Grunewald, S., Compeyrot-Lacassagne, S., Muntoni, F., Pilkington, C., Quinlivan, R., Thapar, N., Wallis, C., Wedderburn, L.R., Worth, A., Bueser, T., Compton, C., Deshpande, C., Fassihi, H., Haque, E., Izatt, L., Josifova, D., Mohammed, S., Robert, L., Rose, S., Ruddy, D., Sarkany, R., Say, G., Shaw, A.C., Wolejko, A., Habib, B., Burns, G., Hunter, S., Grocock, R.J., Humphray, S.J., Robinson, P.N., Haendel, M., Simpson, M.A., Banka, S., Clayton-Smith, J., Douzgou, S., Hall, G., Thomas, H.B., O'Keefe, R.T., Michaelides, M., Moore, A.T., Malka, S., Pontikos, N., Browning, A.C., Straub, V., Gorman, G.S., Horvath, R., Quinton, R., Schaefer, A.M., Yu-Wai-Man, P., Turnbull, D.M., McFarland, R., Taylor, R.W., O'Connor, E., Yip, J., Newland, K., Morris, H.R., Polke, J., Wood, N.W., Campbell, C., Camps, C., Gibson, K., Koelling, N., Lester, T., Németh, A.H., Palles, C., Patel, S., Roy, N.B.A., Sen, A., Taylor, J., Cacheiro, P., Jacobsen, J.O., Seaby, E.G., Davison, V., Chitty, L., Douglas, A., Naresh, K., McMullan, D., Ellard, S., Temple, I.K., Mumford, A.D., Wilson, G., Beales, P., Bitner-Glindzic, M., Black, G., Bradley, J.R., Brennan, P., Burn, J., Chinnery, P.F., Elliott, P., Flinter, F., Houlden, H., Irving, M., Newman, W., Rahman, S., Sayer, J.A., Taylor, J.C., Webster, A.R., Wilkie, A.O.M., Ouwehand, W.H., Raymond, F.L., Chisholm, J., Hill, S., Bentley, D., Scott, R.H., Fowler, T., Rendon, A., Caulfield, M., 2021. 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *N Engl J Med* 385, 1868–1880. <https://doi.org/10.1056/NEJMoa2035790>
- Abeysooriya, M., Soria, M., Kasu, M.S., Ziemann, M., 2021. Gene name errors: Lessons not learned. *PLoS Comput Biol* 17, e1008984. <https://doi.org/10.1371/journal.pcbi.1008984>
- Abramson, J., Adler, J., Dunger, J., Evans, R., Green, T., Pritch, A., Ronneberger, O., Willmore, L., Ballard, A.J., Bambrick, J., Bodenstein, S.W., Evans, D.A., Hung, C.-C., O'Neill, M., Reiman, D., Tunyasuvunakool, K., Wu, Z., Žemgulytė, A., Arvaniti, E., Beattie, C., Bertolli, O., Bridgland, A., Cherepanov, A., Congreve, M., Cowen-Rivers, A.I., Cowie, A., Figurnov, M., Fuchs, F.B., Gladman, H., Jain, R., Khan, Y.A., Low, C.M.R., Perlin, K., Potapenko, A., Savy, P., Singh, S., Stecula, A., Thillaisundaram, A., Tong, C., Yakneen, S., Zhong, E.D., Zielinski, M., Žídek, A., Bapst, V., Kohli, P., Jaderberg, M., Hassabis, D., Jumper, J.M., 2024. Accurate structure prediction of biomolecular interactions with AlphaFold 3. *Nature* 630, 493–500. <https://doi.org/10.1038/s41586-024-07487-w>

Abul-Husn, N.S., Manickam, K., Jones, L.K., Wright, E.A., Hartzel, D.N., Gonzaga-Jauregui, C., O'Dushlaine, C., Leader, J.B., Lester Kirchner, H., Lindbuchler, D.M., Barr, M.L., Giovanni, M.A., Ritchie, M.D., Overton, J.D., Reid, J.G., Metpally, R.P.R., Wardeh, A.H., Borecki, I.B., Yancopoulos, G.D., Baras, A., Shuldiner, A.R., Gottesman, O., Ledbetter, D.H., Carey, D.J., Dewey, F.E., Murray, M.F., 2016. Genetic identification of familial hypercholesterolemia within a single U.S. health care system. *Science* 354.

<https://doi.org/10.1126/science.aaf7000>

Adams, D., Gonzalez-Duarte, A., O'Riordan, W.D., Yang, C.-C., Ueda, M., Kristen, A.V., Tournev, I., Schmidt, H.H., Coelho, T., Berk, J.L., Lin, K.-P., Vita, G., Attarian, S., Planté-Bordeneuve, V., Mezei, M.M., Campistol, J.M., Buades, J., Brannagan, T.H., Kim, B.J., Oh, J., Parman, Y., Sekijima, Y., Hawkins, P.N., Solomon, S.D., Polydefkis, M., Dyck, P.J., Gandhi, P.J., Goyal, S., Chen, J., Strahs, A.L., Nochur, S.V., Sweetser, M.T., Garg, P.P., Vaishnav, A.K., Gollob, J.A., Suhr, O.B., 2018. Patisiran, an RNAi Therapeutic, for Hereditary Transthyretin Amyloidosis. *N Engl J Med* 379, 11–21.

<https://doi.org/10.1056/NEJMoa1716153>

Adler-Wagstyl, S., 2018. Making science better: openness, reproducibility and teamwork | UCL PhD journeys at Great Ormond Street Institute of Child Health. University College London. URL <https://blogs.ucl.ac.uk/phd-research-gosich/2018/11/14/making-science-better-openness-reproducibility-and-teamwork/> (accessed 5.9.21).

Ahmad, A., Khan, S.H., Khan, Z., 2022. The CRISPR/Cas Tool Kit for Genome Editing.

Ahmad, S.M., Ganai, N.A., Bhat, B., 2025. Bioinformatics Essentials: Core Principles and Techniques. Academic Press.

Albers, D.J., Elhadad, N., Claassen, J., Perotte, R., Goldstein, A., Hripcsak, G., 2018. Estimating summary statistics for electronic health record laboratory data for use in high-throughput phenotyping algorithms. *J Biomed Inform* 78, 87–101.

<https://doi.org/10.1016/j.jbi.2018.01.004>

Alix-Panabières, C., Pantel, K., 2025. Advances in liquid biopsy: From exploration to practical application. *Cancer Cell* 43, 161–165. <https://doi.org/10.1016/j.ccr.2024.11.009>

Alterovitz, G., Heale, B., Jones, J., Kreda, D., Lin, F., Liu, L., Liu, X., Mandl, K.D., Poloway, D.W., Ramoni, R., Wagner, A., Warner, J.L., 2020. FHIR Genomics: enabling standardization for precision medicine use cases. *NPJ Genom Med* 5, 13.

<https://doi.org/10.1038/s41525-020-0115-6>

Altman, R.B., 2012. Translational bioinformatics: linking the molecular world to the clinical world. *Clin Pharmacol Ther* 91, 994–1000. <https://doi.org/10.1038/clpt.2012.49>

Altman, R.B., Klein, T.E., 2002. Challenges for biomedical informatics and pharmacogenomics. *Annu Rev Pharmacol Toxicol* 42, 113–133.

<https://doi.org/10.1146/annurev.pharmtox.42.082401.140850>

Altmann, D.M., Rasmussen, A.L., 2025. How to respond when biomedical science and global health is under existential threat. *Nat Rev Immunol* 25, 313–314.

<https://doi.org/10.1038/s41577-025-01166-1>

Amaral, P., Carbonell-Sala, S., De La Vega, F.M., Faial, T., Frankish, A., Gingeras, T., Guigo, R., Harrow, J.L., Hatzigeorgiou, A.G., Johnson, R., Murphy, T.D., Pertea, M., Pruitt, K.D., Pujar, S., Takahashi, H., Ulitsky, I., Varabyou, A., Wells, C.A., Yandell, M., Carninci, P., Salzberg, S.L., 2023. The status of the human gene catalogue. *Nature* 622, 41–47.

<https://doi.org/10.1038/s41586-023-06490-x>

- André, F., Rassy, E., Marabelle, A., Michiels, S., Besse, B., 2024. Forget lung, breast or prostate cancer: why tumour naming needs to change. *Nature* 626, 26–29.
<https://doi.org/10.1038/d41586-024-00216-3>
- Annaratone, L., De Palma, G., Bonizzi, G., Sapino, A., Botti, G., Berrino, E., Mannelli, C., Arcella, P., Di Martino, S., Steffan, A., Daidone, M.G., Canzonieri, V., Parodi, B., Paradiso, A.V., Barberis, M., Marchiò, C., Alleanza Contro il Cancro (ACC) Pathology and Biobanking Working Group, 2021. Basic principles of biobanking: from biological samples to precision medicine for patients. *Virchows Arch* 479, 233–246.
<https://doi.org/10.1007/s00428-021-03151-0>
- Annual Economic Report, 2025. . United For Medical Research. URL
<https://www.unitedformedicalresearch.org/annual-economic-report/> (accessed 5.7.25).
- Artin, M.G., Stiles, D., Kiryluk, K., Chung, W.K., 2019. Cases in Precision Medicine: When Patients Present With Direct-to-Consumer Genetic Test Results. *Ann Intern Med* 170, 643–650. <https://doi.org/10.7326/M18-2356>
- Ashley, E.A., Butte, A.J., Wheeler, M.T., Chen, R., Klein, T.E., Dewey, F.E., Dudley, J.T., Ormond, K.E., Pavlovic, A., Morgan, A.A., Pushkarev, D., Neff, N.F., Hudgins, L., Gong, L., Hodges, L.M., Berlin, D.S., Thorn, C.F., Sangkuhl, K., Hebert, J.M., Woon, M., Sagreiya, H., Whaley, R., Knowles, J.W., Chou, M.F., Thakuria, J.V., Rosenbaum, A.M., Zaranek, A.W., Church, G.M., Greely, H.T., Quake, S.R., Altman, R.B., 2010. Clinical assessment incorporating a personal genome. *Lancet* 375, 1525–1535.
[https://doi.org/10.1016/S0140-6736\(10\)60452-7](https://doi.org/10.1016/S0140-6736(10)60452-7)
- Bakken, S., Sang, E., de Brujin, B., 2024. Returning value to communities from the All of Us Research Program through innovative approaches for data use, analysis, dissemination, and research capacity building. *J Am Med Inform Assoc* 31, 2773–2780.
<https://doi.org/10.1093/jamia/ocae276>
- Balwani, M., Sardh, E., Ventura, P., Peiró, P.A., Rees, D.C., Stölzel, U., Bissell, D.M., Bonkovsky, H.L., Windyga, J., Anderson, K.E., Parker, C., Silver, S.M., Keel, S.B., Wang, J.-D., Stein, P.E., Harper, P., Vassiliou, D., Wang, B., Phillips, J., Ivanova, A., Langendonk, J.G., Kauppinen, R., Minder, E., Horie, Y., Penz, C., Chen, J., Liu, S., Ko, J.J., Sweetser, M.T., Garg, P., Vaishnav, A., Kim, J.B., Simon, A.R., Gouya, L., ENVISION Investigators, 2020. Phase 3 Trial of RNAi Therapeutic Givosiran for Acute Intermittent Porphyria. *N Engl J Med* 382, 2289–2301. <https://doi.org/10.1056/NEJMoa1913147>
- Banks, M.A., 2020. Sequencing effort in Africa fine-tunes data collection. *Nat Med* 26, 3–4.
<https://doi.org/10.1038/s41591-019-0704-z>
- Bastarache, L., 2021. Using Phecodes for Research with the Electronic Health Record: From PheWAS to PheRS. *Annu Rev Biomed Data Sci* 4, 1–19. <https://doi.org/10.1146/annurev-biodatasci-122320-112352>
- Bastarache, L., Denny, J.C., Roden, D.M., 2022. Phenome-Wide Association Studies. *JAMA* 327, 75–76. <https://doi.org/10.1001/jama.2021.20356>
- Baxevanis, A.D., Bader, G.D., Wishart, D.S. (Eds.), 2020. Bioinformatics: A Practical Guide to the Analysis of Genes and Proteins. Wiley.
- Beaulieu-Jones, B.K., Wu, Z.S., Williams, C., Lee, R., Bhavnani, S.P., Byrd, J.B., Greene, C.S., 2019. Privacy-Preserving Generative Deep Neural Networks Support Clinical Data Sharing. *Circ Cardiovasc Qual Outcomes* 12, e005122.
<https://doi.org/10.1161/CIRCOUTCOMES.118.005122>

- Beitelshees, A.L., Veenstra, D.L., 2011. Evolving research and stakeholder perspectives on pharmacogenomics. *JAMA* 306, 1252–1253. <https://doi.org/10.1001/jama.2011.1343>
- Benegas, G., Ye, C., Albors, C., Li, J.C., Song, Y.S., 2024. Genomic Language Models: Opportunities and Challenges. <https://doi.org/10.48550/arXiv.2407.11435>
- Bennett, T.D., Moffitt, R.A., Hajagos, J.G., Amor, B., Anand, A., Bissell, M.M., Bradwell, K.R., Bremer, C., Byrd, J.B., Denham, A., DeWitt, P.E., Gabriel, D., Garibaldi, B.T., Girvin, A.T., Guinney, J., Hill, E.L., Hong, S.S., Jimenez, H., Kavuluru, R., Kostka, K., Lehmann, H.P., Levitt, E., Mallipattu, S.K., Manna, A., McMurry, J.A., Morris, M., Muschelli, J., Neumann, A.J., Palchuk, M.B., Pfaff, E.R., Qian, Z., Qureshi, N., Russell, S., Spratt, H., Walden, A., Williams, A.E., Wooldridge, J.T., Yoo, Y.J., Zhang, X.T., Zhu, R.L., Austin, C.P., Saltz, J.H., Gersing, K.R., Haendel, M.A., Chute, C.G., National COVID Cohort Collaborative (N3C) Consortium, 2021. Clinical Characterization and Prediction of Clinical Severity of SARS-CoV-2 Infection Among US Adults Using Data From the US National COVID Cohort Collaborative. *JAMA Netw Open* 4, e2116901. <https://doi.org/10.1001/jamanetworkopen.2021.16901>
- Bernstam, E.V., Hersh, W.R., Johnson, S.B., Chute, C.G., Nguyen, H., Sim, I., Nahm, M., Weiner, M.G., Miller, P., DiLaura, R.P., Overcash, M., Lehmann, H.P., Eichmann, D., Athey, B.D., Scheuermann, R.H., Anderson, N., Starren, J., Harris, P.A., Smith, J.W., Barbour, E., Silverstein, J.C., Krusch, D.A., Nagarajan, R., Becich, M.J., CTSA Biomedical Informatics Key Function Committee, 2009. Synergies and distinctions between computational disciplines in biomedical research: perspective from the Clinical and Translational Science Award programs. *Acad Med* 84, 964–970. <https://doi.org/10.1097/ACM.0b013e3181a8144d>
- Bierer, B.E., Barnes, M., Lynch, H.F., 2017a. Revised “Common Rule” Shapes Protections For Research Participants. *Health Aff (Millwood)* 36, 784–788. <https://doi.org/10.1377/hlthaff.2017.0307>
- Bierer, B.E., Crosas, M., Pierce, H.H., 2017b. Data Authorship as an Incentive to Data Sharing. *N Engl J Med* 376, 1684–1687. <https://doi.org/10.1056/NEJMsb1616595>
- Bourn, D., 2022. Diagnostic Genetic Testing. Springer.
- Boyle, E.A., Li, Y.I., Pritchard, J.K., 2017. An Expanded View of Complex Traits: From Polygenic to Omnipotent. *Cell* 169, 1177–1186. <https://doi.org/10.1016/j.cell.2017.05.038>
- Bradwell, K.R., Wooldridge, J.T., Amor, B., Bennett, T.D., Anand, A., Bremer, C., Yoo, Y.J., Qian, Z., Johnson, S.G., Pfaff, E.R., Girvin, A.T., Manna, A., Niehaus, E.A., Hong, S.S., Zhang, X.T., Zhu, R.L., Bissell, M., Qureshi, N., Saltz, J., Haendel, M.A., Chute, C.G., Lehmann, H.P., Moffitt, R.A., 2022. Harmonizing units and values of quantitative data elements in a very large nationally pooled electronic health record (EHR) dataset. *J Am Med Inform Assoc* ocac054. <https://doi.org/10.1093/jamia/ocac054>
- Brennan, P.F., 2016. The National Library of Medicine: Accelerating Discovery, Delivering Information, Improving Health. *Ann Intern Med* 165, 808–809. <https://doi.org/10.7326/M16-1737>
- Brieger, K., Zajac, G.J.M., Pandit, A., Foerster, J.R., Li, K.W., Annis, A.C., Schmidt, E.M., Clark, C.P., McMorrow, K., Zhou, W., Yang, J., Kwong, A.M., Boughton, A.P., Wu, J., Scheller, C., Parikh, T., de la Vega, A., Brazel, D.M., Frieser, M., Rea-Sandin, G., Fritzsche, L.G., Vrieze, S.I., Abecasis, G.R., 2019. Genes for Good: Engaging the Public in Genetics Research via Social Media. *Am J Hum Genet* 105, 65–77. <https://doi.org/10.1016/j.ajhg.2019.05.006>

- Buchanan, A.H., Lester Kirchner, H., Schwartz, M.L.B., Kelly, M.A., Schmidlen, T., Jones, L.K., Hallquist, M.L.G., Rocha, H., Betts, M., Schwiter, R., Butry, L., Lazzeri, A.L., Frisbie, L.R., Rahm, A.K., Hao, J., Willard, H.F., Martin, C.L., Ledbetter, D.H., Williams, M.S., Sturm, A.C., 2020. Clinical outcomes of a genomic screening program for actionable genetic conditions. *Genet Med* 22, 1874–1882. <https://doi.org/10.1038/s41436-020-0876-4>
- Budnik, B., Amirkhani, H., Forouzanfar, M.H., Afshin, A., 2024. Novel proteomics-based plasma test for early detection of multiple cancers in the general population. *BMJ Oncology* 3. <https://doi.org/10.1136/bmjonc-2023-000073>
- Buergel, T., Steinfeldt, J., Ruyoga, G., Pietzner, M., Bizzarri, D., Vojinovic, D., Upmeier Zu Belzen, J., Loock, L., Kittner, P., Christmann, L., Hollmann, N., Strangalies, H., Braunger, J.M., Wild, B., Chiesa, S.T., Spranger, J., Klostermann, F., van den Akker, E.B., Trompet, S., Mooijaart, S.P., Sattar, N., Jukema, J.W., Lavrijssen, B., Kavousi, M., Ghanbari, M., Ikram, M.A., Slagboom, E., Kivimaki, M., Langenberg, C., Deanfield, J., Eils, R., Landmesser, U., 2022. Metabolomic profiles predict individual multidisease outcomes. *Nat Med* 28, 2309–2320. <https://doi.org/10.1038/s41591-022-01980-3>
- Burd, A., Levine, R.L., Ruppert, A.S., Mims, A.S., Borate, U., Stein, E.M., Patel, P., Baer, M.R., Stock, W., Deininger, M., Blum, W., Schiller, G., Olin, R., Litzow, M., Foran, J., Lin, T.L., Ball, B., Boyiadzis, M., Traer, E., Odenike, O., Arellano, M., Walker, A., Duong, V.H., Kovacsics, T., Collins, R., Shoben, A.B., Heerema, N.A., Foster, M.C., Vergilio, J.-A., Brennan, T., Vietz, C., Severson, E., Miller, M., Rosenberg, L., Marcus, S., Yocum, A., Chen, T., Stefanos, M., Druker, B., Byrd, J.C., 2020. Precision medicine treatment in acute myeloid leukemia using prospective genomic profiling: feasibility and preliminary efficacy of the Beat AML Master Trial. *Nat Med* 26, 1852–1858. <https://doi.org/10.1038/s41591-020-1089-8>
- Burke, W., Parens, E., Chung, W.K., Berger, S.M., Appelbaum, P.S., 2022. The Challenge of Genetic Variants of Uncertain Clinical Significance : A Narrative Review. *Ann Intern Med*. <https://doi.org/10.7326/M21-4109>
- Burke, W., Trinidad, S.B., 2016. The Deceptive Appeal of Direct-to-Consumer Genetics. *Ann Intern Med* 164, 564–565. <https://doi.org/10.7326/M16-0257>
- Burotto, M., Manasanch, E.E., Wilkerson, J., Fojo, T., 2015. Gefitinib and erlotinib in metastatic non-small cell lung cancer: a meta-analysis of toxicity and efficacy of randomized clinical trials. *Oncologist* 20, 400–410. <https://doi.org/10.1634/theoncologist.2014-0154>
- Bycroft, C., Freeman, C., Petkova, D., Band, G., Elliott, L.T., Sharp, K., Motyer, A., Vukcevic, D., Delaneau, O., O'Connell, J., Cortes, A., Welsh, S., Young, A., Effingham, M., McVean, G., Leslie, S., Allen, N., Donnelly, P., Marchini, J., 2018. The UK Biobank resource with deep phenotyping and genomic data. *Nature* 562, 203–209. <https://doi.org/10.1038/s41586-018-0579-z>
- Campion, T.R., Craven, C.K., Dorr, D.A., Knosp, B.M., 2020. Understanding enterprise data warehouses to support clinical and translational research. *J Am Med Inform Assoc* 27, 1352–1358. <https://doi.org/10.1093/jamia/ocaa089>
- Cardoso, F., van't Veer, L.J., Bogaerts, J., Slaets, L., Viale, G., Delaloge, S., Pierga, J.-Y., Brain, E., Causeret, S., DeLorenzi, M., Glas, A.M., Golfinopoulos, V., Goulioti, T., Knox, S., Matos, E., Meulemans, B., Neijenhuis, P.A., Nitz, U., Passalacqua, R., Ravdin, P., Rubio, I.T., Saghatelian, M., Smilde, T.J., Sotiriou, C., Stork, L., Straehle, C., Thomas, G., Thompson, A.M., van der Hoeven, J.M., Vuylsteke, P., Bernards, R., Tryfonidis, K., Rutgers, E., Piccart, M., MINDACT Investigators, 2016. 70-Gene Signature as an Aid to

- Treatment Decisions in Early-Stage Breast Cancer. *N Engl J Med* 375, 717–729.
<https://doi.org/10.1056/NEJMoa1602253>
- Carmi, S., Greely, H.T., Mitchell, K.J., 2025. Human embryo editing against disease is unsafe and unproven - despite rosy predictions. *Nature* 637, 554–556.
<https://doi.org/10.1038/d41586-024-04105-7>
- Carter, A.B., Abruzzo, L.V., Hirschhorn, J.W., Jones, D., Jordan, D.C., Nassiri, M., Ogino, S., Patel, N.R., Suciu, C.G., Temple-Smolkin, R.L., Zehir, A., Roy, S., 2022. Electronic Health Records and Genomics: Perspectives from the Association for Molecular Pathology Electronic Health Record (EHR) Interoperability for Clinical Genomics Data Working Group. *J Mol Diagn* 24, 1–17. <https://doi.org/10.1016/j.jmoldx.2021.09.009>
- Cavalli, G., Heard, E., 2019. Advances in epigenetics link genetics to the environment and disease. *Nature* 571, 489–499. <https://doi.org/10.1038/s41586-019-1411-0>
- Cerezo, M., Sollis, E., Ji, Y., Lewis, E., Abid, A., Bircan, K.O., Hall, P., Hayhurst, J., John, S., Mosaku, A., Ramachandran, S., Foreman, A., Ibrahim, A., McLaughlin, J., Pendlington, Z., Stefancsik, R., Lambert, S.A., McMahon, A., Morales, J., Keane, T., Inouye, M., Parkinson, H., Harris, L.W., 2025. The NHGRI-EBI GWAS Catalog: standards for reusability, sustainability and diversity. *Nucleic Acids Res* 53, D998–D1005.
<https://doi.org/10.1093/nar/gkae1070>
- Chakravarty, D., Solit, D.B., 2021. Clinical cancer genomic profiling. *Nat Rev Genet.* <https://doi.org/10.1038/s41576-021-00338-8>
- Chen, J., Chun, D., Patel, M., Chiang, E., James, J., 2019. The validity of synthetic clinical data: a validation study of a leading synthetic data generator (Synthea) using clinical quality measures. *BMC Med Inform Decis Mak* 19, 44. <https://doi.org/10.1186/s12911-019-0793-0>
- Chen, L., Liu, H., Friedman, C., 2005. Gene name ambiguity of eukaryotic nomenclatures. *Bioinformatics* 21, 248–256. <https://doi.org/10.1093/bioinformatics/bth496>
- Chen, L., Liu, P., Evans, T.C., Ettwiller, L.M., 2017. DNA damage is a pervasive cause of sequencing errors, directly confounding variant identification. *Science* 355, 752–756.
<https://doi.org/10.1126/science.aai8690>
- Chen, R.J., Lu, M.Y., Williamson, D.F.K., Chen, T.Y., Lipkova, J., Noor, Z., Shaban, M., Shady, M., Williams, M., Joo, B., Mahmood, F., 2022. Pan-cancer integrative histology-genomic analysis via multimodal deep learning. *Cancer Cell* 40, 865–878.e6.
<https://doi.org/10.1016/j.ccr.2022.07.004>
- Cheng, A.C., Duda, S.N., Taylor, R., Delacqua, F., Lewis, A.A., Bosler, T., Johnson, K.B., Harris, P.A., 2021. REDCap on FHIR: Clinical Data Interoperability Services. *J Biomed Inform* 121, 103871. <https://doi.org/10.1016/j.jbi.2021.103871>
- Chin, L., Hahn, W.C., Getz, G., Meyerson, M., 2011. Making sense of cancer genomic data. *Genes Dev* 25, 534–555. <https://doi.org/10.1101/gad.2017311>
- Choi, E., Biswal, S., Malin, B., Duke, J., Stewart, W.F., Sun, J., 2017. Generating Multi-label Discrete Patient Records using Generative Adversarial Networks, in: Machine Learning for Healthcare. Presented at the Machine Learning for Healthcare, PMLR, pp. 286–305.
- Claussnitzer, M., Cho, J.H., Collins, R., Cox, N.J., Dermitzakis, E.T., Hurles, M.E., Kathiresan, S., Kenny, E.E., Lindgren, C.M., MacArthur, D.G., North, K.N., Plon, S.E., Rehm, H.L., Risch, N., Rotimi, C.N., Shendure, J., Soranzo, N., McCarthy, M.I., 2020. A brief history of human disease genetics. *Nature* 577, 179–189. <https://doi.org/10.1038/s41586-019-1879-7>

- Cline, H., Coolen, L., de Vries, S., Hyman, S., Segal, R., Steward, O., 2020. Recognizing Team Science Contributions in Academic Hiring, Promotion, and Tenure. *J Neurosci* 40, 6662–6663. <https://doi.org/10.1523/JNEUROSCI.1139-20.2020>
- Cohen, S.A., Liu, M.C., Aleshin, A., 2023. Practical recommendations for using ctDNA in clinical decision making. *Nature* 619, 259–268. <https://doi.org/10.1038/s41586-023-06225-y>
- Collins, F.S., 2009. The Language of Life: DNA and the Revolution in Personalized Medicine, Illustrated edition. ed. HarperCollins e-books.
- Collins, F.S., Doudna, J.A., Lander, E.S., Rotimi, C.N., 2021a. Human Molecular Genetics and Genomics — Important Advances and Exciting Possibilities. *New England Journal of Medicine* 0, null. <https://doi.org/10.1056/NEJMmp2030694>
- Collins, F.S., Hudson, K.L., Briggs, J.P., Lauer, M.S., 2014. PCORnet: turning a dream into reality. *J Am Med Inform Assoc* 21, 576–577. <https://doi.org/10.1136/amiajnl-2014-002864>
- Collins, F.S., Morgan, M., Patrinos, A., 2003. The Human Genome Project: lessons from large-scale biology. *Science* 300, 286–290. <https://doi.org/10.1126/science.1084564>
- Collins, F.S., Schwetz, T.A., Tabak, L.A., Lander, E.S., 2021b. ARPA-H: Accelerating biomedical breakthroughs. *Science* 373, 165–167. <https://doi.org/10.1126/science.abj8547>
- Collins, F.S., Varmus, H., 2015. A new initiative on precision medicine. *N Engl J Med* 372, 793–795. <https://doi.org/10.1056/NEJMmp1500523>
- Coorey, G., Figtree, G.A., Fletcher, D.F., Snelson, V.J., Vernon, S.T., Winlaw, D., Grieve, S.M., McEwan, A., Yang, J.Y.H., Qian, P., O'Brien, K., Orchard, J., Kim, J., Patel, S., Redfern, J., 2022. The health digital twin to tackle cardiovascular disease-a review of an emerging interdisciplinary field. *NPJ Digit Med* 5, 126. <https://doi.org/10.1038/s41746-022-00640-7>
- Crawford, D.C., Crosslin, D.R., Tromp, G., Kullo, I.J., Kuivaniemi, H., Hayes, M.G., Denny, J.C., Bush, W.S., Haines, J.L., Roden, D.M., McCarty, C.A., Jarvik, G.P., Ritchie, M.D., 2014. eMERGEing progress in genomics-the first seven years. *Front Genet* 5, 184. <https://doi.org/10.3389/fgene.2014.00184>
- Crellin, E., McClaren, B., Nisselle, A., Best, S., Gaff, C., Metcalfe, S., 2019. Preparing Medical Specialists to Practice Genomic Medicine: Education an Essential Part of a Broader Strategy. *Front Genet* 10, 789. <https://doi.org/10.3389/fgene.2019.00789>
- Dagliati, A., Malovini, A., Tibollo, V., Bellazzi, R., 2021. Health informatics and EHR to support clinical research in the COVID-19 pandemic: an overview. *Brief Bioinform* 22, 812–822. <https://doi.org/10.1093/bib/bbaa418>
- Dalerba, P., Sahoo, D., Paik, S., Guo, X., Yothers, G., Song, N., Wilcox-Fogel, N., Forgó, E., Rajendran, P.S., Miranda, S.P., Hisamori, S., Hutchison, J., Kalisky, T., Qian, D., Wolmark, N., Fisher, G.A., van de Rijn, M., Clarke, M.F., 2016. CDX2 as a Prognostic Biomarker in Stage II and Stage III Colon Cancer. *N Engl J Med* 374, 211–222. <https://doi.org/10.1056/NEJMoa1506597>
- Dalton, K.P., Rehm, H.L., Wright, M.W., Mandell, M.E., Krysiak, K., Babb, L., Riehle, K., Nelson, T., Wagner, A.H., 2023. Accessing clinical-grade genomic classification data through the ClinGen Data Platform. *Pac Symp Biocomput* 28, 531–535.
- Dance, A., 2024. Computational technologies of the Human Cell Atlas. *Nature* 635, 773–775. <https://doi.org/10.1038/d41586-024-03762-y>
- Daniels, R., Dzau, V., 2018. Supporting the Next Generation of Biomedical Researchers. *JAMA* 320, 29–30. <https://doi.org/10.1001/jama.2018.7902>

- Davis, W., 2023. Microsoft fixes the Excel feature that was wrecking scientific data [WWW Document]. The Verge. URL <https://www.theverge.com/2023/10/21/23926585/microsoft-excel-misreading-dates-human-genes-conversion-fixed> (accessed 5.9.24).
- de Vries, P.S., Kavousi, M., Ligthart, S., Uitterlinden, A.G., Hofman, A., Franco, O.H., Dehghan, A., 2015. Incremental predictive value of 152 single nucleotide polymorphisms in the 10-year risk prediction of incident coronary heart disease: the Rotterdam Study. *Int J Epidemiol* 44, 682–688. <https://doi.org/10.1093/ije/dyv070>
- Denny, J.C., Bastarache, L., Roden, D.M., 2016. Phenome-Wide Association Studies as a Tool to Advance Precision Medicine. *Annu Rev Genomics Hum Genet* 17, 353–373. <https://doi.org/10.1146/annurev-genom-090314-024956>
- Denny, J.C., Ritchie, M.D., Basford, M.A., Pulley, J.M., Bastarache, L., Brown-Gentry, K., Wang, D., Masys, D.R., Roden, D.M., Crawford, D.C., 2010. PheWAS: demonstrating the feasibility of a phenome-wide scan to discover gene-disease associations. *Bioinformatics* 26, 1205–1210. <https://doi.org/10.1093/bioinformatics/btq126>
- Denny, J.C., Rutter, J.L., Goldstein, D.B., Philippakis, A., Smoller, J.W., Jenkins, G., Dishman, E., 2019. The “All of Us” Research Program. *N Engl J Med* 381, 668–676. <https://doi.org/10.1056/NEJMsr1809937>
- Dewey, F.E., Murray, M.F., Overton, J.D., Habegger, L., Leader, J.B., Fetterolf, S.N., O’Dushlaine, C., Van Hout, C.V., Staples, J., Gonzaga-Jauregui, C., Metpally, R., Pendergrass, S.A., Giovanni, M.A., Kirchner, H.L., Balasubramanian, S., Abul-Husn, N.S., Hartzel, D.N., Lavage, D.R., Kost, K.A., Packer, J.S., Lopez, A.E., Penn, J., Mukherjee, S., Gosalia, N., Kanagaraj, M., Li, A.H., Mitnaul, L.J., Adams, L.J., Person, T.N., Praveen, K., Marcketta, A., Lebo, M.S., Austin-Tse, C.A., Mason-Suarez, H.M., Bruse, S., Mellis, S., Phillips, R., Stahl, N., Murphy, A., Economides, A., Skelding, K.A., Still, C.D., Elmore, J.R., Borecki, I.B., Yancopoulos, G.D., Davis, F.D., Fauci, W.A., Gottesman, O., Ritchie, M.D., Shuldiner, A.R., Reid, J.G., Ledbetter, D.H., Baras, A., Carey, D.J., 2016. Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. *Science* 354. <https://doi.org/10.1126/science.aaf6814>
- Dickson, D., Johnson, J., Bergan, R., Owens, R., Subbiah, V., Kurzrock, R., 2020. The Master Observational Trial: A New Class of Master Protocol to Advance Precision Medicine. *Cell* 180, 9–14. <https://doi.org/10.1016/j.cell.2019.12.009>
- Dolin, R.H., Heale, B.S.E., Alterovitz, G., Gupta, R., Aronson, J., Boxwala, A., Gothi, S.R., Haines, D., Hermann, A., Hongsermeier, T., Husami, A., Jones, J., Naeymi-Rad, F., Rapchak, B., Ravishankar, C., Shalaby, J., Terry, M., Xie, N., Zhang, P., Chamala, S., 2023. Introducing HL7 FHIR Genomics Operations: a developer-friendly approach to genomics-EHR integration. *J Am Med Inform Assoc* 30, 485–493. <https://doi.org/10.1093/jamia/ocac246>
- Dorr, D.A., Adams, L., Embí, P., 2023. Harnessing the Promise of Artificial Intelligence Responsibly. *JAMA* 329, 1347–1348. <https://doi.org/10.1001/jama.2023.2771>
- Doudna, J.A., 2020. The promise and challenge of therapeutic genome editing. *Nature* 578, 229–236. <https://doi.org/10.1038/s41586-020-1978-5>
- Dougherty, D., Conway, P.H., 2008. The “3T’s” road map to transform US health care: the “how” of high-quality care. *JAMA* 299, 2319–2321. <https://doi.org/10.1001/jama.299.19.2319>
- Druker, B.J., 2008. Translation of the Philadelphia chromosome into therapy for CML. *Blood* 112, 4808–4817. <https://doi.org/10.1182/blood-2008-07-077958>

- Duda, S.N., Kennedy, N., Conway, D., Cheng, A.C., Nguyen, V., Zayas-Cabán, T., Harris, P.A., 2022. HL7 FHIR-based tools and initiatives to support clinical research: a scoping review. *J Am Med Inform Assoc* 29, 1642–1653. <https://doi.org/10.1093/jamia/ocac105>
- Dzau, V.J., Shambaugh, E.L., Laitner, M.H., 2024. Crossing the Equity Chasm: Addressing a Second Valley of Death in Biomedical Innovation. *JAMA* 332, 1781–1782. <https://doi.org/10.1001/jama.2024.20677>
- El Emam, K., Mosquera, L., Jonker, E., Sood, H., 2021. Evaluating the utility of synthetic COVID-19 case data. *JAMIA Open* 4, ooab012. <https://doi.org/10.1093/jamiaopen/ooab012>
- Embi, P.J., Payne, P.R.O., 2013. Evidence generating medicine: redefining the research-practice relationship to complete the evidence cycle. *Med Care* 51, S87-91. <https://doi.org/10.1097/MLR.0b013e31829b1d66>
- Embi, P.J., Richesson, R., Tenenbaum, J., Kannry, J., Friedman, C., Sarkar, I.N., Smith, J., members of 2016 AMIA Policy Invitational Planning Committee, 2019. Reimagining the research-practice relationship: policy recommendations for informatics-enabled evidence-generation across the US health system. *JAMIA Open* 2, 2–9. <https://doi.org/10.1093/jamiaopen/ooy056>
- eMERGE Clinical Annotation Working Group, 2020. Frequency of genomic secondary findings among 21,915 eMERGE network participants. *Genet Med* 22, 1470–1477. <https://doi.org/10.1038/s41436-020-0810-9>
- eMERGE Consortium. Electronic address: agibbs@bcm.edu, eMERGE Consortium, 2019. Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *Am J Hum Genet* 105, 588–605. <https://doi.org/10.1016/j.ajhg.2019.07.018>
- eMERGE Consortiumdavid.crosslin@gmail.com, 2021. Lessons learned from the eMERGE Network: balancing genomics in discovery and practice. *HGG Adv* 2, 100018. <https://doi.org/10.1016/j.xhgg.2020.100018>
- ENCODE Project Consortium, 2012. An integrated encyclopedia of DNA elements in the human genome. *Nature* 489, 57–74. <https://doi.org/10.1038/nature11247>
- ENCODE Project Consortium, Birney, E., Stamatoyannopoulos, J.A., Dutta, A., Guigó, R., Gingeras, T.R., Margulies, E.H., Weng, Z., Snyder, M., Dermitzakis, E.T., Thurman, R.E., Kuehn, M.S., Taylor, C.M., Neph, S., Koch, C.M., Asthana, S., Malhotra, A., Adzhubei, I., Greenbaum, J.A., Andrews, R.M., Flicek, P., Boyle, P.J., Cao, H., Carter, N.P., Clelland, G.K., Davis, S., Day, N., Dhami, P., Dillon, S.C., Dorschner, M.O., Fiegler, H., Giresi, P.G., Goldy, J., Hawrylycz, M., Haydock, A., Humbert, R., James, K.D., Johnson, B.E., Johnson, E.M., Frum, T.T., Rosenzweig, E.R., Karnani, N., Lee, K., Lefebvre, G.C., Navas, P.A., Neri, F., Parker, S.C.J., Sabo, P.J., Sandstrom, R., Shafer, A., Vetrie, D., Weaver, M., Wilcox, S., Yu, M., Collins, F.S., Dekker, J., Lieb, J.D., Tullius, T.D., Crawford, G.E., Sunyaev, S., Noble, W.S., Dunham, I., Denoeud, F., Reymond, A., Kapranov, P., Rozowsky, J., Zheng, D., Castelo, R., Frankish, A., Harrow, J., Ghosh, S., Sandelin, A., Hofacker, I.L., Baertsch, R., Keefe, D., Dike, S., Cheng, J., Hirsch, H.A., Sekinger, E.A., Lagarde, J., Abril, J.F., Shahab, A., Flamm, C., Fried, C., Hackermüller, J., Hertel, J., Lindemeyer, M., Missal, K., Tanzer, A., Washietl, S., Korbel, J., Emanuelsson, O., Pedersen, J.S., Holroyd, N., Taylor, R., Swarbreck, D., Matthews, N., Dickson, M.C., Thomas, D.J., Weirauch, M.T., Gilbert, J., Drenkow, J., Bell, I., Zhao, X., Srinivasan, K.G., Sung, W.-K., Ooi, H.S., Chiu, K.P., Foissac, S., Alioto, T., Brent, M., Pachter, L., Tress, M.L., Valencia, A., Choo, S.W., Choo, C.Y., UCLA, C., Manzano, C., Wyss, C., Cheung, E., Clark, T.G., Brown, J.B., Ganesh, M., Patel, S., Tammana, H., Chrast, J., Henrichsen, C.N.,

Kai, C., Kawai, J., Nagalakshmi, U., Wu, J., Lian, Z., Lian, J., Newburger, P., Zhang, Xueqing, Bickel, P., Mattick, J.S., Carninci, P., Hayashizaki, Y., Weissman, S., Hubbard, T., Myers, R.M., Rogers, J., Stadler, P.F., Lowe, T.M., Wei, C.-L., Ruan, Y., Struhl, K., Gerstein, M., Antonarakis, S.E., Fu, Y., Green, E.D., Karaöz, U., Siepel, A., Taylor, J., Liefer, L.A., Wetterstrand, K.A., Good, P.J., Feingold, E.A., Guyer, M.S., Cooper, G.M., Asimenos, G., Dewey, C.N., Hou, M., Nikolaev, S., Montoya-Burgos, J.I., Löytynoja, A., Whelan, S., Pardi, F., Massingham, T., Huang, H., Zhang, N.R., Holmes, I., Mullikin, J.C., Ureta-Vidal, A., Paten, B., Seringhaus, M., Church, D., Rosenbloom, K., Kent, W.J., Stone, E.A., NISC Comparative Sequencing Program, Baylor College of Medicine Human Genome Sequencing Center, Washington University Genome Sequencing Center, Broad Institute, Children's Hospital Oakland Research Institute, Batzoglou, S., Goldman, N., Hardison, R.C., Haussler, D., Miller, W., Sidow, A., Trinklein, N.D., Zhang, Z.D., Barrera, L., Stuart, R., King, D.C., Ameur, A., Enroth, S., Bieda, M.C., Kim, J., Bhinge, A.A., Jiang, N., Liu, J., Yao, F., Vega, V.B., Lee, C.W.H., Ng, P., Shahab, A., Yang, A., Moqtaderi, Z., Zhu, Z., Xu, X., Squazzo, S., Oberley, M.J., Inman, D., Singer, M.A., Richmond, T.A., Munn, K.J., Rada-Iglesias, A., Wallerman, O., Komorowski, J., Fowler, J.C., Couttet, P., Bruce, A.W., Dovey, O.M., Ellis, P.D., Langford, C.F., Nix, D.A., Euskirchen, G., Hartman, S., Urban, A.E., Kraus, P., Van Calcar, S., Heintzman, N., Kim, T.H., Wang, K., Qu, C., Hon, G., Luna, R., Glass, C.K., Rosenfeld, M.G., Aldred, S.F., Cooper, S.J., Halees, A., Lin, J.M., Shulha, H.P., Zhang, Xiaoling, Xu, M., Haidar, J.N.S., Yu, Y., Ruan, Y., Iyer, V.R., Green, R.D., Wadelius, C., Farnham, P.J., Ren, B., Harte, R.A., Hinrichs, A.S., Trumbower, H., Clawson, H., Hillman-Jackson, J., Zweig, A.S., Smith, K., Thakkapallayil, A., Barber, G., Kuhn, R.M., Karolchik, D., Armengol, L., Bird, C.P., de Bakker, P.I.W., Kern, A.D., Lopez-Bigas, N., Martin, J.D., Stranger, B.E., Woodroffe, A., Davydov, E., Dimas, A., Eyras, E., Hallgrímsdóttir, I.B., Huppert, J., Zody, M.C., Abecasis, G.R., Estivill, X., Bouffard, G.G., Guan, X., Hansen, N.F., Idol, J.R., Maduro, V.V.B., Maskeri, B., McDowell, J.C., Park, M., Thomas, P.J., Young, A.C., Blakesley, R.W., Muzny, D.M., Sodergren, E., Wheeler, D.A., Worley, K.C., Jiang, H., Weinstock, G.M., Gibbs, R.A., Graves, T., Fulton, R., Mardis, E.R., Wilson, R.K., Clamp, M., Cuff, J., Gnerre, S., Jaffe, D.B., Chang, J.L., Lindblad-Toh, K., Lander, E.S., Koriabine, M., Nefedov, M., Osoegawa, K., Yoshinaga, Y., Zhu, B., de Jong, P.J., 2007. Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature* 447, 799–816. <https://doi.org/10.1038/nature05874>

Estiri, H., Stephens, K.A., Klann, J.G., Murphy, S.N., 2018. Exploring completeness in clinical data research networks with DQe-c. *J Am Med Inform Assoc* 25, 17–24. <https://doi.org/10.1093/jamia/ocx109>

Evans, J.P., Dale, D.C., Fomous, C., 2010. Preparing for a consumer-driven genomic age. *N Engl J Med* 363, 1099–1103. <https://doi.org/10.1056/NEJMp1006202>

Evolution: Library: A Mutation Story [WWW Document], 2001.. WGBH Educational Foundation. URL http://www.pbs.org/wgbh/evolution/library/01/2/l_012_02.html (accessed 5.13.21).

Fadnes, L.T., Celis-Morales, C., Økland, J.-M., Parra-Soto, S., Livingstone, K.M., Ho, F.K., Pell, J.P., Balakrishna, R., Javadi Arjmand, E., Johansson, K.A., Haaland, Ø.A., Mathers, J.C., 2023. Life expectancy can increase by up to 10 years following sustained shifts towards healthier diets in the United Kingdom. *Nat Food* 4, 961–965. <https://doi.org/10.1038/s43016-023-00868-w>

- Fatumo, S., Chikowore, T., Choudhury, A., Ayub, M., Martin, A.R., Kuchenbaecker, K., 2022. A roadmap to increase diversity in genomic studies. *Nat Med* 28, 243–250. <https://doi.org/10.1038/s41591-021-01672-4>
- Flaherty, K.T., Gray, R., Chen, A., Li, S., Patton, D., Hamilton, S.R., Williams, P.M., Mitchell, E.P., Iafrate, A.J., Sklar, J., Harris, L.N., McShane, L.M., Rubinstein, L.V., Sims, D.J., Routbort, M., Coffey, B., Fu, T., Zwiebel, J.A., Little, R.F., Marinucci, D., Catalano, R., Magnan, R., Kibbe, W., Weil, C., Tricoli, J.V., Alexander, B., Kumar, S., Schwartz, G.K., Meric-Bernstam, F., Lih, C.-J., McCaskill-Stevens, W., Caimi, P., Takebe, N., Datta, V., Arteaga, C.L., Abrams, J.S., Comis, R., O'Dwyer, P.J., Conley, B.A., NCI-MATCH Team, 2020. The Molecular Analysis for Therapy Choice (NCI-MATCH) Trial: Lessons for Genomic Trial Design. *J Natl Cancer Inst* 112, 1021–1029. <https://doi.org/10.1093/jnci/djz245>
- Fleurence, R.L., Curtis, L.H., Califff, R.M., Platt, R., Selby, J.V., Brown, J.S., 2014. Launching PCORnet, a national patient-centered clinical research network. *J Am Med Inform Assoc* 21, 578–582. <https://doi.org/10.1136/amiajnl-2014-002747>
- Foà, R., 2025. Ph-Positive Acute Lymphoblastic Leukemia — 25 Years of Progress. *New England Journal of Medicine* 392, 1941–1952. <https://doi.org/10.1056/NEJMra2405573>
- Foote, H.P., Hong, C., Anwar, M., Borentain, M., Bugin, K., Dreyer, N., Fessel, J., Goyal, N., Hanger, M., Hernandez, A.F., Hornik, C.P., Jackman, J.G., Lindsay, A.C., Matheny, M.E., Ozer, K., Seidel, J., Stockbridge, N., Embi, P.J., Lindsell, C.J., 2025. Embracing Generative Artificial Intelligence in Clinical Research and Beyond: Opportunities, Challenges, and Solutions. *JACC Adv* 4, 101593. <https://doi.org/10.1016/j.jacadv.2025.101593>
- Foraker, R.E., Yu, S.C., Gupta, A., Michelson, A.P., Pineda Soto, J.A., Colvin, R., Loh, F., Kollef, M.H., Maddox, T., Evanoff, B., Dror, H., Zamstein, N., Lai, A.M., Payne, P.R.O., 2020. Spot the difference: comparing results of analyses from real patient data and synthetic derivatives. *JAMIA Open* 3, 557–566. <https://doi.org/10.1093/jamiaopen/ooaa060>
- Forrest, C.B., McTigue, K.M., Hernandez, A.F., Cohen, L.W., Cruz, H., Haynes, K., Kaushal, R., Kho, A.N., Marsolo, K.A., Nair, V.P., Platt, R., Puro, J.E., Rothman, R.L., Shenkman, E.A., Waitman, L.R., Williams, N.A., Carton, T.W., 2021. PCORnet® 2020: current state, accomplishments, and future directions. *J Clin Epidemiol* 129, 60–67. <https://doi.org/10.1016/j.jclinepi.2020.09.036>
- Fort, D.G., Herr, T.M., Shaw, P.L., Gutzman, K.E., Starren, J.B., 2017. Mapping the evolving definitions of translational research. *J Clin Transl Sci* 1, 60–66. <https://doi.org/10.1017/cts.2016.10>
- Franklin, J.B., Marra, C., Abebe, K.Z., Butte, A.J., Cook, D.J., Esserman, L., Fleisher, L.A., Grossman, C.I., Kass, N.E., Krumholz, H.M., Rowan, K., Abernethy, A.P., JAMA Summit on Clinical Trials Participants, 2024. Modernizing the Data Infrastructure for Clinical Research to Meet Evolving Demands for Evidence. *JAMA*. <https://doi.org/10.1001/jama.2024.0268>
- Galkina Cleary, E., Beierlein, J.M., Khanuja, N.S., McNamee, L.M., Ledley, F.D., 2018. Contribution of NIH funding to new drug approvals 2010-2016. *Proc Natl Acad Sci U S A* 115, 2329–2334. <https://doi.org/10.1073/pnas.1715368115>
- Garcia, S., 2022. Sharing Genomic Data for Patient Care – Sync for Genes Phase 4 Wraps Up [WWW Document]. Health IT Buzz. URL <https://www.healthit.gov/buzz-blog/health-it/sharing-genomic-data-for-patient-care-sync-for-genes-phase-4-wraps-up> (accessed 5.7.23).

- Garcia, S.J., Zayas-Cabán, T., Freimuth, R.R., 2020. Sync for Genes: Making Clinical Genomics Available for Precision Medicine at the Point-of-Care. *Appl Clin Inform* 11, 295–302. <https://doi.org/10.1055/s-0040-1708051>
- Gaziano, J.M., 2010. The evolution of population science: advent of the mega cohort. *JAMA* 304, 2288–2289. <https://doi.org/10.1001/jama.2010.1691>
- Gaziano, J.M., Concato, J., Brophy, M., Fiore, L., Pyarajan, S., Breeling, J., Whitbourne, S., Deen, J., Shannon, C., Humphries, D., Guarino, P., Aslan, M., Anderson, D., LaFleur, R., Hammond, T., Schaa, K., Moser, J., Huang, G., Muralidhar, S., Przygodzki, R., O’Leary, T.J., 2016. Million Veteran Program: A mega-biobank to study genetic influences on health and disease. *J Clin Epidemiol* 70, 214–223. <https://doi.org/10.1016/j.jclinepi.2015.09.016>
- Gene Ontology Consortium, 2021. The Gene Ontology resource: enriching a GOld mine. *Nucleic Acids Res* 49, D325–D334. <https://doi.org/10.1093/nar/gkaa1113>
- Gerlinger, M., Rowan, A.J., Horswell, S., Math, M., Larkin, J., Endesfelder, D., Gronroos, E., Martinez, P., Matthews, N., Stewart, A., Tarpey, P., Varella, I., Phillimore, B., Begum, S., McDonald, N.Q., Butler, A., Jones, D., Raine, K., Latimer, C., Santos, C.R., Nohadani, M., Eklund, A.C., Spencer-Dene, B., Clark, G., Pickering, L., Stamp, G., Gore, M., Szallasi, Z., Downward, J., Futreal, P.A., Swanton, C., 2012. Intratumor heterogeneity and branched evolution revealed by multiregion sequencing. *N Engl J Med* 366, 883–892. <https://doi.org/10.1056/NEJMoa1113205>
- Gerstung, M., Jolly, C., Leshchiner, I., Dentro, S.C., Gonzalez, S., Rosebrock, D., Mitchell, T.J., Rubanova, Y., Anur, P., Yu, K., Tarabichi, M., Deshwar, A., Wintersinger, J., Kleinheinz, K., Vázquez-García, I., Haase, K., Jerman, L., Sengupta, S., Macintyre, G., Malikic, S., Donmez, N., Livitz, D.G., Cmero, M., Demeulemeester, J., Schumacher, S., Fan, Y., Yao, X., Lee, J., Schlesner, M., Boutros, P.C., Bowtell, D.D., Zhu, H., Getz, G., Imielinski, M., Beroukhim, R., Sahinalp, S.C., Ji, Y., Peifer, M., Markowetz, F., Mustonen, V., Yuan, K., Wang, W., Morris, Q.D., PCAWG Evolution & Heterogeneity Working Group, Spellman, P.T., Wedge, D.C., Van Loo, P., PCAWG Consortium, 2020. The evolutionary history of 2,658 cancers. *Nature* 578, 122–128. <https://doi.org/10.1038/s41586-019-1907-7>
- Glas, A.M., Floore, A., Delahaye, L.J.M.J., Witteveen, A.T., Pover, R.C.F., Bakx, N., Lahti-Domenici, J.S.T., Bruinsma, T.J., Warmoes, M.O., Bernards, R., Wessels, L.F.A., Van’t Veer, L.J., 2006. Converting a breast cancer microarray signature into a high-throughput diagnostic test. *BMC Genomics* 7, 278. <https://doi.org/10.1186/1471-2164-7-278>
- Glicksberg, B.S., Johnson, K.W., Dudley, J.T., 2018. The next generation of precision medicine: observational studies, electronic health records, biobanks and continuous monitoring. *Hum Mol Genet* 27, R56–R62. <https://doi.org/10.1093/hmg/ddy114>
- Goldfeder, R.L., Priest, J.R., Zook, J.M., Grove, M.E., Waggett, D., Wheeler, M.T., Salit, M., Ashley, E.A., 2016. Medical implications of technical accuracy in genome sequencing. *Genome Med* 8, 24. <https://doi.org/10.1186/s13073-016-0269-0>
- Goldhaber, N.H., Jacobs, M.B., Laurent, L.C., Knight, R., Zhu, W., Pham, D., Tran, A., Patel, S.P., Hogarth, M., Longhurst, C.A., 2024. Integrating clinical research into electronic health record workflows to support a learning health system. *JAMIA Open* 7, ooae023. <https://doi.org/10.1093/jamiaopen/ooae023>
- Gomez, F., Danos, A.M., Del Fiol, G., Madabhushi, A., Tiwari, P., McMichael, J.F., Bakas, S., Bian, J., Davatzikos, C., Fertig, E.J., Kalpathy-Cramer, J., Kenney, J., Savova, G.K., Yetisgen, M., Van Allen, E.M., Warner, J.L., Prior, F., Griffith, M., Griffith, O.L., 2024. A

- New Era of Data-Driven Cancer Research and Care: Opportunities and Challenges. *Cancer Discov* 14, 1774–1778. <https://doi.org/10.1158/2159-8290.CD-24-1130>
- Goodwin, S., McPherson, J.D., McCombie, W.R., 2016. Coming of age: ten years of next-generation sequencing technologies. *Nat Rev Genet* 17, 333–351.
<https://doi.org/10.1038/nrg.2016.49>
- Gostin, L.O., 1995. Genetic privacy. *J Law Med Ethics* 23, 320–330.
<https://doi.org/10.1111/j.1748-720x.1995.tb01374.x>
- Grady, C., 2015. Institutional Review Boards: Purpose and Challenges. *Chest* 148, 1148–1155.
<https://doi.org/10.1378/chest.15-0706>
- Grande, D., Mitra, N., Shah, A., Wan, F., Asch, D.A., 2013. Public preferences about secondary uses of electronic health information. *JAMA Intern Med* 173, 1798–1806.
<https://doi.org/10.1001/jamainternmed.2013.9166>
- Grassadonia, A., Caporale, M., Tinari, N., Zilli, M., DeTursi, M., Gamucci, T., Vici, P., Natoli, C., 2015. Effect of targeted agents on the endocrine response of breast cancer in the neoadjuvant setting: a systematic review. *J Cancer* 6, 575–582.
<https://doi.org/10.7150/jca.11566>
- Grebe, T.A., Khushf, G., Chen, M., Bailey, D., Brenman, L.M., Williams, M.S., Seaver, L.H., ACMG Social, Ethical and Legal Issues Committee, 2020. The interface of genomic information with the electronic health record: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 22, 1431–1436. <https://doi.org/10.1038/s41436-020-0841-2>
- Green, R.C., Lautenbach, D., McGuire, A.L., 2015. GINA, genetic discrimination, and genomic medicine. *N Engl J Med* 372, 397–399. <https://doi.org/10.1056/NEJMp1404776>
- Green, R.C., Shah, N., Genetti, C.A., Yu, T., Zettler, B., Uveges, M.K., Ceyhan-Birsoy, O., Lebo, M.S., Pereira, S., Agrawal, P.B., Parad, R.B., McGuire, A.L., Christensen, K.D., Schwartz, T.S., Rehm, H.L., Holm, I.A., Beggs, A.H., BabySeq Project Team, 2023. Actionability of unanticipated monogenic disease risks in newborn genomic screening: Findings from the BabySeq Project. *Am J Hum Genet* 110, 1034–1045.
<https://doi.org/10.1016/j.ajhg.2023.05.007>
- Greenbaum, D., Gerstein, M., 2022. GATTACA is still pertinent 25 years later. *Nat Genet* 54, 1758–1760. <https://doi.org/10.1038/s41588-022-01242-5>
- Griffiths, J., 2008. A brief history of mass spectrometry. *Anal Chem* 80, 5678–5683.
<https://doi.org/10.1021/ac8013065>
- Gropman, A.L., Komor, A.C., 2025. Personalized Gene Editing to Treat an Inborn Error of Metabolism. *N Engl J Med*. <https://doi.org/10.1056/NEJMc2505721>
- Guinney, J., Saez-Rodriguez, J., 2018. Alternative models for sharing confidential biomedical data. *Nat Biotechnol* 36, 391–392. <https://doi.org/10.1038/nbt.4128>
- Gwinn, M., MacCannell, D., Armstrong, G.L., 2019. Next-Generation Sequencing of Infectious Pathogens. *JAMA* 321, 893–894. <https://doi.org/10.1001/jama.2018.21669>
- Győrffy, B., Hatzis, C., Sanft, T., Hofstatter, E., Aktas, B., Pusztai, L., 2015. Multigene prognostic tests in breast cancer: past, present, future. *Breast Cancer Res* 17, 11.
<https://doi.org/10.1186/s13058-015-0514-2>
- Haendel, M., Su, A., McMurry, J., 2016. FAIR-TLC: Metrics to Assess Value of Biomedical Digital Repositories: Response to RFI NOT-OD-16-133. Zenodo.
<https://doi.org/10.5281/zenodo.203295>

- Haendel, M.A., Chute, C.G., Bennett, T.D., Eichmann, D.A., Guinney, J., Kibbe, W.A., Payne, P.R.O., Pfaff, E.R., Robinson, P.N., Saltz, J.H., Spratt, H., Suver, C., Wilbanks, J., Wilcox, A.B., Williams, A.E., Wu, C., Blacketer, C., Bradford, R.L., Cimino, J.J., Clark, M., Colmenares, E.W., Francis, P.A., Gabriel, D., Graves, A., Hemadri, R., Hong, S.S., Hripcak, G., Jiao, D., Klann, J.G., Kostka, K., Lee, A.M., Lehmann, H.P., Lingrey, L., Miller, R.T., Morris, M., Murphy, S.N., Natarajan, K., Palchuk, M.B., Sheikh, U., Solbrig, H., Visweswaran, S., Walden, A., Walters, K.M., Weber, G.M., Zhang, X.T., Zhu, R.L., Amor, B., Girvin, A.T., Manna, A., Qureshi, N., Kurilla, M.G., Michael, S.G., Portilla, L.M., Rutter, J.L., Austin, C.P., Gersing, K.R., N3C Consortium, 2021. The National COVID Cohort Collaborative (N3C): Rationale, design, infrastructure, and deployment. *J Am Med Inform Assoc* 28, 427–443. <https://doi.org/10.1093/jamia/ocaa196>
- Hakenberg, J., Cheng, W.-Y., Thomas, P., Wang, Y.-C., Uzilov, A.V., Chen, R., 2016. Integrating 400 million variants from 80,000 human samples with extensive annotations: towards a knowledge base to analyze disease cohorts. *BMC Bioinformatics* 17, 24. <https://doi.org/10.1186/s12859-015-0865-9>
- Halldorsson, B.V., Eggertsson, H.P., Moore, K.H.S., Hauswedell, H., Eiriksson, O., Ulfarsson, M.O., Palsson, G., Hardarson, M.T., Oddsson, A., Jensson, B.O., Kristmundsdottir, S., Sigurpalsdottir, B.D., Stefansson, O.A., Beyter, D., Holley, G., Tragante, V., Gylfason, A., Olason, P.I., Zink, F., Asgeirsdottir, M., Sverrisson, S.T., Sigurdsson, B., Gudjonsson, S.A., Sigurdsson, G.T., Halldorsson, G.H., Sveinbjornsson, G., Norland, K., Styrkarsdottir, U., Magnusdottir, D.N., Snorradottir, S., Kristinsson, K., Sobech, E., Jonsson, Helgi, Geirsson, A.J., Olafsson, I., Jonsson, P., Pedersen, O.B., Erikstrup, C., Brunak, S., Ostrowski, S.R., DBDS Genetic Consortium, Thorleifsson, G., Jonsson, F., Melsted, P., Jonsdottir, I., Rafnar, T., Holm, H., Stefansson, H., Saemundsdottir, J., Gudbjartsson, D.F., Magnusson, O.T., Masson, G., Thorsteinsdottir, U., Helgason, A., Jonsson, Hakon, Sulem, P., Stefansson, K., 2022. The sequences of 150,119 genomes in the UK Biobank. *Nature* 607, 732–740. <https://doi.org/10.1038/s41586-022-04965-x>
- Hanahan, D., Weinberg, R.A., 2011. Hallmarks of cancer: the next generation. *Cell* 144, 646–674. <https://doi.org/10.1016/j.cell.2011.02.013>
- Hanahan, D., Weinberg, R.A., 2000. The hallmarks of cancer. *Cell* 100, 57–70. [https://doi.org/10.1016/s0092-8674\(00\)81683-9](https://doi.org/10.1016/s0092-8674(00)81683-9)
- Handel, A.E., Ramagopalan, S.V., 2010. Is Lamarckian evolution relevant to medicine? *BMC Med Genet* 11, 73. <https://doi.org/10.1186/1471-2350-11-73>
- Hanley, M., Limb, S., Purvis, R., Saya, S., James, P.A., Forrest, L.E., 2025. The development and evaluation of Polygenic Risk Score reports: A systematised review of the literature. *Genet Med* 101426. <https://doi.org/10.1016/j.gim.2025.101426>
- Hao, L., Kraft, P., Berriz, G.F., Hynes, E.D., Koch, C., Korategere V Kumar, P., Parpattedar, S.S., Steeves, M., Yu, W., Antwi, A.A., Brunette, C.A., Danowski, M., Gala, M.K., Green, R.C., Jones, N.E., Lewis, A.C.F., Lubitz, S.A., Natarajan, P., Vassy, J.L., Lebo, M.S., 2022. Development of a clinical polygenic risk score assay and reporting workflow. *Nat Med* 28, 1006–1013. <https://doi.org/10.1038/s41591-022-01767-6>
- Harris, P.A., Taylor, R., Minor, B.L., Elliott, V., Fernandez, M., O’Neal, L., McLeod, L., Delacqua, G., Delacqua, F., Kirby, J., Duda, S.N., REDCap Consortium, 2019. The REDCap consortium: Building an international community of software platform partners. *J Biomed Inform* 95, 103208. <https://doi.org/10.1016/j.jbi.2019.103208>

- Haukoos, J.S., Lewis, R.J., 2015. The Propensity Score. *JAMA* 314, 1637–1638.
<https://doi.org/10.1001/jama.2015.13480>
- Haynes, W.A., Tomczak, A., Khatri, P., 2018. Gene annotation bias impedes biomedical research. *Sci Rep* 8, 1362. <https://doi.org/10.1038/s41598-018-19333-x>
- He, T., Belouali, A., Patricoski, J., Lehmann, H., Ball, R., Anagnostou, V., Kreimeyer, K., Botsis, T., 2023. Trends and opportunities in computable clinical phenotyping: A scoping review. *J Biomed Inform* 140, 104335. <https://doi.org/10.1016/j.jbi.2023.104335>
- Heale, B.S.E., Overby, C.L., Del Fiol, G., Rubinstein, W.S., Maglott, D.R., Nelson, T.H., Milosavljevic, A., Martin, C.L., Goehringer, S.R., Freimuth, R., Williams, M.S., 2016. Integrating Genomic Resources with Electronic Health Records using the HL7 Infobutton Standard. *Appl Clin Inform* 7, 817–831. <https://doi.org/10.4338/ACI-2016-04-RA-0058>
- Heard, E., Martienssen, R.A., 2014. Transgenerational epigenetic inheritance: myths and mechanisms. *Cell* 157, 95–109. <https://doi.org/10.1016/j.cell.2014.02.045>
- Hebbring, S.J., 2014. The challenges, advantages and future of genome-wide association studies. *Immunology* 141, 157–165. <https://doi.org/10.1111/imm.12195>
- Heifetz, A., 2022. Artificial Intelligence in Drug Design. Springer.
- Hernán, M.A., Dahabreh, I.J., Dickerman, B.A., Swanson, S.A., 2025. The Target Trial Framework for Causal Inference From Observational Data: Why and When Is It Helpful? *Ann Intern Med* 178, 402–407. <https://doi.org/10.7326/ANNALS-24-01871>
- Hernandez, J., 2025. 23andMe is filing for bankruptcy. Here's what it means for your genetic data. *NPR*.
- Hoadley, K.A., Yau, C., Wolf, D.M., Cherniack, A.D., Tamborero, D., Ng, S., Leiserson, M.D.M., Niu, B., McLellan, M.D., Uzunangelov, V., Zhang, J., Kandoth, C., Akbani, R., Shen, H., Omberg, L., Chu, A., Margolin, A.A., Van't Veer, L.J., Lopez-Bigas, N., Laird, P.W., Raphael, B.J., Ding, L., Robertson, A.G., Byers, L.A., Mills, G.B., Weinstein, J.N., Van Waes, C., Chen, Z., Collisson, E.A., Cancer Genome Atlas Research Network, Benz, C.C., Perou, C.M., Stuart, J.M., 2014. Multiplatform analysis of 12 cancer types reveals molecular classification within and across tissues of origin. *Cell* 158, 929–944.
<https://doi.org/10.1016/j.cell.2014.06.049>
- Robert, O., 2008. Gene regulation by transcription factors and microRNAs. *Science* 319, 1785–1786. <https://doi.org/10.1126/science.1151651>
- Hodge, J.G., Gostin, L.O., 2017. Revamping the US Federal Common Rule: Modernizing Human Participant Research Regulations. *JAMA* 317, 1521–1522.
<https://doi.org/10.1001/jama.2017.1633>
- Holmes, J.H., Beinlich, J., Boland, M.R., Bowles, K.H., Chen, Y., Cook, T.S., Demiris, G., Draugelis, M., Fluharty, L., Gabriel, P.E., Grundmeier, R., Hanson, C.W., Herman, D.S., Himes, B.E., Hubbard, R.A., Kahn, C.E., Kim, D., Koppel, R., Long, Q., Mirkovic, N., Morris, J.S., Mowery, D.L., Ritchie, M.D., Urbanowicz, R., Moore, J.H., 2021. Why Is the Electronic Health Record So Challenging for Research and Clinical Care? *Methods Inf Med* 60, 32–48. <https://doi.org/10.1055/s-0041-1731784>
- Hong, J., Lee, D., Hwang, A., Kim, T., Ryu, H.-Y., Choi, J., 2024. Rare disease genomics and precision medicine. *Genomics Inform* 22, 28. <https://doi.org/10.1186/s44342-024-00032-1>
- Horie, T., Ono, K., 2024. VERVE-101: a promising CRISPR-based gene editing therapy that reduces LDL-C and PCSK9 levels in HeFH patients. *Eur Heart J Cardiovasc Pharmacother* 10, 89–90. <https://doi.org/10.1093/ehjcvp/pvad103>

- Hornbrook, M.C., Hart, G., Ellis, J.L., Bachman, D.J., Ansell, G., Greene, S.M., Wagner, E.H., Pardee, R., Schmidt, M.M., Geiger, A., Butani, A.L., Field, T., Fouayzi, H., Miroshnik, I., Liu, L., Diseker, R., Wells, K., Krajenta, R., Lamerato, L., Neslund Dudas, C., 2005. Building a virtual cancer research organization. *J Natl Cancer Inst Monogr* 12–25. <https://doi.org/10.1093/jncimonographs/lgi033>
- Hripcsak, G., Albers, D.J., 2018. High-fidelity phenotyping: richness and freedom from bias. *J Am Med Inform Assoc* 25, 289–294. <https://doi.org/10.1093/jamia/ocx110>
- Hripcsak, G., Albers, D.J., 2013. Next-generation phenotyping of electronic health records. *J Am Med Inform Assoc* 20, 117–121. <https://doi.org/10.1136/amiajnl-2012-001145>
- Hripcsak, G., Duke, J.D., Shah, N.H., Reich, C.G., Huser, V., Schuemie, M.J., Suchard, M.A., Park, R.W., Wong, I.C.K., Rijnbeek, P.R., van der Lei, J., Pratt, N., Norén, G.N., Li, Y.-C., Stang, P.E., Madigan, D., Ryan, P.B., 2015. Observational Health Data Sciences and Informatics (OHDSI): Opportunities for Observational Researchers. *Stud Health Technol Inform* 216, 574–578.
- Hripcsak, G., Ryan, P.B., Duke, J.D., Shah, N.H., Park, R.W., Huser, V., Suchard, M.A., Schuemie, M.J., DeFalco, F.J., Perotte, A., Banda, J.M., Reich, C.G., Schilling, L.M., Matheny, M.E., Meeker, D., Pratt, N., Madigan, D., 2016. Characterizing treatment pathways at scale using the OHDSI network. *Proc Natl Acad Sci U S A* 113, 7329–7336. <https://doi.org/10.1073/pnas.1510502113>
- Huang, Y.-N., Munteanu, V., Love, M.I., Ronkowski, C.F., Deshpande, D., Wong-Beringer, A., Corbett-Detig, R., Dimian, M., Moore, J.H., Garmire, L.X., Reddy, T.B.K., Butte, A.J., Robinson, M.D., Eskin, E., Abedalthagafi, M.S., Mangul, S., 2025. Perceptual and technical barriers in sharing and formatting metadata accompanying omics studies. *Cell Genom* 100845. <https://doi.org/10.1016/j.xgen.2025.100845>
- Hudson, K.L., Collins, F.S., 2017. The 21st Century Cures Act - A View from the NIH. *N Engl J Med* 376, 111–113. <https://doi.org/10.1056/NEJMp1615745>
- Hull, L.E., Gold, N.B., Armstrong, K.A., 2020. Revisiting the Roles of Primary Care Clinicians in Genetic Medicine. *JAMA* 324, 1607–1608. <https://doi.org/10.1001/jama.2020.18745>
- Humphreys, B.L., Logan, R.A., Miller, R.A., Siegel, E.R. (Eds.), 2022. Transforming Biomedical Informatics and Health Information Access - Don Lindberg and the U.S. National Library of Medicine.
- Hunter, D.J., Drazen, J.M., 2019. Has the Genome Granted Our Wish Yet? *N Engl J Med* 380, 2391–2393. <https://doi.org/10.1056/NEJMp1904511>
- Insel, T.R., 2017. Digital Phenotyping: Technology for a New Science of Behavior. *JAMA* 318, 1215–1216. <https://doi.org/10.1001/jama.2017.11295>
- Insel, T.R., Volkow, N.D., Li, T.-K., Battey, J.F., Landis, S.C., 2003. Neuroscience networks: data-sharing in an information age. *PLoS Biol* 1, E17. <https://doi.org/10.1371/journal.pbio.0000017>
- Institute of Medicine, 2012. Best Care at Lower Cost: The Path to Continuously Learning Health Care in America. <https://doi.org/10.17226/13444>
- Institute of Medicine, 2011. Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease. <https://doi.org/10.17226/13284>
- Institute of Medicine, 2009. Beyond the HIPAA Privacy Rule: Enhancing Privacy, Improving Health Through Research. <https://doi.org/10.17226/12458>

- International Consortium of Investigators for Fairness in Trial Data Sharing, Devereaux, P.J., Guyatt, G., Gerstein, H., Connolly, S., Yusuf, S., 2016. Toward Fairness in Data Sharing. *N Engl J Med* 375, 405–407. <https://doi.org/10.1056/NEJMp1605654>
- Ioannidis, J.P.A., n.d. What meta-research has taught us about research and changes to research practices. *Journal of Economic Surveys* n/a. <https://doi.org/10.1111/joes.12666>
- İşik, E.B., Brazas, M.D., Schwartz, R., Gaeta, B., Palagi, P.M., van Gelder, C.W.G., Suravajhala, P., Singh, H., Morgan, S.L., Zahrooh, H., Ling, M., Satagopam, V.P., McGrath, A., Nakai, K., Tan, T.W., Gao, G., Mulder, N., Schönbach, C., Zheng, Y., De Las Rivas, J., Khan, A.M., 2023. Grand challenges in bioinformatics education and training. *Nat Biotechnol* 41, 1171–1174. <https://doi.org/10.1038/s41587-023-01891-9>
- Jackson, B.R., Kaplan, B., Schreiber, R., DeMuro, P.R., Nichols-Johnson, V., Ozeran, L., Solomonides, A., Koppel, R., 2025. Ethical Dimensions of Clinical Data Sharing by U.S. Health Care Organizations for Purposes beyond Direct Patient Care: Interviews with Health Care Leaders. *Appl Clin Inform* 16, 90–100. <https://doi.org/10.1055/a-2432-0329>
- Jacobsen, J.O.B., Baudis, M., Baynam, G.S., Beckmann, J.S., Beltran, S., Buske, O.J., Callahan, T.J., Chute, C.G., Courtot, M., Danis, D., Elemento, O., Essewanger, A., Freimuth, R.R., Gargano, M.A., Groza, T., Hamosh, A., Harris, N.L., Kaliyaperumal, R., Lloyd, K.C.K., Khalifa, A., Krawitz, P.M., Köhler, S., Laraway, B.J., Lehväläaho, H., Matalonga, L., McMurry, J.A., Metke-Jimenez, A., Mungall, C.J., Munoz-Torres, M.C., Ogishima, S., Papakonstantinou, A., Piscia, D., Pontikos, N., Queralt-Rosinach, N., Roos, M., Sass, J., Schofield, P.N., Seelow, D., Siapos, A., Smedley, D., Smith, L.D., Steinhaus, R., Sundaramurthi, J.C., Swietlik, E.M., Thun, S., Vasilevsky, N.A., Wagner, A.H., Warner, J.L., Weiland, C., GAGH Phenopacket Modeling Consortium, Haendel, M.A., Robinson, P.N., 2022. The GA4GH Phenopacket schema defines a computable representation of clinical data. *Nat Biotechnol* 40, 817–820. <https://doi.org/10.1038/s41587-022-01357-4>
- Jagadeesh, K.A., Wu, D.J., Birgmeier, J.A., Boneh, D., Bejerano, G., 2017. Deriving genomic diagnoses without revealing patient genomes. *Science* 357, 692–695. <https://doi.org/10.1126/science.aam9710>
- Jameson, J.L., Longo, D.L., 2015. Precision medicine--personalized, problematic, and promising. *N Engl J Med* 372, 2229–2234. <https://doi.org/10.1056/NEJMsb1503104>
- Je, G., Sd, G., K, S., Td, J., Dg, F., Me, G., E, S., T, P., J, M., G, B., Ja, B., S, C., Pc, C., Jw, C., H, C., Kp, D., K, D., Dr, G., J, G., Jw, K., A, K., M, M., T, M., M, N., M, P., Mrz, R., M, S., A, S., C, W., Cj, W., K, X., T, Z., M, J., Fj, S., A, C., B, P., Ea, A., 2022. Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. *The New England journal of medicine* 386. <https://doi.org/10.1056/NEJMc2112090>
- Jensson, B.O., Arnadottir, G.A., Katrinardottir, H., Fridriksdottir, R., Helgason, H., Oddsson, A., Sveinbjornsson, G., Eggertsson, H.P., Halldorsson, G.H., Atlason, B.A., Jonsson, H., Oskarsson, G.R., Sturluson, A., Gudjonsson, S.A., Thorisson, G.A., Zink, F., Moore, K.H.S., Palsson, G., Sigurdsson, A., Jonasdottir, Adalbjorg, Jonasdottir, Aslaug, Magnusson, M.K., Helgadottir, A., Steinhorsdottir, V., Gudmundsson, J., Stacey, S.N., Hilmarsson, R., Olafsson, I., Johannsson, O.T., Arnar, D.O., Saemundsdottir, J., Magnusson, O.T., Masson, G., Halldorsson, B.V., Helgason, A., Stefansson, H., Jonsdottir, I., Holm, H., Rafnar, T., Thorsteinsdottir, U., Gudbjartsson, D.F., Stefansson, K., Sulem, P., 2023. Actionable Genotypes and Their Association with Life Span in Iceland. *N Engl J Med* 389, 1741–1752. <https://doi.org/10.1056/NEJMoa2300792>

- Jiang, G., Kiefer, R.C., Sharma, D.K., Prud'hommeaux, E., Solbrig, H.R., 2017. A Consensus-Based Approach for Harmonizing the OHDSI Common Data Model with HL7 FHIR. *Stud Health Technol Inform* 245, 887–891.
- Johansen Taber, K.A., Dickinson, B.D., Wilson, M., 2014. The promise and challenges of next-generation genome sequencing for clinical care. *JAMA Intern Med* 174, 275–280.
<https://doi.org/10.1001/jamainternmed.2013.12048>
- Johnson, D., Del Fiol, G., Kawamoto, K., Romagnoli, K.M., Sanders, N., Isaacson, G., Jenkins, E., Williams, M.S., 2024. Genetically guided precision medicine clinical decision support tools: a systematic review. *J Am Med Inform Assoc* 31, 1183–1194.
<https://doi.org/10.1093/jamia/ocae033>
- Joseph, A., 2023a. Updated data show long-term benefits of CRISPR treatment for sickle cell, beta thalassemia. STAT. URL <https://www.statnews.com/2023/06/09/vertex-crispr-sickle-cell-thalassemia/> (accessed 5.7.24).
- Joseph, A., 2023b. With latest tranche, U.K. Biobank has genome sequences from 500,000 people available for research. STAT. URL <https://www.statnews.com/2023/11/29/uk-biobank-genome-sequences-500000-people-research/> (accessed 5.7.24).
- Joyner, M.J., Paneth, N., 2019. Promises, promises, and precision medicine. *J Clin Invest* 129, 946–948. <https://doi.org/10.1172/JCI126119>
- Joyner, M.J., Paneth, N., Ioannidis, J.P.A., 2016. What Happens When Underperforming Big Ideas in Research Become Entrenched? *JAMA* 316, 1355–1356.
<https://doi.org/10.1001/jama.2016.11076>
- Jukarainen, S., Kiiskinen, T., Kuitunen, S., Havulinna, A.S., Karjalainen, J., Cordioli, M., Rämö, J.T., Mars, N., FinnGen, Samocha, K.E., Ollila, H.M., Pirinen, M., Ganna, A., 2022. Genetic risk factors have a substantial impact on healthy life years. *Nat Med* 28, 1893–1901. <https://doi.org/10.1038/s41591-022-01957-2>
- Jump, J., Evans, R., Pritzel, A., Green, T., Figurnov, M., Ronneberger, O., Tunyasuvunakool, K., Bates, R., Žídek, A., Potapenko, A., Bridgland, A., Meyer, C., Kohl, S.A.A., Ballard, A.J., Cowie, A., Romera-Paredes, B., Nikolov, S., Jain, R., Adler, J., Back, T., Petersen, S., Reiman, D., Clancy, E., Zieliński, M., Steinegger, M., Pacholska, M., Berghammer, T., Bodenstein, S., Silver, D., Vinyals, O., Senior, A.W., Kavukcuoglu, K., Kohli, P., Hassabis, D., 2021. Highly accurate protein structure prediction with AlphaFold. *Nature* 596, 583–589. <https://doi.org/10.1038/s41586-021-03819-2>
- Jung, A.W., Holm, P.C., Gaurav, K., Hjaltelin, J.X., Placido, D., Mortensen, L.H., Birney, E., Brunak, S.R., Gerstung, M., 2024. Multi-cancer risk stratification based on national health data: a retrospective modelling and validation study. *Lancet Digit Health* 6, e396–e406.
[https://doi.org/10.1016/S2589-7500\(24\)00062-1](https://doi.org/10.1016/S2589-7500(24)00062-1)
- Kaiser, J., 2015. What does a disease deserve? *Science* 350, 900–902.
<https://doi.org/10.1126/science.350.6263.900>
- Kappelmann-Fenzl, M. (Ed.), 2021. Next Generation Sequencing and Data Analysis, 1st ed. 2021 edition. ed. Springer, Cham.
- Karasikov, M., Mustafa, H., Danciu, D., Zimmermann, M., Barber, C., Rätsch, G., Kahles, A., 2024. Indexing All Life's Known Biological Sequences.
<https://doi.org/10.1101/2020.10.01.322164>
- Kashyap, M., Seneviratne, M., Banda, J.M., Falconer, T., Ryu, B., Yoo, S., Hripcak, G., Shah, N.H., 2020. Development and validation of phenotype classifiers across multiple sites in the

- observational health data sciences and informatics network. J Am Med Inform Assoc 27, 877–883. <https://doi.org/10.1093/jamia/ocaa032>
- Katsch, F., Hussein, R., Stamm, T., Duftschmid, G., 2025. Converting Health Level 7 Clinical Document Architecture (CDA) documents to Observational Medical Outcomes Partnership Common Data Model (OMOP CDM) by leveraging CDA Template definitions. JAMIA Open 8, ooaf022. <https://doi.org/10.1093/jamiaopen/ooaf022>
- Katz, S.J., Jaggi, R., Morrow, M., 2018. Reducing Overtreatment of Cancer With Precision Medicine: Just What the Doctor Ordered. JAMA 319, 1091–1092. <https://doi.org/10.1001/jama.2018.0018>
- Keller, M.S., Roberts, P., Japardi, K., Ebinger, J.E., Davis, T., Pevnick, J., Chevvuri, S., Stuck, H., Huang, S.-C., Kowalewski, E., Lin, A., Sanapanya, A., Tomines, A., SooHoo, S., 2025. The Honest Enterprise Research Broker: Facilitating Ethical, Efficient, and Secure Access to Health Data for Research. Appl Clin Inform 16, 362–368. <https://doi.org/10.1055/a-2499-4090>
- Keloth, V.K., Banda, J.M., Gurley, M., Heider, P.M., Kennedy, G., Liu, H., Liu, F., Miller, T., Natarajan, K., Patterson, O., Peng, Y., Raja, K., Reeves, R.M., Rouhizadeh, M., Shi, J., Wang, X., Wang, Y., Wei, W.-Q., Williams, A.E., Zhang, R., Belenkaya, R., Reich, C., Blacketer, C., Ryan, P., Hripcsak, G., Elhadad, N., Xu, H., 2023. Representing and Utilizing Clinical Textual Data for Real World Studies: An OHDSI Approach. J Biomed Inform 104343. <https://doi.org/10.1016/j.jbi.2023.104343>
- Kesselheim, A.S., Avorn, J., 2017. New “21st Century Cures” Legislation: Speed and Ease vs Science. JAMA 317, 581–582. <https://doi.org/10.1001/jama.2016.20640>
- Kesselheim, A.S., Tan, Y.T., Avorn, J., 2015. The roles of academia, rare diseases, and repurposing in the development of the most transformative drugs. Health Aff (Millwood) 34, 286–293. <https://doi.org/10.1377/hlthaff.2014.1038>
- Khera, A.V., Kathiresan, S., 2017. Is Coronary Atherosclerosis One Disease or Many? Setting Realistic Expectations for Precision Medicine. Circulation 135, 1005–1007. <https://doi.org/10.1161/CIRCULATIONAHA.116.026479>
- Khoury, M.J., 2017. No Shortcuts on the Long Road to Evidence-Based Genomic Medicine. JAMA 318, 27–28. <https://doi.org/10.1001/jama.2017.6315>
- Kiessling, P., Kuppe, C., 2024. Spatial multi-omics: novel tools to study the complexity of cardiovascular diseases. Genome Med 16, 14. <https://doi.org/10.1186/s13073-024-01282-y>
- Kilbride, M.K., Bradbury, A.R., 2020. The Need to Improve the Clinical Utility of Direct-to-Consumer Genetic Tests: Either Too Narrow or Too Broad. JAMA 323, 1443–1444. <https://doi.org/10.1001/jama.2019.22504>
- Kim, J., Kim, H., Bell, E., Bath, T., Paul, P., Pham, A., Jiang, X., Zheng, K., Ohno-Machado, L., 2019. Patient Perspectives About Decisions to Share Medical Data and Biospecimens for Research. JAMA Netw Open 2, e199550. <https://doi.org/10.1001/jamanetworkopen.2019.9550>
- Kim, M., Costello, J., 2017. DNA methylation: an epigenetic mark of cellular memory. Exp Mol Med 49, e322. <https://doi.org/10.1038/emm.2017.10>
- Kim, M.S., Shim, I., Fahed, A.C., Do, R., Park, W.-Y., Natarajan, P., Khera, A.V., Won, H.-H., 2024. Association of genetic risk, lifestyle, and their interaction with obesity and obesity-related morbidities. Cell Metab 36, 1494–1503.e3. <https://doi.org/10.1016/j.cmet.2024.06.004>

- Klann, J.G., Abend, A., Raghavan, V.A., Mandl, K.D., Murphy, S.N., 2016. Data interchange using i2b2. *J Am Med Inform Assoc* 23, 909–915. <https://doi.org/10.1093/jamia/ocv188>
- Klann, J.G., Joss, M.A.H., Embree, K., Murphy, S.N., 2019. Data model harmonization for the All Of Us Research Program: Transforming i2b2 data into the OMOP common data model. *PLoS One* 14, e0212463. <https://doi.org/10.1371/journal.pone.0212463>
- Klann, J.G., Phillips, L.C., Herrick, C., Joss, M.A.H., Wagholarikar, K.B., Murphy, S.N., 2018. Web services for data warehouses: OMOP and PCORnet on i2b2. *J Am Med Inform Assoc* 25, 1331–1338. <https://doi.org/10.1093/jamia/ocy093>
- Kohane, I.S., Churchill, S.E., Murphy, S.N., 2012. A translational engine at the national scale: informatics for integrating biology and the bedside. *J Am Med Inform Assoc* 19, 181–185. <https://doi.org/10.1136/amiajnl-2011-000492>
- Köhler, S., Gargano, M., Matentzoglu, N., Carmody, L.C., Lewis-Smith, D., Vasilevsky, N.A., Danis, D., Balagura, G., Baynam, G., Brower, A.M., Callahan, T.J., Chute, C.G., Est, J.L., Galer, P.D., Ganesan, S., Griese, M., Haimel, M., Pazmandi, J., Hanauer, M., Harris, N.L., Hartnett, M.J., Hastreiter, M., Hauck, F., He, Y., Jeske, T., Kearney, H., Kindle, G., Klein, C., Knoflach, K., Krause, R., Lagorce, D., McMurry, J.A., Miller, J.A., Munoz-Torres, M.C., Peters, R.L., Rapp, C.K., Rath, A.M., Rind, S.A., Rosenberg, A.Z., Segal, M.M., Seidel, M.G., Smedley, D., Talmy, T., Thomas, Y., Wiafe, S.A., Xian, J., Yüksel, Z., Helbig, I., Mungall, C.J., Haendel, M.A., Robinson, P.N., 2021. The Human Phenotype Ontology in 2021. *Nucleic Acids Res* 49, D1207–D1217. <https://doi.org/10.1093/nar/gkaa1043>
- Kolata, G., Belluck, P., 2018. Why Are Scientists So Upset About the First Crispr Babies? *The New York Times*.
- Kolker, E., Özdemir, V., Martens, L., Hancock, W., Anderson, G., Anderson, N., Aynacioglu, S., Baranova, A., Campagna, S.R., Chen, R., Choiniere, J., Dearth, S.P., Feng, W.-C., Ferguson, L., Fox, G., Frishman, D., Grossman, R., Heath, A., Higdon, R., Hutz, M.H., Janko, I., Jiang, L., Joshi, S., Kel, A., Kemnitz, J.W., Kohane, I.S., Kolker, N., Lancet, D., Lee, E., Li, W., Lisitsa, A., Llerena, A., Macnealy-Koch, C., Marshall, J.-C., Masuzzo, P., May, A., Mias, G., Monroe, M., Montague, E., Mooney, S., Nesvizhskii, A., Noronha, S., Omenn, G., Rajasimha, H., Ramamoorthy, P., Sheehan, J., Smarr, L., Smith, C.V., Smith, T., Snyder, M., Rapole, S., Srivastava, S., Stanberry, L., Stewart, E., Toppo, S., Uetz, P., Verheggen, K., Voy, B.H., Warnich, L., Wilhelm, S.W., Yandl, G., 2014. Toward more transparent and reproducible omics studies through a common metadata checklist and data publications. *OMICS* 18, 10–14. <https://doi.org/10.1089/omi.2013.0149>
- Kosicki, M., Tomberg, K., Bradley, A., 2018. Repair of double-strand breaks induced by CRISPR-Cas9 leads to large deletions and complex rearrangements. *Nat Biotechnol* 36, 765–771. <https://doi.org/10.1038/nbt.4192>
- Kovaka, S., Ou, S., Jenike, K.M., Schatz, M.C., 2023. Approaching complete genomes, transcriptomes and epi-omes with accurate long-read sequencing. *Nat Methods* 20, 12–16. <https://doi.org/10.1038/s41592-022-01716-8>
- Kush, R., Goldman, M., 2014. Fostering responsible data sharing through standards. *N Engl J Med* 370, 2163–2165. <https://doi.org/10.1056/NEJMp1401444>
- Laganà, A., 2022. Computational Methods for Precision Oncology.
- Laine, C., Chang, S., Chopra, V., Cotton, D., Guallar, E., Wee, C., Williams, S., 2025. Damage Control in the Wake of Political Action That Threatens the Integrity of Medical Research. *Ann Intern Med*. <https://doi.org/10.7326/ANNALS-25-00985>

- Laitner, M.H., Huang, A.M., Takala-Harrison, S. (Eds.), 2024. The State of the U.S. Biomedical and Health Research Enterprise: Strategies for Achieving a Healthier America. National Academies Press, Washington, D.C. <https://doi.org/10.17226/27588>
- Lambert, S.A., Gil, L., Jupp, S., Ritchie, S.C., Xu, Y., Buniello, A., McMahon, A., Abraham, G., Chapman, M., Parkinson, H., Danesh, J., MacArthur, J.A.L., Inouye, M., 2021. The Polygenic Score Catalog as an open database for reproducibility and systematic evaluation. *Nat Genet* 53, 420–425. <https://doi.org/10.1038/s41588-021-00783-5>
- Lambert, S.A., Wingfield, B., Gibson, J.T., Gil, L., Ramachandran, S., Yvon, F., Saverimuttu, S., Tinsley, E., Lewis, E., Ritchie, S.C., Wu, J., Cánovas, R., McMahon, A., Harris, L.W., Parkinson, H., Inouye, M., 2024. Enhancing the Polygenic Score Catalog with tools for score calculation and ancestry normalization. *Nat Genet* 56, 1989–1994. <https://doi.org/10.1038/s41588-024-01937-x>
- Lander, E.S., Linton, L.M., Birren, B., Nusbaum, C., Zody, M.C., Baldwin, J., Devon, K., Dewar, K., Doyle, M., FitzHugh, W., Funke, R., Gage, D., Harris, K., Heaford, A., Howland, J., Kann, L., Lehoczky, J., LeVine, R., McEwan, P., McKernan, K., Meldrim, J., Mesirov, J.P., Miranda, C., Morris, W., Naylor, J., Raymond, C., Rosetti, M., Santos, R., Sheridan, A., Sougnez, C., Stange-Thomann, Y., Stojanovic, N., Subramanian, A., Wyman, D., Rogers, J., Sulston, J., Ainscough, R., Beck, S., Bentley, D., Burton, J., Clee, C., Carter, N., Coulson, A., Deadman, R., Deloukas, P., Dunham, A., Dunham, I., Durbin, R., French, L., Grafham, D., Gregory, S., Hubbard, T., Humphray, S., Hunt, A., Jones, M., Lloyd, C., McMurray, A., Matthews, L., Mercer, S., Milne, S., Mullikin, J.C., Mungall, A., Plumb, R., Ross, M., Shownkeen, R., Sims, S., Waterston, R.H., Wilson, R.K., Hillier, L.W., McPherson, J.D., Marra, M.A., Mardis, E.R., Fulton, L.A., Chinwalla, A.T., Pepin, K.H., Gish, W.R., Chissoe, S.L., Wendl, M.C., Delehaunty, K.D., Miner, T.L., Delehaunty, A., Kramer, J.B., Cook, L.L., Fulton, R.S., Johnson, D.L., Minx, P.J., Clifton, S.W., Hawkins, T., Branscomb, E., Predki, P., Richardson, P., Wenning, S., Slezak, T., Doggett, N., Cheng, J.F., Olsen, A., Lucas, S., Elkin, C., Uberbacher, E., Frazier, M., Gibbs, R.A., Muzny, D.M., Scherer, S.E., Bouck, J.B., Sodergren, E.J., Worley, K.C., Rives, C.M., Gorrell, J.H., Metzker, M.L., Naylor, S.L., Kucherlapati, R.S., Nelson, D.L., Weinstock, G.M., Sakaki, Y., Fujiyama, A., Hattori, M., Yada, T., Toyoda, A., Itoh, T., Kawagoe, C., Watanabe, H., Totoki, Y., Taylor, T., Weissenbach, J., Heilig, R., Saurin, W., Artiguenave, F., Brottier, P., Bruls, T., Pelletier, E., Robert, C., Wincker, P., Smith, D.R., Doucette-Stamm, L., Rubenfield, M., Weinstock, K., Lee, H.M., Dubois, J., Rosenthal, A., Platzer, M., Nyakatura, G., Taudien, S., Rump, A., Yang, H., Yu, J., Wang, J., Huang, G., Gu, J., Hood, L., Rowen, L., Madan, A., Qin, S., Davis, R.W., Federspiel, N.A., Abola, A.P., Proctor, M.J., Myers, R.M., Schmutz, J., Dickson, M., Grimwood, J., Cox, D.R., Olson, M.V., Kaul, R., Raymond, C., Shimizu, N., Kawasaki, K., Minoshima, S., Evans, G.A., Athanasiou, M., Schultz, R., Roe, B.A., Chen, F., Pan, H., Ramser, J., Lehrach, H., Reinhardt, R., McCombie, W.R., de la Bastide, M., Dedhia, N., Blöcker, H., Hornischer, K., Nordsiek, G., Agarwala, R., Aravind, L., Bailey, J.A., Bateman, A., Batzoglou, S., Birney, E., Bork, P., Brown, D.G., Burge, C.B., Cerutti, L., Chen, H.C., Church, D., Clamp, M., Copley, R.R., Doerks, T., Eddy, S.R., Eichler, E.E., Furey, T.S., Galagan, J., Gilbert, J.G., Harmon, C., Hayashizaki, Y., Haussler, D., Hermjakob, H., Hokamp, K., Jang, W., Johnson, L.S., Jones, T.A., Kasif, S., Kaspryzk, A., Kennedy, S., Kent, W.J., Kitts, P., Koonin, E.V., Korf, I., Kulp, D., Lancet, D., Lowe, T.M., McLysaght, A., Mikkelsen, T., Moran, J.V., Mulder, N., Pollara, V.J., Ponting, C.P., Schuler, G., Schultz, J., Slater, G., Smit, A.F., Stupka, E.,

- Szustakowki, J., Thierry-Mieg, D., Thierry-Mieg, J., Wagner, L., Wallis, J., Wheeler, R., Williams, A., Wolf, Y.I., Wolfe, K.H., Yang, S.P., Yeh, R.F., Collins, F., Guyer, M.S., Peterson, J., Felsenfeld, A., Wetterstrand, K.A., Patrinos, A., Morgan, M.J., de Jong, P., Catanese, J.J., Osoegawa, K., Shizuya, H., Choi, S., Chen, Y.J., Szustakowki, J., International Human Genome Sequencing Consortium, 2001. Initial sequencing and analysis of the human genome. *Nature* 409, 860–921. <https://doi.org/10.1038/35057062>
- Langenberg, C., Sharp, S.J., Franks, P.W., Scott, R.A., Deloukas, P., Forouhi, N.G., Froguel, P., Groop, L.C., Hansen, T., Palla, L., Pedersen, O., Schulze, M.B., Tormo, M.-J., Wheeler, E., Agnoli, C., Arriola, L., Barricarte, A., Boeing, H., Clarke, G.M., Clavel-Chapelon, F., Duell, E.J., Fagherazzi, G., Kaaks, R., Kerrison, N.D., Key, T.J., Khaw, K.T., Kröger, J., Lajous, M., Morris, A.P., Navarro, C., Nilsson, P.M., Overvad, K., Palli, D., Panico, S., Quirós, J.R., Rolandsson, O., Sacerdote, C., Sánchez, M.-J., Slimani, N., Spijkerman, A.M.W., Tumino, R., van der A, D.L., van der Schouw, Y.T., Barroso, I., McCarthy, M.I., Riboli, E., Wareham, N.J., 2014. Gene-lifestyle interaction and type 2 diabetes: the EPIC interact case-cohort study. *PLoS Med* 11, e1001647. <https://doi.org/10.1371/journal.pmed.1001647>
- Le, D.T., Durham, J.N., Smith, K.N., Wang, H., Bartlett, B.R., Aulakh, L.K., Lu, S., Kemberling, H., Wilt, C., Luber, B.S., Wong, F., Azad, N.S., Rucki, A.A., Laheru, D., Donehower, R., Zaheer, A., Fisher, G.A., Crocenzi, T.S., Lee, J.J., Greten, T.F., Duffy, A.G., Ciombor, K.K., Eyring, A.D., Lam, B.H., Joe, A., Kang, S.P., Holdhoff, M., Danilova, L., Cope, L., Meyer, C., Zhou, S., Goldberg, R.M., Armstrong, D.K., Bever, K.M., Fader, A.N., Taube, J., Housseau, F., Spetzler, D., Xiao, N., Pardoll, D.M., Papadopoulos, N., Kinzler, K.W., Eshleman, J.R., Vogelstein, B., Anders, R.A., Diaz, L.A., 2017. Mismatch repair deficiency predicts response of solid tumors to PD-1 blockade. *Science* 357, 409–413. <https://doi.org/10.1126/science.aan6733>
- Le Tourneau, C., Delord, J.-P., Gonçalves, A., Gavoille, C., Dubot, C., Isambert, N., Campone, M., Trédan, O., Massiani, M.-A., Mauborgne, C., Armanet, S., Servant, N., Bièche, I., Bernard, V., Gentien, D., Jezequel, P., Attignon, V., Boyault, S., Vincent-Salomon, A., Servois, V., Sablin, M.-P., Kamal, M., Paoletti, X., SHIVA investigators, 2015. Molecularly targeted therapy based on tumour molecular profiling versus conventional therapy for advanced cancer (SHIVA): a multicentre, open-label, proof-of-concept, randomised, controlled phase 2 trial. *Lancet Oncol* 16, 1324–1334. [https://doi.org/10.1016/S1470-2045\(15\)00188-6](https://doi.org/10.1016/S1470-2045(15)00188-6)
- Lee, I., Wallace, Z.S., Wang, Y., Park, S., Nam, H., Majithia, A.R., Ideker, T., 2025. A genotype-phenotype transformer to assess and explain polygenic risk. <https://doi.org/10.1101/2024.10.23.619940>
- Lemery, S., Keegan, P., Pazdur, R., 2017. First FDA Approval Agnostic of Cancer Site - When a Biomarker Defines the Indication. *N Engl J Med* 377, 1409–1412. <https://doi.org/10.1056/NEJMp1709968>
- LeMieux, J., 2021. H3Africa Funds Scientists, Advances Genomics, and Fosters Collaboration in Africa. GEN - Genetic Engineering and Biotechnology News. URL <https://www.genengnews.com/topics/omics/h3africa-funds-scientists-advances-genomics-and-fosters-collaboration-in-africa/> (accessed 5.29.22).
- Lennon, N.J., Kottyan, L.C., Kachulis, C., Abul-Husn, N.S., Arias, J., Belbin, G., Below, J.E., Berndt, S.I., Chung, W.K., Cimino, J.J., Clayton, E.W., Connolly, J.J., Crosslin, D.R., Dikilitas, O., Velez Edwards, D.R., Feng, Q., Fisher, M., Freimuth, R.R., Ge, T., GIANT

Consortium, All of Us Research Program, Glessner, J.T., Gordon, A.S., Patterson, C., Hakonarson, H., Harden, M., Harr, M., Hirschhorn, J.N., Hoggart, C., Hsu, L., Irvin, M.R., Jarvik, G.P., Karlson, E.W., Khan, A., Khera, A., Kiryluk, K., Kullo, I., Larkin, K., Limdi, N., Linder, J.E., Loos, R.J.F., Luo, Y., Malolepsza, E., Manolio, T.A., Martin, L.J., McCarthy, L., McNally, E.M., Meigs, J.B., Mersha, T.B., Mosley, J.D., Musick, A., Namjou, B., Pai, N., Pesce, L.L., Peters, U., Peterson, J.F., Prows, C.A., Puckelwartz, M.J., Rehm, H.L., Roden, D.M., Rosenthal, E.A., Rowley, R., Sawicki, K.T., Schaid, D.J., Smit, R.A.J., Smith, J.L., Smoller, J.W., Thomas, M., Tiwari, H., Toledo, D.M., Vaitinadin, N.S., Veenstra, D., Walunas, T.L., Wang, Z., Wei, W.-Q., Weng, C., Wiesner, G.L., Yin, X., Kenny, E.E., 2024. Selection, optimization and validation of ten chronic disease polygenic risk scores for clinical implementation in diverse US populations. *Nat Med* 30, 480–487.

<https://doi.org/10.1038/s41591-024-02796-z>

Lesk, A., 2025. Introduction to Genomics 4e, 4th edition. ed. Oxford University Press, Oxford.

Levy, S., Sutton, G., Ng, P.C., Feuk, L., Halpern, A.L., Walenz, B.P., Axelrod, N., Huang, J., Kirkness, E.F., Denisov, G., Lin, Y., MacDonald, J.R., Pang, A.W.C., Shago, M., Stockwell, T.B., Tsiamouri, A., Bafna, V., Bansal, V., Kravitz, S.A., Busam, D.A., Beeson, K.Y., McIntosh, T.C., Remington, K.A., Abril, J.F., Gill, J., Borman, J., Rogers, Y.-H., Frazier, M.E., Scherer, S.W., Strausberg, R.L., Venter, J.C., 2007. The diploid genome sequence of an individual human. *PLoS Biol* 5, e254.

<https://doi.org/10.1371/journal.pbio.0050254>

Lewis, A.C.F., Knoppers, B.M., Green, R.C., 2021. An international policy on returning genomic research results. *Genome Med* 13, 115. <https://doi.org/10.1186/s13073-021-00928-5>

Lewis, A.C.F., Perez, E.F., Prince, A.E.R., Flaxman, H.R., Gomez, L., Brockman, D.G., Chandler, P.D., Kerman, B.J., Lebo, M.S., Smoller, J.W., Weiss, S.T., Blout Zawatsky, C.L., Meigs, J.B., Green, R.C., Vassy, J.L., Karlson, E.W., 2022. Patient and provider perspectives on polygenic risk scores: implications for clinical reporting and utilization. *Genome Med* 14, 114. <https://doi.org/10.1186/s13073-022-01117-8>

Li, R., Chen, Y., Ritchie, M.D., Moore, J.H., 2020. Electronic health records and polygenic risk scores for predicting disease risk. *Nat Rev Genet* 21, 493–502.

<https://doi.org/10.1038/s41576-020-0224-1>

Li, R., Romano, J.D., Chen, Y., Moore, J.H., 2024. Centralized and Federated Models for the Analysis of Clinical Data. *Annu Rev Biomed Data Sci* 7, 179–199.

<https://doi.org/10.1146/annurev-biodatasci-122220-115746>

Li, X., Whan, A., McNeil, M., Andrew, S.C., Dai, X., Fechner, M., Paris, C., Sucheki, R., 2025. GeneWhisperer: Enhancing manual genome annotation with large language models.

<https://doi.org/10.1101/2025.03.30.646211>

Liang, P., Xu, Y., Zhang, X., Ding, C., Huang, R., Zhang, Z., Lv, J., Xie, X., Chen, Y., Li, Y., Sun, Y., Bai, Y., Songyang, Z., Ma, W., Zhou, C., Huang, J., 2015. CRISPR/Cas9-mediated gene editing in human tripromuclear zygotes. *Protein Cell* 6, 363–372.

<https://doi.org/10.1007/s13238-015-0153-5>

Liao, W.-W., Asri, M., Ebler, J., Doerr, D., Haukness, M., Hickey, G., Lu, S., Lucas, J.K., Monlong, J., Abel, H.J., Buonaiuto, S., Chang, X.H., Cheng, H., Chu, J., Colonna, V., Eizenga, J.M., Feng, X., Fischer, C., Fulton, R.S., Garg, S., Groza, C., Guarracino, A., Harvey, W.T., Heumos, S., Howe, K., Jain, M., Lu, T.-Y., Markello, C., Martin, F.J., Mitchell, M.W., Munson, K.M., Mwaniki, M.N., Novak, A.M., Olsen, H.E., Pesout, T., Porubsky, D., Prins, P., Sibbesen, J.A., Sirén, J., Tomlinson, C., Villani, F., Vollger, M.R.,

- Antonacci-Fulton, L.L., Baid, G., Baker, C.A., Belyaeva, A., Billis, K., Carroll, A., Chang, P.-C., Cody, S., Cook, D.E., Cook-Deegan, R.M., Cornejo, O.E., Diekhans, M., Ebert, P., Fairley, S., Fedrigo, O., Felsenfeld, A.L., Formenti, G., Frankish, A., Gao, Y., Garrison, N.A., Giron, C.G., Green, R.E., Haggerty, L., Hoekzema, K., Hourlier, T., Ji, H.P., Kenny, E.E., Koenig, B.A., Kolesnikov, A., Korbel, J.O., Kordosky, J., Koren, S., Lee, H., Lewis, A.P., Magalhães, H., Marco-Sola, S., Marijon, P., McCartney, A., McDaniel, J., Mountcastle, J., Nattestad, M., Nurk, S., Olson, N.D., Popejoy, A.B., Puiu, D., Rautiainen, M., Regier, A.A., Rhie, A., Sacco, S., Sanders, A.D., Schneider, V.A., Schultz, B.I., Shafin, K., Smith, M.W., Sofia, H.J., Abou Tayoun, A.N., Thibaud-Nissen, F., Tricomi, F.F., Wagner, J., Walenz, B., Wood, J.M.D., Zimin, A.V., Bourque, G., Chaisson, M.J.P., Flieck, P., Phillippy, A.M., Zook, J.M., Eichler, E.E., Haussler, D., Wang, T., Jarvis, E.D., Miga, K.H., Garrison, E., Marschall, T., Hall, I.M., Li, H., Paten, B., 2023. A draft human pangenome reference. *Nature* 617, 312–324. <https://doi.org/10.1038/s41586-023-05896-x>
- Lih, C.-J., Harrington, R.D., Sims, D.J., Harper, K.N., Bouk, C.H., Datta, V., Yau, J., Singh, R.R., Routbort, M.J., Luthra, R., Patel, K.P., Mantha, G.S., Krishnamurthy, S., Ronski, K., Walther, Z., Finberg, K.E., Canosa, S., Robinson, H., Raymond, A., Le, L.P., McShane, L.M., Polley, E.C., Conley, B.A., Doroshow, J.H., Iafrate, A.J., Sklar, J.L., Hamilton, S.R., Williams, P.M., 2017. Analytical Validation of the Next-Generation Sequencing Assay for a Nationwide Signal-Finding Clinical Trial: Molecular Analysis for Therapy Choice Clinical Trial. *J Mol Diagn* 19, 313–327. <https://doi.org/10.1016/j.jmoldx.2016.10.007>
- Lin, D., McAuliffe, M., Pruitt, K.D., Gururaj, A., Melchior, C., Schmitt, C., Wright, S.N., 2024. Biomedical Data Repository Concepts and Management Principles. *Sci Data* 11, 622. <https://doi.org/10.1038/s41597-024-03449-z>
- Lippert, C., Sabatini, R., Maher, M.C., Kang, E.Y., Lee, S., Arikan, O., Harley, A., Bernal, A., Garst, P., Lavrenko, V., Yocum, K., Wong, T., Zhu, M., Yang, W.-Y., Chang, C., Lu, T., Lee, C.W.H., Hicks, B., Ramakrishnan, S., Tang, H., Xie, C., Piper, J., Brewerton, S., Turpaz, Y., Telenti, A., Roby, R.K., Och, F.J., Venter, J.C., 2017. Identification of individuals by trait prediction using whole-genome sequencing data. *Proc Natl Acad Sci U S A* 114, 10166–10171. <https://doi.org/10.1073/pnas.1711125114>
- Liu, C., Zeinomar, N., Chung, W.K., Kiryluk, K., Gharavi, A.G., Hripcak, G., Crew, K.D., Shang, N., Khan, A., Fasel, D., Manolio, T.A., Jarvik, G.P., Rowley, R., Justice, A.E., Rahm, A.K., Fullerton, S.M., Smoller, J.W., Larson, E.B., Crane, P.K., Dikilitas, O., Wiesner, G.L., Bick, A.G., Terry, M.B., Weng, C., 2021. Generalizability of Polygenic Risk Scores for Breast Cancer Among Women With European, African, and Latinx Ancestry. *JAMA Netw Open* 4, e2119084. <https://doi.org/10.1001/jamanetworkopen.2021.19084>
- Liu, D., Li, Z.-H., Shen, D., Zhang, P.-D., Song, W.-Q., Zhang, W.-T., Huang, Q.-M., Chen, P.-L., Zhang, X.-R., Mao, C., 2022. Association of Sugar-Sweetened, Artificially Sweetened, and Unsweetened Coffee Consumption With All-Cause and Cause-Specific Mortality : A Large Prospective Cohort Study. *Ann Intern Med*. <https://doi.org/10.7326/M21-2977>
- Liu, X., Zhang, X., Jiang, S., Mo, M., Wang, Q., Wang, Yanli, Zhou, L., Hu, S., Yang, H., Hou, Y., Chen, Y., Lu, X., Wang, Yu, Zhou, X., Li, W., Chang, C., Yang, X., Chen, K., Cao, J., Xu, Q., Sun, Y., Luo, J., Luo, Z., Hu, X., 2024. Site-specific therapy guided by a 90-gene expression assay versus empirical chemotherapy in patients with cancer of unknown primary (Fudan CUP-001): a randomised controlled trial. *Lancet Oncol* 25, 1092–1102. [https://doi.org/10.1016/S1470-2045\(24\)00313-9](https://doi.org/10.1016/S1470-2045(24)00313-9)

- Lo Faro, V., Johansson, T., Johansson, Å., 2024. The risk of venous thromboembolism in oral contraceptive users: the role of genetic factors-a prospective cohort study of 240,000 women in the UK Biobank. *Am J Obstet Gynecol* 230, 360.e1-360.e13.
<https://doi.org/10.1016/j.ajog.2023.09.012>
- Luo, J., Juul Rasmussen, I., Thomassen, J.Q., Frikke-Schmidt, R., 2024. Modifiable Risk Factors for Dementia: Causal Estimates on Individual-Level Data.
<https://doi.org/10.2139/ssrn.4986344>
- Ma, H., Marti-Gutierrez, N., Park, S.-W., Wu, J., Lee, Y., Suzuki, K., Koski, A., Ji, D., Hayama, T., Ahmed, R., Darby, H., Van Dyken, C., Li, Y., Kang, E., Park, A.-R., Kim, D., Kim, S.-T., Gong, J., Gu, Y., Xu, X., Battaglia, D., Krieg, S.A., Lee, D.M., Wu, D.H., Wolf, D.P., Heitner, S.B., Belmonte, J.C.I., Amato, P., Kim, J.-S., Kaul, S., Mitalipov, S., 2017. Correction of a pathogenic gene mutation in human embryos. *Nature* 548, 413–419.
<https://doi.org/10.1038/nature23305>
- Malcomson, F.C., Parra-Soto, S., Ho, F.K., Lu, L., Celis-Morales, C., Sharp, L., Mathers, J.C., 2023. Adherence to the 2018 World Cancer Research Fund (WCRF)/American Institute for Cancer Research (AICR) Cancer Prevention Recommendations and risk of 14 lifestyle-related cancers in the UK Biobank prospective cohort study. *BMC Med* 21, 407.
<https://doi.org/10.1186/s12916-023-03107-y>
- Mandel, J.C., Kreda, D.A., Mandl, K.D., Kohane, I.S., Ramoni, R.B., 2016. SMART on FHIR: a standards-based, interoperable apps platform for electronic health records. *J Am Med Inform Assoc* 23, 899–908. <https://doi.org/10.1093/jamia/ocv189>
- Mandl, K.D., Gottlieb, D., Ellis, A., 2019. Beyond One-Off Integrations: A Commercial, Substitutable, Reusable, Standards-Based, Electronic Health Record-Connected App. *J Med Internet Res* 21, e12902. <https://doi.org/10.2196/12902>
- Mars, N., Lindbohm, J.V., Della Briotta Parolo, P., Widén, E., Kaprio, J., Palotie, A., FinnGen, Ripatti, S., 2022. Systematic comparison of family history and polygenic risk across 24 common diseases. *Am J Hum Genet* 109, 2152–2162.
<https://doi.org/10.1016/j.ajhg.2022.10.009>
- Martin, S., Wagner, J., Lupulescu-Mann, N., Ramsey, K., Cohen, A., Graven, P., Weiskopf, N.G., Dorr, D.A., 2017. Comparison of EHR-based diagnosis documentation locations to a gold standard for risk stratification in patients with multiple chronic conditions. *Appl Clin Inform* 8, 794–809. <https://doi.org/10.4338/ACI-2016-12-RA-0210>
- Massard, C., Michiels, S., Ferté, C., Le Deley, M.-C., Lacroix, L., Hollebecque, A., Verlingue, L., Ileana, E., Rosellini, S., Ammari, S., Ngo-Camus, M., Bahleda, R., Gazzah, A., Varga, A., Postel-Vinay, S., Loriot, Y., Even, C., Breuskin, I., Auger, N., Job, B., De Baere, T., Deschamps, F., Vielh, P., Scoazec, J.-Y., Lazar, V., Richon, C., Ribrag, V., Deutsch, E., Angevin, E., Vassal, G., Eggermont, A., André, F., Soria, J.-C., 2017. High-Throughput Genomics and Clinical Outcome in Hard-to-Treat Advanced Cancers: Results of the MOSCATO 01 Trial. *Cancer Discov* 7, 586–595. <https://doi.org/10.1158/2159-8290.CD-16-1396>
- Master, H., Annis, J., Huang, S., Beckman, J.A., Ratsimbazafy, F., Marginean, K., Carroll, R., Natarajan, K., Harrell, F.E., Roden, D.M., Harris, P., Brittain, E.L., 2022. Association of step counts over time with the risk of chronic disease in the All of Us Research Program. *Nat Med* 28, 2301–2308. <https://doi.org/10.1038/s41591-022-02012-w>
- Masys, D.R., Jarvik, G.P., Abernethy, N.F., Anderson, N.R., Papanicolaou, G.J., Paltoo, D.N., Hoffman, M.A., Kohane, I.S., Levy, H.P., 2012. Technical desiderata for the integration of

- genomic data into Electronic Health Records. *J Biomed Inform* 45, 419–422.
<https://doi.org/10.1016/j.jbi.2011.12.005>
- Maxmen, A., 2021. One million coronavirus sequences: popular genome site hits mega milestone. *Nature* 593, 21. <https://doi.org/10.1038/d41586-021-01069-w>
- Mayo, K.R., Basford, M.A., Carroll, R.J., Dillon, M., Fullen, H., Leung, J., Master, H., Rura, S., Sulieman, L., Kennedy, N., Banks, E., Bernick, D., Gauchan, A., Lichtenstein, L., Mapes, B.M., Marginean, K., Nyemba, S.L., Ramirez, A., Rotundo, C., Wolfe, K., Xia, W., Azuine, R.E., Cronin, R.M., Denny, J.C., Kho, A., Lunt, C., Malin, B., Natarajan, K., Wilkins, C.H., Xu, H., Hripcsak, G., Roden, D.M., Philippakis, A.A., Glazer, D., Harris, P.A., 2023. The All of Us Data and Research Center: Creating a Secure, Scalable, and Sustainable Ecosystem for Biomedical Research. *Annu Rev Biomed Data Sci* 6, 443–464.
<https://doi.org/10.1146/annurev-biodatasci-122120-104825>
- McArthur, E., Bastarache, L., Capra, J.A., 2023. Linking rare and common disease vocabularies by mapping between the human phenotype ontology and phecodes. *JAMIA Open* 6, ooad007. <https://doi.org/10.1093/jamiaopen/oad007>
- McBrien, K.A., Souris, S., Symonds, N.E., Rouhi, A., Letebe, B.C., Williamson, T.S., Garies, S., Birtwhistle, R., Quan, H., Fabreau, G.E., Ronksley, P.E., 2018. Identification of validated case definitions for medical conditions used in primary care electronic medical record databases: a systematic review. *J Am Med Inform Assoc* 25, 1567–1578.
<https://doi.org/10.1093/jamia/ocy094>
- McMurry, A.J., Murphy, S.N., MacFadden, D., Weber, G., Simons, W.W., Orechia, J., Bickel, J., Wattanasin, N., Gilbert, C., Trevett, P., Churchill, S., Kohane, I.S., 2013. SHRINE: enabling nationally scalable multi-site disease studies. *PLoS One* 8, e55811.
<https://doi.org/10.1371/journal.pone.0055811>
- Mello, M.M., Lieou, V., Goodman, S.N., 2018. Clinical Trial Participants' Views of the Risks and Benefits of Data Sharing. *N Engl J Med* 378, 2202–2211.
<https://doi.org/10.1056/NEJMsa1713258>
- Melov, S., Tarnopolsky, M.A., Beckman, K., Felkey, K., Hubbard, A., 2007. Resistance exercise reverses aging in human skeletal muscle. *PLoS One* 2, e465.
<https://doi.org/10.1371/journal.pone.0000465>
- Meric-Bernstam, F., Johnson, A., Holla, V., Bailey, A.M., Brusco, L., Chen, K., Routbort, M., Patel, K.P., Zeng, J., Kopetz, S., Davies, M.A., Piha-Paul, S.A., Hong, D.S., Eterovic, A.K., Tsimberidou, A.M., Broaddus, R., Bernstam, E.V., Shaw, K.R., Mendelsohn, J., Mills, G.B., 2015. A decision support framework for genomically informed investigational cancer therapy. *J Natl Cancer Inst* 107. <https://doi.org/10.1093/jnci/djv098>
- Method of the Year 2022: long-read sequencing, 2023. . *Nat Methods* 20, 1.
<https://doi.org/10.1038/s41592-022-01759-x>
- Meyerson, M., Gabriel, S., Getz, G., 2010. Advances in understanding cancer genomes through second-generation sequencing. *Nat Rev Genet* 11, 685–696.
<https://doi.org/10.1038/nrg2841>
- Meystre, S.M., Lovis, C., Bürkle, T., Tognola, G., Budrionis, A., Lehmann, C.U., 2017. Clinical Data Reuse or Secondary Use: Current Status and Potential Future Progress. *Yearb Med Inform* 26, 38–52. <https://doi.org/10.15265/IY-2017-007>
- Miller, D.T., Lee, K., Abul-Husn, N.S., Amendola, L.M., Brothers, K., Chung, W.K., Gollob, M.H., Gordon, A.S., Harrison, S.M., Hershberger, R.E., Klein, T.E., Richards, C.S., Stewart, D.R., Martin, C.L., ACMG Secondary Findings Working Group. Electronic

- address: documents@acmg.net, 2023. ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 25, 100866. <https://doi.org/10.1016/j.gim.2023.100866>
- Misra, B.B., Langefeld, C.D., Olivier, M., Cox, L.A., 2018. Integrated Omics: Tools, Advances, and Future Approaches. *J Mol Endocrinol.* <https://doi.org/10.1530/JME-18-0055>
- Mohanty, A., Fatrekar, A.P., Krishnan, S., Vernekar, A.A., 2021. A Concise Discussion on the Potential Spectral Tools for the Rapid COVID-19 Diagnosis. *Results Chem* 100138. <https://doi.org/10.1016/j.rechem.2021.100138>
- Moodley, K., Cengiz, N., Domingo, A., Nair, G., Obasa, A.E., Lessells, R.J., de Oliveira, T., 2022. Ethics and governance challenges related to genomic data sharing in southern Africa: the case of SARS-CoV-2. *Lancet Glob Health* 10, e1855–e1859. [https://doi.org/10.1016/S2214-109X\(22\)00417-X](https://doi.org/10.1016/S2214-109X(22)00417-X)
- Mukherjee, S., 2016. The Gene: An Intimate History, 1st edition. ed. Scribner, New York.
- Mulder, N., Abimiku, A., Adebamowo, S.N., de Vries, J., Matimba, A., Olowoyo, P., Ramsay, M., Skelton, M., Stein, D.J., 2018. H3Africa: current perspectives. *Pharmgenomics Pers Med* 11, 59–66. <https://doi.org/10.2147/PGPM.S141546>
- Mulder, N.J., Adebiyi, E., Adebiyi, M., Adeyemi, S., Ahmed, A., Ahmed, R., Akanle, B., Alibi, M., Armstrong, D.L., Aron, S., Ashano, E., Baichoo, S., Benkahla, A., Brown, D.K., Chimusa, E.R., Fadlelmola, F.M., Falola, D., Fatumo, S., Ghedira, K., Ghouila, A., Hazelhurst, S., Isewon, I., Jung, S., Kassim, S.K., Kayondo, J.K., Mbiyavanga, M., Meintjes, A., Mohammed, S., Mosaku, A., Moussa, A., Muhammd, M., Mungloo-Dilmohamud, Z., Nashiru, O., Odia, T., Okafor, A., Oladipo, O., Osamor, V., Oyelade, J., Sadki, K., Salifu, S.P., Soyemi, J., Panji, S., Radouani, F., Souiai, O., Tastan Bishop, Ö., H3ABioNet Consortium, as members of the H3Africa Consortium, 2017. Development of Bioinformatics Infrastructure for Genomics Research. *Glob Heart* 12, 91–98. <https://doi.org/10.1016/j.gheart.2017.01.005>
- Murray, M.F., Evans, J.P., Khoury, M.J., 2020. DNA-Based Population Screening: Potential Suitability and Important Knowledge Gaps. *JAMA* 323, 307–308. <https://doi.org/10.1001/jama.2019.18640>
- Musunuru, K., Grandinette, S.A., Wang, X., Hudson, T.R., Briseno, K., Berry, A.M., Hacker, J.L., Hsu, A., Silverstein, R.A., Hille, L.T., Ogul, A.N., Robinson-Garvin, N.A., Small, J.C., McCague, S., Burke, S.M., Wright, C.M., Bick, S., Indurthi, V., Sharma, S., Jepperson, M., Vakulskas, C.A., Collingwood, M., Keogh, K., Jacobi, A., Sturgeon, M., Brommel, C., Schmaljohn, E., Kurgan, G., Osborne, T., Zhang, H., Kinney, K., Rettig, G., Barbosa, C.J., Semple, S.C., Tam, Y.K., Lutz, C., George, L.A., Kleinstiver, B.P., Liu, D.R., Ng, K., Kassim, S.H., Giannikopoulos, P., Alameh, M.-G., Urnov, F.D., Ahrens-Nicklas, R.C., 2025. Patient-Specific In Vivo Gene Editing to Treat a Rare Genetic Disease. *N Engl J Med.* <https://doi.org/10.1056/NEJMoa2504747>
- Naderian, M., Norland, K., Schaid, D.J., Kullo, I.J., 2025. Development and Evaluation of a Comprehensive Prediction Model for Incident Coronary Heart Disease Using Genetic, Social, and Lifestyle-Psychological Factors: A Prospective Analysis of the UK Biobank. *Ann Intern Med* 178, 1–10. <https://doi.org/10.7326/ANNALS-24-00716>
- National Academies of Sciences, Engineering, and Medicine, 2019. Reproducibility and Replicability in Science. <https://doi.org/10.17226/25303>

National Academies of Sciences, Engineering, and Medicine, 2018a. The Next Generation of Biomedical and Behavioral Sciences Researchers: Breaking Through.

<https://doi.org/10.17226/25008>

National Academies of Sciences, Engineering, and Medicine, 2018b. Open Science by Design: Realizing a Vision for 21st Century Research. <https://doi.org/10.17226/25116>

National Academies of Sciences, Engineering, and Medicine; Division of Behavioral and Social Sciences and Education; Health and Medicine Division; Committee on Population; Board on Health Sciences Policy; Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research, 2023. Using Population Descriptors in Genetics and Genomics Research: A New Framework for an Evolving Field. National Academies Press (US), Washington (DC).

Naudet, F., Sakarovitch, C., Janiaud, P., Cristea, I., Fanelli, D., Moher, D., Ioannidis, J.P.A., 2018. Data sharing and reanalysis of randomized controlled trials in leading biomedical journals with a full data sharing policy: survey of studies published in The BMJ and PLOS Medicine. *BMJ* 360, k400. <https://doi.org/10.1136/bmj.k400>

Nelson, H.D., Pappas, M., Cantor, A., Haney, E., Holmes, R., 2019. Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer in Women: Updated Evidence Report and Systematic Review for the US Preventive Services Task Force. *JAMA* 322, 666–685. <https://doi.org/10.1001/jama.2019.8430>

Nguyen, J.Q., Crews, K.R., Moore, B.T., Kornegay, N.M., Baker, D.K., Hasan, M., Campbell, P.K., Dean, S.M., Relling, M.V., Hoffman, J.M., Haidar, C.E., 2022. Clinician adherence to pharmacogenomics prescribing recommendations in clinical decision support alerts. *J Am Med Inform Assoc* 30, 132–138. <https://doi.org/10.1093/jamia/ocac187>

Nishimura, A.A., Shirts, B.H., Dorschner, M.O., Amendola, L.M., Smith, J.W., Jarvik, G.P., Tarczy-Hornoch, P., 2015. Development of clinical decision support alerts for pharmacogenomic incidental findings from exome sequencing. *Genet Med* 17, 939–942. <https://doi.org/10.1038/gim.2015.5>

Nurk, S., Koren, S., Rhie, A., Rautiainen, M., Bzikadze, A.V., Mikheenko, A., Vollger, M.R., Altemose, N., Uralsky, L., Gershman, A., Aganezov, S., Hoyt, S.J., Diekhans, M., Logsdon, G.A., Alonge, M., Antonarakis, S.E., Borchers, M., Bouffard, G.G., Brooks, S.Y., Caldas, G.V., Chen, N.-C., Cheng, H., Chin, C.-S., Chow, W., de Lima, L.G., Dishuck, P.C., Durbin, R., Dvorkina, T., Fiddes, I.T., Formenti, G., Fulton, R.S., Fungtammasan, A., Garrison, E., Grady, P.G.S., Graves-Lindsay, T.A., Hall, I.M., Hansen, N.F., Hartley, G.A., Haukness, M., Howe, K., Hunkapiller, M.W., Jain, C., Jain, M., Jarvis, E.D., Kerpedjiev, P., Kirsche, M., Kolmogorov, M., Korlach, J., Kremitzki, M., Li, H., Maduro, V.V., Marschall, T., McCartney, A.M., McDaniel, J., Miller, D.E., Mullikin, J.C., Myers, E.W., Olson, N.D., Paten, B., Peluso, P., Pevzner, P.A., Porubsky, D., Potapova, T., Rogaev, E.I., Rosenfeld, J.A., Salzberg, S.L., Schneider, V.A., Sedlazeck, F.J., Shafin, K., Shew, C.J., Shumate, A., Sims, Y., Smit, A.F.A., Soto, D.C., Sović, I., Storer, J.M., Streets, A., Sullivan, B.A., Thibaud-Nissen, F., Torrance, J., Wagner, J., Walenz, B.P., Wenger, A., Wood, J.M.D., Xiao, C., Yan, S.M., Young, A.C., Zarate, S., Surti, U., McCoy, R.C., Dennis, M.Y., Alexandrov, I.A., Gerton, J.L., O'Neill, R.J., Timp, W., Zook, J.M., Schatz, M.C., Eichler, E.E., Miga, K.H., Phillippy, A.M., 2022. The complete sequence of a human genome. *Science* 376, 44–53. <https://doi.org/10.1126/science.abj6987>

Obeid, J.S., Beskow, L.M., Rape, M., Gouripeddi, R., Black, R.A., Cimino, J.J., Embi, P.J., Weng, C., Marnocha, R., Buse, J.B., 2017. A survey of practices for the use of electronic

- health records to support research recruitment. *J Clin Transl Sci* 1, 246–252.
<https://doi.org/10.1017/cts.2017.301>
- Obeid, J.S., Shoaibi, A., Oates, J.C., Habrat, M.L., Hughes-Halbert, C., Lenert, L.A., 2018. Research participation preferences as expressed through a patient portal: implications of demographic characteristics. *JAMIA Open* 1, 202–209.
<https://doi.org/10.1093/jamiaopen/ooy034>
- O'Dwyer, P.J., Gray, R.J., Flaherty, K.T., Chen, A.P., Li, S., Wang, V., McShane, L.M., Patton, D.R., Tricoli, J.V., Williams, P.M., Iafrate, A.J., Sklar, J., Mitchell, E.P., Takebe, N., Sims, D.J., Coffey, B., Fu, T., Routbort, M., Rubinstein, L.V., Little, R.F., Arteaga, C.L., Marinucci, D., Hamilton, S.R., Conley, B.A., Harris, L.N., Doroshow, J.H., 2023. The NCI-MATCH trial: lessons for precision oncology. *Nat Med* 29, 1349–1357.
<https://doi.org/10.1038/s41591-023-02379-4>
- Offord, C., 2024. “Google for DNA” indexes 10% of world’s known sequence data. *Science* 384, 1053–1054. <https://doi.org/10.1126/science.adq8853>
- Ogbunugafor, C.B., Edge, M.D., 2022. Gattaca as a lens on contemporary genetics: marking 25 years into the film’s “not-too-distant” future. *Genetics* 222, iyac142.
<https://doi.org/10.1093/genetics/iyac142>
- O’Rahilly, S., 2023. Academic clinician-scientists risk becoming an endangered species. *Nat Med* 29, 2989. <https://doi.org/10.1038/s41591-023-02626-8>
- Pacheco, J.A., Rasmussen, L.V., Wiley, K., Person, T.N., Cronkite, D.J., Sohn, S., Murphy, S., Gundelach, J.H., Gainer, V., Castro, V.M., Liu, C., Mentch, F., Lingren, T., Sundaresan, A.S., Eickelberg, G., Willis, V., Furmanchuk, A., Patel, R., Carrell, D.S., Deng, Y., Walton, N., Satterfield, B.A., Kullo, I.J., Dikilitas, O., Smith, J.C., Peterson, J.F., Shang, N., Kiryluk, K., Ni, Y., Li, Y., Nadkarni, G.N., Rosenthal, E.A., Walunas, T.L., Williams, M.S., Karlson, E.W., Linder, J.E., Luo, Y., Weng, C., Wei, W., 2023. Evaluation of the portability of computable phenotypes with natural language processing in the eMERGE network. *Sci Rep* 13, 1971. <https://doi.org/10.1038/s41598-023-27481-y>
- Pacheco, J.M., Gao, D., Smith, D., Purcell, T., Hancock, M., Bunn, P., Robin, T., Liu, A., Karam, S., Gaspar, L., Kavanagh, B., Rusthoven, C., Aisner, D., Doebele, R., Camidge, D.R., 2019. Natural History and Factors Associated with Overall Survival in Stage IV ALK-Rearranged Non-Small Cell Lung Cancer. *J Thorac Oncol* 14, 691–700.
<https://doi.org/10.1016/j.jtho.2018.12.014>
- Papaemmanuil, E., Gerstung, M., Bullinger, L., Gaidzik, V.I., Paschka, P., Roberts, N.D., Potter, N.E., Heuser, M., Thol, F., Bolli, N., Gundem, G., Van Loo, P., Martincorena, I., Ganly, P., Mudie, L., McLaren, S., O’Meara, S., Raine, K., Jones, D.R., Teague, J.W., Butler, A.P., Greaves, M.F., Ganser, A., Döhner, K., Schlenk, R.F., Döhner, H., Campbell, P.J., 2016. Genomic Classification and Prognosis in Acute Myeloid Leukemia. *N Engl J Med* 374, 2209–2221. <https://doi.org/10.1056/NEJMoa1516192>
- Papez, V., Moinat, M., Voss, E.A., Bazakou, S., Van Winzum, A., Peviani, A., Payralbe, S., Kallfelz, M., Asselbergs, F.W., Prieto-Alhambra, D., Dobson, R.J.B., Denaxas, S., 2022. Transforming and evaluating the UK Biobank to the OMOP Common Data Model for COVID-19 research and beyond. *J Am Med Inform Assoc* 30, 103–111.
<https://doi.org/10.1093/jamia/ocac203>
- Patel, A.P., Wang, M., Ruan, Y., Koyama, S., Clarke, S.L., Yang, X., Tcheandjieu, C., Agrawal, S., Fahed, A.C., Ellinor, P.T., Genes & Health Research Team; the Million Veteran Program, Tsao, P.S., Sun, Y.V., Cho, K., Wilson, P.W.F., Assimes, T.L., van Heel, D.A.,

- Butterworth, A.S., Aragam, K.G., Natarajan, P., Khera, A.V., 2023. A multi-ancestry polygenic risk score improves risk prediction for coronary artery disease. *Nat Med* 29, 1793–1803. <https://doi.org/10.1038/s41591-023-02429-x>
- Pearson, B. (Ed.), n.d. *Handbook of Medical Genomics*.
- Peltonen, L., McKusick, V.A., 2001. Genomics and medicine. Dissecting human disease in the postgenomic era. *Science* 291, 1224–1229. <https://doi.org/10.1126/science.291.5507.1224>
- Pennisi, E., 2007. Genomics. DNA study forces rethink of what it means to be a gene. *Science* 316, 1556–1557. <https://doi.org/10.1126/science.316.5831.1556>
- Pennisi, E., 2003. Bioinformatics. Gene counters struggle to get the right answer. *Science* 301, 1040–1041. <https://doi.org/10.1126/science.301.5636.1040>
- Petersen, C., Austin, R.R., Backonja, U., Campos, H., Chung, A.E., Hekler, E.B., Hsueh, P.-Y.S., Kim, K.K., Pho, A., Salmi, L., Solomonides, A., Valdez, R.S., 2020. Citizen science to further precision medicine: from vision to implementation. *JAMIA Open* 3, 2–8. <https://doi.org/10.1093/jamiaopen/ooz060>
- Peterson, K.A., Delaney, B.C., Arvanitis, T.N., Taweele, A., Sandberg, E.A., Speedie, S., Richard Hobbs, F.D., 2012. A model for the electronic support of practice-based research networks. *Ann Fam Med* 10, 560–567. <https://doi.org/10.1370/afm.1434>
- Prasad, V., 2016. Perspective: The precision-oncology illusion. *Nature* 537, S63. <https://doi.org/10.1038/537S63a>
- Precision Medicine Initiative (PMI) Data Security Principles Implementation Guide, 2016. . Office of the National Coordinator for Health Information Technology.
- Price, A.L., Spencer, C.C.A., Donnelly, P., 2015. Progress and promise in understanding the genetic basis of common diseases. *Proc Biol Sci* 282, 20151684. <https://doi.org/10.1098/rspb.2015.1684>
- Pritchard, J.K., 2023. An Owner's Guide to the Human Genome [WWW Document]. Stanford University. URL <https://web.stanford.edu/group/pritchardlab/HGbook.html> (accessed 5.8.24).
- Pujol Gualdo, N., Džigurski, J., Rukins, V., Pajuste, F.-D., Wolford, B.N., Võsa, M., Golob, M., Haug, L., Alver, M., Läll, K., Peters, M., Brumpton, B.M., Estonian Biobank Research Team, Palta, P., Mägi, R., Laisk, T., 2025. Atlas of genetic and phenotypic associations across 42 female reproductive health diagnoses. *Nat Med*. <https://doi.org/10.1038/s41591-025-03543-8>
- Pulley, J.M., Denny, J.C., Peterson, J.F., Bernard, G.R., Vnencak-Jones, C.L., Ramirez, A.H., Delaney, J.T., Bowton, E., Brothers, K., Johnson, K., Crawford, D.C., Schildcrout, J., Masys, D.R., Dilks, H.H., Wilke, R.A., Clayton, E.W., Shultz, E., Laposata, M., McPherson, J., Jirjis, J.N., Roden, D.M., 2012. Operational implementation of prospective genotyping for personalized medicine: the design of the Vanderbilt PREDICT project. *Clin Pharmacol Ther* 92, 87–95. <https://doi.org/10.1038/clpt.2011.371>
- Purvis, R., Forrest, L.E., Young, M.-A., Limb, S., James, P., Taylor, N., 2025. Defining next steps in the clinical implementation of polygenic scores: A landscape analysis of professional groups' perspectives. *Genet Med* 27, 101414. <https://doi.org/10.1016/j.gim.2025.101414>
- Ramirez, A.H., Sulieman, L., Schlueter, D.J., Halvorson, A., Qian, J., Ratsimbazafy, F., Loperena, R., Mayo, K., Basford, M., Deflaux, N., Muthuraman, K.N., Natarajan, K., Kho, A., Xu, H., Wilkins, C., Anton-Culver, H., Boerwinkle, E., Cicek, M., Clark, C.R., Cohn, E., Ohno-Machado, L., Schully, S.D., Ahmedani, B.K., Argos, M., Cronin, R.M.,

O'Donnell, C., Fouad, M., Goldstein, D.B., Greenland, P., Hebbring, S.J., Karlson, E.W., Khatri, P., Korf, B., Smoller, J.W., Sodeke, S., Wilbanks, J., Hentges, J., Mockrin, S., Lunt, C., Devaney, S.A., Gebo, K., Denny, J.C., Carroll, R.J., Glazer, D., Harris, P.A., Hripcak, G., Philippakis, A., Roden, D.M., All of Us Research Program, 2022. The All of Us Research Program: Data quality, utility, and diversity. Patterns (N Y) 3, 100570. <https://doi.org/10.1016/j.patter.2022.100570>

- Rao, S., Pitel, B., Wagner, A.H., Boca, S.M., McCoy, M., King, I., Gupta, S., Park, B.H., Warner, J.L., Chen, J., Rogan, P.K., Chakravarty, D., Griffith, M., Griffith, O.L., Madhavan, S., 2020. Collaborative, Multidisciplinary Evaluation of Cancer Variants Through Virtual Molecular Tumor Boards Informs Local Clinical Practices. JCO Clin Cancer Inform 4, 602–613. <https://doi.org/10.1200/CCI.19.00169>
- Rehm, H.L., Berg, J.S., Brooks, L.D., Bustamante, C.D., Evans, J.P., Landrum, M.J., Ledbetter, D.H., Maglott, D.R., Martin, C.L., Nussbaum, R.L., Plon, S.E., Ramos, E.M., Sherry, S.T., Watson, M.S., ClinGen, 2015. ClinGen--the Clinical Genome Resource. N Engl J Med 372, 2235–2242. <https://doi.org/10.1056/NEJMsr1406261>

Rehm, H.L., Page, A.J.H., Smith, L., Adams, J.B., Alterovitz, G., Babb, L.J., Barkley, M.P., Baudis, M., Beauvais, M.J.S., Beck, T., Beckmann, J.S., Beltran, S., Bernick, D., Bernier, A., Bonfield, J.K., Boughtwood, T.F., Bourque, G., Bowers, S.R., Brookes, A.J., Brudno, M., Brush, M.H., Bujold, D., Burdett, T., Buske, O.J., Cabilio, M.N., Cameron, D.L., Carroll, R.J., Casas-Silva, E., Chakravarty, D., Chaudhari, B.P., Chen, S.H., Cherry, J.M., Chung, J., Cline, M., Clissold, H.L., Cook-Deegan, R.M., Courtot, M., Cunningham, F., Cupak, M., Davies, R.M., Denisko, D., Doerr, M.J., Dolman, L.I., Dove, E.S., Dursi, L.J., Dyke, S.O.M., Eddy, J.A., Eilbeck, K., Ellrott, K.P., Fairley, S., Fakhro, K.A., Firth, H.V., Fitzsimons, M.S., Fiume, M., Flicek, P., Fore, I.M., Freeberg, M.A., Freimuth, R.R., Fromont, L.A., Fuerth, J., Gaff, C.L., Gan, W., Ghanaim, E.M., Glazer, D., Green, R.C., Griffith, M., Griffith, O.L., Grossman, R.L., Groza, T., Auvin, J.M.G., Guigó, R., Gupta, D., Haendel, M.A., Hamosh, A., Hansen, D.P., Hart, R.K., Hartley, D.M., Haussler, D., Hendricks-Sturup, R.M., Ho, C.W.L., Hobb, A.E., Hoffman, M.M., Hofmann, O.M., Holub, P., Hsu, J.S., Hubaux, J.-P., Hunt, S.E., Husami, A., Jacobsen, J.O., Jamuar, S.S., Janes, E.L., Jeanson, F., Jené, A., Johns, A.L., Joly, Y., Jones, S.J.M., Kanitz, A., Kato, K., Keane, T.M., Kekesi-Lafrance, K., Kelleher, J., Kerry, G., Khor, S.-S., Knoppers, B.M., Konopko, M.A., Kosaki, K., Kuba, M., Lawson, J., Leinonen, R., Li, S., Lin, M.F., Linden, M., Liu, X., Udara Liyanage, I., Lopez, J., Lucassen, A.M., Lukowski, M., Mann, A.L., Marshall, J., Mattioni, M., Metke-Jimenez, A., Middleton, A., Milne, R.J., Molnár-Gábor, F., Mulder, N., Munoz-Torres, M.C., Nag, R., Nakagawa, H., Nasir, J., Navarro, A., Nelson, T.H., Niewielska, A., Nisselle, A., Niu, J., Nyrönen, T.H., O'Connor, B.D., Oesterle, S., Ogishima, S., Wang, V.O., Paglione, L.A.D., Palumbo, E., Parkinson, H.E., Philippakis, A.A., Pizarro, A.D., Prlic, A., Rambla, J., Rendon, A., Rider, R.A., Robinson, P.N., Rodarmer, K.W., Rodriguez, L.L., Rubin, A.F., Rueda, M., Rushton, G.A., Ryan, R.S., Saunders, G.I., Schuilenburg, H., Schwede, T., Scollen, S., Senf, A., Sheffield, N.C., Skantharajah, N., Smith, A.V., Sofia, H.J., Spalding, D., Spurdle, A.B., Stark, Z., Stein, L.D., Suematsu, M., Tan, P., Tedds, J.A., Thomson, A.A., Thorogood, A., Tickle, T.L., Tokunaga, K., Törnroos, J., Torrents, D., Upchurch, S., Valencia, A., Guimera, R.V., Vamathevan, J., Varma, S., Vears, D.F., Viner, C., Voisin, C., Wagner, A.H., Wallace, S.E., Walsh, B.P., Williams, M.S., Winkler, E.C., Wold, B.J., Wood, G.M., Woolley, J.P., Yamasaki, C., Yates, A.D., Yung, C.K., Zass, L.J., Zaytseva, K., Zhang, J., Goodhand, P.,

- North, K., Birney, E., 2021. GA4GH: International policies and standards for data sharing across genomic research and healthcare. *Cell Genom* 1, 100029.
<https://doi.org/10.1016/j.xgen.2021.100029>
- Reich, C., Ostropolets, A., Ryan, P., Rijnbeek, P., Schuemie, M., Davydov, A., Dymshyts, D., Hripcak, G., 2024. OHDSI Standardized Vocabularies-a large-scale centralized reference ontology for international data harmonization. *J Am Med Inform Assoc* 31, 583–590.
<https://doi.org/10.1093/jamia/ocad247>
- Reiner Benaim, A., Almog, R., Gorelik, Y., Hochberg, I., Nassar, L., Mashiach, T., Khamaisi, M., Lurie, Y., Azzam, Z.S., Khoury, J., Kurnik, D., Beyar, R., 2020. Analyzing Medical Research Results Based on Synthetic Data and Their Relation to Real Data Results: Systematic Comparison From Five Observational Studies. *JMIR Med Inform* 8, e16492.
<https://doi.org/10.2196/16492>
- Rhie, A., Nurk, S., Cechova, M., Hoyt, S.J., Taylor, D.J., Altemose, N., Hook, P.W., Koren, S., Rautiainen, M., Alexandrov, I.A., Allen, J., Asri, M., Bzikadze, A.V., Chen, N.-C., Chin, C.-S., Diekhans, M., Flieck, P., Formenti, G., Fungtammasan, A., Garcia Giron, C., Garrison, E., Gershman, A., Gerton, J.L., Grady, P.G.S., Guaracino, A., Haggerty, L., Halabian, R., Hansen, N.F., Harris, R., Hartley, G.A., Harvey, W.T., Haukness, M., Heinz, J., Hourlier, T., Hubley, R.M., Hunt, S.E., Hwang, S., Jain, M., Kesharwani, R.K., Lewis, A.P., Li, H., Logsdon, G.A., Lucas, J.K., Makalowski, W., Markovic, C., Martin, F.J., Mc Cartney, A.M., McCoy, R.C., McDaniel, J., McNulty, B.M., Medvedev, P., Mikheenko, A., Munson, K.M., Murphy, T.D., Olsen, H.E., Olson, N.D., Paulin, L.F., Porubsky, D., Potapova, T., Ryabov, F., Salzberg, S.L., Sauria, M.E.G., Sedlazeck, F.J., Shafin, K., Shepelev, V.A., Shumate, A., Storer, J.M., Surapaneni, L., Taravella Oill, A.M., Thibaud-Nissen, F., Timp, W., Tomaszkiewicz, M., Vollger, M.R., Walenz, B.P., Watwood, A.C., Weissensteiner, M.H., Wenger, A.M., Wilson, M.A., Zarate, S., Zhu, Y., Zook, J.M., Eichler, E.E., O'Neill, R.J., Schatz, M.C., Miga, K.H., Makova, K.D., Phillippy, A.M., 2023. The complete sequence of a human Y chromosome. *Nature* 621, 344–354.
<https://doi.org/10.1038/s41586-023-06457-y>
- Richesson, R., Andrews, J.E., Hollis, K.F. (Eds.), 2023. Clinical Research Informatics, 3rd ed. 2023 edition. ed. Springer.
- Ridley, M., 1999. Genome: The Autobiography of a Species in 23 Chapters, Reprint edition. ed. Harper Perennial.
- Ritchie, M.D., Denny, J.C., Zuvich, R.L., Crawford, D.C., Schildcrout, J.S., Bastarache, L., Ramirez, A.H., Mosley, J.D., Pulley, J.M., Basford, M.A., Bradford, Y., Rasmussen, L.V., Pathak, J., Chute, C.G., Kullo, I.J., McCarty, C.A., Chisholm, R.L., Kho, A.N., Carlson, C.S., Larson, E.B., Jarvik, G.P., Sotoodehnia, N., Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) QRS Group, Manolio, T.A., Li, R., Masys, D.R., Haines, J.L., Roden, D.M., 2013. Genome- and phenome-wide analyses of cardiac conduction identifies markers of arrhythmia risk. *Circulation* 127, 1377–1385.
<https://doi.org/10.1161/CIRCULATIONAHA.112.000604>
- Robinson, J.R., Wei, W.-Q., Roden, D.M., Denny, J.C., 2018. Defining Phenotypes from Clinical Data to Drive Genomic Research. *Annual Review of Biomedical Data Science* 1, 69–92. <https://doi.org/10.1146/annurev-biodatasci-080917-013335>
- Rocher, L., Hendrickx, J.M., de Montjoye, Y.-A., 2019. Estimating the success of re-identifications in incomplete datasets using generative models. *Nat Commun* 10, 3069.
<https://doi.org/10.1038/s41467-019-10933-3>

- Rockhold, F., Nisen, P., Freeman, A., 2016. Data Sharing at a Crossroads. *N Engl J Med* 375, 1115–1117. <https://doi.org/10.1056/NEJMp1608086>
- Rogers, J.R., Liu, C., Hripcsak, G., Cheung, Y.K., Weng, C., 2021. Comparison of Clinical Characteristics Between Clinical Trial Participants and Nonparticipants Using Electronic Health Record Data. *JAMA Netw Open* 4, e214732. <https://doi.org/10.1001/jamanetworkopen.2021.4732>
- Rogith, D., Yusuf, R.A., Hovick, S.R., Peterson, S.K., Burton-Chase, A.M., Li, Y., Meric-Bernstam, F., Bernstam, E.V., 2014. Attitudes regarding privacy of genomic information in personalized cancer therapy. *J Am Med Inform Assoc* 21, e320-325. <https://doi.org/10.1136/amiajnl-2013-002579>
- Rood, J.E., Wynne, S., Robson, L., Hupalowska, A., Randell, J., Teichmann, S.A., Regev, A., 2025. The Human Cell Atlas from a cell census to a unified foundation model. *Nature* 637, 1065–1071. <https://doi.org/10.1038/s41586-024-08338-4>
- Ross, J., Tu, S., Carini, S., Sim, I., 2010. Analysis of eligibility criteria complexity in clinical trials. *Summit Transl Bioinform* 2010, 46–50.
- Ross, J.S., Krumholz, H.M., 2013. Ushering in a new era of open science through data sharing: the wall must come down. *JAMA* 309, 1355–1356. <https://doi.org/10.1001/jama.2013.1299>
- Rubin, R., 2023. It Takes an Average of 17 Years for Evidence to Change Practice—the Burgeoning Field of Implementation Science Seeks to Speed Things Up. *JAMA* 329, 1333–1336. <https://doi.org/10.1001/jama.2023.4387>
- Sadybekov, A.V., Katritch, V., 2023. Computational approaches streamlining drug discovery. *Nature* 616, 673–685. <https://doi.org/10.1038/s41586-023-05905-z>
- Safran, C., Bloomrosen, M., Hammond, W.E., Labkoff, S., Markel-Fox, S., Tang, P.C., Detmer, D.E., Expert Panel, null, 2007. Toward a national framework for the secondary use of health data: an American Medical Informatics Association White Paper. *J Am Med Inform Assoc* 14, 1–9. <https://doi.org/10.1197/jamia.M2273>
- Saha, K., Sontheimer, E.J., Brooks, P.J., Dwinell, M.R., Gersbach, C.A., Liu, D.R., Murray, S.A., Tsai, S.Q., Wilson, R.C., Anderson, D.G., Asokan, A., Banfield, J.F., Bankiewicz, K.S., Bao, G., Bulte, J.W.M., Bursac, N., Campbell, J.M., Carlson, D.F., Chaikof, E.L., Chen, Z.-Y., Cheng, R.H., Clark, K.J., Curiel, D.T., Dahlman, J.E., Deverman, B.E., Dickinson, M.E., Doudna, J.A., Ekker, S.C., Emborg, M.E., Feng, G., Freedman, B.S., Gamm, D.M., Gao, G., Ghiran, I.C., Glazer, P.M., Gong, S., Heaney, J.D., Hennebold, J.D., Hinson, J.T., Khvorova, A., Kiani, S., Lagor, W.R., Lam, K.S., Leong, K.W., Levine, J.E., Lewis, J.A., Lutz, C.M., Ly, D.H., Maragh, S., McCray, P.B., McDevitt, T.C., Mirochnitchenko, O., Morizane, R., Murthy, N., Prather, R.S., Ronald, J.A., Roy, Subhajit, Roy, Sushmita, Sabbisetti, V., Saltzman, W.M., Santangelo, P.J., Segal, D.J., Shimoyama, M., Skala, M.C., Tarantal, A.F., Tilton, J.C., Truskey, G.A., Vandsburger, M., Watts, J.K., Wells, K.D., Wolfe, S.A., Xu, Q., Xue, W., Yi, G., Zhou, J., SCGE Consortium, 2021. The NIH Somatic Cell Genome Editing program. *Nature* 592, 195–204. <https://doi.org/10.1038/s41586-021-03191-1>
- Sanchez-Pinto, L.N., Mosa, A.S.M., Fultz-Hollis, K., Tachinardi, U., Barnett, W.K., Embi, P.J., 2017. The Emerging Role of the Chief Research Informatics Officer in Academic Health Centers. *Appl Clin Inform* 8, 845–853. <https://doi.org/10.4338/ACI-2017-04-RA-0062>
- Sarumi, O.A., Heider, D., 2024. Large language models and their applications in bioinformatics. *Comput Struct Biotechnol J* 23, 3498–3505. <https://doi.org/10.1016/j.csbj.2024.09.031>

- Savova, G.K., Tseytlin, E., Finan, S., Castine, M., Miller, T., Medvedeva, O., Harris, D., Hochheiser, H., Lin, C., Chavan, G., Jacobson, R.S., 2017. DeepPhe: A Natural Language Processing System for Extracting Cancer Phenotypes from Clinical Records. *Cancer Res* 77, e115–e118. <https://doi.org/10.1158/0008-5472.CAN-17-0615>
- Sayers, E.W., Beck, J., Bolton, E.E., Brister, J.R., Chan, J., Connor, R., Feldgarden, M., Fine, A.M., Funk, K., Hoffman, J., Kannan, S., Kelly, C., Klimke, W., Kim, S., Lathrop, S., Marchler-Bauer, A., Murphy, T.D., O’Sullivan, C., Schmieder, E., Skripchenko, Y., Stine, A., Thibaud-Nissen, F., Wang, J., Ye, J., Zellers, E., Schneider, V.A., Pruitt, K.D., 2025. Database resources of the National Center for Biotechnology Information in 2025. *Nucleic Acids Res* 53, D20–D29. <https://doi.org/10.1093/nar/gkae979>
- Schilsky, R.L., Longo, D.L., 2022. Closing the Gap in Cancer Genomic Testing. *N Engl J Med* 387, 2107–2110. <https://doi.org/10.1056/NEJMmp2210638>
- Schneeweiss, S., Brown, J.S., Bate, A., Trifirò, G., Bartels, D.B., 2020. Choosing Among Common Data Models for Real-World Data Analyses Fit for Making Decisions About the Effectiveness of Medical Products. *Clin Pharmacol Ther* 107, 827–833. <https://doi.org/10.1002/cpt.1577>
- Schneider, C.V., Schneider, K.M., Teumer, A., Rudolph, K.L., Hartmann, D., Rader, D.J., Strnád, P., 2022. Association of Telomere Length With Risk of Disease and Mortality. *JAMA Intern Med* 182, 291–300. <https://doi.org/10.1001/jamainternmed.2021.7804>
- Schoeler, T., Speed, D., Porcu, E., Pirastu, N., Pingault, J.-B., Kutalik, Z., 2023. Participation bias in the UK Biobank distorts genetic associations and downstream analyses. *Nat Hum Behav*. <https://doi.org/10.1038/s41562-023-01579-9>
- Schork, N.J., 2015. Personalized medicine: Time for one-person trials. *Nature* 520, 609–611. <https://doi.org/10.1038/520609a>
- Sebastian, A., Carroll, J.C., Oldfield, L.E., Mighton, C., Shickh, S., Uleryk, E., Bombard, Y., 2021. Effect of genetics clinical decision support tools on health-care providers’ decision making: a mixed-methods systematic review. *Genet Med* 23, 593–602. <https://doi.org/10.1038/s41436-020-01045-1>
- Shang, N., Weng, C., Hripcsak, G., 2018. A conceptual framework for evaluating data suitability for observational studies. *J Am Med Inform Assoc* 25, 248–258. <https://doi.org/10.1093/jamia/ocx095>
- Sharpless, N.E., Singer, D.S., 2021. Progress and potential: the Cancer Moonshot. *Cancer Cell*. <https://doi.org/10.1016/j.ccr.2021.04.015>
- Shaywitz, D., 2018. Will Real World Performance Replace RCTs As Healthcare’s Most Important Standard? *Forbes*.
- Sherman, M.A., Yaari, A.U., Priebe, O., Dietlein, F., Loh, P.-R., Berger, B., 2022. Genome-wide mapping of somatic mutation rates uncovers drivers of cancer. *Nat Biotechnol* 40, 1634–1643. <https://doi.org/10.1038/s41587-022-01353-8>
- Sherman, R.L., Firth, A.U., Henley, S.J., Siegel, R.L., Negoita, S., Sung, H., Kohler, B.A., Anderson, R.N., Cucinelli, J., Scott, S., Benard, V.B., Richardson, L.C., Jemal, A., Cronin, K.A., 2025. Annual Report to the Nation on the Status of Cancer, featuring state-level statistics after the onset of the COVID-19 pandemic. *Cancer* 131, e35833. <https://doi.org/10.1002/cncr.35833>
- Sherman, R.M., Salzberg, S.L., 2020. Pan-genomics in the human genome era. *Nat Rev Genet* 21, 243–254. <https://doi.org/10.1038/s41576-020-0210-7>

- Shi, X., Wu, X., 2017. An overview of human genetic privacy. *Ann N Y Acad Sci* 1387, 61–72. <https://doi.org/10.1111/nyas.13211>
- Simmons, S., Sahinalp, C., Berger, B., 2016. Enabling Privacy-Preserving GWASs in Heterogeneous Human Populations. *Cell Syst* 3, 54–61. <https://doi.org/10.1016/j.cels.2016.04.013>
- Siu, L.L., Lawler, M., Haussler, D., Knoppers, B.M., Lewin, J., Vis, D.J., Liao, R.G., Andre, F., Banks, I., Barrett, J.C., Caldas, C., Camargo, A.A., Fitzgerald, R.C., Mao, M., Mattison, J.E., Pao, W., Sellers, W.R., Sullivan, P., Teh, B.T., Ward, R.L., ZenKlusen, J.C., Sawyers, C.L., Voest, E.E., 2016. Facilitating a culture of responsible and effective sharing of cancer genome data. *Nat Med* 22, 464–471. <https://doi.org/10.1038/nm.4089>
- Slomski, A., 2011. The National Library of Medicine: 175 years of advancing biomedical knowledge. *JAMA* 305, 2158–2161. <https://doi.org/10.1001/jama.2011.703>
- Solomon, B.J., Liu, G., Felip, E., Mok, T.S.K., Soo, R.A., Mazieres, J., Shaw, A.T., de Marinis, F., Goto, Y., Wu, Y.-L., Kim, D.-W., Martini, J.-F., Messina, R., Paolini, J., Polli, A., Thomaidou, D., Toffalorio, F., Bauer, T.M., 2024. Lorlatinib Versus Crizotinib in Patients With Advanced ALK-Positive Non-Small Cell Lung Cancer: 5-Year Outcomes From the Phase III CROWN Study. *J Clin Oncol* 42, 3400–3409. <https://doi.org/10.1200/JCO.24.00581>
- Son, J.H., Xie, G., Yuan, C., Ena, L., Li, Z., Goldstein, A., Huang, L., Wang, L., Shen, F., Liu, H., Mehl, K., Groopman, E.E., Marasa, M., Kiryluk, K., Gharavi, A.G., Chung, W.K., Hripcak, G., Friedman, C., Weng, C., Wang, K., 2018. Deep Phenotyping on Electronic Health Records Facilitates Genetic Diagnosis by Clinical Exomes. *Am J Hum Genet* 103, 58–73. <https://doi.org/10.1016/j.ajhg.2018.05.010>
- Stark, R., Grzelak, M., Hadfield, J., 2019. RNA sequencing: the teenage years. *Nat Rev Genet* 20, 631–656. <https://doi.org/10.1038/s41576-019-0150-2>
- Stellmach, C., Sass, J., Auber, B., Boeker, M., Wienker, T., Heidel, A.J., Benary, M., Schumacher, S., Ossowski, S., Klauschen, F., Möller, Y., Schmutzler, R., Ustjanzew, A., Werner, P., Tomczak, A., Höltner, T., Thun, S., 2023. Creation of a structured molecular genomics report for Germany as a local adaption of HL7's Genomic Reporting Implementation Guide. *J Am Med Inform Assoc* ocad061. <https://doi.org/10.1093/jamia/ocad061>
- Strom, B.L., Buyse, M.E., Hughes, J., Knoppers, B.M., 2016. Data Sharing - Is the Juice Worth the Squeeze? *N Engl J Med* 375, 1608–1609. <https://doi.org/10.1056/NEJMp1610336>
- Subramanian, I., Verma, S., Kumar, S., Jere, A., Anamika, K., 2020. Multi-omics Data Integration, Interpretation, and Its Application. *Bioinform Biol Insights* 14, 1177932219899051. <https://doi.org/10.1177/1177932219899051>
- Sud, A., Kinnersley, B., Houlston, R.S., 2017. Genome-wide association studies of cancer: current insights and future perspectives. *Nat Rev Cancer* 17, 692–704. <https://doi.org/10.1038/nrc.2017.82>
- Sudlow, C., Gallacher, J., Allen, N., Beral, V., Burton, P., Danesh, J., Downey, P., Elliott, P., Green, J., Landray, M., Liu, B., Matthews, P., Ong, G., Pell, J., Silman, A., Young, A., Sprosen, T., Peakman, T., Collins, R., 2015. UK biobank: an open access resource for identifying the causes of a wide range of complex diseases of middle and old age. *PLoS Med* 12, e1001779. <https://doi.org/10.1371/journal.pmed.1001779>
- Sugrue, L.P., Desikan, R.S., 2019. What Are Polygenic Scores and Why Are They Important? *JAMA* 321, 1820–1821. <https://doi.org/10.1001/jama.2019.3893>

- Sulieman, L., Cronin, R.M., Carroll, R.J., Natarajan, K., Marginean, K., Mapes, B., Roden, D., Harris, P., Ramirez, A., 2022. Comparing medical history data derived from electronic health records and survey answers in the All of Us Research Program. *J Am Med Inform Assoc* 29, 1131–1141. <https://doi.org/10.1093/jamia/ocac046>
- Supriami, K., Urbut, S.M., Tello-Ayala, J.R., Unlu, O., Friedman, S.F., Abou-Karam, R., Koyama, S., Uddin, M.M., Pomerantsev, E., Lu, M.T., Honigberg, M.C., Aragam, K.G., Doshi-Velez, F., Patel, A.P., Natarajan, P., Ellinor, P.T., Fahed, A.C., 2025. Genomic Drivers of Coronary Artery Disease and Risk of Future Outcomes After Coronary Angiography. *JAMA Netw Open* 8, e2455368. <https://doi.org/10.1001/jamanetworkopen.2024.55368>
- Sutherland, J.P., Zhou, A., Hyppönen, E., 2022. Vitamin D Deficiency Increases Mortality Risk in the UK Biobank : A Nonlinear Mendelian Randomization Study. *Ann Intern Med* 175, 1552–1559. <https://doi.org/10.7326/M21-3324>
- Swanson, K., Liu, G., Catacutan, D.B., Arnold, A., Zou, J., Stokes, J.M., 2024. Generative AI for designing and validating easily synthesizable and structurally novel antibiotics. *Nat Mach Intell* 6, 338–353. <https://doi.org/10.1038/s42256-024-00809-7>
- Swen, J.J., van der Wouden, C.H., Manson, L.E., Abdullah-Koolmees, H., Blagec, K., Blagus, T., Böhringer, S., Cambon-Thomsen, A., Cecchin, E., Cheung, K.-C., Deneer, V.H., Dupui, M., Ingelman-Sundberg, M., Jonsson, S., Joefield-Roka, C., Just, K.S., Karlsson, M.O., Konta, L., Koopmann, R., Kriek, M., Lehr, T., Mitropoulou, C., Rial-Sebbag, E., Rollinson, V., Roncato, R., Samwald, M., Schaeffeler, E., Skokou, M., Schwab, M., Steinberger, D., Stingl, J.C., Tremmel, R., Turner, R.M., van Rhenen, M.H., Dávila Fajardo, C.L., Dolžan, V., Patrinos, G.P., Pirmohamed, M., Sunder-Plassmann, G., Toffoli, G., Guchelaar, H.-J., Ubiquitous Pharmacogenomics Consortium, 2023. A 12-gene pharmacogenetic panel to prevent adverse drug reactions: an open-label, multicentre, controlled, cluster-randomised crossover implementation study. *Lancet* 401, 347–356. [https://doi.org/10.1016/S0140-6736\(22\)01841-4](https://doi.org/10.1016/S0140-6736(22)01841-4)
- Taichman, D.B., Sahni, P., Pinborg, A., Peiperl, L., Laine, C., James, A., Hong, S.-T., Haileamlak, A., Gollogly, L., Godlee, F., Frizelle, F.A., Florenzano, F., Drazen, J.M., Bauchner, H., Baethge, C., Backus, J., 2017. Data Sharing Statements for Clinical Trials: A Requirement of the International Committee of Medical Journal Editors. *JAMA* 317, 2491–2492. <https://doi.org/10.1001/jama.2017.6514>
- Tam, V., Patel, N., Turcotte, M., Bossé, Y., Paré, G., Meyre, D., 2019. Benefits and limitations of genome-wide association studies. *Nat Rev Genet* 20, 467–484. <https://doi.org/10.1038/s41576-019-0127-1>
- Tang, A.S., Woldemariam, S.R., Miramontes, S., Norgeot, B., Oskotsky, T.T., Sirota, M., 2024. Harnessing EHR data for health research. *Nat Med* 30, 1847–1855. <https://doi.org/10.1038/s41591-024-03074-8>
- Tarabichi, Y., Frees, A., Honeywell, S., Huang, C., Naidech, A.M., Moore, J.H., Kaelber, D.C., 2021. The Cosmos Collaborative: A Vendor-Facilitated Electronic Health Record Data Aggregation Platform. *ACI open* 5, e36–e46. <https://doi.org/10.1055/s-0041-1731004>
- Tenenbaum, J.D., 2016. Translational Bioinformatics: Past, Present, and Future. *Genomics Proteomics Bioinformatics* 14, 31–41. <https://doi.org/10.1016/j.gpb.2016.01.003>
- Tenenbaum, J.D., Avillach, P., Benham-Hutchins, M., Breitenstein, M.K., Crowgey, E.L., Hoffman, M.A., Jiang, X., Madhavan, S., Mattison, J.E., Nagarajan, R., Ray, B., Shin, D., Visweswaran, S., Zhao, Z., Freimuth, R.R., 2016. An informatics research agenda to

- support precision medicine: seven key areas. *J Am Med Inform Assoc* 23, 791–795. <https://doi.org/10.1093/jamia/ocv213>
- Tiwary, B.K., 2022. Bioinformatics and Computational Biology.
- Topaloglu, U., Palchuk, M.B., 2018. Using a Federated Network of Real-World Data to Optimize Clinical Trials Operations. *JCO Clin Cancer Inform* 2, 1–10. <https://doi.org/10.1200/CCI.17.00067>
- Topol, E., 2012. The Creative Destruction of Medicine: How the Digital Revolution Will Create Better Health Care, 1st edition. ed. Basic Books, New York.
- Tucker, A., Wang, Z., Rotalinti, Y., Myles, P., 2020. Generating high-fidelity synthetic patient data for assessing machine learning healthcare software. *NPJ Digit Med* 3, 147. <https://doi.org/10.1038/s41746-020-00353-9>
- Turning the tide of early cancer detection, 2024. . *Nat Med* 30, 1217. <https://doi.org/10.1038/s41591-024-03046-y>
- Tyner, J.W., 2014. Functional genomics for personalized cancer therapy. *Sci Transl Med* 6, 243fs26. <https://doi.org/10.1126/scitranslmed.3009586>
- Tyner, J.W., Tognon, C.E., Bottomly, D., Wilmot, B., Kurtz, S.E., Savage, S.L., Long, N., Schultz, A.R., Traer, E., Abel, M., Agarwal, A., Blucher, A., Borate, U., Bryant, J., Burke, R., Carlos, A., Carpenter, R., Carroll, J., Chang, B.H., Coblenz, C., d'Almeida, A., Cook, R., Danilov, A., Dao, K.-H.T., Degnin, M., Devine, D., Dibb, J., Edwards, D.K., Eide, C.A., English, I., Glover, J., Henson, R., Ho, H., Jemal, A., Johnson, K., Johnson, R., Junio, B., Kaempf, A., Leonard, J., Lin, C., Liu, S.Q., Lo, P., Loriaux, M.M., Luty, S., Macey, T., MacManiman, J., Martinez, J., Mori, M., Nelson, D., Nichols, C., Peters, J., Ramsdill, J., Rofelty, A., Schuff, R., Searles, R., Segerdell, E., Smith, R.L., Spurgeon, S.E., Sweeney, T., Thapa, A., Visser, C., Wagner, J., Watanabe-Smith, K., Werth, K., Wolf, J., White, L., Yates, A., Zhang, H., Cogle, C.R., Collins, R.H., Connolly, D.C., Deininger, M.W., Drusbosky, L., Hourigan, C.S., Jordan, C.T., Kropf, P., Lin, T.L., Martinez, M.E., Medeiros, B.C., Pallapati, R.R., Polleyea, D.A., Swords, R.T., Watts, J.M., Weir, S.J., Wiest, D.L., Winters, R.M., McWeeney, S.K., Druker, B.J., 2018. Functional genomic landscape of acute myeloid leukaemia. *Nature* 562, 526–531. <https://doi.org/10.1038/s41586-018-0623-z>
- Underwood, B.R., Lourida, I., Gong, J., Tamburin, S., Tang, E.Y.H., Sidhom, E., Tai, X.Y., Betts, M.J., Ranson, J.M., Zachariou, M., Olaleye, O.E., Das, S., Oxtoby, N.P., Chen, S., Llewellyn, D.J., Deep Dementia Phenotyping (DEMON) Network, 2025. Data-driven discovery of associations between prescribed drugs and dementia risk: A systematic review. *Alzheimers Dement (N Y)* 11, e70037. <https://doi.org/10.1002/trc2.70037>
- Utz, P.J., Jain, M.K., Cheung, V.G., Kobilka, B.K., Lefkowitz, R., Yamada, T., Dzau, V.J., 2022. Translating science to medicine: The case for physician-scientists. *Sci Transl Med* 14, eabg7852. <https://doi.org/10.1126/scitranslmed.abg7852>
- van de Vijver, M.J., He, Y.D., van't Veer, L.J., Dai, H., Hart, A.A.M., Voskuil, D.W., Schreiber, G.J., Peterse, J.L., Roberts, C., Marton, M.J., Parrish, M., Atsma, D., Witteveen, A., Glas, A., Delahaye, L., van der Velde, T., Bartelink, H., Rodenhuis, S., Rutgers, E.T., Friend, S.H., Bernards, R., 2002. A gene-expression signature as a predictor of survival in breast cancer. *N Engl J Med* 347, 1999–2009. <https://doi.org/10.1056/NEJMoa021967>
- Van Driest, S.L., Wells, Q.S., Stallings, S., Bush, W.S., Gordon, A., Nickerson, D.A., Kim, J.H., Crosslin, D.R., Jarvik, G.P., Carrell, D.S., Ralston, J.D., Larson, E.B., Bielinski, S.J., Olson, J.E., Ye, Z., Kullo, I.J., Abul-Husn, N.S., Scott, S.A., Bottinger, E., Almoguera, B.,

Connolly, J., Chiavacci, R., Hakonarson, H., Rasmussen-Torvik, L.J., Pan, V., Persell, S.D., Smith, M., Chisholm, R.L., Kitchner, T.E., He, M.M., Brilliant, M.H., Wallace, J.R., Doheny, K.F., Shoemaker, M.B., Li, R., Manolio, T.A., Callis, T.E., Macaya, D., Williams, M.S., Carey, D., Kapplinger, J.D., Ackerman, M.J., Ritchie, M.D., Denny, J.C., Roden, D.M., 2016. Association of Arrhythmia-Related Genetic Variants With Phenotypes Documented in Electronic Medical Records. *JAMA* 315, 47–57.

<https://doi.org/10.1001/jama.2015.17701>

Varadi, M., Anyango, S., Deshpande, M., Nair, S., Natassia, C., Yordanova, G., Yuan, D., Stroe, O., Wood, G., Laydon, A., Žídek, A., Green, T., Tunyasuvunakool, K., Petersen, S., Jumper, J., Clancy, E., Green, R., Vora, A., Lutfi, M., Figurnov, M., Cowie, A., Hobbs, N., Kohli, P., Kleywegt, G., Birney, E., Hassabis, D., Velankar, S., 2022. AlphaFold Protein Structure Database: massively expanding the structural coverage of protein-sequence space with high-accuracy models. *Nucleic Acids Res* 50, D439–D444.

<https://doi.org/10.1093/nar/gkab1061>

Vashisht, R., Patel, A., Dahm, L., Han, C., Medders, K.E., Mowers, R., Byington, C.L., Koliwad, S.K., Butte, A.J., 2023. Second-Line Pharmaceutical Treatments for Patients with Type 2 Diabetes. *JAMA Netw Open* 6, e2336613.

<https://doi.org/10.1001/jamanetworkopen.2023.36613>

Vasilevsky, N., Zhang, A., Gourdin, J.-P., Yates, A., Haendel, M., Robinson, P., 2018. LOINC2HPO: Improving Translational Informatics by Standardizing EHR Phenotypic Data Using the Human Phenotype Ontology, in: Rocky Bioinformatics 2018. figshare.

<https://doi.org/10.6084/m9.figshare.7482125.v1>

Vassy, J.L., Christensen, K.D., Schonman, E.F., Blout, C.L., Robinson, J.O., Krier, J.B., Diamond, P.M., Lebo, M., Machini, K., Azzariti, D.R., Dukhovny, D., Bates, D.W., MacRae, C.A., Murray, M.F., Rehm, H.L., McGuire, A.L., Green, R.C., MedSeq Project, 2017. The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients: A Pilot Randomized Trial. *Ann Intern Med* 167, 159–169.

<https://doi.org/10.7326/M17-0188>

Vassy, J.L., Hivert, M.-F., Porneala, B., Dauriz, M., Florez, J.C., Dupuis, J., Siscovick, D.S., Fornage, M., Rasmussen-Torvik, L.J., Bouchard, C., Meigs, J.B., 2014. Polygenic type 2 diabetes prediction at the limit of common variant detection. *Diabetes* 63, 2172–2182.

<https://doi.org/10.2337/db13-1663>

Venner, E., Muzny, D., Smith, J.D., Walker, K., Neben, C.L., Lockwood, C.M., Empey, P.E., Metcalf, G.A., Kachulis, C., All of Us Research Program Regulatory Working Group, Mian, S., Musick, A., Rehm, H.L., Harrison, S., Gabriel, S., Gibbs, R.A., Nickerson, D., Zhou, A.Y., Doheny, K., Ozenberger, B., Topper, S.E., Lennon, N.J., 2022. Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program. *Genome Med* 14, 34.

<https://doi.org/10.1186/s13073-022-01031-z>

Venter, J.C., Adams, M.D., Myers, E.W., Li, P.W., Mural, R.J., Sutton, G.G., Smith, H.O., Yandell, M., Evans, C.A., Holt, R.A., Gocayne, J.D., Amanatides, P., Ballew, R.M., Huson, D.H., Wortman, J.R., Zhang, Q., Kodira, C.D., Zheng, X.H., Chen, L., Skupski, M., Subramanian, G., Thomas, P.D., Zhang, J., Gabor Miklos, G.L., Nelson, C., Broder, S., Clark, A.G., Nadeau, J., McKusick, V.A., Zinder, N., Levine, A.J., Roberts, R.J., Simon, M., Slayman, C., Hunkapiller, M., Bolanos, R., Delcher, A., Dew, I., Fasulo, D., Flanigan, M., Florea, L., Halpern, A., Hannenhalli, S., Kravitz, S., Levy, S., Mobarry, C., Reinert, K.,

Remington, K., Abu-Threideh, J., Beasley, E., Biddick, K., Bonazzi, V., Brandon, R., Cargill, M., ChandramouliSwaran, I., Charlab, R., Chaturvedi, K., Deng, Z., Di Francesco, V., Dunn, P., Eilbeck, K., Evangelista, C., Gabrielian, A.E., Gan, W., Ge, W., Gong, F., Gu, Z., Guan, P., Heiman, T.J., Higgins, M.E., Ji, R.R., Ke, Z., Ketchum, K.A., Lai, Z., Lei, Y., Li, Z., Li, J., Liang, Y., Lin, X., Lu, F., Merkulov, G.V., Milshina, N., Moore, H.M., Naik, A.K., Narayan, V.A., Neelam, B., Nusskern, D., Rusch, D.B., Salzberg, S., Shao, W., Shue, B., Sun, J., Wang, Z., Wang, A., Wang, X., Wang, J., Wei, M., Wides, R., Xiao, C., Yan, C., Yao, A., Ye, J., Zhan, M., Zhang, W., Zhang, H., Zhao, Q., Zheng, L., Zhong, F., Zhong, W., Zhu, S., Zhao, S., Gilbert, D., Baumhueter, S., Spier, G., Carter, C., Cravchik, A., Woodage, T., Ali, F., An, H., Awe, A., Baldwin, D., Baden, H., Barnstead, M., Barrow, I., Beeson, K., Busam, D., Carver, A., Center, A., Cheng, M.L., Curry, L., Danaher, S., Davenport, L., Desilets, R., Dietz, S., Dodson, K., Doup, L., Ferriera, S., Garg, N., Gluecksmann, A., Hart, B., Haynes, J., Haynes, C., Heiner, C., Hladun, S., Hostin, D., Houck, J., Howland, T., Ibegwam, C., Johnson, J., Kalush, F., Kline, L., Koduru, S., Love, A., Mann, F., May, D., McCawley, S., McIntosh, T., McMullen, I., Moy, M., Moy, L., Murphy, B., Nelson, K., Pfannkoch, C., Pratts, E., Puri, V., Qureshi, H., Reardon, M., Rodriguez, R., Rogers, Y.H., Romblad, D., Ruhfel, B., Scott, R., Sitter, C., Smallwood, M., Stewart, E., Strong, R., Suh, E., Thomas, R., Tint, N.N., Tse, S., Vech, C., Wang, G., Wetter, J., Williams, S., Williams, M., Windsor, S., Winn-Deen, E., Wolfe, K., Zaveri, J., Zaveri, K., Abril, J.F., Guigó, R., Campbell, M.J., Sjolander, K.V., Karlak, B., Kejariwal, A., Mi, H., Lazareva, B., Hatton, T., Narechania, A., Diemer, K., Muruganujan, A., Guo, N., Sato, S., Bafna, V., Istrail, S., Lippert, R., Schwartz, R., Walenz, B., Yooseph, S., Allen, D., Basu, A., Baxendale, J., Blick, L., Caminha, M., Carnes-Stine, J., Caulk, P., Chiang, Y.H., Coyne, M., Dahlke, C., Mays, A., Dombroski, M., Donnelly, M., Ely, D., Esparham, S., Fosler, C., Gire, H., Glanowski, S., Glasser, K., Glodek, A., Gorokhov, M., Graham, K., Gropman, B., Harris, M., Heil, J., Henderson, S., Hoover, J., Jennings, D., Jordan, C., Jordan, J., Kasha, J., Kagan, L., Kraft, C., Levitsky, A., Lewis, M., Liu, X., Lopez, J., Ma, D., Majoros, W., McDaniel, J., Murphy, S., Newman, M., Nguyen, T., Nguyen, N., Nodell, M., Pan, S., Peck, J., Peterson, M., Rowe, W., Sanders, R., Scott, J., Simpson, M., Smith, T., Sprague, A., Stockwell, T., Turner, R., Venter, E., Wang, M., Wen, M., Wu, D., Wu, M., Xia, A., Zandieh, A., Zhu, X., 2001. The sequence of the human genome. *Science* 291, 1304–1351. <https://doi.org/10.1126/science.1058040>

Verma, A., Lucas, A., Verma, S.S., Zhang, Y., Josyula, N., Khan, A., Hartzel, D.N., Lavage, D.R., Leader, J., Ritchie, M.D., Pendergrass, S.A., 2018. PheWAS and Beyond: The Landscape of Associations with Medical Diagnoses and Clinical Measures across 38,662 Individuals from Geisinger. *Am J Hum Genet* 102, 592–608.

<https://doi.org/10.1016/j.ajhg.2018.02.017>

Versel, N., 2023. Standards, Regulation, Funding Move Bioinformatics in 2022, But Hurdles to Precision Medicine Remain | GenomeWeb [WWW Document]. Genome Web. URL <https://www.genomeweb.com/informatics/standards-regulation-funding-move-bioinformatics-2022-hurdles-precision-medicine-remain> (accessed 5.2.23).

Visscher, P.M., Gyngell, C., Yengo, L., Savulescu, J., 2025. Heritable polygenic editing: the next frontier in genomic medicine? *Nature* 637, 637–645. <https://doi.org/10.1038/s41586-024-08300-4>

Visweswaran, S., Becich, M.J., D’Itri, V.S., Sendro, E.R., MacFadden, D., Anderson, N.R., Allen, K.A., Ranganathan, D., Murphy, S.N., Morrato, E.H., Pincus, H.A., Toto, R.,

- Firestein, G.S., Nadler, L.M., Reis, S.E., 2018. Accrual to Clinical Trials (ACT): A Clinical and Translational Science Award Consortium Network. *JAMIA Open* 1, 147–152.
<https://doi.org/10.1093/jamiaopen/ooy033>
- Vokinger, K.N., Avorn, J., Kesselheim, A.S., 2023. Sources of Innovation in Gene Therapies - Approaches to Achieving Affordable Prices. *N Engl J Med* 388, 292–295.
<https://doi.org/10.1056/NEJMp2211729>
- Walensky, R.P., Walensky, L.D., 2025. US Science in Peril. *JAMA* 333, 933–934.
<https://doi.org/10.1001/jama.2025.1929>
- Walonuski, J., Klaus, S., Granger, E., Hall, D., Gregorowicz, A., Neyarapally, G., Watson, A., Eastman, J., 2020. Synthea™ Novel coronavirus (COVID-19) model and synthetic data set. *Intell Based Med* 1, 100007. <https://doi.org/10.1016/j.ibmed.2020.100007>
- Walonuski, J., Kramer, M., Nichols, J., Quina, A., Moesel, C., Hall, D., Duffett, C., Dube, K., Gallagher, T., McLachlan, S., 2018. Synthea: An approach, method, and software mechanism for generating synthetic patients and the synthetic electronic health care record. *J Am Med Inform Assoc* 25, 230–238. <https://doi.org/10.1093/jamia/ocx079>
- Wan, Z., Hazel, J.W., Clayton, E.W., Vorobeychik, Y., Kantarciooglu, M., Malin, B.A., 2022. Sociotechnical safeguards for genomic data privacy. *Nat Rev Genet* 23, 429–445.
<https://doi.org/10.1038/s41576-022-00455-y>
- Wang, J., Li, Z., Zhang, J., 2022. Visualizing the knowledge structure and evolution of bioinformatics. *BMC Bioinformatics* 23, 404. <https://doi.org/10.1186/s12859-022-04948-9>
- Wang, L., Wen, A., Fu, S., Ruan, X., Huang, M., Li, R., Lu, Q., Lyu, H., Williams, A.E., Liu, H., 2025. A scoping review of OMOP CDM adoption for cancer research using real world data. *NPJ Digit Med* 8, 189. <https://doi.org/10.1038/s41746-025-01581-7>
- Wang, Q., Dhindsa, R.S., Carss, K., Harper, A.R., Nag, A., Tachmazidou, I., Vitsios, D., Deevi, S.V.V., Mackay, A., Muthas, D., Hühn, M., Monkley, S., Olsson, H., AstraZeneca Genomics Initiative, Wasilewski, S., Smith, K.R., March, R., Platt, A., Haefliger, C., Petrovski, S., 2021. Rare variant contribution to human disease in 281,104 UK Biobank exomes. *Nature* 597, 527–532. <https://doi.org/10.1038/s41586-021-03855-y>
- Wang, S., Jiang, X., Singh, S., Marmor, R., Bonomi, L., Fox, D., Dow, M., Ohno-Machado, L., 2017. Genome privacy: challenges, technical approaches to mitigate risk, and ethical considerations in the United States. *Ann N Y Acad Sci* 1387, 73–83.
<https://doi.org/10.1111/nyas.13259>
- Wang, S.V., Schneeweiss, S., RCT-DUPLICATE Initiative, Franklin, J.M., Desai, R.J., Feldman, W., Garry, E.M., Glynn, R.J., Lin, K.J., Paik, J., Patorno, E., Suissa, S., D’Andrea, E., Jawaid, D., Lee, H., Pawar, A., Sreedhara, S.K., Tesfaye, H., Bessette, L.G., Zabotka, L., Lee, S.B., Gautam, N., York, C., Zakoul, H., Concato, J., Martin, D., Paraoan, D., Quinto, K., 2023. Emulation of Randomized Clinical Trials With Nonrandomized Database Analyses: Results of 32 Clinical Trials. *JAMA* 329, 1376–1385.
<https://doi.org/10.1001/jama.2023.4221>
- Wang, S.V., Sreedhara, S.K., Schneeweiss, S., REPEAT Initiative, 2022. Reproducibility of real-world evidence studies using clinical practice data to inform regulatory and coverage decisions. *Nat Commun* 13, 5126. <https://doi.org/10.1038/s41467-022-32310-3>
- Wang, X., Rothen, J., Huang, S., Long, J.B., Soulous, P.R., Goldberg, S.B., Mamtani, R., Presley, C.J., Kunst, N., Ma, S., Wang, S.-Y., Gross, C.P., Dinan, M.A., 2025. Adoption of Broad Genomic Profiling in Patients With Cancer. *JAMA Oncol* e250499.
<https://doi.org/10.1001/jamaoncol.2025.0499>

- Wang, Y., Huang, J., He, H., Zhang, V., Zhou, Y., Hao, X., Ram, P., Qian, L., Xie, Q., Weng, R.-L., Lin, F., Hu, Y., Cui, L., Jiang, X., Xu, H., Hong, N., 2025. CDEMapper: enhancing National Institutes of Health common data element use with large language models. *J Am Med Inform Assoc* ocaf064. <https://doi.org/10.1093/jamia/ocaf064>
- Warner, J.L., Rioth, M.J., Mandl, K.D., Mandel, J.C., Kreda, D.A., Kohane, I.S., Carbone, D., Oretto, R., Wang, L., Zhu, S., Yao, H., Alterovitz, G., 2016. SMART precision cancer medicine: a FHIR-based app to provide genomic information at the point of care. *J Am Med Inform Assoc* 23, 701–710. <https://doi.org/10.1093/jamia/ocw015>
- Weng, C., Friedman, C., Rommel, C.A., Hurdle, J.F., 2019. A two-site survey of medical center personnel's willingness to share clinical data for research: implications for reproducible health NLP research. *BMC Med Inform Decis Mak* 19, 70. <https://doi.org/10.1186/s12911-019-0778-z>
- Westfall, J.M., Mold, J., Fagnan, L., 2007. Practice-based research--"Blue Highways" on the NIH roadmap. *JAMA* 297, 403–406. <https://doi.org/10.1001/jama.297.4.403>
- Whirl-Carrillo, M., McDonagh, E.M., Hebert, J.M., Gong, L., Sangkuhl, K., Thorn, C.F., Altman, R.B., Klein, T.E., 2012. Pharmacogenomics knowledge for personalized medicine. *Clin Pharmacol Ther* 92, 414–417. <https://doi.org/10.1038/clpt.2012.96>
- Wilkinson, M.D., Dumontier, M., Aalbersberg, I.J.J., Appleton, G., Axton, M., Baak, A., Blomberg, N., Boiten, J.-W., da Silva Santos, L.B., Bourne, P.E., Bouwman, J., Brookes, A.J., Clark, T., Crosas, M., Dillo, I., Dumon, O., Edmunds, S., Evelo, C.T., Finkers, R., Gonzalez-Beltran, A., Gray, A.J.G., Groth, P., Goble, C., Grethe, J.S., Heringa, J., 't Hoen, P.A.C., Hooft, R., Kuhn, T., Kok, R., Kok, J., Lusher, S.J., Martone, M.E., Mons, A., Packer, A.L., Persson, B., Rocca-Serra, P., Roos, M., van Schaik, R., Sansone, S.-A., Schultes, E., Sengstag, T., Slater, T., Strawn, G., Swertz, M.A., Thompson, M., van der Lei, J., van Mulligen, E., Velterop, J., Waagmeester, A., Wittenburg, P., Wolstencroft, K., Zhao, J., Mons, B., 2016. The FAIR Guiding Principles for scientific data management and stewardship. *Sci Data* 3, 160018. <https://doi.org/10.1038/sdata.2016.18>
- Williams, M.S., Buchanan, A.H., Davis, F.D., Fauchet, W.A., Hallquist, M.L.G., Leader, J.B., Martin, C.L., McCormick, C.Z., Meyer, M.N., Murray, M.F., Rahm, A.K., Schwartz, M.L.B., Sturm, A.C., Wagner, J.K., Williams, J.L., Willard, H.F., Ledbetter, D.H., 2018. Patient-Centered Precision Health In A Learning Health Care System: Geisinger's Genomic Medicine Experience. *Health Aff (Millwood)* 37, 757–764. <https://doi.org/10.1377/hlthaff.2017.1557>
- Williams, R.W., 2006. Expression genetics and the phenotype revolution. *Mamm Genome* 17, 496–502. <https://doi.org/10.1007/s00335-006-0006-x>
- Wu, F., Zhao, S., Yu, B., Chen, Y.-M., Wang, W., Song, Z.-G., Hu, Y., Tao, Z.-W., Tian, J.-H., Pei, Y.-Y., Yuan, M.-L., Zhang, Y.-L., Dai, F.-H., Liu, Y., Wang, Q.-M., Zheng, J.-J., Xu, L., Holmes, E.C., Zhang, Y.-Z., 2020. A new coronavirus associated with human respiratory disease in China. *Nature* 579, 265–269. <https://doi.org/10.1038/s41586-020-2008-3>
- Wünschiers, R., 2022. Genes, Genomes and Society.
- Xu, J., Lee, H.-J., Zeng, J., Wu, Y., Zhang, Y., Huang, L.-C., Johnson, A., Holla, V., Bailey, A.M., Cohen, T., Meric-Bernstam, F., Bernstam, E.V., Xu, H., 2016. Extracting genetic alteration information for personalized cancer therapy from ClinicalTrials.gov. *J Am Med Inform Assoc* 23, 750–757. <https://doi.org/10.1093/jamia/ocw009>

- Xu, J., Wang, F., Zang, C., Zhang, H., Niotis, K., Liberman, A.L., Stonnington, C.M., Ishii, M., Adekkanattu, P., Luo, Y., Mao, C., Rasmussen, L.V., Xu, Z., Brandt, P., Pacheco, J.A., Peng, Y., Jiang, G., Isaacson, R., Pathak, J., 2023. Comparing the effects of four common drug classes on the progression of mild cognitive impairment to dementia using electronic health records. *Sci Rep* 13, 8102. <https://doi.org/10.1038/s41598-023-35258-6>
- Xu, Y., Vuckovic, D., Ritchie, S.C., Akbari, P., Jiang, T., Grealey, J., Butterworth, A.S., Ouwehand, W.H., Roberts, D.J., Di Angelantonio, E., Danesh, J., Soranzo, N., Inouye, M., 2022. Machine learning optimized polygenic scores for blood cell traits identify sex-specific trajectories and genetic correlations with disease. *Cell Genom* 2, None. <https://doi.org/10.1016/j.xgen.2021.100086>
- Yang, L., Wang, S., Altman, R.B., 2023. POPDx: an automated framework for patient phenotyping across 392 246 individuals in the UK Biobank study. *J Am Med Inform Assoc* 30, 245–255. <https://doi.org/10.1093/jamia/ocac226>
- Yauy, K., Lecoquierre, F., Baert-Desurmont, S., Trost, D., Boughalem, A., Luscan, A., Costa, J.-M., Gerome, V., Raymond, L., Richard, P., Coutant, S., Broutin, M., Lanos, R., Fort, Q., Cackowski, S., Testard, Q., Diallo, A., Soirat, N., Holder, J.-M., Duforest-Frebourg, N., Bouge, A.-L., Beaumeunier, S., Bertrand, D., Audoux, J., Genevieve, D., Mesnard, L., Nicolas, G., Thevenon, J., Philippe, N., 2022. Genome Alert!: A standardized procedure for genomic variant reinterpretation and automated gene-phenotype reassessment in clinical routine. *Genet Med* 24, 1316–1327. <https://doi.org/10.1016/j.gim.2022.02.008>
- Yoon, J., Mizrahi, M., Ghalaty, N.F., Jarvinen, T., Ravi, A.S., Brune, P., Kong, F., Anderson, D., Lee, G., Meir, A., Bandukwala, F., Kanal, E., Arik, S.Ö., Pfister, T., 2023. EHR-Safe: generating high-fidelity and privacy-preserving synthetic electronic health records. *NPJ Digit Med* 6, 141. <https://doi.org/10.1038/s41746-023-00888-7>
- Yu, S., Ma, Y., Gronsbell, J., Cai, Tianrun, Ananthakrishnan, A.N., Gainer, V.S., Churchill, S.E., Szolovits, P., Murphy, S.N., Kohane, I.S., Liao, K.P., Cai, Tianxi, 2018. Enabling phenotypic big data with PheNorm. *J Am Med Inform Assoc* 25, 54–60. <https://doi.org/10.1093/jamia/ocx111>
- Yu, Y., Zong, N., Wen, A., Liu, S., Stone, D.J., Knaack, D., Chamberlain, A.M., Pfaff, E., Gabriel, D., Chute, C.G., Shah, N., Jiang, G., 2022. Developing an ETL tool for converting the PCORnet CDM into the OMOP CDM to facilitate the COVID-19 data integration. *J Biomed Inform* 127, 104002. <https://doi.org/10.1016/j.jbi.2022.104002>
- Zarrei, M., MacDonald, J.R., Merico, D., Scherer, S.W., 2015. A copy number variation map of the human genome. *Nat Rev Genet* 16, 172–183. <https://doi.org/10.1038/nrg3871>
- Zeng, C., Schlueter, D.J., Tran, T.C., Babbar, A., Cassini, T., Bastarache, L.A., Denny, J.C., 2024. Comparison of phenomic profiles in the All of Us Research Program against the US general population and the UK Biobank. *J Am Med Inform Assoc* 31, 846–854. <https://doi.org/10.1093/jamia/ocad260>
- Zerhouni, E.A., 2007. Translational research: moving discovery to practice. *Clin Pharmacol Ther* 81, 126–128. <https://doi.org/10.1038/sj.cpt.6100029>
- Zhang, B., Thacker, D., Zhou, T., Zhang, D., Lei, Y., Chen, J., Chrischilles, E.A., Christakis, D.A., Fernandez, S., Garg, V., Kim, S., Mosa, A.S.M., Sills, M.R., Taylor, B.W., Williams, D.A., Wu, Q., Forrest, C.B., Chen, Y., 2025. Cardiovascular post-acute sequelae of SARS-CoV-2 in children and adolescents: cohort study using electronic health records. *Nat Commun* 16, 3445. <https://doi.org/10.1038/s41467-025-56284-0>

- Zhang, M.M., Bahal, R., Rasmussen, T.P., Manautou, J.E., Zhong, X.-B., 2021. The growth of siRNA-based therapeutics: Updated clinical studies. *Biochem Pharmacol* 114432.
<https://doi.org/10.1016/j.bcp.2021.114432>
- Zhang, Y., Yang, H., Li, S., Li, W.-D., Wang, Y., 2021. Consumption of coffee and tea and risk of developing stroke, dementia, and poststroke dementia: A cohort study in the UK Biobank. *PLoS Med* 18, e1003830. <https://doi.org/10.1371/journal.pmed.1003830>
- Zhang, Z., Wang, P., liu, J.-L., 2022. CRISPR.
- Zhou, W., Kanai, M., Wu, K.-H.H., Rasheed, H., Tsuo, K., Hirbo, J.B., Wang, Y., Bhattacharya, A., Zhao, H., Namba, S., Surakka, I., Wolford, B.N., Lo Faro, V., Lopera-Maya, E.A., Läll, K., Favé, M.-J., Partanen, J.J., Chapman, S.B., Karjalainen, J., Kurki, M., Maasha, M., Brumpton, B.M., Chavan, S., Chen, T.-T., Daya, M., Ding, Y., Feng, Y.-C.A., Guare, L.A., Gignoux, C.R., Graham, S.E., Hornsby, W.E., Ingold, N., Ismail, S.I., Johnson, R., Laisk, T., Lin, K., Lv, J., Millwood, I.Y., Moreno-Grau, S., Nam, K., Palta, P., Pandit, A., Preuss, M.H., Saad, C., Setia-Verma, S., Thorsteinsdottir, U., Uzunovic, J., Verma, A., Zawistowski, M., Zhong, X., Afifi, N., Al-Dabholani, K.M., Al Thani, A., Bradford, Y., Campbell, A., Crooks, K., de Bock, G.H., Damrauer, S.M., Douville, N.J., Finer, S., Fritsche, L.G., Fthenou, E., Gonzalez-Arroyo, G., Griffiths, C.J., Guo, Y., Hunt, K.A., Ioannidis, A., Jansonius, N.M., Konuma, T., Lee, M.T.M., Lopez-Pineda, A., Matsuda, Y., Marioni, R.E., Moatamed, B., Nava-Aguilar, M.A., Numakura, K., Patil, S., Rafaels, N., Richmond, A., Rojas-Muñoz, A., Shortt, J.A., Straub, P., Tao, R., Vanderwerff, B., Vernekar, M., Veturi, Y., Barnes, K.C., Boezen, M., Chen, Z., Chen, C.-Y., Cho, J., Smith, G.D., Finucane, H.K., Franke, L., Gamazon, E.R., Ganna, A., Gaunt, T.R., Ge, T., Huang, H., Huffman, J., Katsanis, N., Koskela, J.T., Lajonchere, C., Law, M.H., Li, L., Lindgren, C.M., Loos, R.J.F., MacGregor, S., Matsuda, K., Olsen, C.M., Porteous, D.J., Shavit, J.A., Snieder, H., Takano, T., Trembath, R.C., Vonk, J.M., Whiteman, D.C., Wicks, S.J., Wijmenga, C., Wright, J., Zheng, J., Zhou, X., Awadalla, P., Boehnke, M., Bustamante, C.D., Cox, N.J., Fatumo, S., Geschwind, D.H., Hayward, C., Hveem, K., Kenny, E.E., Lee, S., Lin, Y.-F., Mbarek, H., Mägi, R., Martin, H.C., Medland, S.E., Okada, Y., Palotie, A.V., Pasaniuc, B., Rader, D.J., Ritchie, M.D., Sanna, S., Smoller, J.W., Stefansson, K., van Heel, D.A., Walters, R.G., Zöllner, S., Biobank of the Americas, Biobank Japan Project, BioMe, BioVU, CanPath - Ontario Health Study, China Kadoorie Biobank Collaborative Group, Colorado Center for Personalized Medicine, deCODE Genetics, Estonian Biobank, FinnGen, Generation Scotland, Genes & Health Research Team, LifeLines, Mass General Brigham Biobank, Michigan Genomics Initiative, National Biobank of Korea, Penn Medicine BioBank, Qatar Biobank, QSkin Sun and Health Study, Taiwan Biobank, HUNT Study, UCLA ATLAS Community Health Initiative, Uganda Genome Resource, UK Biobank, Martin, A.R., Willer, C.J., Daly, M.J., Neale, B.M., 2022. Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease. *Cell Genom* 2, 100192. <https://doi.org/10.1016/j.xgen.2022.100192>
- Zhou, Z., Wu, Q., Yan, Z., Zheng, H., Chen, C.-J., Liu, Y., Qi, Z., Calandrelli, R., Chen, Z., Chien, S., Su, H.I., Zhong, S., 2019. Extracellular RNA in a single droplet of human serum reflects physiologic and disease states. *Proc Natl Acad Sci U S A* 116, 19200–19208. <https://doi.org/10.1073/pnas.1908252116>
- Ziegler, A., Koval-Burt, C., Kay, D.M., Suchy, S.F., Begtrup, A., Langley, K.G., Hernan, R., Amendola, L.M., Boyd, B.M., Bradley, J., Brandt, T., Cohen, L.L., Coffey, A.J., Devaney, J.M., Dygulsk, B., Friedman, B., Fuleihan, R.L., Gyimah, A., Hahn, S., Hofherr, S.,

Hruska, K.S., Hu, Z., Jeanne, M., Jin, G., Johnson, D.A., Kavus, H., Leibel, R.L., Lobritto, S.J., McGee, S., Milner, J.D., McWalter, K., Monaghan, K.G., Orange, J.S., Pimentel Soler, N., Quevedo, Y., Ratner, S., Retterer, K., Shah, A., Shapiro, N., Sicko, R.J., Silver, E.S., Strom, S., Torene, R.I., Williams, O., Ustach, V.D., Wynn, J., Taft, R.J., Kruszka, P., Caggana, M., Chung, W.K., 2025. Expanded Newborn Screening Using Genome Sequencing for Early Actionable Conditions. *JAMA* 333, 232–240.

<https://doi.org/10.1001/jama.2024.19662>

Ziemann, M., Eren, Y., El-Osta, A., 2016. Gene name errors are widespread in the scientific literature. *Genome Biol* 17, 177. <https://doi.org/10.1186/s13059-016-1044-7>