

Reference List

- 1000 Genomes Project Consortium, Abecasis, G. R., Auton, A., Brooks, L. D., DePristo, M. A., Durbin, R. M., et al. (2012). An integrated map of genetic variation from 1,092 human genomes. *Nature*, *491*(7422), 56–65. doi:10.1038/nature11632
- Bauer, S., Köhler, S., Schulz, M. H., & Robinson, P. N. (2012). Bayesian ontology querying for accurate and noise-tolerant semantic searches. *Bioinformatics*, *28*(19), 2502–2508. doi:10.1093/bioinformatics/bts471
- Bauer-Mehren, A., van Mullingen, E. M., Avillach, P., Carrascosa, M. D. C., Garcia-Serna, R., Piñero, J., et al. (2012). Automatic filtering and substantiation of drug safety signals. *PLoS Computational Biology*, *8*(4), e1002457. doi:10.1371/journal.pcbi.1002457
- Biesecker, L. G., Burke, W., Kohane, I., Plon, S. E., & Zimmern, R. (2012). Next-generation sequencing in the clinic: are we ready? *Nature Publishing Group*, *13*(11), 818–824. doi:10.1038/nrg3357
- Califano, A., Butte, A. J., Friend, S., Ideker, T., & Schadt, E. (2012). Leveraging models of cell regulation and GWAS data in integrative network-based association studies. *Nature Genetics*, *44*(8), 841–847. doi:10.1038/ng.2355
- Cassa, C. A., Miller, R. A., & Mandl, K. D. (2013). A novel, privacy-preserving cryptographic approach for sharing sequencing data. *Journal of the American Medical Informatics Association*, *20*(1), 69–76. doi:10.1136/amiajnl-2012-001366
- Cassa, C. A., Savage, S. K., Taylor, P. L., Green, R. C., McGuire, A. L., & Mandl, K. D. (2012). Disclosing pathogenic genetic variants to research participants: quantifying an emerging ethical responsibility. *Genome Research*, *22*(3), 421–428. doi:10.1101/gr.127845.111
- Chen, R., Mias, G. I., Li-Pook-Than, J., Jiang, L., Lam, H. Y. K., Chen, R., et al. (2012). Personal omics profiling reveals dynamic molecular and medical phenotypes. *Cell*, *148*(6), 1293–1307. doi:10.1016/j.cell.2012.02.009
- Crockett, D. K., Lyon, E., Williams, M. S., Narus, S. P., Facelli, J. C., & Mitchell, J. A. (2012). Utility of gene-specific algorithms for predicting pathogenicity of uncertain gene variants. *Journal of the American Medical Informatics Association*, *19*(2), 207–211. doi:10.1136/amiajnl-2011-000309
- Cross-Disorder Group of the Psychiatric Genomics Consortium. (2013). Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis. *Lancet*. doi:10.1016/S0140-6736(12)62129-1
- Delaney, J. T., Ramirez, A. H., Bowton, E., Pulley, J. M., Basford, M. A., Schildcrout, J. S., et al. (2012). Predicting clopidogrel response using DNA samples linked to an electronic health record. *Clinical Pharmacology & Therapeutics*, *91*(2), 257–263. doi:10.1038/clpt.2011.221
- Divoli, A., Mendonça, E. A., Evans, J. A., & Rzhetsky, A. (2011). Conflicting biomedical

assumptions for mathematical modeling: the case of cancer metastasis. *PLoS Computational Biology*, 7(10), e1002132. doi:10.1371/journal.pcbi.1002132

Do, C. B., Hinds, D. A., Francke, U., & Eriksson, N. (2012). Comparison of family history and SNPs for predicting risk of complex disease. *PLoS genetics*, 8(10), e1002973. doi:10.1371/journal.pgen.1002973

Duke, J. D., Han, X., Wang, Z., Subhadarshini, A., Karnik, S. D., Li, X., et al. (2012). Literature based drug interaction prediction with clinical assessment using electronic medical records: novel myopathy associated drug interactions. (J. D. Duke, X. Han, Z. Wang, A. Subhadarshini, S. D. Karnik, X. Li, et al., Eds.) *PLoS Computational Biology*, 8(8), e1002614. doi:10.1371/journal.pcbi.1002614.t006

Ellis, M. J., Ding, L., Shen, D., Luo, J., Suman, V. J., Wallis, J. W., et al. (2012). Whole-genome analysis informs breast cancer response to aromatase inhibition. *Nature*, 486(7403), 353–360. doi:10.1038/nature11143

ENCODE Project Consortium, Dunham, I., Kundaje, A., Aldred, S. F., Collins, P. J., Davis, C. A., et al. (2012). An integrated encyclopedia of DNA elements in the human genome. *Nature*, 489(7414), 57–74. doi:10.1038/nature11247

Garnett, M. J., Edelman, E. J., Heidorn, S. J., Greenman, C. D., Dastur, A., Lau, K. W., et al. (2012). Systematic identification of genomic markers of drug sensitivity in cancer cells. *Nature*, 483(7391), 570–575. doi:10.1038/nature11005

Gerstein, M. B., Kundaje, A., Hariharan, M., Landt, S. G., Yan, K.-K., Cheng, C., et al. (2012). Architecture of the human regulatory network derived from ENCODE data. *Nature*, 489(7414), 91–100. doi:10.1038/nature11245

Gilman, S. R., Chang, J., Xu, B., Bawa, T. S., Gogos, J. A., Karayiorgou, M., & Vitkup, D. (2012). Diverse types of genetic variation converge on functional gene networks involved in schizophrenia. *Nature neuroscience*, 15(12), 1723–1728. doi:10.1038/nn.3261

Gu, H. H., Elhanan, G., Perl, Y., Hripcsak, G., Cimino, J. J., Xu, J., et al. (2012). A study of terminology auditors' performance for UMLS semantic type assignments. *JOURNAL OF BIOMEDICAL INFORMATICS*, 45(6), 1042–1048. doi:10.1016/j.jbi.2012.05.006

Gymrek, M., McGuire, A. L., Golan, D., Halperin, E., & Erlich, Y. (2013). Identifying personal genomes by surname inference. *Science*, 339(6117), 321–324. doi:10.1126/science.1229566

Hicks, J. K., Crews, K. R., Hoffman, J. M., Kornegay, N. M., Wilkinson, M. R., Lorier, R., et al. (2012). A clinician-driven automated system for integration of pharmacogenetic interpretations into an electronic medical record. *Clinical Pharmacology & Therapeutics*, 92(5), 563–566. doi:10.1038/clpt.2012.140

Jing, X., Kay, S., Marley, T., Hardiker, N. R., & Cimino, J. J. (2012). Incorporating personalized gene sequence variants, molecular genetics knowledge, and health knowledge into an EHR prototype based on the Continuity of Care Record standard. *JOURNAL OF BIOMEDICAL INFORMATICS*, 45(1), 82–92. doi:10.1016/j.jbi.2011.09.001

Karr, J. R., Sanghvi, J. C., Macklin, D. N., Gutschow, M. V., Jacobs, J. M., Bolival, B., et al. (2012). A whole-cell computational model predicts phenotype from genotype. *Cell*, 150(2),

Li, H., Lee, Y., Chen, J. L., Rebman, E., Li, J., & Lussier, Y. A. (2012a). Complex-disease networks of trait-associated single-nucleotide polymorphisms (SNPs) unveiled by information theory. *Journal of the American Medical Informatics Association*, *19*(2), 295–305. doi:10.1136/amiajnl-2011-000482

Li, J., & Lu, Z. (2012). Systematic identification of pharmacogenomics information from clinical trials. *JOURNAL OF BIOMEDICAL INFORMATICS*, *45*(5), 870–878. doi:10.1016/j.jbi.2012.04.005

Li, Z.-H., Liu, Y.-F., Li, K.-N., Duanmu, H.-Z., Chang, Z.-Q., Li, Z.-Q., et al. (2012b). Analysis of functional and pathway association of differential co-expressed genes: a case study in drug addiction. *JOURNAL OF BIOMEDICAL INFORMATICS*, *45*(1), 30–36. doi:10.1016/j.jbi.2011.08.014

Liu, D. J., & Leal, S. M. (2012). Estimating genetic effects and quantifying missing heritability explained by identified rare-variant associations. *American journal of human genetics*, *91*(4), 585–596. doi:10.1016/j.ajhg.2012.08.008

Prabhu, S., & Pe'er, I. (2012). Ultrafast genome-wide scan for SNP-SNP interactions in common complex disease. *Genome Research*, *22*(11), 2230–2240. doi:10.1101/gr.137885.112

Pulley, J. M., Denny, J. C., Peterson, J. F., Bernard, G. R., Vnencak-Jones, C. L., Ramirez, A. H., et al. (2012). Operational implementation of prospective genotyping for personalized medicine: the design of the Vanderbilt PREDICT project. *Clinical Pharmacology & Therapeutics*, *92*(1), 87–95. doi:10.1038/clpt.2011.371

Roberts, N. J., Vogelstein, J. T., Parmigiani, G., Kinzler, K. W., Vogelstein, B., & Velculescu, V. E. (2012). The predictive capacity of personal genome sequencing. *Science Translational Medicine*, *4*(133), 133ra58. doi:10.1126/scitranslmed.3003380

Samwald, M., & Adlassnig, K.-P. (2013). Pharmacogenomics in the pocket of every patient? A prototype based on quick response codes. *Journal of the American Medical Informatics Association*. doi:10.1136/amiajnl-2012-001275

Sanseau, P., Agarwal, P., Barnes, M. R., Pastinen, T., Richards, J. B., Cardon, L. R., & Mooser, V. (2012). Use of genome-wide association studies for drug repositioning. *Nature Biotechnology*, *30*(4), 317–320. doi:10.1038/nbt.2151

Sarkar, I. N. (2012). A vector space model approach to identify genetically related diseases. *Journal of the American Medical Informatics Association*, *19*(2), 249–254. doi:10.1136/amiajnl-2011-000480

Snitkin, E. S., Zelazny, A. M., Thomas, P. J., Stock, F., NISC Comparative Sequencing Program Group, Henderson, D. K., et al. (2012). Tracking a hospital outbreak of carbapenem-resistant *Klebsiella pneumoniae* with whole-genome sequencing. *Science Translational Medicine*, *4*(148), 148ra116. doi:10.1126/scitranslmed.3004129

Tenenbaum, J., James, A., & Paulyson-Nuñez, K. (2012). An Altered Treatment Plan Based on Direct to Consumer (DTC) Genetic Testing: Personalized Medicine from the Patient/Pin-cushion Perspective. *Journal of Personalized Medicine*, *2*(4), 192–200. doi:10.3390/jpm2040192

Voight, B. F., Peloso, G. M., Orho-Melander, M., Frikke-Schmidt, R., Barbalic, M., Jensen, M. K., et al. (2012). Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. *Lancet*, *380*(9841), 572–580. doi:10.1016/S0140-6736(12)60312-2

Wen, Z., Liu, Z.-P., Liu, Z., Zhang, Y., & Chen, L. (2012). An integrated approach to identify causal network modules of complex diseases with application to colorectal cancer. *Journal of the American Medical Informatics Association*. doi:10.1136/amiajnl-2012-001168

Yuan, Y., Failmezger, H., Rueda, O. M., Ali, H. R., Gräf, S., Chin, S.-F., et al. (2012). Quantitative image analysis of cellular heterogeneity in breast tumors complements genomic profiling. *Science Translational Medicine*, *4*(157), 157ra143. doi:10.1126/scitranslmed.3004330

Zuk, O., Hechter, E., Sunyaev, S. R., & Lander, E. S. (2012). The mystery of missing heritability: Genetic interactions create phantom heritability. *Proceedings of the National Academy of Sciences*, *109*(4), 1193–1198. doi:10.1073/pnas.1119675109