Supplementary References

for "Genomic Analysis Tools for Precision Medicine: Challenges and Future Perspectives in Oncology"

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This document lists the **31 genomic analysis tools** referenced in the paper.

References

- [1] Tobin, R. M., Singh, S., Kumar, S., & Miura, S. (2025). GenoPath: A pipeline to infer tumor clone composition, mutational history, and metastatic cell migration events from tumor DNA sequencing data. Frontiers in Bioinformatics, 5, 1615834. https://doi.org/10.3389/fbinf.2025.1615834
- [2] Pepe, G., Notturno Granieri, C., Appierdo, R., Ausiello, G., Helmer-Citterich, M., & Gherardini, P. F. (2025). PANDA: PAN Cancer Data Analysis Web Tool. Journal of Molecular Biology, 437(15), 169158. https://doi.org/10.1016/j.jmb.2025.169158
- [3] El-Kamand, S., Quinn, J. M. W., Sareen, H., Becker, T. M., Wong-Erasmus, M., & Cowley, M. J. (2024). CRUX: A platform for visualising, exploring and analysing cancer genome cohort data. NAR Genomics and Bioinformatics, 6(1), lqae003. https://doi.org/10.1093/nargab/lqae003
- [4] Deyell, R. J., et al. (2024). Whole genome and transcriptome integrated analyses guide clinical care of pediatric poor prognosis cancers. Nature Communications, 15, 48363. https://doi.org/10.1038/s41467-024-48363-5
- [5] Behera, S., Catreux, S., Rossi, M., Truong, S., Huang, Z., Ruehle, M., Visvanath, A., Parnaby, G., Roddey, C., Onuchic, V., Finocchio, A., Cameron, D. L., English, A., Mehtalia, S., Han, J., Mehio, R., & Sedlazeck, F. J. (2025). Comprehensive genome analysis and variant detection at scale using DRAGEN. Nature Biotechnology, 43(7), 1177–1191. https://doi.org/10.1038/s41587-024-02382-1

- [6] O'Connell, K. A., et al. (2023). Accelerating genomic workflows using NVIDIA Parabricks. BMC Bioinformatics, 24, 342. https://doi.org/10.1186/ s12859-023-05292-2
- [7] The Galaxy Community. (2022). The Galaxy platform for accessible, reproducible and collaborative biomedical analyses: 2022 update. Nucleic Acids Research, 50(W1), W345–W351. https://doi.org/10.1093/nar/gkac247
- [8] Anilkumar Sithara, A., et al. (2022). *iCOMIC: A graphical interface-driven bioinfor-matics pipeline for analyzing cancer omics data*. NAR Genomics and Bioinformatics, 4(3), lqac053. https://doi.org/10.1093/nargab/lqac053
- [9] Danecek, P., et al. (2021). Twelve years of SAMtools and BCFtools. GigaScience, 10(2), giab008. https://doi.org/10.1093/gigascience/giab008
- [10] He, X., et al. (2021). DIVIS: Integrated and Customizable Pipeline for Cancer Genome Sequencing Analysis and Interpretation. Frontiers in Oncology, 11, 672597. https://doi.org/10.3389/fonc.2021.672597
- [11] Mölder, F., et al. (2021). Sustainable data analysis with Snakemake. F1000Research, 10, 33. https://doi.org/10.12688/f1000research.29032.2
- [12] Ewels, P. A., et al. (2020). The nf-core framework for community-curated bioinformatics pipelines. Nature Biotechnology, 38(3), 276–278. https://doi.org/10. 1038/s41587-020-0439-x
- [13] Korhonen, P. K., Hall, R. S., Young, N. D., & Gasser, R. B. (2019). Common workflow language (CWL)-based software pipeline for de novo genome assembly from long- and short-read data. GigaScience, 8(4), giz014. https://doi.org/10.1093/ gigascience/giz014
- [14] Singer, J., et al. (2018). NGS-pipe: A flexible, easily extendable and highly configurable framework for NGS analysis. Bioinformatics, 34(1), 107–108. https://doi.org/10.1093/bioinformatics/btx540
- [15] Wingett, S. W., & Andrews, S. (2018). FastQ Screen: A tool for multi-genome mapping and quality control. F1000Research, 7, 1338. https://doi.org/10.12688/ f1000research.15931.2
- [16] Poplin, R., et al. (2018). A universal SNP and small-indel variant caller using deep neural networks. Nature Biotechnology, 36(10), 983–987. https://doi.org/ 10.1038/nbt.4235

- [17] Vivian, J., et al. (2017). Toil enables reproducible, open source, big biomedical data analyses. Nature Biotechnology, 35(4), 314–316. https://doi.org/10.1038/nbt. 3772
- [18] Lau, J. W., et al. (2017). The Cancer Genomics Cloud: Collaborative, Reproducible, and Democratized—A New Paradigm in Large-Scale Computational Research. Cancer Research, 77(21), e3–e6. https://doi.org/10.1158/0008-5472.CAN-17-0387
- [19] McLaren, W., et al. (2016). The Ensembl Variant Effect Predictor. Genome Biology, 17, 122. https://doi.org/10.1186/s13059-016-0974-4
- [20] Ewels, P., Magnusson, M., Lundin, S., & Käller, M. (2016). *MultiQC: Summarize analysis results for multiple tools and samples in a single report.* Bioinformatics, 32(19), 3047–3048. https://doi.org/10.1093/bioinformatics/btw354
- [21] Kim, D., Langmead, B., & Salzberg, S. L. (2015). HISAT: A fast spliced aligner with low memory requirements. Nature Methods, 12(4), 357–360. https://doi.org/10. 1038/nmeth.3317
- [22] Bolger, A. M., Lohse, M., & Usadel, B. (2014). Trimmomatic: A flexible trimmer for Illumina sequence data. Bioinformatics, 30(15), 2114–2120. https://doi.org/10.1093/bioinformatics/btu170
- [23] Van der Auwera, G. A., et al. (2013). From FastQ Data to High-Confidence Variant Calls: The Genome Analysis Toolkit Best Practices Pipeline. Current Protocols in Bioinformatics, 43, 11.10.1–11.10.33. https://doi.org/10.1002/0471250953.bi1110s43
- [24] Thorvaldsdottir, H., Robinson, J. T., & Mesirov, J. P. (2013). Integrative Genomics Viewer (IGV): High-performance genomics data visualization and exploration. Briefings in Bioinformatics, 14(2), 178–192. https://doi.org/10.1093/bib/bbs017
- [25] Dobin, A., et al. (2013). STAR: Ultrafast universal RNA-seq aligner. Bioinformatics, 29(1), 15–21. https://doi.org/10.1093/bioinformatics/bts635
- [26] Cingolani, P., et al. (2012). A program for annotating and predicting the effects of single nucleotide polymorphisms, SnpEff: SNPs in the genome of Drosophila melanogaster strain w1118; iso-2; iso-3. Fly, 6(2), 80-92. https://doi.org/10.4161/fly.19695
- [27] Langmead, B., & Salzberg, S. L. (2012). Fast gapped-read alignment with Bowtie 2. Nature Methods, 9(4), 357–359. https://doi.org/10.1038/nmeth.1923

- [28] Martin, M. (2011). Cutadapt removes adapter sequences from high-throughput sequencing reads. EMBnet.journal, 17(1), 10-12. https://doi.org/10.14806/ej.17.1.200
- [29] Wang, K., Li, M., & Hakonarson, H. (2010). ANNOVAR: Functional annotation of genetic variants from high-throughput sequencing data. Nucleic Acids Research, 38(16), e164. https://doi.org/10.1093/nar/gkq603
- [30] Li, H., & Durbin, R. (2009). Fast and accurate short read alignment with Burrows-Wheeler transform. Bioinformatics, 25(14), 1754-1760. https://doi.org/10.1093/bioinformatics/btp324
- [31] Li, H., et al. (2009). The Sequence Alignment/Map format and SAMtools. Bioinformatics, 25(16), 2078–2079. https://doi.org/10.1093/bioinformatics/btp352