

Sources for Technical Overview: What I'm Learning

Organized to match the sections on the Pasciro Labs technical overview page.

Genomics vs. genetics

- National Human Genome Research Institute (NHGRI). (2018, September 7). Genetics vs. Genomics fact sheet. <https://www.genome.gov/about-genomics/fact-sheets/Genetics-vs-Genomics>

DNA sequencing and what a “variant” means

- National Human Genome Research Institute (NHGRI). (2023, June 27). DNA sequencing fact sheet. <https://www.genome.gov/about-genomics/fact-sheets/DNA-Sequencing-Fact-Sheet>
- Richards, S., Aziz, N., Bale, S., et al. (2015). Standards and guidelines for the interpretation of sequence variants: A joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine. <https://www.nature.com/articles/gim201530>
- National Center for Biotechnology Information (NCBI). (2024, December 14). What is ClinVar? <https://www.ncbi.nlm.nih.gov/clinvar/intro/>
- Karczewski, K. J., Francioli, L. C., Tiao, G., et al. (2020). The mutational constraint spectrum quantified from variation in 141,456 humans. Nature. <https://www.nature.com/articles/s41586-020-2308-7>

Cell regulation and apoptosis (background concept)

- Alberts, B., Johnson, A., Lewis, J., Raff, M., Roberts, K., & Walter, P. (2002). Programmed cell death (apoptosis). In Molecular Biology of the Cell (4th ed.). NCBI Bookshelf. <https://www.ncbi.nlm.nih.gov/books/NBK26873/>

CRISPR basics and DNA repair pathways (NHEJ vs. HDR)

- Jinek, M., Chylinski, K., Fonfara, I., Hauer, M., Doudna, J. A., & Charpentier, E. (2012). A programmable dual-RNA-guided DNA endonuclease in adaptive bacterial immunity. Science. <https://www.science.org/doi/10.1126/science.1225829>
- Hsu, P. D., Lander, E. S., & Zhang, F. (2014). Development and applications of CRISPR-Cas9 for genome engineering. Cell, 157(6), 1262-1278. <https://doi.org/10.1016/j.cell.2014.05.010>
- Lieber, M. R. (2010). The mechanism of double-strand DNA break repair by the nonhomologous DNA end-joining pathway. Annual Review of Biochemistry, 79, 181-211. <https://doi.org/10.1146/annurev.biochem.052308.093131>

- National Academies of Sciences, Engineering, and Medicine. (2017). The basic science of genome editing (NHEJ and HDR overview). NCBI Bookshelf.
<https://www.ncbi.nlm.nih.gov/books/NBK447276/>

Off-target effects and uncertainty

- Tsai, S. Q., Zheng, Z., Nguyen, N. T., et al. (2015). GUIDE-seq enables genome-wide profiling of off-target cleavage by CRISPR-Cas nucleases. Nature Biotechnology.
<https://www.nature.com/articles/nbt.3117>
- Zhang, X.-H., Tee, L. Y., Wang, X.-G., Huang, Q.-S., & Yang, S.-H. (2015). Off-target effects in CRISPR/Cas9-mediated genome engineering. Molecular Therapy - Nucleic Acids.
<https://pubmed.ncbi.nlm.nih.gov/26575098/>

Delivery and real-world constraints

- Lino, C. A., Harper, J. C., Carney, J. P., & Timlin, J. A. (2018). Delivering CRISPR: A review of the challenges and approaches. Drug Delivery. <https://pubmed.ncbi.nlm.nih.gov/29801422/>

Case study: beta-thalassemia and gene editing

- Frangoul, H., Altshuler, D., Cappellini, M. D., et al. (2021). CRISPR-Cas9 gene editing for sickle cell disease and beta-thalassemia. New England Journal of Medicine, 384(3), 252-260.
<https://www.nejm.org/doi/full/10.1056/NEJMoa2031054>

Ethics, governance, and access

- National Academies of Sciences, Engineering, and Medicine. (2017). Human Genome Editing: Science, Ethics, and Governance. The National Academies Press.
<https://nap.nationalacademies.org/catalog/24623/human-genome-editing-science-ethics-and-governance>
- World Health Organization. (2021). Human genome editing: A framework for governance.
<https://www.who.int/publications/i/item/9789240030060>

Note: This list is intended for educational purposes and for documenting sources used to explain concepts in plain language.