

CHAPTER VII

REFERENCES

PCR Primer Design Guidelines. (n.d.). Retrieved September 8, 2019, from

http://www.premierbiosoft.com/tech_notes/PCR_Primer_Design.html

Troubleshooting Guide for Cloning. (n.d.). Retrieved July 12, 2019, from <https://www.neb.com/tools-and-resources/troubleshooting-guides/troubleshooting-guide-for-cloning>

Abdelhamed, Z. A., Wheway, G., Szymanska, K., Natarajan, S., Toomes, C., Inglehearn, C., & Johnson, C. A. (2013). Variable expressivity of ciliopathy neurological phenotypes that encompass Meckel–Gruber syndrome and Joubert syndrome is caused by complex de-regulated ciliogenesis, Shh and Wnt signalling defects. *Human Molecular Genetics*, 22(7), 1358–1372.

<https://doi.org/10.1093/hmg/dds546>

Alberts, B. (2015). Molecular Biology of the Cell, Sixth Edition. In International Journal of Molecular Sciences (Vol. 16). <https://doi.org/10.3390/ijms161226074>

Alexiev, B. A., Lin, X., Sun, C.-C., & Brenner, D. S. (2006). Meckel-Gruber syndrome: pathologic manifestations, minimal diagnostic criteria, and differential diagnosis. *Archives of Pathology & Laboratory Medicine*, 130(8), 1236–1238. [https://doi.org/10.1043/1543-2165\(2006\)130\[1236:MS\]2.0.CO;2](https://doi.org/10.1043/1543-2165(2006)130[1236:MS]2.0.CO;2)

Arts, H. H., Doherty, D., van Beersum, S. E. C., Parisi, M. A., Letteboer, S. J. F., Gorden, N. T., ... Roepman, R. (2007). Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. *Nature Genetics*, 39(7), 882–888. <https://doi.org/10.1038/ng2069>

- Basten, S. G., & Giles, R. H. (2013). Functional aspects of primary cilia in signaling, cell cycle and tumorigenesis. *Cilia*, 2(1), 6. <https://doi.org/10.1186/2046-2530-2-6>
- Berg, J., Tymoczko, J., & Stryer, L. (2002). Biochemistry (5th ed.). New York: W H Freeman.
- Bergmann, C., Fliegauf, M., Brüchle, N. O., Frank, V., Olbrich, H., Kirschner, J., ... Omran, H. (2008). Loss of Nephrocystin-3 Function Can Cause Embryonic Lethality, Meckel-Gruber-like Syndrome, Situs Inversus, and Renal-Hepatic-Pancreatic Dysplasia. *The American Journal of Human Genetics*, 82(4), 959–970. <https://doi.org/10.1016/j.ajhg.2008.02.017>
- Brancati, F., Dallapiccola, B., & Valente, E. (2010). Joubert Syndrome and related disorders. *Orphanet Journal of Rare Diseases*, 5(1), 20. <https://doi.org/10.1186/1750-1172-5-20>
- Bzymek, M., & Lovett, S. T. (2002). Instability of repetitive DNA sequences: The role of replication in multiple mechanisms. *Proceedings of the National Academy of Sciences*, 98(15), 8319–8325. <https://doi.org/10.1073/pnas.111008398>
- Chan, V., Dreolini, L. F., Flintoff, K. A., Lloyd, S. J., & Mattenley, A. A. (2002). The Effect of Increasing Plasmid Size on Transformation Efficiency in Escherichia coli. 2(April), 207–223.
- Christensen, S. T., Clement, C. A., Satir, P., & Pedersen, L. B. (2012). Primary cilia and coordination of receptor tyrosine kinase (RTK)-signaling. *J Pathol*, 226.
- Clevers, H., & Nusse, R. (2012). Wnt/β-Catenin Signaling and Disease. *Cell*, 149(6), 1192–1205. <https://doi.org/10.1016/j.cell.2012.05.012>
- Dutra, B. E., Sutera, V. A., & Lovett, S. T. (2007). RecA-independent recombination is efficient but limited by exonucleases. *Proceedings of the National Academy of Sciences*, 104(1), 216–221. <https://doi.org/10.1073/pnas.0608293104>
- Fliegauf, M., Benzing, T., & Omran, H. (2007). When cilia go bad: cilia defects and ciliopathies. *Nat Rev Mol Cell Biol*, 8.

- Gerhardt, C., Lier, J. M., Burmühl, S., Struchtrup, A., Deutschmann, K., Vetter, M., ... Rüther, U. (2015). The transition zone protein Rpgrip1l regulates proteasomal activity at the primary cilium. *The Journal of Cell Biology*, 210(1), 115–133. <https://doi.org/10.1083/jcb.201408060>
- Gherman, A., Davis, E. E., & Katsanis, N. (2006). The ciliary proteome database: an integrated community resource for the genetic and functional dissection of cilia. *Nature Genetics*, 38(9), 961–962. <https://doi.org/10.1038/ng0906-961>
- Guruharsha, K. G., Kankel, M. W., & Artavanis-Tsakonas, S. (2012). The Notch signalling system: recent insights into the complexity of a conserved pathway. *Nature Reviews Genetics*, 13(9), 654–666. <https://doi.org/10.1038/nrg3272>
- Hansen, K. (n.d.). Ligation of DNA DNA Ligase. 225–230. <https://doi.org/10.1385/0-89603-064-4:225>
- Horii, T., Ogawa, T., Nakatani, T., Hase, T., Matsubara, H., & Ogawa, H. (1981). Regulation of SOS functions: Purification of *E. coli* LexA protein and determination of its specific site cleaved by the RecA protein. *Cell*, 27(3), 515–522. [https://doi.org/10.1016/0092-8674\(81\)90393-7](https://doi.org/10.1016/0092-8674(81)90393-7)
- Huang, P., & Schier, A. F. (2009). Dampened Hedgehog signaling but normal Wnt signaling in zebrafish without cilia. *Development*, 136(18), 3089–3098. <https://doi.org/10.1242/dev.041343>
- Lee, J. E., & Gleeson, J. G. (2011). A systems-biology approach to understanding the ciliopathy disorders. *Genome Medicine*, 3(9), 59. <https://doi.org/10.1186/gm275>
- Logan, C. V., Abdel-Hamed, Z., & Johnson, C. A. (2011). Molecular Genetics and Pathogenic Mechanisms for the Severe Ciliopathies: Insights into Neurodevelopment and Pathogenesis of Neural Tube Defects. *Molecular Neurobiology*, 43(1), 12–26. <https://doi.org/10.1007/s12035-010-8154-0>
- Lu, Q., Insinna, C., Ott, C., Stauffer, J., Pintado, P. A., Rahajeng, J., ... Westlake, C. J. (2015). Early steps in primary cilium assembly require EHD1/EHD3-dependent ciliary vesicle formation. *Nature Cell Biology*, 17(3), 228–240. <https://doi.org/10.1038/ncb3109>

Morgan, K., Juchheimm, M. A., & Patrick, M. (2014). Plasmids 101: The promotor region (Vol. 2017).

Retrieved from www.addgene.org

Ocbina, P. J. R., Tuson, M., & Anderson, K. V. (2009). Primary Cilia Are Not Required for Normal

Canonical Wnt Signaling in the Mouse Embryo. *PLoS ONE*, 4(8), e6839.

<https://doi.org/10.1371/journal.pone.0006839>

Parisi, M. A. (2009). Clinical and molecular features of Joubert syndrome and related disorders.

American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 151C(4), 326–340.

<https://doi.org/10.1002/ajmg.c.30229>

Patnaik, S. R., Raghupathy, R. K., Zhang, X., Mansfield, D., & Shu, X. (2015). The Role of RPGR and Its

Interacting Proteins in Ciliopathies. *Journal of Ophthalmology*, 2015, 1–10.

<https://doi.org/10.1155/2015/414781>

Pedersen, L. B., Veland, I. R., Schrøder, J. M., & Christensen, S. T. (2008). Assembly of primary cilia.

Developmental Dynamics, 237(8), 1993–2006. <https://doi.org/10.1002/dvdy.21521>

Rahimzadeh, M., Sadeghizadeh, M., Najafi, F., Arab, S., Mobasher, H., Modares, T., ... Modares, T.

(2016). Impact of heat shock step on bacterial transformation efficiency. 5(4), 257–261.

Roberts, R. J. (2005). How restriction enzymes became the workhorses of molecular biology.

Proceedings of the National Academy of Sciences, 102(17), 5905–5908.

<https://doi.org/10.1073/pnas.0500923102>

Salonen, R., Kestilä, M., & Bergmann, C. (2011). Clinical utility gene card for: Meckel syndrome.

European Journal of Human Genetics, 19(7), 832–832. <https://doi.org/10.1038/ejhg.2010.255>

Satir, P., Pedersen, L. B., & Christensen, S. T. (2010). The primary cilium at a glance. *Journal of Cell*

Science, 123(4), 499–503. <https://doi.org/10.1242/jcs.050377>

- Simons, M., Gloy, J., Ganner, A., Bullerkotte, A., Bashkurov, M., Krönig, C., ... Walz, G. (2005). Inversin, the gene product mutated in nephronophthisis type II, functions as a molecular switch between Wnt signaling pathways. *Nature Genetics*, 37(5), 537–543.
<https://doi.org/10.1038/ng1552>
- Vincze, T. (2003). NEBcutter: a program to cleave DNA with restriction enzymes. *Nucleic Acids Research*, 31(13), 3688–3691. <https://doi.org/10.1093/nar/gkg526>
- Waters, A. M., & Beales, P. L. (2011). Ciliopathies: an expanding disease spectrum. *Pediatric Nephrology*, 26(7), 1039–1056. <https://doi.org/10.1007/s00467-010-1731-7>
- Wheway, G., Nazlamova, L., & Hancock, J. T. (2018). Signaling through the Primary Cilium. *Frontiers in Cell and Developmental Biology*, 6. <https://doi.org/10.3389/fcell.2018.00008>
- Wiegering, A., Rüther, U., & Gerhardt, C. (2018). The ciliary protein Rpgrip1l in development and disease. *Developmental Biology*, 442(1), 60–68. <https://doi.org/10.1016/j.ydbio.2018.07.024>
- Williantarra, I. (2018). Pengaruh Protein RPGRIP1L pada Pembentukan Silia Primer sebagai Kandidat Target Terapi Gen Penyakit Siliopati. *Cdk*, 45(3), 181–185.
- Yu, S. L., Tang, Y. Q., Li, Y., Zhang, H., & Wu, X. L. (2010). Gradient decrement of elongation time can improve PCR with fluorescent-labeled primers. *Journal of Bioscience and Bioengineering*, 110(4), 500–504. <https://doi.org/10.1016/j.jbiosc.2010.05.005>