

# LQN100

## Human Genetics

[View Online](#)

Ada Hamosh MD, MPH. (2016). Clinical Case Studies Illustrating Genetic Principles. In Thompson & Thompson Genetics in Medicine (Eighth Edition, pp. 394–395). Elsevier.  
<https://www-clinicalkey-com-au.ezp01.library.qut.edu.au/#!/content/book/3-s2.0-B9781437706963000200?scrollTo=%23sc0015>

Augui, S., Nora, E. P., & Heard, E. (2011). Regulation of X-chromosome inactivation by the X-inactivation centre. *Nature Reviews Genetics*, 12(6), 429–442.  
<https://doi.org/10.1038/nrg2987>

Bennett, Robin LFrench, Kathryn SteinhausResta, Robert GDoyle, Debra Lochner. (n.d.). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, 17, 424–433.  
<https://www.proquest.com/docview/218649564?accountid=13380>

Boyd, J. D., & Hamilton, W. J. (1967). Foetus--or Fetus? *BMJ*, 1(5537), 425–425.  
<https://doi.org/10.1136/bmj.1.5537.425>

Brown, Carolyn JBallabio, Andreaet al. (n.d.). A Gene from the Region of the Human X Inactivation Centre Is Expressed Exclusively from the Inactive X Chromosome. *Nature*, 349 (9), 38–44. <https://www.proquest.com/docview/204459124?accountid=13380>

D. Peter Snustad , and Michael J. Simmons. (2015a). Chapter 15: Genomics (chapter portion). In *Principles of genetics* (7th ed., pp. 387–395). Wiley.  
<https://ebookcentral.proquest.com/lib/qut/reader.action?docID=4806596&ppg=401>

D. Peter Snustad , and Michael J. Simmons. (2015b). Chapter 15: Genomics (chapter portion). In *Principles of genetics* (7th ed., pp. 387–395). Wiley.  
<https://ebookcentral.proquest.com/lib/qut/reader.action?docID=4806596&ppg=401>

Disteche, Christine MBerleth, Joel B. (2015). X-chromosome inactivation and escape. *Journal of Genetics*, 94(4), 591–599.  
<https://www.proquest.com/docview/1754491907?accountid=13380>

Dobyns, W. B., Filauro, A., Tomson, B. N., Chan, A. S., Ho, A. W., Ting, N. T., Oosterwijk, J. C., & Ober, C. (2004). Inheritance of most X-linked traits is not dominant or recessive, just X-linked. *American Journal of Medical Genetics*, 129A(2), 136–143.  
<https://doi.org/10.1002/ajmg.a.30123>

Esteller, Manel. (n.d.). Non-coding RNAs in human disease. *Nature Reviews Genetics*, 12, 861–874. <https://www.proquest.com/docview/905092976?accountid=13380>

Frenette, Paul SAtweh, George F. (n.d.). Sickle cell disease: old discoveries, new concepts, and future promise. 117(7), 850–858.

<https://www.proquest.com/docview/200538792?accountid=13380>

Hamamy, H., Antonarakis, S. E., Cavalli-Sforza, L. L., Temtamy, S., Romeo, G., Kate, L. P. T., Bennett, R. L., Shaw, A., Megarbane, A., van Duijn, C., Bathija, H., Fokstuen, S., Engel, E., Zlotogora, J., Dermitzakis, E., Bottani, A., Dahoun, S., Morris, M. A., Arsenault, S., ... Bittles, A. H. (2011). Consanguineous marriages, pearls and perils: Geneva International Consanguinity Workshop Report. *Genetics in Medicine*, 13(9), 841–847.

<https://doi.org/10.1097/GIM.0b013e318217477f>

Harrow, J., Frankish, A., Gonzalez, J. M., Tapanari, E., Diekhans, M., Kokocinski, F., Aken, B. L., Barrell, D., Zadissa, A., Searle, S., Barnes, I., Bignell, A., Boychenko, V., Hunt, T., Kay, M., Mukherjee, G., Rajan, J., Despacio-Reyes, G., Saunders, G., ... Hubbard, T. J. (2012). GENCODE: The reference human genome annotation for The ENCODE Project. *Genome Research*, 22(9), 1760–1774. <https://doi.org/10.1101/gr.135350.111>

Houtgraaf, J. H., Versmissen, J., & van der Giessen, W. J. (2006). A concise review of DNA damage checkpoints and repair in mammalian cells. *Cardiovascular Revascularization Medicine*, 7(3), 165–172. <https://doi.org/10.1016/j.carrev.2006.02.002>

Jensen, Ole N. (n.d.). Interpreting the protein language using proteomics. *Nature Reviews Molecular Cell Biology*, 7(6), 391–403.

<https://www.proquest.com/docview/224665544?accountid=13380>

Jesse D. Riordan & Joseph H. Nadeau. (2017). From Peas to Disease: Modifier Genes, Network Resilience, and the Genetics of Health. *American Journal of Human Genetics*, 101 (2), 177–191. <https://doi.org/10.1016/j.ajhg.2017.06.004>

Knudsen, G. P. (2009). Gender bias in autoimmune diseases. *Journal of the Neurological Sciences*, 286(1-2), 43–46. <https://doi.org/10.1016/j.jns.2009.04.022>

Korf, B. R., & Irons, M. B. (2013). The human genome. In *Human Genetics and Genomics* (4th Revised edition, pp. 77–82). John Wiley and Sons Ltd.

Lam, Jenny K WChow, Michael Y TZhang, YuLeung, Susan W S. (n.d.). siRNA Versus miRNA as Therapeutics for Gene Silencing. *Molecular Therapy. Nucleic Acids*, 4, 745–747. <https://www.proquest.com/docview/1796352097?accountid=13380>

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., ... Grahama, D. (2001). Initial sequencing and analysis of the human genome. *Nature*, 409(6822), 860–921. <https://doi.org/10.1038/35057062>

Maria Kousi & Nicholas Katsanis. (2015). Genetic Modifiers and Oligogenic Inheritance. *Cold Spring Harbor Perspectives in Medicine*, 5(6 (Article 017145)). <https://doi.org/10.1101/cshperspect.a017145>

Nussbaum, Robert, L., McInness, Roderick, R., & Willard, Huntington, F. (2016). Introduction to the Human Genome. In *Thompson & Thompson Genetics in Medicine* (8th ed., pp. 3–20). Elsevier. <https://www.clinicalkey.com.au/#!/content/book/3-s2.0-B9781437706963000029>

- Pan, Q., Shai, O., Lee, L. J., Frey, B. J., & Blencowe, B. J. (2008a). Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nature Genetics*, 40(12), 1413–1415. <https://doi.org/10.1038/ng.259>
- Pan, Q., Shai, O., Lee, L. J., Frey, B. J., & Blencowe, B. J. (2008b). Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nature Genetics*, 40(12), 1413–1415. <https://doi.org/10.1038/ng.259>
- Pink, R. C., Wicks, K., Caley, D. P., Punch, E. K., Jacobs, L., & Francisco Carter, D. R. (2011). Pseudogenes: Pseudo-functional or key regulators in health and disease? *RNA*, 17(5), 792–798. <https://doi.org/10.1261/rna.2658311>
- Qun Pan,Ofer Shai,Leo J. Lee,Brendan J. Frey,Benjamin J. Blencowe. (n.d.). Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nature Genetics*.  
<http://go.galegroup.com/ps/i.do?p=HRCA&u=qut&id=GALE|A192498260&v=2.1&it=r&sid=summon&authCount=1#>
- Robert L. Nussbaum MD, FACP, FACMG. (2016a). Patterns of Single-Gene Inheritance. In Thompson & Thompson Genetics in Medicine (8th ed., pp. 107–132). Elsevier.  
<https://www.clinicalkey.com.au/#!/content/book/3-s2.0-B9781437706963000078>
- Robert L. Nussbaum MD, FACP, FACMG. (2016b). The Human Genome: Gene Structure and Function. In Thompson & Thompson Genetics in Medicine (Eighth Edition, pp. 21–32). Elsevier.  
<https://www-clinicalkey-com-au.ezp01.library.qut.edu.au/#!/content/book/3-s2.0-B9781437706963000030>
- Rodenhiser, D. (2006). Epigenetics and human disease: translating basic biology into clinical applications. *Canadian Medical Association Journal*, 174(3), 341–348.  
<https://doi.org/10.1503/cmaj.050774>
- Schoenwolf, G. C., Bleyl, S. B., Brauer, P. R., Francis-West, P. H., & Larsen, W. J. (n.d.). Larsen's human embryology (Fifth edition).  
[http://qut.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package\\_service\\_id=2724006090004001&institutionId=4001&customerId=4000](http://qut.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=2724006090004001&institutionId=4001&customerId=4000)
- Snustad, D. P., & Simmons, M. J. (2015). Chapter 3: Mendelism: The basic principles of inheritance. In Principles of Genetics (7th ed., pp. 40–61). Wiley.  
<https://ebookcentral.proquest.com/lib/qut/reader.action?docID=4806596&ppg=62&c=UERG>
- Snustad, D. P., & Simmons, M. J. (2016). Chapter 4: Extensions of Mendelism. In Principles of Genetics (7th ed., pp. 62–87). Wiley.  
<https://ebookcentral.proquest.com/lib/qut/reader.action?docID=4806596&ppg=84&c=UERG>
- The ENCODE (ENCylopedia Of DNA Elements) Project. (2004). *Science*, 306(5696), 636–640. <https://doi.org/10.1126/science.1105136>