Genetics & Molecular Biology

Year 1 — Bachelor of Dental Surgery

Reference and Resource List — Chapters 1 to 12

Supporting the MCQ, SBA, SAQ and flashcard question bank

BM Education FZE

Disclaimer

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References are presented in Vancouver style. Part 1 lists the core sources that apply across the whole subject; Part 2 lists, for each chapter, the relevant sections of those core texts together with additional chapter-specific reading. Numbers in square brackets in Part 2 refer to the numbered entries in Part 1.

PART 1 — CORE REFERENCES (ALL CHAPTERS)

1.1 Principal genetics and molecular biology textbooks

1. Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in Medicine. 8th ed. Philadelphia: Elsevier; 2016.
2. Cohn RD, Scherer SW, Hamosh A, editors. Thompson & Thompson Genetics and Genomics in Medicine. 9th ed. Philadelphia: Elsevier; 2017.
3. Turnpenny PD, Ellard S, Cleaver R. Emery's Elements of Medical Genetics and Genomics. 16th ed. Edinburgh: Elsevier; 2021.
4. Strachan T, Read A. Human Molecular Genetics. 5th ed. Boca Raton (FL): CRC Press/Garland Science; 2018.
5. Griffiths AJF, Doebley J, Peichel C, Wassarman DA. Introduction to Genetic Analysis. 12th ed. New York: W.H. Freeman; 2020.
6. Krebs JE, Goldstein ES, Kilpatrick ST. Lewin's Genes XII. 12th ed. Burlington (MA): Jones & Bartlett Learning; 2018.

1.2 Cell biology and biochemistry for molecular foundations

7. Alberts B, Heald R, Johnson A, Morgan D, Raff M, Roberts K, et al. Molecular Biology of the Cell. 7th ed. New York: W.W. Norton; 2022.
8. Lodish H, Berk A, Kaiser CA, Krieger M, Bretscher A, Ploegh H, et al. Molecular Cell Biology. 9th ed. New York: W.H. Freeman/Macmillan Learning; 2021.
9. Nelson DL, Cox MM, Hoskins AA. Lehninger Principles of Biochemistry. 8th ed. New York: W.H. Freeman/Macmillan Learning; 2021.

10. Kennelly PJ, Botham KM, McGuinness OP, Rodwell VW, Weil PA. Harper's Illustrated Biochemistry. 33rd ed. New York: McGraw Hill; 2026.
11. Vasudevan DM, Sreekumari S, Vaidyanathan K. Textbook of Biochemistry for Dental Students. 5th ed. New Delhi: Jaypee Brothers Medical Publishers; 2025.

1.3 Human and medical genetics for clinical application

12. Jorde LB, Carey JC, Bamshad MJ. Medical Genetics. 6th ed. Philadelphia: Elsevier; 2020.
13. Read A, Donnai D. New Clinical Genetics. 3rd ed. Banbury: Scion Publishing; 2015.
14. Gardner RJM, Amor DJ. Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling. 5th ed. New York: Oxford University Press; 2018.
15. Firth HV, Hurst JA. Oxford Desk Reference: Clinical Genetics and Genomics. 2nd ed. Oxford: Oxford University Press; 2017.
16. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, et al. 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. Genet Med. 2015;17(5):405–24.
17. Hamosh A, Scott AF, Amberger JS, Bocchini CA, McKusick VA. Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. Nucleic Acids Res. 2005;33(Database issue):D514–7.
18. Hardy GH. Mendelian proportions in a mixed population. Science. 1908;28(706):49–50.
19. Lyon MF. Gene action in the X-chromosome of the mouse (Mus musculus L.). Nature. 1961;190:372–3.

1.4 Molecular techniques, genomics and bioinformatics

20. Brown TA. Gene Cloning and DNA Analysis: An Introduction. 8th ed. Hoboken (NJ): Wiley-Blackwell; 2021.
21. Brown TA. Genomes 5. 5th ed. Boca Raton (FL): CRC Press; 2023.
22. Green MR, Sambrook J. Molecular Cloning: A Laboratory Manual. 4th ed. Cold Spring Harbor (NY): Cold Spring Harbor Laboratory Press; 2012.
23. Pevsner J. Bioinformatics and Functional Genomics. 3rd ed. Hoboken (NJ): Wiley-Blackwell; 2015.
24. International Human Genome Sequencing Consortium. Initial sequencing and analysis of the human genome. Nature. 2001;409(6822):860–921.
25. Nurk S, Koren S, Rhie A, Rautiainen M, Bzikadze AV, Mikheenko A, et al. The complete sequence of a human genome. Science. 2022;376(6588):44–53.

1.5 Oral biology, craniofacial development and dental application

26. Nanci A. Ten Cate's Oral Histology: Development, Structure, and Function. 10th ed. St Louis: Elsevier; 2024.
27. Berkovitz BKB, Holland GR, Moxham BJ. Oral Anatomy, Histology and Embryology. 5th ed. Edinburgh: Elsevier; 2017.
28. Sperber GH, Sperber SM. Craniofacial Embryogenetics and Development. 3rd ed. Raleigh (NC): PMPH USA; 2018.
29. Odell EW, editor. Cawson's Essentials of Oral Pathology and Oral Medicine. 9th ed. Edinburgh: Elsevier; 2017.

30. Cameron AC, Widmer RP, editors. Handbook of Pediatric Dentistry. 5th ed. Edinburgh: Elsevier; 2021.
31. Lamont RJ, Hajishengallis GN, Koo H, Jenkinson HF, editors. Oral Microbiology and Immunology. 3rd ed. Washington (DC): ASM Press; 2019.
32. Thesleff I. Epithelial-mesenchymal signalling regulating tooth morphogenesis. J Cell Sci. 2003;116(Pt 9):1647–8.

1.6 Standards, guidelines and professional requirements

33. General Dental Council. The Safe Practitioner: a framework of behaviours and outcomes for dental professional education. London: GDC; 2023. Available from: https://www.gdc-uk.org
34. National Dental Commission Act, 2023. New Delhi: Government of India; 2023. Available from: https://www.indiacode.nic.in
35. Field JC, Cowpe JG, Walmsley AD. The graduating European dentist: a new undergraduate curriculum framework. Eur J Dent Educ. 2017;21 Suppl 1:2–10.
36. Turnbull C, Scott RH, Thomas E, Jones L, Murugaesu N, Pretty FB, et al. The 100 000 Genomes Project: bringing whole genome sequencing to the NHS. BMJ. 2018;361:k1687.
37. NHS England. NHS Genomic Medicine Service. Available from: https://www.england.nhs.uk/genomics/
38. United Nations Educational, Scientific and Cultural Organization. Universal Declaration on the Human Genome and Human Rights. Paris: UNESCO; 1997. Available from: https://www.unesco.org
39. Nuffield Council on Bioethics. Genome editing and human reproduction: social and ethical issues. London: Nuffield Council on Bioethics; 2018. Available from: https://www.nuffieldbioethics.org

1.7 Key journals for currency

Nature Reviews Genetics · Nature Genetics · American Journal of Human Genetics · Genetics in Medicine · Journal of Medical Genetics · Human Molecular Genetics · European Journal of Human Genetics · Journal of Dental Research · Archives of Oral Biology · Oral Diseases · British Dental Journal · Clinical Genetics

PART 2 — CHAPTER-SPECIFIC REFERENCES

Chapter 1 — Introduction to Genetics & Molecular Biology

1.1 Scope and relevance to dentistry · 1.2 Basic biological organization · 1.3 Cell structure with genetic focus

Core texts: [3] Ch. 1 · [2] Ch. 1 · [7] chapters on cells and genomes · [11] introductory chapters on molecular biology · [26] Ch. 1

Additional

- Avery OT, MacLeod CM, McCarty M. Studies on the chemical nature of the substance inducing transformation of pneumococcal types. J Exp Med. 1944;79(2):137–58.
- Hershey AD, Chase M. Independent functions of viral protein and nucleic acid in growth of bacteriophage. J Gen Physiol. 1952;36(1):39–56.
- Mendel G. Versuche über Pflanzen-Hybriden. Verhandlungen des naturforschenden Vereines in Brünn. 1866;4:3–47.
- Collins FS, Varmus H. A new initiative on precision medicine. N Engl J Med. 2015;372(9):793–5.
- Townsend G, Bockmann M, Hughes T, Brook A. Genetic, environmental and epigenetic influences on variation in human tooth number, size and shape. Odontology. 2012;100(1):1–9.

- Brook AH. Multilevel complex interactions between genetic, epigenetic and environmental factors in the aetiology of anomalies of dental development. Arch Oral Biol. 2009;54 Suppl 1:S3–17.

Chapter 2 — DNA, RNA & Chromosomes — The Molecular Basis of Heredity

2.1 Chemical nature of nucleic acids · 2.2 DNA structure · 2.3 Types of DNA · 2.4 RNA structure and types · 2.5 Chromatin and chromosomes

Core texts: [2] Ch. 2–3 · [4] chapters on nucleic acid structure and chromosome organization · [7] chapters on DNA and chromosomes · [10] chapters on nucleotides and nucleic acids · [6] chapters on genes and genomes

Additional

- Watson JD, Crick FHC. Molecular structure of nucleic acids: a structure for deoxyribose nucleic acid. Nature. 1953;171(4356):737–8.
- Franklin RE, Gosling RG. Molecular configuration in sodium thymonucleate. Nature. 1953;171(4356):740–1.
- Kornberg RD. Chromatin structure: a repeating unit of histones and DNA. Science. 1974;184(4139):868–71.
- Luger K, Mäder AW, Richmond RK, Sargent DF, Richmond TJ. Crystal structure of the nucleosome core particle at 2.8 Å resolution. Nature. 1997;389(6648):251–60.
- Taanman JW. The mitochondrial genome: structure, transcription, translation and replication. Biochim Biophys Acta. 1999;1410(2):103–23.
- Cech TR, Steitz JA. The noncoding RNA revolution — trashing old rules to forge new ones. Cell. 2014;157(1):77–94.

Chapter 3 — DNA Replication, Repair & Recombination

3.1 DNA replication — basic concepts · 3.2 Key enzymes in replication · 3.3 Replication in prokaryotes and eukaryotes · 3.4 DNA damage and repair · 3.5 Genetic recombination

Core texts: [3] Ch. 2 · [7] chapters on DNA replication, repair and recombination · [4] chapters on DNA repair and instability · [6] chapters on replication and repair

Additional

- Meselson M, Stahl FW. The replication of DNA in Escherichia coli. Proc Natl Acad Sci U S A. 1958;44(7):671–82.
- Okazaki R, Okazaki T, Sakabe K, Sugimoto K, Sugino A. Mechanism of DNA chain growth. I. Possible discontinuity and unusual secondary structure of newly synthesized chains. Proc Natl Acad Sci U S A. 1968;59(2):598–605.
- Lindahl T. Instability and decay of the primary structure of DNA. Nature. 1993;362(6422):709–15.
- Jackson SP, Bartek J. The DNA-damage response in human biology and disease. Nature. 2009;461(7267):1071–8.
- Hoeijmakers JHJ. DNA damage, aging, and cancer. N Engl J Med. 2009;361(15):1475–85.
- Lynch HT, de la Chapelle A. Hereditary colorectal cancer. N Engl J Med. 2003;348(10):919–32.

Chapter 4 — Gene Expression: Transcription & Translation

4.1 Central dogma · 4.2 Transcription in prokaryotes and eukaryotes · 4.3 RNA processing in eukaryotes · 4.4 Genetic code · 4.5 Translation

Core texts: [7] chapters on how cells read the genome · [8] chapters on transcriptional control and protein synthesis · [4] chapters on transcription, RNA processing and translation · [6] chapters on gene expression · [9] chapters on protein synthesis and the genetic code

Additional

- Crick F. Central dogma of molecular biology. Nature. 1970;227(5258):561–3.
- Nirenberg MW, Matthaei JH. The dependence of cell-free protein synthesis in E. coli upon naturally occurring or synthetic polyribonucleotides. Proc Natl Acad Sci U S A. 1961;47(10):1588–602.
- Berget SM, Moore C, Sharp PA. Spliced segments at the 5′ terminus of adenovirus 2 late mRNA. Proc Natl Acad Sci U S A. 1977;74(8):3171–5.
- Nilsen TW, Graveley BR. Expansion of the eukaryotic proteome by alternative splicing. Nature. 2010;463(7280):457–63.
- Ramakrishnan V. Ribosome structure and the mechanism of translation. Cell. 2002;108(4):557–72.
- Baltimore D. RNA-dependent DNA polymerase in virions of RNA tumour viruses. Nature. 1970;226(5252):1209–11.

Chapter 5 — Regulation of Gene Expression

5.1 Regulation in prokaryotes · 5.2 Regulation in eukaryotes · 5.3 Tissue-specific gene expression

Core texts: [2] Ch. 8 · [7] chapters on the control of gene expression · [4] chapters on epigenetics and regulatory RNA · [26] and [27] chapters on tooth development

Additional

- Jacob F, Monod J. Genetic regulatory mechanisms in the synthesis of proteins. J Mol Biol. 1961;3:318–56.
- Bird A. DNA methylation patterns and epigenetic memory. Genes Dev. 2002;16(1):6–21.
- Jaenisch R, Bird A. Epigenetic regulation of gene expression: how the genome integrates intrinsic and environmental signals. Nat Genet. 2003;33 Suppl:245–54.
- Bartel DP. MicroRNAs: genomics, biogenesis, mechanism, and function. Cell. 2004;116(2):281–97.
- Thesleff I, Sharpe P. Signalling networks regulating dental development. Mech Dev. 1997;67(2):111–23.
- Jernvall J, Thesleff I. Reiterative signaling and patterning during mammalian tooth morphogenesis. Mech Dev. 2000;92(1):19–29.

Chapter 6 — Cell Cycle, Mitosis, Meiosis & Their Genetic Significance

6.1 Cell cycle overview · 6.2 Mitosis · 6.3 Meiosis · 6.4 Clinical relevance

Core texts: [3] Ch. 3 · [2] Ch. 5 · [7] chapters on the cell cycle and cell division · [12] chapters on cytogenetics and cell division

Additional

- Hartwell LH, Weinert TA. Checkpoints: controls that ensure the order of cell cycle events. Science. 1989;246(4930):629–34.
- Nurse P. A long twentieth century of the cell cycle and beyond. Cell. 2000;100(1):71–8.
- Morgan DO. The Cell Cycle: Principles of Control. London: New Science Press; 2007.
- Hassold T, Hunt P. To err (meiotically) is human: the genesis of human aneuploidy. Nat Rev Genet. 2001;2(4):280–91.

- Nagaoka SI, Hassold TJ, Hunt PA. Human aneuploidy: mechanisms and new insights into an age-old problem. Nat Rev Genet. 2012;13(7):493–504.
- Antonarakis SE, Skotko BG, Rafii MS, Strydom A, Pape SE, Bianchi DW, et al. Down syndrome. Nat Rev Dis Primers. 2020;6(1):9.

Chapter 7 — Classical (Mendelian) Genetics & Patterns of Inheritance

7.1 Mendelian principles · 7.2 Modes of inheritance · 7.3 Non-Mendelian inheritance · 7.4 Pedigree analysis

Core texts: [3] Ch. 6 · [2] Ch. 7 · [1] chapters on single-gene inheritance · [12] chapters on patterns of inheritance · [19] on X-inactivation · [5] chapters on Mendelian analysis

Additional

- Bennett RL, French KS, Resta RG, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. J Genet Couns. 2008;17(5):424–33.
- Antonarakis SE, Beckmann JS. Mendelian disorders deserve more attention. Nat Rev Genet. 2006;7(4):277–82.
- Yamamoto F, Clausen H, White T, Marken J, Hakomori S. Molecular genetic basis of the histo-blood group ABO system. Nature. 1990;345(6272):229–33.
- Taylor RW, Turnbull DM. Mitochondrial DNA mutations in human disease. Nat Rev Genet. 2005;6(5):389–402.
- Zlotogora J. Penetrance and expressivity in the molecular age. Genet Med. 2003;5(5):347–52.
- Gorlin RJ, Cohen MM Jr, Hennekam RCM. Syndromes of the Head and Neck. 4th ed. New York: Oxford University Press; 2001.

Chapter 8 — Population Genetics (Introductory)

8.1 Basic concepts · 8.2 Hardy–Weinberg principle · 8.3 Applications to disease

Core texts: [3] Ch. 7–8 · [2] Ch. 10 · [12] chapters on population genetics · [18] the original statement of the equilibrium · [5] chapters on population genetics

Additional

- Weinberg W. Über den Nachweis der Vererbung beim Menschen. Jahreshefte des Vereins für vaterländische Naturkunde in Württemberg. 1908;64:369–82.
- Wigginton JE, Cutler DJ, Abecasis GR. A note on exact tests of Hardy-Weinberg equilibrium. Am J Hum Genet. 2005;76(5):887–93.
- Allison AC. Protection afforded by sickle-cell trait against subtertian malarial infection. Br Med J. 1954;1(4857):290–4.
- Modell B, Darlison M. Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ. 2008;86(6):480–7.
- Bittles AH, Black ML. Consanguinity, human evolution, and complex diseases. Proc Natl Acad Sci U S A. 2010;107 Suppl 1:1779–86.
- 1000 Genomes Project Consortium. A global reference for human genetic variation. Nature. 2015;526(7571):68–74.

Chapter 9 — Human Genetic Disorders — With Oral & Craniofacial Relevance

9.1 Chromosomal abnormalities · 9.2 Single-gene (monogenic) disorders · 9.3 Multifactorial and complex disorders · 9.4 Oral manifestations of systemic genetic disease

Core texts: [2] Ch. 6 and 12 · [3] Ch. 10 and 16 · [13] case-based chapters on genetic disease · [14] on chromosome abnormalities and counselling · [17] as the catalogue of Mendelian phenotypes · [29] chapters on developmental and genetic disorders of the teeth and jaws · [30] chapters on the medically compromised child

Additional

- Smith CEL, Poulter JA, Antanaviciute A, Kirkham J, Brookes SJ, Inglehearn CF, et al. Amelogenesis imperfecta; genes, proteins, and pathways. Front Physiol. 2017;8:435.
- de La Dure-Molla M, Philippe Fournier B, Berdal A. Isolated dentinogenesis imperfecta and dentin dysplasia: revision of the classification. Eur J Hum Genet. 2015;23(4):445–51.
- Shields ED, Bixler D, El-Kafrawy AM. A proposed classification for heritable human dentine defects with a description of a new entity. Arch Oral Biol. 1973;18(4):543–53.
- Marini JC, Forlino A, Bächinger HP, Bishop NJ, Byers PH, De Paepe A, et al. Osteogenesis imperfecta. Nat Rev Dis Primers. 2017;3:17052.
- Elborn JS. Cystic fibrosis. Lancet. 2016;388(10059):2519–31.
- Loos BG, Van Dyke TE. The role of inflammation and genetics in periodontal disease. Periodontol 2000. 2020;83(1):26–39.
- Vieira AR, Modesto A, Marazita ML. Caries: review of human genetics research. Caries Res. 2014;48(5):491–506.

Chapter 10 — Molecular Biology Techniques — Foundation for Clinical & Dental Applications

10.1 Nucleic acid isolation and analysis · 10.2 Polymerase chain reaction · 10.3 DNA sequencing · 10.4 Molecular markers · 10.5 Cloning and recombinant DNA

Core texts: [20] chapters on cloning vectors, restriction enzymes, PCR and sequencing · [22] laboratory protocols · [3] Ch. 5 · [16] on interpreting sequence variants · [39] on genome editing

Additional

- Mullis KB, Faloona FA. Specific synthesis of DNA in vitro via a polymerase-catalyzed chain reaction. Methods Enzymol. 1987;155:335–50.
- Saiki RK, Gelfand DH, Stoffel S, Scharf SJ, Higuchi R, Horn GT, et al. Primer-directed enzymatic amplification of DNA with a thermostable DNA polymerase. Science. 1988;239(4839):487–91.
- Sanger F, Nicklen S, Coulson AR. DNA sequencing with chain-terminating inhibitors. Proc Natl Acad Sci U S A. 1977;74(12):5463–7.
- Southern EM. Detection of specific sequences among DNA fragments separated by gel electrophoresis. J Mol Biol. 1975;98(3):503–17.
- Goodwin S, McPherson JD, McCombie WR. Coming of age: ten years of next-generation sequencing technologies. Nat Rev Genet. 2016;17(6):333–51.
- Jinek M, Chylinski K, Fonfara I, Hauer M, Doudna JA, Charpentier E. A programmable dual-RNA-guided DNA endonuclease in adaptive bacterial immunity. Science. 2012;337(6096):816–21.

Chapter 11 — Genomics, Proteomics & Bioinformatics (Awareness Level)

11.1 Genomics · 11.2 Proteomics · 11.3 Basic bioinformatics · 11.4 Precision and personalised medicine

Core texts: [21] chapters on genome anatomies and genome function · [23] chapters on sequence databases and analysis · [24] and [25] the human genome sequence · [36] and [37] on genomic medicine services · [31] on the oral microbiome · [38] on the human genome and human rights

Additional

- Venter JC, Adams MD, Myers EW, Li PW, Mural RJ, Sutton GG, et al. The sequence of the human genome. Science. 2001;291(5507):1304–51.
- Aebersold R, Mann M. Mass-spectrometric exploration of proteome structure and function. Nature. 2016;537(7620):347–55.
- Sayers EW, Cavanaugh M, Clark K, Ostell J, Pruitt KD, Karsch-Mizrachi I. GenBank. Nucleic Acids Res. 2020;48(D1):D84–6.
- Ashley EA. Towards precision medicine. Nat Rev Genet. 2016;17(9):507–22.
- Dewhirst FE, Chen T, Izard J, Paster BJ, Tanner ACR, Yu WH, et al. The human oral microbiome. J Bacteriol. 2010;192(19):5002–17.
- Wong DT. Salivaomics. J Am Dent Assoc. 2012;143(10 Suppl):19S–24S.

Chapter 12 — Genetics & Molecular Biology in Dentistry

12.1 Genetics of craniofacial development · 12.2 Genetic basis of dental anomalies · 12.3 Genetics of oral cancer · 12.4 Pharmacogenetics in dentistry

Core texts: [28] chapters on craniofacial morphogenesis and the neural crest · [26] chapters on tooth development · [32] on signalling in tooth morphogenesis · [29] chapters on oral cancer and potentially malignant disorders · [3] Ch. 15 · [15] entries on craniofacial syndromes

Additional

- Dixon MJ, Marazita ML, Beaty TH, Murray JC. Cleft lip and palate: understanding genetic and environmental influences. Nat Rev Genet. 2011;12(3):167–78.
- Mossey PA, Little J, Munger RG, Dixon MJ, Shaw WC. Cleft lip and palate. Lancet. 2009;374(9703):1773–85.
- Twigg SRF, Wilkie AOM. A genetic-pathophysiological framework for craniosynostosis. Am J Hum Genet. 2015;97(3):359–77.
- Vastardis H, Karimbux N, Guthua SW, Seidman JG, Seidman CE. A human MSX1 homeodomain missense mutation causes selective tooth agenesis. Nat Genet. 1996;13(4):417–21.
- Stockton DW, Das P, Goldenberg M, D'Souza RN, Patel PI. Mutation of PAX9 is associated with oligodontia. Nat Genet. 2000;24(1):18–9.
- Leemans CR, Snijders PJF, Brakenhoff RH. The molecular landscape of head and neck cancer. Nat Rev Cancer. 2018;18(5):269–82.
- Crews KR, Monte AA, Huddart R, Caudle KE, Kharasch ED, Gaedigk A, et al. Clinical Pharmacogenetics Implementation Consortium guideline for CYP2D6, OPRM1, and COMT genotypes and select opioid therapy. Clin Pharmacol Ther. 2021;110(4):888–96.

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