

Acknowledgement

We would like to thank Dr. Andrew Rosenberg for his outstanding contribution to previous editions of this chapter.

SUGGESTED READINGS

Basic Structure and Biology of Bone

- Cohen MM Jr: The new bone biology: pathologic, molecular, and clinical correlates. *Am J Med Genet A* 140:2646–706, 2006.
- Kogianni G, Noble BS: The biology of osteocytes. *Curr Osteoporos Rep* 5:81–6, 2007.
- Olsen BR, Reginato AM, Wang W: Bone development. *Annu Rev Cell Dev Biol* 16:191–220, 2000.
- Raisz LG: Physiology and pathophysiology of bone remodeling. *Clin Chem* 45:1353–8, 1999.
- Zaidi M: Skeletal remodeling in health and disease. *Nat Med* 13:791–801, 2007.

Skeletal Dysplasias

- Askmyr MK, Fasth A, Richter J: Towards a better understanding and new therapeutics of osteopetrosis. *Br J Haematol* 140:597–609, 2008.
- Kornak U, Kasper D, BÄpsl MR, et al: Loss of the CIC-7 Chloride Channel Leads to Osteopetrosis in Mice and Man. *Cell* 104:205–15, 2001.
- Krakow D, Rimoin DL: The skeletal dysplasias. *Genet Med* 12:327–41, 2010.
- Martin E, Shapiro JR: Osteogenesis imperfecta: epidemiology and pathophysiology. *Curr Osteoporos Rep* 5:91–7, 2007.
- Van Dijk FS, Pals G, Van Rijn RR, et al: Classification of Osteogenesis Imperfecta revisited. *Eur J Med Genet* 53:1–5, 2010.

Osteoporosis

- Anderson GL, Limacher M, Assaf AR, et al: Effects of conjugated equine estrogen in postmenopausal women with hysterectomy: the Women's Health Initiative randomized controlled trial. *JAMA* 291:1701–12, 2004.
- Mosekilde L: Mechanisms of age-related bone loss. *Novartis Found Symp* 235:150–66; discussion 66–71, 2001.
- Russell RG, Watts NB, Ebetino FH, et al: Mechanisms of action of bisphosphonates: similarities and differences and their potential influence on clinical efficacy. *Osteoporos Int* 19:733–59, 2008.
- Styrkarsdottir U, Halldorsson BV, Gretarsdottir S, et al: Multiple genetic loci for bone mineral density and fractures. *N Engl J Med* 358:2355–65, 2008.

Paget Disease

- Roodman GD, Windle JJ: Paget disease of bone. *J Clin Invest* 115:200–8, 2005.
- Singer FR: The Etiology of Paget's Disease of Bone: Viral and Genetic Interactions. *Cell Metabolism* 13:5–6, 2011.
- Whyte MP: Clinical practice. Paget's disease of bone. *N Engl J Med* 355:593–600, 2006.

Metabolic Bone Diseases

- Mazzaferro S, Pasquali M, Pirrò G, et al: The bone and the kidney. *Archives of Biochemistry and Biophysics* 503:95–102, 2010.
- Schwarz C, Sulzbacher I, Oberbauer R: Diagnosis of renal osteodystrophy. *Eur J Clin Invest* 36(Suppl 2):13–22, 2006.

Osteogenic Tumors

- Klein MJ, Siegal GP: Osteosarcoma: anatomic and histologic variants. *Am J Clin Pathol* 125:555–81, 2006.
- Lee EH, Shafi M, Hui JH: Osteoid osteoma: a current review. *J Pediatr Orthop* 26:695–700, 2006.
- Wagner ER, Luther G, Zhu G, et al: Defective osteogenic differentiation in the development of osteosarcoma. *Sarcoma* 2011:325238, 2011.

Chondrogenic Tumors

- Bovee JV, Hogendoorn PC, Wunder JS, et al: Cartilage tumours and bone development: molecular pathology and possible therapeutic targets. *Nat Rev Cancer* 10:481–8, 2010.
- Pansuriya TC, van Eijk R, d'Adamo P, et al: Somatic mosaic IDH1 and IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. *Nat Genet* 43:1256–61, 2011.
- Wuyts W, Van Hul W: Molecular basis of multiple exostoses: mutations in the EXT1 and EXT2 genes. *Human Mutation* 15:220–7, 2000.

Ewing Sarcoma

- Erkank HV, Uversky VN, Toretsky JA: Oncogenic partnerships: EWS-FLI1 protein interactions initiate key pathways of Ewing's sarcoma. *Clin Cancer Res* 16:4077–83, 2010.
- Liang H, Mao X, Olejniczak ET, et al: Solution structure of the ets domain of Fli-1 when bound to DNA. *Nat Struct Biol* 1:871–5, 1994.
- Pinto A, Dickman P, Parham D: Pathobiologic markers of the ewing sarcoma family of tumors: state of the art and prediction of behaviour. *Sarcoma* 2011:856190, 2011.

Giant Cell Tumor of Bone

- Robinson D, Einhorn TA: Giant cell tumor of bone: a unique paradigm of stromal-hematopoietic cellular interactions. *J Cell Biochem* 55:300–3, 1994.
- Salerno M, Avnet S, Alberghini M, et al: Histogenetic characterization of giant cell tumor of bone. *Clin Orthop Relat Res* 466:2081–91, 2008.

Aneurysmal Bone Cyst

- Oliveira AM, Chou MM: The TRE17/USP6 oncogene: a riddle wrapped in a mystery inside an enigma. *Front Biosci (Schol Ed)* 4:321–34, 2012.

Fibrous Dysplasia

- Riminucci M, Robey PG, Saggio I, et al: Skeletal progenitors and the GNAS gene: fibrous dysplasia of bone read through stem cells. *J Mol Endocrinol* 45:355–64, 2010.

Osteoarthritis

- Goldring MB, Goldring SR: Articular cartilage and subchondral bone in the pathogenesis of osteoarthritis. *Ann N Y Acad Sci* 1192:230–7, 2010.
- Valdes AM, Spector TD: Genetic epidemiology of hip and knee osteoarthritis. *Nat Rev Rheumatol* 7:23–32, 2011.
- Yelin E, Callahan LF: The economic cost and social and psychological impact of musculoskeletal conditions. National Arthritis Data Work Groups. *Arthritis Rheum* 38:1351–62, 1995.

Rheumatoid Arthritis and Related Conditions

- Fox DA, Gizinski A, Morgan R, et al: Cell-cell interactions in rheumatoid arthritis synovium. *Rheum Dis Clin North Am* 36:311–23, 2010.
- Imboden JB: The immunopathogenesis of rheumatoid arthritis. *Annu Rev Pathol* 4:417–34, 2009.
- Petty RE, Southwood TR, Manners P, et al: International League of Associations for Rheumatology classification of juvenile idiopathic arthritis: second revision, Edmonton, 2001. *J Rheumatol* 31:390–2, 2004.
- Scott DL, Wolfe F, Huizinga TW: Rheumatoid arthritis. *Lancet* 376:1094–108, 2011.
- Thomas GP, Brown MA: Genetics and genomics of ankylosing spondylitis. *Immunol Rev* 233:162–80, 2010.

Infectious Arthritis

- Iliopoulou BP, Huber BT: Infectious arthritis and immune dysregulation: lessons from Lyme disease. *Curr Opin Rheumatol* 22:451–5, 2010.
- Rosenberg AE, Nielsen GP, Reith J: Surgical pathology of joint prostheses. *Semin Diagn Pathol* 28:65–72, 2011.
- Steere AC, Drouin EE, Glickstein LJ: Relationship between immunity to *Borrelia burgdorferi* outer-surface protein A (OspA) and Lyme arthritis. *Clin Infect Dis* 52(Suppl 3):S259–65, 2011.