Χαρακτηριστικά:

- Identifies and analyzes the genetic basis of bone disorders in humans and demonstrates the utility of mouse models in furthering the knowledge of mechanisms and evaluation of treatments
- Demonstrates how the interactions between bone and joint biology, physiology, and genetics have greatly enhanced the understanding of normal bone function
- Summarizes the clinical, genetic, molecular, animal model, molecular pathology, diagnostic, counseling, and treatment aspects of each disorder

Περιεχόμενα:

PART 1: GENERAL BACKGROUND TO GENETICS AND 'OMICS

1. Introduction to Genetics of Skeletal and Mineral Metabolic Diseases
2. Genome-Sequencing and Big data analysis
3. Genome-Wide Association Studies
4. Epigenetics
5. Multi-omics approaches in bone research
6. Functional Genomics
7. Organ-on-Chips
8. Mouse Models: Approaches to Generate In Vivo Models for Hereditary Disorders of Mineral and Skeletal
9. Emerging therapeutic approaches (including gene therapy) for Skeletal Diseases
10. Pharmacogenetics and Pharmacogenomics of Osteoporosis: Personalized Medicine Outlook
11. Drug discovery in the era of cryo-electron microscopy (cryoEM)

PART 2: GENERAL BACKGROUND TO BONE BIOLOGY

12. Biology of Bone and Cartilage
13. Overview of Bone Structure and Strength
14. Overview of Joint and Cartilage Biology
15. Osteocyte Biology
16. Skeletal Stem Cells/Bone Marrow Stromal Cells
17. Osteoimmunology
18. Senescence in Bone Disease
19. Chronobiology and clock genes, relevance to musculoskeletal health
20. Mechano-sensing in skeletal biology
21. Hypoxia signalling bone and cartilage
22. Integrating Endocrine and Paracrine Influences on Bone; Lessons from Parathyroid Hormone and Parathyroid
23. Genetics of Bone Fat and Energy Regulation
24. The Cross Talk Between the Central Nervous System, Bone, and Energy Metabolism
25. Fetal Control of Calcium and Phosphate Homeostasis
26. Control of Mineral and Skeletal Homeostasis During Pregnancy and Lactation

PART 3: DISORDERS OF BONE AND JOINT

27. Gene discoveries and novel therapies in monogenic low and high bone mass disorders.
28. Osteoporosis
29. Osteogenesis Imperfecta
30. Osteoarthritis: Genetic Studies of Monogenic and Complex Forms
31. Mendelian Disorders of RANKL/OPG/RANK/NF-?B Signalling
32. Skeletal Dysplasias
33. Hypophosphatasia and How Alkaline Phosphatase Promotes Mineralization
34. Sclerosing Bone Dysplasias
35. Melorheostosis
36. Fibrodysplasia (Myositis) Ossificans Progressiva
37. Craniosynostosis
38. Disorders and Mechanisms of Ectopic Calcification
39. Thyroid Hormone in Bone and Joint Disorders

PART 4: PARATHYROID AND RELATED DISORDER

40. Hyperparathyroidism
41. Hypoparathyroidism
42. Gsa, Pseudohypoparathyroidism, Fibrous Dysplasia, and McCune–Albright Syndrome
43. Genetic Disorders Caused by Mutations in the PTH/PTHrP Receptor, its Ligands, and Downstream Effector Molecules
44. Genetically Determined Disorders of Extracellular Calcium (Cao2+) Sensing and Cao2+ Homeostasis
45. Multiple Endocrine Neoplasia Syndromes

PART 5: VITAMIN D AND RENAL DISORDERS

46. Genetic Disorders of Vitamin D Synthesis and Action
47. X-linked hypophosphataemia
48. Heritable Renal Phosphate Wasting Disorders
49. Genetics of Hypercalciuria and Kidney Stones