

1. Record Nr.	UNINA9910918594803321
Autore	Rossi Antonio
Titolo	The Extracellular Matrix in Genetic Skeletal Disorders // edited by Antonio Rossi, Frank Zaucke
Pubbl/distr/stampa	Cham : , : Springer Nature Switzerland : , : Imprint : Springer, , 2024
ISBN	9783031708350 3031708350
Edizione	[1st ed. 2024.]
Descrizione fisica	1 online resource (414 pages)
Collana	Biology of Extracellular Matrix, , 2191-1959 ; ; 16
Altri autori (Persone)	ZauckeFrank
Disciplina	571.6
Soggetti	Cytology Musculoskeletal system Clinical biochemistry Diseases Cell Biology Medical Biochemistry Mechanisms of Disease
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di contenuto	Part I: ECM proteins and skeletal disorders -- Chapter 1: Extracellular matrix and skeletal dysplasias -- Chapter 2: Osteogenesis imperfecta and related diseases – collagen I alterations in skeletal and extraskeletal tissues -- Chapter 3: Bone involvement in the Ehlers-Danlos syndromes -- Chapter 4: Collagen II mutations in inherited cartilage diseases: our current understanding of genotype-phenotype correlations -- Chapter 5: Skeletal disorders linked to GAG synthesis -- Chapter 6: GAG sulfation and skeletal disorders -- Chapter 7: The fibrillinopathies – fibrillin-dependent skeletal dysplasias affecting long bone growth -- Part II: ECM signaling and skeletal disorders -- Chapter 8: FGF signaling: a key pathway during skeletal development -- Chapter 9: Impaired Wnt signaling as a cause of skeletal disorders -- Chapter 10: The skeletal ciliopathies -- Part III: Intracellular trafficking of ECM proteins and related skeletal disorders -- Chapter 11: Microtubule-associated motor proteins in skeletal development and health -- Chapter 12: COG4 variant and Saul-Wilson Syndrome -- 13:

Sommario/riassunto

This book explores how defects in extracellular matrix (ECM) proteins, their post-translational modifications, and intracellular trafficking impact cartilage and bone integrity. It underscores the ECM's role in providing structural support, establishing morphogenetic gradients, and interacting with cell surface receptors in musculoskeletal tissues. The book delves into the structure and biology of the ECM in the skeleton, discussing skeletal disorders caused by mutations in genes associated with ECM proteins, synthesis, turnover, and signal transduction. Authored by experts who have made significant discoveries in the molecular mechanisms of skeletal disorders and are developing therapeutic strategies, this book is an invaluable research for both scientists and clinicians seeking a comprehensive understanding of this growing and exciting field. The series Biology of Extracellular Matrix is published in collaboration with the American Society for Matrix Biology and the International Society for Matrix Biology.

---