1. Record Nr. UNINA9910143216403321

Titolo	Connective tissue and it heritable disorders [[electronic resource]] : molecular, genetic, and medical aspects / / edited by Peter M. Royce and Beat Steinmann
Pubbl/distr/stampa	New York, : Wiley-Liss, c2002
ISBN	1-280-36667-2 9786610366675 0-470-35006-7 0-471-46117-2 0-471-22192-9
Edizione	[2nd ed.]
Descrizione fisica	1 online resource (1221 p.)
Altri autori (Persone)	RoycePeter M SteinmannBeat U
Disciplina	616.77042
Soggetti	Connective tissues - Diseases - Genetic aspects Connective tissues - Diseases - Molecular aspects Electronic books.
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	CONNECTIVE TISSUE AND ITS HERITABLE DISORDERS SECOND EDITION; Contents; Preface; Acknowledgments; Preface to the First Edition; Contributors; Introduction Research on Collagen in the Author's Laboratory, 1952-1982; Introduction Heritable Disorders of Connective Tissue: A Personal Account of the Origins, Evolution, Validation, and Expansion of a Concept; BIOLOGY OF EXTRACELLULAR MATRIX; Chapter 1 Morphology and Chemical Composition of Connective Tissue; Part I Structure of the Skin and Tendon; Part II Cartilage; Part III Bone; Part IV The Cardiovascular System; Part V The Eye Chapter 2 CollagenPart I The Collagen Family: Structure, Assembly, and Organization in the Extracellular Matrix; Part II Gene Structure; Chapter 3 Elastin and the Microfibrillar Apparatus; Chapter 4 Glycosylated Matrix Proteins; Chapter 5 Adhesive Glycoproteins; Chapter 6 Keratins; Chapter 7 Extracellular Matrix Degradation; HERITABLE DISORDERS OF CONNECTIVE TISSUE; Chapter 8 Osteogenesis Imperfecta; Chapter 9

The Ehlers-Danlos Syndrome; Chapter 10 Cutis Laxa and Premature Aging Syndromes; Chapter 11 Pseudoxanthoma Elasticum; Chapter 12 The Marfan Syndrome and Other Microfibrillar Disorders
Chapter 13 The HomocystinuriasChapter 14 Menkes Disease and the Occipital Horn Syndrome; Chapter 15 Epidermolysis Bullosa; Chapter 16 Prolidase Deficiency; Chapter 17 a(1)-Antitrypsin Deficiency; Chapter 18 Heritable Forms of Rickets and Osteomalacia; Chapter 19 Osteopetrosis; Chapter 20 Alkaptonuria; Chapter 21 Fibrodysplasia Ossificans Progressiva; Chapter 22 Disorders of Lysosomal Enzymes; Part I General Considerations; Part II Clinical Phenotypes; Chapter 23 Skeletal Dysplasias; Part I Chondrodysplasias: General Concepts and Diagnostic and Management Considerations
Part II Chondrodysplasias: Disorders of Cartilage Matrix ProteinsPart III Skeletal Dysplasias Related to Defects in Sulfate Metabolism; Part IV Craniosynostosis Syndromes and Skeletal Dysplasias Caused by Mutations in Fibroblast Growth Factor Receptor Genes; Part V Defects in Skeletal Morphogenesis; Chapter 24 Disorders of Keratinization; Chapter 25 Alport Syndrome; Chapter 26 Miscellaneous Disorders; Part I Corneal Dystrophies Due to Mutations in the Kerato-Epithelin Gene (big-h3); Part II Progressive Pseudorheumatoid Dysplasia
Part III The Camptodactyly-Arthropathy-Coxa Vara-Pericarditis SyndromePart IV Bruck Syndrome; Part V Osteoporosis-Pseudoglioma Syndrome; Part VI Myopathies Due to Defects in Collagen VI; Part VII Knobloch Syndrome; Appendix I International Nomenclature of Constitutional Disorders of Bone: Osteochondrodysplasias; Appendix II Extracellular Collagen Metabolites in Body Fluids; Index

Sommario/riassunto

The Second Edition of Connective Tissue and Its Heritable Disorders: Molecular, Genetic, and Medical Aspects is the definitive reference text in its field, with over 40% more pages on the nature, diagnosis, and treatment of disease than its predecessor. Collecting new research on disorders detailed in the first edition as well as on those previously excluded, editors Peter Royce and Beat Steinmann provide the most up-to-date clinical and scientific information for medical specialists treating affected individuals. Features of this revised and updated volume include detailed reviews of the cl