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Aging Syndromes; Chapter 11 Pseudoxanthoma Elasticum; Chapter 12 The Marfan Syndrome and Other Microfibrillar Disorders Chapter 13 The Homocystinurias Chapter 14 Menkes Disease and the Occipital Horn Syndrome; Chapter 15 Epidermolysis Bullosa; Chapter 16 Prolidase Deficiency; Chapter 17 α (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 Proteins Part 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 Syndrome Part 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
