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Autore	Pyeritz Reed E
Titolo	Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics : Clinical Principles and Applications
Pubbl/distr/stampa	San Diego : , : Elsevier Science & Technology, , 2018 ©2019
ISBN	0-12-812684-1
Edizione	[7th ed.]
Descrizione fisica	1 online resource : illustrations (some color)
Altri autori (Persone)	KorfBruce R GrodyWayne W
Disciplina	616.042
Soggetti	Medical genetics Genomics Genetics, Medical Genetic Diseases, Inborn
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Companion volume to Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics. Foundations / Edited by Reed E. Pyeritz, Bruce R. Korf, Wayne W. Grody. Seventh edition. [2019].
Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	A Clinical approach to the dysmorphic child -- Clinical teratology -- Neurodevelopmental disabilities: global developmental delay, intellectual disability, and autism -- Abnormal body size and proportion -- Cytogenetic analysis -- Diagnostic molecular genetics -- Therapies for lysosomal storage diseases -- Transplantation genetics -- Genetic evaluation for common, chronic disorders of adulthood 265 -- Carrier screening and heterozygote testing -- Circadian rhythms and disease -- The Genomic health record: current status and vision for the future -- Ethical and social issues in clinical genetics -- Genetics and genomics in public health -- Implementation of genomic medicine: an international perspective.
Sommario/riassunto	The Seventh Edition of this classic reference includes the latest information on seminal topics such as prenatal diagnosis, genome and exome sequencing, public health genetics, genetic counseling, and management and treatment strategies to complete its coverage of this growing field for medical students, residents, physicians, and

researchers involved in the care of patients with genetic conditions. This comprehensive yet practical resource emphasizes theory and research fundamentals related to applications of medical genetics across the full spectrum of inherited disorders and applications to medicine more broadly. Clinical Principles and Applications thoroughly addresses general methods and approaches to genetic counseling, genetic diagnostics, treatment pathways, and drug discovery. Additionally, new and updated chapters explore the clinical implementation of genomic technologies, analytics, and therapeutics, with special attention paid to developing technologies, common challenges, patient care, and ethical and legal aspects. Examines key topics and developing areas within clinical genomics, including genetic evaluation of patients, clinical trials and drug discovery, genetic health records, cytogenetic analysis, diagnostic molecular genetics, small molecule genetic therapeutics, gene product replacement, clinical teratology, transplantation genetics, and ethical and legal aspects of genomic medicine. Includes color images supporting identification, concept illustration, and method processing. Features contributions by leading international researchers and practitioners of medical genetics.

-- Publisher
