Record Nr. UNINA9910154626103321 Autore Turnpenny Peter D. Titolo Emery's elements of medical genetics / / Peter D. Turnpenny, Sian Ellard Pubbl/distr/stampa [Saint Louis, Missouri]:,: Elsevier,, 2017 ©2017 **ISBN** 0-7020-6689-3 Edizione [Fifteenth Edition] Descrizione fisica 1 online resource (1,017 pages): illustrations (some color), photographs, tables Disciplina 616.042 Genetics, Medical Soggetti Genetic Diseases, Inborn Medical genetics Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Note generali Includes index. Nota di bibliografia Includes bibliographical references and index. Nota di contenuto The history and impact of genetics in medicine -- Section A: The scientific basis of human genetics -- The cellular and molecular basis of inheritance -- Chromosomes and cell division -- Finding the cause of monogenic disorders by identifying disease genes -- Laboratory techniques for diagnosis of monogenic disorders -- Patterns of inheritance -- Population and mathematical genetics -- Risk calculation -- Developmental genetics -- Section B: Genetics in medicine and genomic medicine -- Common disease, polygenic and multifactorial genetics -- Screening for genetic disease -- Hemoglobin and the hemoglobinopathies -- Immunogenetics -- The genetics of cancer and cancer genetics -- Pharmacogenetics, personalized medicine and the treatment of genetic disease -- Section C: Clinical genetics, counseling, and ethics -- Congenital, abnormalities, dysmorphic syndromes, and learning disability -- Chromosome disorders -- Inborn errors of metabolism -- Mainstream monogenetic disorders -- Prenatal testing and reproductive genetics -- Genetic

Sommario/riassunto

"Everything a student needs to know about medical genetics is here in the 15th ediction of this award-winning textbook. Thoroughly updated and revised throughout to map a fast-moving area, the 15th edition

counseling -- Ethical and legal issues in medical genetics.

continues Emery's enviable reputation for successfully balancing up-todateness in a rapidly developing field with a strong basis in practical clinical genetics for medical students." --Back Cover.