Record Nr. UNISALENTO991003654569707536 Autore Dione, Djibril Titolo Histoire de l'Afrique [par] Djibril Dione, Sekene Mody Cissoko Paris, Presence africaine[1973?] Pubbl/distr/stampa Descrizione fisica 191 p. ill.; 25 cm Altri autori (Persone) Cissoko, Sekene Modyauthor 960 Disciplina Africa Storia Soggetti Lingua di pubblicazione Francese **Formato** Materiale a stampa Livello bibliografico Monografia Record Nr. UNINA9910583376903321 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

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Nota di contenuto

Includes bibliographical references.

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