1. Record Nr. UNINA9910674359403321

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Titolo	Human chromosome atlas : introduction to diagnostics of structural aberrations / / Claudia Behrend [and four others]
Pubbl/distr/stampa	Cham, Switzerland : , : Springer, , 2023
ISBN	3-031-10588-5
Edizione	[Second edition.]
Descrizione fisica	1 online resource (385 pages)
Disciplina	611.01816
Soggetti	Human chromosomes
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	Introduction: Social Attitude towards Disabled People in Different Areas and Cultures -- Role of Ethics Committees -- International Quality Control in the Field of Diagnostics -- Structural Chromosom Abarrations -- Mutations in Non-coding DNA Regions. Single Case Presentations of Intrachromosomal Rearrangements: Inversions -- Deletions -- Duplications -- Ring Chromosomes -- Isochromosomes. Single Case Presentations of Interchromosomal Rearrangements: Translocations -- Insertions -- Complex Chromosome Rearrangements (CCR) -- Marker Chromosomes -- Cases of Intra- and Interchromosomal Mutation in Polymorphic Regions. Guidelines to Clinical Questions and Practical Approaches: Genetic Counselling Procedures -- Life of Persons with Chromosomal Disorders -- Support Groups for Chromosomal Disorders -- Diagnosis and Therapy -- Outlook.
Sommario/riassunto	Now in its second edition, this atlas serves as an easy-to-use diagnostic guide for the analysis of the human karyotype. Split in four parts, it starts with a comprehensive introduction covering the molecular cytogenetic basics, the role of ethic committees and international quality control in the field of diagnostics. The main parts II and III, demonstrate the spectrum of the different types of chromosome abnormalities by a combination of karyogram and ideogram, it compares the expressiveness of different banding techniques, and it gives the karyotype formula and describes morphological peculiarities of each presented case. The final part,

provides a detailed description of variants of non-coding DNA and focuses on potential problems in detecting aberrations and mentions necessary additional investigations and peculiarities, which have to be taken into account when counseling carriers of a chromosome aberration or their relatives. Given its comprehensive scope and practical approach, this atlas is an indispensable resource for researchers, clinicians and practitioners working in the field of cytogenetics and clinical genetics.