Record Nr.	UNINA9910132169203321
Autore	Zneimer Susan Mahler
Titolo	Cytogenetic abnormalities : chromosomal, FISH, and microarray-based clinical reporting / / Susan Mahler Zneimer
Pubbl/distr/stampa	Chichester, England : , : Wiley Blackwell, , 2014 ©2014
ISBN	1-118-41256-7 1-118-41260-5 1-118-41257-5
Descrizione fisica	1 online resource (665 p.)
Disciplina	572.87
Soggetti	Chromosomes Chromatin
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Cytogenetic Abnormalities; Copyright; Contents; Preface; Acknowledgments; Introduction; Part 1 Constitutional Analyses; Section 1 Chromosome Analysis; Chapter 1 Components of a standard cytogenetics report, normal results and culture failures; 1.1 Components of a standard cytogenetics report; 1.2 Prenatal normal results; 1.3 Neonatal normal results; 1.4 Normal variants in the population; 1.5 Disclaimers and recommendations; 1.6 Culture failures; 1.7 Contamination; Bibliography; Chapter 2 Mosaicism; 2.1 Normal results with 30-50 cells examined; 2.2 Normal and abnormal cell lines 2.3 Two or more abnormal cell lines Bibliography; Chapter 3 Autosomal trisomies - prenatal and livebirths; 3.1 Introduction; 3.2 Trisomy 21 - Down syndrome; 3.3 Mosaic trisomy 21 - mosaic Down syndrome; 3.4 Trisomy 13 - Patau syndrome; 3.5 Trisomy 18 - Edwards syndrome; 3.6 Trisomy 8 - mosaic; 3.7 Trisomy 9 - mosaic; 3.8 Trisomy 20 - mosaic, prenatal; 3.9 Trisomy 22 - mosaic, prenatal; Bibliography; Chapter 4 Translocations; 4.1 Reciprocal (balanced) translocations; 4.2 Robertsonian translocations; Bibliography; Chapter 5 Inversions and recombinant chromosomes

1.

5.1 Risks of spontaneous abortions and live born abnormal offspring

	 5.2 Pericentric inversions and their recombinants; 5.3 Paracentric inversions and their recombinants; Bibliography; Chapter 6 Visible deletions, duplications and insertions; 6.1 Definitions; 6.2 Visible duplications; 6.3 Balanced Insertions; Bibliography; Chapter 7 Unidentifiable marker chromosomes, derivative chromosomes, chromosomes with additional material and rings; 7.1 Marker chromosomes; 7.2 Derivative chromosomes; 7.3 Chromosomes with additional material; 7.4 Ring chromosomes; 7.5 Homogenously staining regions Bibliography Chapter 8 Isochromosomes, dicentric chromosomes and pseudodicentric chromosomes; 8.1 Isochromosomes; Bibliography; Chapter 9 Composite karyotypes and other complex rearrangements; 9.1 Composite karyotypes; 9.2 Complex rearrangements; Bibliography; Chapter 10 Sex chromosome abnormalities; 10.1 X chromosome aneuploidies - female phenotypes; 10.2 X and Y chromosome aneuploidies - female phenotypes; 10.3 X chromosome structural abnormalities; 10.4 Y chromosome tructural abnormalities; 10.5 46,XX males and 46,XY females 10.6 X chromosome translocations Bibliography; Chapter 11 undefined; 11.1 Aneuploid rate; 11.2 Confined placental mosaicism; 11.3 Hydatidiform moles; 11.4 Monosomy X in a fetus; 11.5 Trisomies in a fetus; 11.6 Double trisomy; 11.7 Triploidy; 11.8 Tetraploidy; Bibliography; Chapter 12 Uniparental disomy of chromosome 15; 12.3 Uniparental disomy of chromosome 14 p15; Bibliography; Section 2 Fluorescence In Situ Hybridization (FISH) Analysis; Chapter 13 Metaphase analysis; 13.1 Introduction; 13.2 Reporting normal results 13.3 Common disclaimers
Sommario/riassunto	"This guide discusses chromosomal abnormalities and how best to report and communicate lab findings in research and clinical settings. Providing a standard approach to writing cytogenetic laboratory reports, the guide further covers useful guidance on implementing International System for Human Cytogenetic Nomenclature in reports. Part one of the guide explores chromosomal, FISH, and microarray analysis in constitutional cytogenetic analyses, while part two looks at acquired abnormalities in cancers. Both sections provide illustrative examples of chromosomal abnormalities and how to communicate these findings in standardized laboratory reports"Provided by publisher.