1. Record Nr. UNINA9910781939103321 Autore Gardner R. J. M. Titolo Chromosome abnormalities and genetic counseling [[electronic resource] /] / R.J. McKinlay Gardner, Grant R. Sutherland, Lisa G. Shaffer Oxford,: Oxford University Press, c2012 Pubbl/distr/stampa **ISBN** 0-19-997517-5 1-283-42706-0 9786613427069 0-19-974915-9 Edizione [4th ed.] Descrizione fisica 1 online resource (649 p.) Oxford monographs on medical genetics;; no. 61 Collana Altri autori (Persone) SutherlandGrant R ShafferLisa G Disciplina 616/.042 Soggetti Genetic counseling Human chromosome abnormalities - Patients - Counseling of Chromosome abnormalities Lingua di pubblicazione Inglese **Formato** Materiale a stampa Monografia Livello bibliografico Description based upon print version of record. Note generali Nota di bibliografia Includes bibliographical references and index. Nota di contenuto Cover; Contents; PART ONE: BASIC CONCEPTS; 1. Elements of Medical Cytogenetics; 2. Chromosome Analysis; 3. The Origins and Consequences of Chromosome Pathology: 4. Deriving and Using a Risk Figure; PART TWO: PARENT WITH A CHROMOSOMAL ABNORMALITY; 5. Autosomal Reciprocal Translocations; 6. Sex Chromosome Translocations: 7. Robertsonian Translocations: 8. Centromere Fissions, Complementary Isochromosomes, Telomeric Fusions, Balancing Supernumerary Chromosomes, and Jumping Translocations; 9. Inversions: 10. Insertions: 11. Autosomal Ring Chromosomes: 12. Complex Rearrangements 13. Parental Sex Chromosome Aneuploidy14. Parental Autosomal Aneuploidy: 15. The Fragile X Syndromes; PART THREE: VARIANTS; 16. Variant Chromosomes and Abnormalities of No Phenotypic Consequence; 17. Copy Number Changes; PART FOUR: NORMAL PARENTS WITH A CHROMOSOMALLY ABNORMAL CHILD; 18. Down

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## Sommario/riassunto

Chromosome abnormalities have been known for over 50 years, though the methods of analysis have become increasing more sophisticated and precise. Surprisingly, the questions that parents and families raise in genetic counseling have changed little over that period. Questions like, ""Why did an abnormality happen? Why did it cause the problems we see in our child? Would it happen again in a future child? How could we avoid it happening again?"" are common concerns for families. This new edition of Chromosome Abnormalities and Genetic Counseling deals with these universal questions, and in the c