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Nota di contenuto	Part I Introduction -- 1. Historical Perspective of Human Ring Chromosomes -- 2. Diagnostic Methods for Ring Chromosomes -- 3. Genetic Databases and Online Ring Chromosome Registry -- 4. Advocate Activities and Patient-centered Approaches -- Part II Constitutional Ring Chromosomes -- 5. Ring Chromosome 1 -- 6. Ring Chromosome 2 -- 7. Ring Chromosome 3 -- 8. Ring Chromosome 4 -- 9. Ring Chromosome 5 -- 10. Ring Chromosome 6 -- 11. Ring Chromosome 7 -- 12. Ring Chromosome 8 -- 13. Ring Chromosome 9 -- 14. Ring Chromosome 10 -- 15. Ring Chromosome 11 -- 16. Ring Chromosome 12 -- 17. Ring Chromosome 13 -- 18. Ring Chromosome 14 -- 19. Ring Chromosome 15 -- 20 -- Ring Chromosome 16 -- 21. Ring Chromosome 17 -- 22. Ring Chromosome 18 -- 23. Ring Chromosome 19 -- 24. Ring Chromosome 20 -- 25. Ring Chromosome 21 -- 26. Ring Chromosome 22 -- 27. Ring Chromosome X -- 28. Ring Chromosome Y -- 29. Supernumerary Small Ring Chromosomes -- 30. Ring Chromosomes from Patients' Perspective -- Part III Somatic Ring

Chromosomes -- 31. Acquired Ring Chromosomes in Tumors of Hematopoietic and Lymphoid Tissues -- 32. Acquired Ring Chromosomes in Solid Tumors -- Part IV Ring Chromosome Research -- 33. Molecular Mechanisms of Ring Chromosome Formation and Instability -- 34. iPSC Models of Ring Chromosomes, Genome Editing, and Chromosome Therapy -- 35. Genetic Mosaic Analysis in Model Organisms.

Sommario/riassunto

This book presents chromosome-wise clinical cases following an evidence-based protocol, in addition to providing the scientific background on the mechanisms of human ring chromosome (RC) formation. Presence of RCs in a genome can lead to several rare genetic diseases. This book, edited by the leading experts Prof. Peining Li and Prof. Thomas Liehr, is the first comprehensive book on this topic. Over the past 60 years, banding cytogenetics, fluorescence in situ hybridization, chromosome microarray analysis, and whole genome sequencing have been used to diagnose cases with a RC. Ring syndrome of sever growth retardation and variable intellectual disability has been considered a common clinical feature for all RCs. Clinical heterogeneity of chromosome-specific deletion and duplication syndromes, gene-related organ and tissue defects, cancer predisposition to different types of tumors, and reproduction failure has been reported in the literature. However, the cases of RCs reported in the literature account for less than 1% of its real occurrence. Current diagnostic practice lacks laboratory standards for analyzing cellular behavior and genomic imbalances of RCs to evaluate its compound effects on patients. The under-representation of clinical cases and the lack of comprehensive diagnostic analysis make challenging to establish accurate clinico-cytogenomic correlations. Given recent advances in genomic technology and organized efforts from peer experts, standardized cytogenomic diagnosis and evidence-based clinical management could be envisioned for all patients with RCs. Furthermore, supernumerary small ring chromosomes and the patient’s perspective are addressed—the latter by including family stories of RC-carrier relatives. Acquired RCs in various cancers are also discussed, as well as the potential role of RCs in research applications like iPSC cellular modeling and genomic editing. This book is a valuable reference for clinical geneticists, personnel in cytogenetics and molecular genetics laboratories, genetic counselors, and researchers in related fields.