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Nota di contenuto	1. Epidemiology of Neurofibromatosis Type 1 -- 2. Genetics and Pathway in Neurofibromatosis Type 1 -- 3. Molecular Diagnosis for NF1 -- 4. Diagnosis in NF1, Old and New -- 5. Clinical Features of NF1 in the Skin -- 6. Ocular Manifestations in Neurofibromatosis Type 1 -- 7. Skeletal Manifestations in NF1 -- 8. NF1 in Other Organs -- 9. Genomics of Peripheral Nerve Sheath Tumors Associated to Neurofibromatosis Type 1 -- 10. Mechanotransduction and Nf1 Loss--Partner in Crime: New Hints for Neurofibroma Genesis -- 11. Diagnosis

and Management of Benign Nerve Sheath Tumors in NF1: Evolution from Plexiform to Atypical Neurofibroma and Novel Treatment Approaches -- 12. Diagnosis and Management of Malignant Tumors in NF1: Evolution from Atypical Neurofibromas to Malignant Peripheral Nerve Sheath Tumors and Treatment Options -- 13. Neurological Complications in NF -- 14. Learning Disabilities and Behavior in Neurofibromatosis Type 1 Patients -- 15. Mosaic NF1 -- 16. Legius Syndrome, Other Café -au-lait Diseases and Differential Diagnosis of NF1 -- 17. Cancer Risk and Spectrum in Individuals with RASopathies -- 18. Therapeutical Approaches for NF1 -- 19. Medical Follow up in Neurofibromatosis Type 1 -- 20. Brief Notes on Pregnancy, Prenatal Diagnosis and Preimplantation Procedures in NF1 -- 21. Proposal of New Diagnostic Criteria.

Sommario/riassunto

This volume offers an update of the clinical signs, diagnostic criteria (including molecular diagnosis) and targeted therapies for a particular type of genodermatosis, providing a handy and unique tool for early diagnosis. In recent years, our understanding of genodermatosis and neurocutaneous syndromes has increased, but although Type 1 Neurofibromatosis (NF1) is the most common neuroectodermal disorder and involves a large number of patients and medical disciplines, this syndrome remains underestimated, often misdiagnosed thus leading to inaccurate treament. The literature on the molecular and pathogenetic aspects is ample, but current clinical approaches, classification, diagnostic criteria and treatment protocols are outdated, creating difficulties in early diagnosis and treatment. As such, a chapter is devoted renewing current diagnostic criteria; it includes clinical and molecular data, to offer a sound, updated discussion basis for a consensus conference. NF1 is a “time-dependent” disorder, meaning that the onset of clinical signs are closely linked to patient age and the book discusses this particularly neglected aspect extensively, as well as the latest molecular diagnosis techniques, which are highly sensitive have not been included in the diagnostic criteria. It also explains the role of the RAS-MAPK pathway and genotype-phenotype correlations. In addition it explores new concepts concerning the pathogenesis of neurofibromas and other hamarthomas and their relevance for a modern therapeutical approach with targeted molecular drugs, as well as newly discovered aspects of NF1 in all internal organs, together with their diagnostic counterparts. A chapter on mosaic neurofibromatosis is also included. There is a particular focus on differential diagnosis (i.e. other diseases with café-au-lait macules), and the recently described Legius syndrome will be presented directly by Prof Eric Legius. All chapters are easy-to-understand, up-to-date, comprehensive and concise tools and are intended for a wide range of professionals involved with genetic disorders of the skin and neurocutaneous diseases: dermatologists, pediatricians, neurologists, oncologists and general practitioners.