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BOOK REVIEW

D. Kaufmann (ed) Neurofibromatosis

Karger, Basel, 2008. 192 pp, 28 figures, 14 in color, 15 tables, US$190.00, ISBN 978-3-8055-8520-0

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Neurofibromatosis can be considered as systemic disease due to the involvement of different organs and systems by the disease process. There are multiple clinical entities under the large umbrella called "neurofibromatosis." Generally, neurofibromas are considered to be hamartomas but they show differing growth potentials and clinical course in different patients. In the past two decades, great strides have been made by laboratory research, which has increased the understanding of the genetic basis of this disease, but there remain large voids to be filled.

The editor has done an excellent job of selecting different topics and compiling this monograph. Introduction to the clinical presentation in the second article is well organized and elaborated considering the diversity of the scenarios encountered by clinicians dealing with this disease.

The proposed classification is informative in terms of clinical presentation and genetic basis but does not guide in management and counseling in such patients. It does however differentiate between neurofibromin 2 (NF2) and Schwannomatosis using radiologic investigations.

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A chapter on treatment and management of NF1 lays stress on the multidisciplinary approach in this clinical entity and, rightly, the need for dedicated NF clinics for better patient care and education of general practitioners involved in the care of such patients.

Four chapters have been dedicated to the structure, characteristics, and mutational spectrum of the genes involved in the occurrence of this disease process. The clinical phenotype and its correlation with the genotype shed light on pathogenesis of specific features.

It is intriguing to understand the role of neurofibromin in the pathogenesis of neurofibromas. Haploinsufficiency and the role of Merlin in tumor genesis and clinical behavior are interesting. The molecular basis of this condition appears to be an area of future research to find possible cell-directed management.

The genetic basis of neurofibromatosis is well explained and many chapters include portions devoted to increase the understanding of clinicians. Light has been shed on Schwannomatosis in addition to other related tumors.

Most of the contributors exhibit vast experience in human genetics and neurobiology. A remarkable attempt has been made to cover the available literature on this subject. This book will provide a good guide and reference volume for any clinician and geneticist dealing with the, as yet, poorly understood entity of neurofibromatosis.