Neurofibromatoses (Monographs in Human Genetics, Vol. 16)

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The neurofibromatoses are autosomal-dominant genetic disorders of the nervous system that primarily affect the development and growth of neural cell tissue. These disorders cause tumors to grow on nerves and produce other abnormalities such as skin changes and bone deformities. In recent years, the genes and mutations causing neurofibromatoses have been identified. The main types of neurofibromatoses, type 1 (NF1) and type 2 (NF2), have been shown to be distinctive disorders both clinically and genetically. More recently, allelic and non-allelic subtypes of NF1 have been defined as well as the NF2-related condition schwannomatosis. Many of the complex molecular mechanisms leading to the neurofibromatoses have been elucidated, resulting in a growing body of publications which are difficult to keep up with. This volume provides an important overview of recent findings on the neurofibromatoses. It focuses on the genetics and molecular biology underlying these diseases, but also covers their clinical features, diagnosis and treatment, stressing the need for interdisciplinary medical care. With contributions by the foremost investigators in the field, this timely book will appeal to geneticists, genetic counselors, pediatricians, neurologists and oncologists.

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