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Neurofibromatosis Type I: From Genotype to Phenotype (Human Molecular Genetics)

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Neurofibromatosis type 1 (NF1) is one of the most common dominantly inherited neurogenetic disorders. Affecting 1 in 4000 individuals worldwide. Symptoms include facial and body disfigurement, mental retardation and abnormalities of the cardiovascular, renal and endocrine systems.
This book is written for a broad readership ranging from students to professionals. It provides the reader with a basic overview of the disease before discussing the most recent research and therapeutic developments in detail. It is essential reading for medical geneticists, molecular biologists, dermatologists, oncologists, neurologists, genetic counsellors, general practitioners and research students.
The volume attempts to cover clinical aspects of the disease; gene structure, expression and mutation; structural and functional aspects of the encoded protein, neurofibromin; the role of the *NF1*gene as a tumour suppressor, the emerging genotype-phenotype relationship for NF1, animal models of the disease and future prospects for disease treatment and prevention.

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