Preface

Over the last 20 years there has been a rapid increase in our understanding of the disease mechanisms underlying neurofibromatosis 1 (NF1) and neurofibromatosis 2 (NF2), and related disorders. The neurofibromatoses are inherited diseases that involve the nervous system predominantly, but are distinct on both clinical and genetic grounds. Advances in molecular biology and mouse models have paved the way for clinical trials to combat benign and malignant tumors that characterize both diseases.

NF1 and NF2 are well documented in the medical literature, but partly due to the nomenclature, the distinction between the two conditions is blurred by clinicians. Furthermore the characterization of related and overlapping disorders has added to the complexity of diagnosis and management. The media has focused inexorably on people with NF1 who have extreme disfigurement, aiming to titillate rather than educate us, while NF2 is largely unknown outside of hospital practice.

In this book we aim to provide an accessible, up-to-date guide for nonspecialists on the diagnosis, management, and long-term care of people with NF1 and NF2. We emphasize the referral pathways to specialist centers for individuals with complex disease and highlight the available support networks. Above all we wish to show that coping with the neurofibromatoses relies on a partnership between patient and clinician, based on mutual trust and an ability to listen to the needs and choice of the individual.

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