The first edition of this book was published in 1990. It was written during an exciting time in the neurofibromatosis community: the genes for NF1 and NF2 had just been mapped and the NIH recently had held its consensus conference on diagnosis and management of neurofibromatosis. In fact, neurofibromatosis research was then just turning a corner. It had been about 100 years since von Recklinghausen’s seminal work that defined the disorder. Many refinements had been made in clinical care, especially in surgery and imaging, but the 1990 approach to diagnosis and management would, in many ways, have been familiar to a 19th century physician such as von Recklinghausen. The ensuing 15 years, however, have seen dramatic changes. These have been a source of great hope for those who must deal with neurofibromatosis, whether as a patient, a family member, or a physician.

Many of the changes are the result of general advances in medicine, not specific to neurofibromatosis. The use of magnetic resonance imaging (MRI) is a dramatic example of a technology that was just emerging when the first edition of this book was published but which is now routine. It is hard to imagine taking care of a person with NF1 or NF2 without the benefit of MRI to precisely define the extent and rate of growth of major tumors. Approaches to management have also improved, including better surgical techniques and new forms of chemotherapy. For example, substitution of watchful waiting or chemotherapy for radiation therapy has vastly improved the quality of care for children with NF1 and optic glioma.

Other advances—the ones that have generated the most excitement—are those that have revealed the basic mechanisms by which the neurofibromatoses exert their effects on the body. These advances flow from the ability to study the neurofibromatosis genes, the proteins encoded by those genes, and the way these proteins behave in cells and in animal models. Although much remains to be learned about the disorders,
x  Preface

The cellular pathways that are disturbed in individuals with neurofibromatosis are rapidly coming into focus. This has spawned genetic tests and is beginning to generate insights that may lead to new treatments. Families now have a realistic expectation that an individual who has NF might be eligible to participate in a clinical trial, a notion that was unheard of in 1990.

As rapidly as advances are being made, they can never happen fast enough for those who are affected by neurofibromatosis. In spite of the power of modern medical research, there is still a significant lag between discovery of a gene or cellular mechanism and the ability to use this information for treatment. The more that is learned, the greater the respect that scientists have for the complexity of any biological system. We cannot yet say that light is visible at the end of the neurofibromatosis tunnel, but at least we are now traveling through the tunnel on a high-speed train rather than walking.

The first edition of this book was written originally for patients, families, and health care providers. Since it was published, several books have been written for professionals, but no others for patients and families. This new edition has been completely rewritten, and is specifically targeted towards this latter audience. It was written in the spirit that having a better understanding of neurofibromatosis is the first step towards taking charge and dealing with it. We are grateful to many colleagues who have provided advice, and to many patients and families who have shared their personal stories.

We thank the employees, directors, and members of the National Neurofibromatosis Foundation, Inc., Peter W.R. Bellermann, president; Ann MacDonald, writer; Jane Novak Pugh, editor and project manager, whose efforts made this book possible.

When the first edition was written we expressed the hope in the preface that our foresight would prove to be myopic. It was: research has revealed insights we could not have imagined at the time. It is certain that the next 15 years will produce even more dramatic insights and surprises. No one can say when these will result in new treatments, but the prospects for a person diagnosed today with neurofibromatosis have never been brighter.

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