Preface

Numerous books and an untold number of publications in the scientific literature describe various chromosome abnormalities, both common and rare. One can search for any chromosome abnormality in a book index or an online scientific database and find every possible chromosome change that has been identified. Most of these abnormalities are reported, rightly so, in order to make a correlation with a clinical phenotype, diagnosis or prognosis of a disease. In other words, the emphasis has always been on directly correlating chromosome abnormalities with disease.

However, to date, little emphasis has been placed on standardizing how to designate chromosome abnormalities with their nomenclature and the related interpretation of these results with patient diagnoses. This book attempts to address the lack of standardization for writing the nomenclature of chromosome abnormalities and interpretive comments regarding those abnormalities. With over 250 cytogenetic laboratories in the United States alone, and maybe 500 laboratories worldwide, it is indeed time to develop more established guidelines for writing and interpreting cytogenetic, fluorescence *in situ* hybridization (FISH) and microarray test results.

The International System for Human Cytogenetic Nomenclature (ISCN) is the only resource (albeit a great one) to use in order to designate clear nomenclature in writing test results. Even so, if one is to compare a cytogenetic result from 10 different laboratories in the designation of a marker chromosome, for example, there could easily be five different ways to either write the ISCN nomenclature or interpret the result.

This book will be useful to cytogeneticists who write test results, to other geneticists, physicians, allied professionals and scientists who need to read these reports or use them in their clinical and/or research endeavors, and to those medical and human genetic students who are required to understand cytogenetic test results.

In this book, for each normal and abnormal cytogenetic result, the ISCN nomenclature and an interpretive comment with related recommendations are given. These abnormalities are divided among constitutional disorders and acquired malignancies, and are described by their genetic composition, genetic nomenclature and associated clinical features. Each chapter also contains a bibliography from which the information was obtained as well as pertinent research articles, databases and/or online web addresses for the reader's reference.

This book attempts to discuss as many genetic and malignant diseases as possible that have a known and prominent underlying genetic defect; however, it is not possible to list all the cytogenetic abnormalities in one manuscript. Readers who wish to see other abnormalities not included in the manuscript are encouraged to contact the author for reference in a future edition of the book.