Preface

The idea for this book evolved from discussions with parents, teachers, researchers, and clinicians—discussions for which the common thread was the potential for misinterpretation of phenotype descriptions. While the specific topics of discussion were highly variable, the lesson reinforcing the notion that genetic disorders may have highly variable effects was, perhaps, not so obvious. Indeed, it is the within-group homogeneity observed in each of these populations that is of inherent interest, to both clinicians and researchers. This book was the result of our need to expand the discussion of the *differences* represented in populations with well-characterized phenotypes.

As this story was unfolding, the pervasive emphases on genetics and brain development in science continued and altered the phenotypic description of various developmental disorders. But this increase in knowledge does not automatically translate into increased awareness in practice, even for common disorders, such as fragile X syndrome. One obstacle to early identification of a disorder is the phenotypic variation that differs significantly from a ''typical'' case. For this reason, the emphasis in each chapter of this volume is on reporting the full breadth of phenotypes such that ''subtle'' or ''atypical'' variants are also described.

To address the needs of practitioners and researchers alike, we report on disorders that each have a wide-ranging cognitive phenotype, including relatively common disorders (fragile X, Turner, and Klinefelter syndromes) and other disorders for which a genetic etiology is fairly well understood (as seen in Section I), but we also report on the broader categories of congenital hypothyroidism and metabolic disorders. Together, this combination of disorders has implications for understanding influences on development that apply to all human beings. Indeed, the wide range of phenotypic characteristics within each disorder presented affords the opportunity to study such influences whether via gene mapping studies (such as for Turner syndrome) or gene dosage effects (such as for fragile X syndrome).

Part I includes chapters on the common disorders that have an established etiology. For each disorder, there is an explanation of the genotype leading to the syndrome,

the medical implications, and the behavioral or psychological consequences directly related to the disorder. Within each chapter, there is an emphasis on how much variability is observed across individuals with the disorder as well as changes that occur during development.

Part II deals with broader categories of etiologies: congenital hypothyroidism, metabolic diseases, and environmental neurotoxins. What these distinct categories have in common is their widespread implications for brain and behavioral sequelae.

Part III deals with potential reactions to, and interactions with, diagnostic information, such as the role of genetic counseling after a diagnosis is made, the family's adaptation to a diagnosis that includes mental retardation or learning disability, and navigation of the early intervention options during the preschool or school-age years. Each of these chapters is intended to serve as a resource for the specialist whose expertise may not include these general issues beyond the identification and diagnosis of specific genetic disorder(s) in question.

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