Preface

Due to the generous representation of the afferent visual system within the brain, neurological disease may disrupt vision as a presenting symptom or as a secondary effect of the disease. Conversely, early developmental disturbances of vision often disrupt ocular motor control systems, giving rise to complex disorders such as nystagmus, strabismus, and torticollis. The signs and symptoms of neurological disease are elusive by their very nature, presenting a confounding diagnostic challenge. Neurological medications and neurosurgical treatments can produce neuro-ophthalmological dysfunction that can be difficult to distinguish from disease progression. Affected patients may experience substantial delays in diagnosis and are often subjected to extensive (and expensive) diagnostic testing. Scientific articles pertaining to specific disorders are scattered throughout medical subspecialty journals. These children continue to "fall through the cracks" of our medical education system. The increasing recognition that pediatric neuro-ophthalmology comprises a distinct set of diseases from those seen in adults has led to its emergence as a dedicated field of study.

Since the original publication of *Pediatric Neuro-Ophthalmology* 20 years ago, interest in the field has burgeoned. Pediatric ophthalmology and pediatric neurology subspecialty conferences often include symposia dedicated to recent advances in pediatric neuro-ophthalmology. Technical advances in neuroimaging and optical coherence tomography have given rise to a more integrated mechanistic classification of neuro-ophthalmological disease in children. Our understanding of neurodevelopmental disorders of the visual system has expanded, longstanding monoliths have been dissembled into component parts, basic molecular mechanisms have taken center stage, and genetic underpinnings have become definitional. Evolutionary alterations can now be observed at the level of the gene, adding a new dimension to our understanding of disease pathogenesis. New classifications now encompass clinically disparate conditions. Descriptive definitions have been supplanted by mechanistic ones, and clinical definitions superseded by genetic ones. Our concept of disease pathogenesis has been revised and in some cases overturned. Bearing witness to these remarkable advancements has impelled me to enhance and expand the second edition of *Pediatric Neuro-Ophthalmology* into a comprehensive third edition.

In the first multi-authored edition of this book, our goal was to present the clinical characteristics, diagnostic evaluation, and therapeutic options for the common neuro-ophthalmologic disorders of childhood. In so doing, we designed the book to provide a narrative journey through the thought processes involved in the clinical management of these disorders. In the second edition, I retained the basic narrative format of the original book, while expanding the exploration of these complex visual disorders in the context of the many new scientific advancements and discoveries that had come to light. Despite the proliferation of sophisticated ancillary tests, I generally rely on the clinical examination to diagnose common pediatric conditions such as Horner syndrome, myasthenia gravis, and pseudopapilledema. In the current edition, I have added tables and used italics to denote key clinical signs to "clinch the diagnosis." These conditions are challenging to diagnose, fascinating to understand, and gratifying to manage.

Since the publication of the previous edition, numerous scientific advances have refined our diagnostic armamentarium in pediatric neuro-ophthalmology. For example, optical coherence tomography now plays a pivotal role in the diagnostic evaluation of infantile nystagmus, as

well as in the evaluation of psychogenic or unexplained visual loss. Mitochondrial disease is now recognized as causing many if not most forms of pediatric optic atrophy. At the same time, our notion of pediatric optic neuritis is quickly evolving from a descriptive paradigm to one defined largely by neuroimmunological biomarkers. The melanopsin visual pathways are now implicated in a wide range of symptomatology. Genetic causes have now been assigned to obscure entities such as congenital mydriasis, and benign tonic upgaze of infancy. Batten disease is now diagnosed with a simple genetic blood test rather than by rectal or conjunctival biopsy.

T.S. Eliot asked "Where is the wisdom we have lost in knowledge? Where is the knowledge we have lost in information?" My goal in rewriting this book one last time is to preserve the clinical wisdom that would otherwise be lost in the mists of time. To this end, I have made a conscious decision to retain old references which, even when obsolete, give the reader a historical context to judge the role of seminal contributions in accelerating the evolution of our knowledge over time. Charcot is quoted as saying: "Disease is very old, and nothing about it has changed. It is we who change as we learn to recognize what was formerly imperceptible." Hopefully, this book will accomplish this ambitious goal for its readers. I have made a special effort to retain the time-honored "clinical pearls" that I gathered during my fellowship training with William F. Hoyt, M.D., Creig S. Hoyt, M.D., and Edward G. Buckley, M.D., who endured my endless queries and taught me the art of neuro-ophthalmologic examination. I would never have pursued the field of pediatric neuro-ophthalmology, if I had not "crossed the street" at UCSF during my fellowship to observe Creig Hoyt, a great diagnostician and progenitor of this field. My hope is that the third edition will serve as a useful resource to ophthalmologists, neurologists, neurosurgeons, and pediatricians; and that it will catalyze research into the basic mechanisms of these disorders.

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