Book Review

Am. J. Hum. Genet. 65:581, 1999

Genetic Diseases of the Eye. Edited by Elias I. Traboulsi. New York and Oxford: Oxford University Press, 1998. Pp. 876. $198.50

A large set of inherited disorders clinically manifest themselves as primary disorders of the eye. As every medical student learns, however, the eye can be the site of discovery of numerous generalized diseases. The characteristic vascular changes of diabetes or hypertension and the inflammatory response to infectious agents are just two examples. Although there may be a genetic component to susceptibility to these diseases, the specific ailments of the eye are usually the province of the eye specialists—ophthalmologists and specialists in allied disciplines. Increasingly, however, these seemingly arcane disorders are coming to the forefront of our understanding of medical genetics outside of the eye because of the importance of vision, the numerous inherited disorders of the visual system, and the insights these genetic disorders provide for understanding diseases that affect other organs.

Physicians who encounter such disorders, but are unfamiliar with the eye and its diseases, immediately confront a considerable barrier to understanding even elementary aspects of the disorder: the bewildering array of eponyms and special terms associated with visual-system anatomy and pathology. Take, for example, the term "retinitis pigmentosa," a disease associated, in 24% of affected American kindreds, with mutations of the *Rhodopsin* gene. The disease is not an inflammatory lesion, and so its name—retained for historical reasons within the field of ophthalmology—fails to indicate our present understanding of its etiology. The disorder primarily affects rod photoreceptors, but it is the curious rearrangement of the pigmented epithelial cells along fine vessels that accounts for its appearance when viewed with an ophthalmoscope. Retinitis pigmentosa can also be generated by mutations in the gene for another rod protein, Peripherin/RDS, but other clinical entities, such as the formidably named "retinitis punctata albescens," can also arise through mutations in this latter gene. Worse still, mutations in yet another gene, which encodes the retinaldehyde-binding protein RLBP1, can lead to retinitis punctata albescens. My purpose in illustrating this confusion of clinical terms and genetics is to encourage those encountering the eye and its diseases to consult *Genetic Diseases of the Eye.*

In a major undertaking, Elias I. Traboulsi has assembled a large group of clinician-scientists with expertise in eye diseases to author 42 chapters on disorders of nearly every component of the eye. Accordingly, there are chapters relating to the cornea, lens, retina, and central connections, as well as chapters on diseases involving the interactions of various parts of the eye and/or its development (i.e., glaucoma and aniridia) and disorders affecting subcellular organelles (i.e., mitochondria, lysosomes, and peroxisomes). An excellent index serves the reader well when a topic of interest is not obviously described in a chapter title.

Traboulsi's book covers some topics in greater depth than would a general reference, and it will be of great benefit to medical students looking to supplement their ophthalmology textbooks. Some chapters provide sophisticated lessons for clinicians on how to think about multifactorial genetic conditions, such as Michael B. Gorin's outstanding chapter on "The Genetics of Age-Related Maculopathy." The other audience to consider is the graduate student/fellow or the Ph.D. mentor working in a genetics laboratory who discovers a gene that maps to a locus of a vision disorder. Before turning to the primary literature and confronting the perplexing nomenclature of the field, such researchers would be well advised to consult this book. Starting with the relevant chapters in *Genetic Diseases of the Eye* and the references included, the newcomer should be prepared to conduct the Boolean searches of the literature without getting lost.

Excellent web sites are now available that catalogue a large number of inherited vision disorders, and these resources will be of great assistance to investigators who are armed with the information in this book. New web sites are being created, and a vision research society (Association for Research in Vision and Ophthalmology) is considering creating an electronic link to such sites to help scientists, clinicians, and the lay public to navigate this important field. Unfortunately, however, Traboulsi does not include a list of these web sites, and in general the reference lists in the book have become outdated. Despite the 1998 publication date, most of the chapters in *Genetic Diseases of the Eye* refer to primary studies conducted before 1996. Only a few more recent references appear in some chapters. In addition, although the reproduction of color images is impressive, black-and-white histology images are really below standard in the field, so the original work should be consulted when needed. Still, I would encourage all medical libraries, ophthalmology departments, and training programs in medical genetics to acquire this book.

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