Book Reviews

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Genetic Skin Disorders. By Virginia P. Sybert. New York: Oxford University Press, 1997. Pp. 675. $195.00

In the preface of Dr. Sybert's book she poses the question: "Why another book on inherited skin diseases?" A perusal of this work quickly answers the question. Practitioners of medical genetics are consistently faced with dealing with disorders of the skin in two contexts: as signs or clues for syndromes and in the genetic counseling of the primary heritable skin disorders. Dr. Sybert indicates that she "modeled this book after David W. Smith's incomparable *Recognizable Patterns of Human Malformation*." This reviewer believes that Dr. Sybert was successful in this attempt and has actually added a new volume to the genre of useful catalogs based on specific disease groupings. *Genetic Skin Disorders* can be placed along with Gorlin's books on head and neck syndromes and deafness and Jones's recent versions of Smith's *Recognizable Patterns.*

After the introduction and a brief overview of medical genetics, entitled "Practical Inheritance," the book is divided into the following chapters: "Disorders of the Epidermis," "Disorders of Epidermal Appendages," "Disorders of Pigmentation," "Disorders of the Dermis," "Disorders of Subcutaneous Tissue," "Lymphedema," "Urticaria," "Other Disorders," "Tumors/Hamartomas," "Metabolic Disease," "Premature Aging," "Photosensitivity," and "Immune Deficiency Diseases." The author closes the book with a glossary and a very useful appendix that lists syndromes and disorders according to skin sign. Various categories of cutaneous signs are compiled, and the reader is able to obtain a differential diagnosis by manifestation. For example, the syndromes that have calcinosis cutis as a feature are listed; the conditions of thin/sparse hair and the conditions of curly hair are catalogued and detectable by the reader.

This work has many strengths. The entries are organized into a similar format, are easy to read and follow, and conclude with a selected annotated bibliography. The references are invariably up-to-date and usually include the seminal papers and at least one review. More than half of the photographs in the book are in color, and the quality of the photographs is generally excellent. At least one photograph, black-and-white or color, accompanies each entry in the book. In addition, Dr. Sybert has included, with each entry, the support group related to the disorder in question. During the past decade, referral to support groups has become a routine aspect of genetic care. Here the author has provided us with easy access to these resources. In addition, when it exists, the MIM number is included in the entry. This and other features would enable *Genetic Skin Disorders* to eventually go on-line.

The discussions in each entry are concise but give the reader a sense of the disorder; of its variability, natural history, and pathology; and, when available, knowledge on basic defects. There is little detail on molecular aspects of the basic defect, but comprehensiveness on that topic is not an objective of this work. As mentioned above, the appendix is quite useful in helping the clinician with the differential diagnosis of a sign or feature. This index is modeled closely to the text of Smith's *Recognizable Patterns* and is quite usable to anyone familiar with that commonly used "incomparable" catalog.

This reviewer found that one of the additional strengths of the book was the personal nature of Dr. Sybert's discussions of patients. I felt at times that I was actually having a conversation with her, almost as if we were consulting on the patient together while making rounds. Her comments on some of the clinical aspects of cutaneous features are particularly helpful to those of us who are not trained in dermatology but who commonly evaluate patients in the above-mentioned contexts. I believe that more of this discussion based on her clinical experience and thinking about patients would be welcome. Although, to some extent, it partakes more of the classical expert-based (as opposed to evidence-based) approach, it has the definite feel of traditional clinical pearls.

This reviewer found very few weaknesses, deficiencies, or errors in the book. There was the occasional use of dermatological jargon, which required some paging in order to figure out the meaning. Discussion of the various classical heritable skin disorders, such as epidermolysis bullosa and ichthyosis, would have been improved by a table summarizing the classifications. There were a few conditions that we often think of in clinical genetics, such as the Dubowitz syndrome, that were missing from the entries (but this was included in one differential diagnosis). Otherwise, the book is immediately usable while one is seeing a patient, which is its stated purpose.

Genetic Skin Disorders is strongly recommended for the shelf of anyone involved in direct genetic care. It could be placed along with the other books listed above, for access in clinic settings.

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