

The Bedside Dysmorphologist A Guide to Identifying and Assessing Congenital Malformations Second Edition

Reardon [2015] just barely fits in a white coat pocket. The book itself is organized by organ system and expertly written with two to three pages devoted to a given physical feature. The Preface clearly stresses that there exists a hole in medical dysmorphology literature of good references for the non-specialist, the pediatrician, or the trainee. This book reaches that target audience admirably. It also does not suffer by being too elementary and would be well used by younger geneticists who may be more comfortable with molecular methods than classic dysmorphology.

Reading the book feels like having a wizened attending standing over your shoulder. Doctor Reardon writes in a conversational style and gives concise discussions with multiple teaching points on each specific dysmorphic feature. Unlike *Smith's Recognizable Patterns of Human Malformation* [Jones et al., 2013] which is organized by syndrome, this book is structured around the physical exam in action and goes beyond *Elements of Morphology* [Allanson et al., 2009] which purely defines structure. Common features and syndromes are well represented though also as an example; with cutis aplasia, Dr. Reardon point out the association with maternal use of aspirin. As another example, joint restriction may be the presenting sign of Antley–Bixler syndrome or be due to maternal fluconazole exposure. Apart from well written summaries, this book over 100 glossy color photographs located in line with the text and annotated by syndrome. It is an excellent book to just read.

The Bedside Dysmorphologist has a strong writing style though struggles with balance. There is simply too much content for any one resource to be both comprehensive and equally approachable. The book does not use strict organization like what would be found in a naturalist's field guide, though neither are the chapters written to be read from cover to cover. For each feature, several characteristic syndromes are proposed for consideration though this is not a reference to find complete differentials. More problematic is that when the book does include underlying molecular pathology it does so in a manner that proposes confirmatory studies. The book often states that a FISH probe or single gene sequencing could confirm a syndrome, but it does not mention key information such as pretest probability, or that other more appropriate arrays and panels exist. This makes it an insufficient reference in the work up of a dysmorphic patient though it gives the impression that testing strategies are included.

The Bedside Dysmorphologist also struggles as a standalone teaching text. After the polite acknowledgments it starts right at plagiocephaly without an introduction or general guide to the genetics evaluation. This absence is felt throughout the text which

How to Cite this Article:

Wilson TE. 2016. Book review.

Am J Med Genet Part A 170A:3347.

in many descriptions emphasizes the need to obtain an obstetric history or a thorough family history. These are certainly helpful tips, though they are so often important that this advice can become repetitive across multiple entries. Likewise, in terms of providing a basic description for each feature with appropriate nomenclature it will not replace *Elements of Morphology* [Allanson et al., 2009]. Neither will this guide serve as a replacement for an OMIM search or GeneReviews summary which are still needed for the actual management of children presenting with a suspected genetic disorder as the accompanying text is often not thorough enough.

With those criticisms stated the downsides of *The Bedside Dysmorphologist* are the same challenges facing the profession of clinical genetics. As the molecular side of our field advances there seems to be a steadily increasing amount of content to learn and less teaching dedicated to physical exam findings. This book does an admirable job of defining its own niche and then expertly filling that space. In addition to simply being an enjoyable read, this is a springboard toward finding those other references while literally at the bedside. When the text works well it helps the practitioner; recognize the finding, suspect the diagnosis, and then look just a little harder for additional details to identify a likely syndrome.

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Manuscript Received: 21 July 2016;
Manuscript Accepted: 8 August 2016

Article first published online in Wiley Online Library
(wileyonlinelibrary.com): 12 September 2016
DOI 10.1002/ajmg.a.37955