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BOOK REVIEW

The difficulty to classify complex dysmorphic syndromes on the ward

Syndrome diagnosis on the ward

The Bedside Dysmorphologist. Classic Clinical Signs in Human Malformation Syndromes and Their Diagnostic Significance
Edited by William Readon
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Reviewed by Albert Schinzel.

When confronted with a newborn with multiple congenital developmental defects including a pattern of dysmorphic signs, the clinical dysmorphologist is challenged to find the correct diagnosis out of some 1500 syndromes, if possible directly at the ward. As we unfortunately do not have unlimited memory capacity, we have to evaluate the clinical findings and then go back to the office, to a library or a computer and search in books and genetic programs, which is often a frustrating and time-consuming task.

The idea behind Dr Reardon's book is to enable the clinical dysmorphologist to get help directly by checking specific dysmorphic features and malformations. The author lists dysmorphic features and malformations throughout 300 pages, starting with a short characterization of the sign, followed by a differential diagnosis regarding for which syndromes it is characteristic. Finally, a further diagnostic laboratory workup for confirmation of the proposed syndrome(s) is listed.

The idea is, at the first glance, challenging. However, testing with various syndromes soon shows the limitations of the approach. For frequent signs such as microcephaly, hypertelorism, dysmorphic ears, non-horizontal eye axes and many others, a limitation to 3–5 diagnoses is inevitable, yet unsatisfactory and incomplete. For less-frequent signs, on the other hand, it is often not well known which signs are the most specific ones for a given diagnosis. Therefore, the shortcoming of this book is the same as that of other books and databases: beginners will still be lost and often not able to perform a reliable classification; experienced dysmorphologists will not get much help for their attempts, simply because of the necessary limitation of the contents of the book.

It is unfortunate that the book came out more or less exactly at the time when a consortium of dysmorphologists made proposals for classification and definition of dysmorphic findings which, of course, could not be considered for the text of this edition (for example, for short palpebral fissures *versus* blepharophimosis).

The reviewer tested the book for chromosomal aberrations and found it incomplete. For example, the following differential diagnoses were not listed: larger 13q deletions with anencephaly and hypoplastic thumbs, midface hypoplasia with distal 18q deletions, and micrognathia with unbalanced 11;22 translocations. Although for postaxial polydactyly of the upper limbs trisomy 13 is listed, it is not listed for the lower limbs. Deep plantar creases are mentioned, but deep palmar creases are not. Coffin–Siris syndrome is not mentioned for hypoplastic nails. 45X0 is not the correct cytogenetic nomenclature and should be replaced by 45,X. This is just an arbitrary list, which could be extended.

The illustrations are very helpful, but of quite variable quality. A definition of what can be considered a syndrome is not given. The author sometimes mentions Fanconi syndrome and on other occasions calls the same condition Fanconi anemia (the latter term is far more popular).

Despite these limitations, which are of a fundamental nature and reflect the complexity of syndrome classification, this book, written by a very experienced dysmorphologist, will be helpful if used critically at the ward■

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