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BOOK REVIEW

Congenital Malformations

All I wanted to know about congenital developmental defects

RE Stevenson, JG Hall, Human Malformations and Related Anomalies, Second Edition, Oxford University Press, Oxford, UK, 2005.
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The title of this opus raises high expectations: a book where not only the medical geneticists, clinicians, but also biologists find what they want to know about congenital malformations and other developmental defects in humans. After the 1971 edition of *The Warkany*, there was a gap in this field, which could only partly be covered by chapters in the well-known books about syndromes and clinical genetics.

The book begins with a general overview covering nomenclature, classification, gene and chromosome mutations, teratology, diagnostic procedures, management and genetic advice. The reviewer would have wished to see a longer and more detailed section on teratology because such information is difficult to obtain; however, as the focus of the book is elsewhere, the authors cannot be blamed for keeping this part short. The following chapters deal with organ systems (cardiovascular and respiratory, craniofacial, neuromuscular, skeletal, gastro-intestinal, urogenital, skin, and others including twins, endocrine organs, hemihyper/hypotrophy and umbilical cord).

The reviewer tested the content of the book not only with defects for which it would be very difficult to find an appropriate literature such as agnathia-otocephaly and iniencephaly, but also for more common conditions such as twins, tongue, Moebius syndrome (not rather Moebius sequence) and found the main questions a clinical geneticist could pose answered and the literature updated. This is unique and deserves that the book should receive a high recommendation. There are two types of tables: one listing 'syndromes with…', the other 'occurrence in…'. For some conditions this seems like Sisyphos work, for example, the 24 page-long list of conditions in which microcephaly has already been described – long and by necessity incomplete, but nevertheless useful for special questions. With 568 references, the reader certainly has more than enough sources for further reading; sometimes less could be more.

Errors in such a large field are inevitable. For example, that Table 1–10 (selected malformations and malformation syndromes for which the gene has been identified or localized) contains the cat eye and the Jacobsen syndrome. As in most illustrations in clinical genetic books, the quality of illustrations is suboptimal. Some conditions could be presented or at least cross-referenced in another section, for example, amnion disruptions under limbs. It would be helpful to present some growth curves, but in this case Hall would compete with Hall (her book on growth curves just appeared in a second edition), and if all these extra wishes would be considered, the book would clearly fall beyond the maximal size.

The authors are very right in mentioning that a 'universally acceptable and permanent terminology for anomalies would appear utopian' (p. 6) – a crux for geneticists dealing with unclassifiable congenital developmental defects. This should, however, not imply the inconsistent use of terms themselves, for example Proteus 'syndrome' for a condition that according to their own definition, cannot be named a syndrome (p. 9).

For such an important book and the global interest in the topic, it is regrettable that, with only $1\frac{1}{2}$ exceptions (Passarge, Spranger), exclusively authors from Canada and the United States were considered.

In summary, this book will be of great help for clinical geneticists concerned with congenital developmental defects. It can be highly recommended■

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