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BOOK REVIEW

Robin D Clark & Cynthia J Curry: *Genetic Consultations in the Newborn*

Oxford University Press, ISBN no.: 978-0-1999-9099-3 Price: Hardback: £65.00/$99.95

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"**Genetic Consultation in The Newborn**" is a really innovative, interesting and useful textbook (about 360 pages). In fact, I do believe it is a must to have for every expert people working in this field and for general practitioners too.

Nowadays, the recent techniques of diagnosis, such as array CGH and the next-generation sequence, give the opportunity to analyze minimal chromosomal rearrangements (microdeletions, microduplications, and so contiguous genes' syndromes) and the whole exome/genome in order to discover more and more new genetic causes of disease before misdiagnosed.

Each chapter of this manual is complete and schematic at the same time. That means a physician who needs to know immediately how to manage a newborn with congenital malformations at birth or during infancy can consult it in a very simple and quick manner.

It is structured in an initial clinical case with a defined or suspected genetic disease and then it is described the discussion about the definition, genetic testing, the differential diagnosis, including genetic conditions and the environmental conditions too, the management and follow up. The book is enriched by a lot of illustrative pictures about clinical cases, a very precious collection of years and years of clinical activity of the authors. These pictures defined a clinical atlas very useful to recognize immediately a specific *gestalt.*

The concept of practical "pearls", derived from the experience of the authors and the common consensus gained over time by scientific evidence, is very appreciated and original.

The book is organized in nine parts and an appendix. The first part deals with general conditions can affect a newborn as soon as it was born: hypotonia, intrauterine growth restriction, overgrowth, hydrops, teratogenic anomalies. The part dedicated to the twins is very interesting because highlights the importance of the epigenetic imprinting in monozygotic twins with an identical genome but different phenotype.

Each one of the other eight parts treats specific organs and systems: heart, craniofacial, central nervous system, gastrointestinal, genitourinary, skeletal dysplasia, and skin. For further information, every disease is recognized by a unique OMIM (online mendelian inheritance in man) number and accompanied by links and the most up to date references.

I think that the real strength of this book is the emphasis given to the impact of teratogens on malformations. Sometimes a phenotype due to an environmental factor could be confused with a genetic syndrome (i.e., in the third part the description of a clinical case of a newborn with an initial clinical diagnosis of Treacher Collins syndrome not confirmed by genetic testing where an impact on phenotype due to an immunosuppressant maternal intake during pregnancy has been discovered). The book is full of similar cases and environmental factors have a predominant role in differential diagnosis too.

The appendix of the book includes a description of nineteen well known diseases and the authors enrich it with the last recent news about the related diagnosis and management.

I was very excited to read this book because I found it very useful and for sure I will use it in my daily work.

Compliance with ethical standards

Conflict of interest The author declares that they have no conflict of interest.

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