

Book Review: Genetic Consultations in the Newborn

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Genetic Consultations in the Newborn

First Edition.

Robin D. Clark and Cynthia J. Curry. Oxford University Press,

Hardcover

Published: 27 February 2019

400 Pages

8-1/2 x 11 inches

ISBN: 9780199990993

Also Available As:

Ebook

The practicing Perinatal/Neonatal health care provider today has noted a changing frontier in diagnosing genetic abnormalities and determining prognosis.... if at all possible. The simple words "chromosome analysis" has been replaced by an ever-changing vocabulary. There needs to be an understanding of karyotyping, extended banding chromosome studies, Fluorescence in situ hybridization (FISH) studies, microarray analysis, genomics, epigenetics, and whole exome sequencing. These are just a few of the platforms which the perinatal/neonatal health care provider must be conversant.

"There are 42 chapters in this monograph, formatted in a specific manner. Each chapter begins with the presentation of a 'clinical consult.' "

The authors in the preface have determined that their goal is to " make the evaluation of common neonatal anomalies and genetic syndromes accessible and understandable." A very complex subject needed to be simplified with an approach that would be up to date with the general "new approach" to learning.

There are 42 chapters in this monograph, formatted in a specific manner. Each chapter begins with the presentation of a "clinical consult." This immediately involves the reader in a case, parts of which may be familiar. These clinical consults are short and to the point. Following the clinical presentation, the Sections following g include Definitions, Differential Diagnoses, Evaluation and Management and finally a Suggested Reading section. At the end of each Section, a "Pearl" is presented which focuses the reader to return and re-review some of the presented material. The chapters are presented in a

"System Format". That is, for example, the cardiovascular system, and the central nervous system are the main topics of a specific section.

At the end of the book, there is an Appendix of Syndromes that commonly present in the newborn period. The format of the syndromes is identical to the presentations in earlier chapters.

The illustrations, although only in black and white depict many of the clinical findings which are presented in the text. There is an index which is reasonable and not overly detailed. It allows the reader to easily look up the necessary material and follow a suggested blueprint for diagnosis.

This book is not meant to be a compendium of the thousands of genetic disorders which are now part of our genetic environment. The book services as a guide to today's perinatal/neonatal health care provider to make a more accurate diagnosis and to allow for a better understanding of these diseases so that our colleagues and the family can make educated choices for the future of the child.

Disclosure: The author has no relevant disclosures..

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