Book Review

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Velo-cardio-facial Syndrome: A Model for Understanding Microdeletion Disorders. Edited by Kieran Murphy and Peter Scambler. New York: Cambridge University Press, 2005. Pp. 243. $120.

Velo-cardio-facial Syndrome: A Model for Understanding Microdeletion Disorders is a compilation of chapters that each focus on one aspect of this well-known genetic syndrome. Each chapter is written by a well-known expert in his or her discipline, someone who has done extensive work with individuals with velocardiofacial syndrome (VCFS). The authors are from many different centers, which is different from many similar books in which the authors are all based at a single center. This diversity is a strength but also a weakness, since the chapters lack uniformity in style and presentation.

VCFS is among the most common genetic syndromes but is also one of the most variable. Every organ system can be affected, and developmental, behavioral, and psychiatric problems are very common. It is easy to imagine that individuals with VCFS are cared for by a wide variety of health care providers. Since they are focused on their own disciplines, they may not recognize or appreciate the roles of the many other specialists involved in the care of the patient with VCFS. It is the goal of this book to provide a resource for clinicians on both the range of complications seen in VCFS as well as on the roles of the various specialists in the care of these patients. Meeting this goal is a daunting task, since the intended audience includes professionals with a variable range of medical and scientific backgrounds. In large part, the book accomplishes this goal and would be suitable for individuals with a wide range of backgrounds. This is an ideal reference book for a cleft and craniofacial clinic, for example, where many VCFS patients are seen, just as it would be appropriate for a speech pathologist or a psychologist, as well as for a plastic surgeon or pediatric subspecialist.

The book is organized in 12 chapters that follow a logical sequence. Each chapter focuses on a specific aspect of VCFS and is written by an acknowledged expert on the topic. For example, the first chapter, entitled "Historic Overview," is written by Dr. Robert Shprintzen, the man who first coined the term "velocardiofacial syndrome" and whose work has done much to define the disorder as we know it today. This chapter is among the strongest in the book, providing a broad introduction to the topic so that any reader would have an understanding of how this disorder has come to be considered one of the most important and well-studied genetic syndromes.

The next chapter, by Drs. Scambler and Katrina Prescott, is a detailed description of the molecular genetics of VCFS. It nicely reviews how the deletion occurs and why it is so common—questions that I, as a clinical geneticist, am often asked by parents, as well as by medical colleagues. I will use this reference to explain the answers to these questions in the future, since the chapter is concise and well-illustrated. Mouse models, gene mapping, and molecular embryology are also reviewed in a manner that is easily understandable by readers at different levels. Several pages are devoted to listing every gene known in the typically deleted region.

Chapters 3–6 deal with specific physical complications associated with VCFS: Chapter 3 covers congenital heart defects, chapter 4 covers palatal anomalies and velopharyngeal dysfunction, chapter 5 covers nephrourologic, gastrointestinal, and ophthalmologic findings, and chapter 6 covers immunodeficiency.

The chapter on palatal anomalies and velopharyngeal dysfunction is also very well written, with helpful illustrations. In a relatively short space, Dr. Richard Kushner covers a tremendous amount of material yet provides depth that satisfies even plastic surgeons. Chapters 3, 5, and 6, however, lack sufficient detail to be of interest to the experienced clinician. These chapters are too broad and are not focused on VCFS. They read as stand-alone reviews, repeating information that is covered elsewhere in more detail, and, as a consequence, have less space to address the intended topic.

Chapters 7–10 cover perhaps the least recognized but most interesting aspects of VCFS—namely, the behavior and psychiatric manifestations, cognition, neuroimaging, and speech and language abnormalities. These chapters are each excellent and well written at a level that is appropriate for any health care provider. The discussions are detailed, focusing on the specific topic, with little redundancy between these closely related topics. The figures and illustrations are, in general, helpful, although only those in chapter 8 are in color. One unfortunate, if somewhat minor, issue is that one of these color figures is mislabeled with the legend from the following figure.

Chapter 11, "Genetic Counseling," by Donna McDonald-McGinn and Dr. Elaine Zackai, is an excellent summary of the book. It is perfectly placed—as if to say, this is how one uses all the information in preceding chapters. The chapter covers counseling issues that apply not only to VCFS but to any genetic disorder. The illustrations are very helpful teaching tools, and a figure of 29 pedigrees of VCFS families graphically demonstrates the clinical variability of this disorder in a way that is better than words.

The final chapter, "Family Issues," is a fitting epilogue. Written by a member of The 22q11 Group, a U.K.-based parent support organization for VCFS, it reads as a primer for health care providers on a parent's thought process upon learning

that his or her child has VCFS. If the previous chapters provide an excellent foundation of medical knowledge on VCFS, this chapter is a keen reminder of who the patient is and what this diagnosis means for the child and his or her entire family.

Overall, this is a book I'd recommend to anyone, regardless of background, who cares for children with VCFS. I would recommend it to speech pathologists, psychologists, and medical residents. Of the more experienced providers, no one will find every chapter worthwhile; I would imagine that specialists will find the chapters on their own disciplines relatively simple. However, the remaining chapters will provide just what they need, which is a concise summary of the other clinical and genetic aspects of one of the most common and fascinating genetic syndromes.

NATHANIEL H. ROBIN
Associate Professor of Genetics and Pediatrics
University of Alabama at Birmingham

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