Book Reviews/ Évaluation des livres

Velo-Cardio-Facial Syndrome. Volume 1
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Velo-Cardio-Facial Syndrome, Volume 1 is the fourth book in the "Genetic Syndromes and Communication Disorder Series." The authors, Dr. Shprintzen and Dr. Golding-Kushner, have extensive experience in the diagnosis, treatment and study of VCFS. It was Dr. Shprintzen who first coined the term "Velo-Cardio-Facial syndrome," and he has since contributed much to our understanding of the condition. This volume is the first of two and presents a comprehensive look at the history, phenotype, genetics, growth/feeding issues, and medical management guidelines pertaining to VCFS. The intended audience is defined as "anyone interested in VCFS."

The book is divided into five chapters. The first chapter provides a detailed summary of the history of VCFS from the first suspected published case report in 1955 to the identification of microdeletion 22q11.2 as the etiology to the ongoing current debate about what to call the syndrome. This is a comprehensive and insightful overview. It also provides an understanding into the reasoning behind the many different labels applied to the same disorder and the resultant "nosologic confusion" that continues to puzzle both professionals and the public.

The second chapter provides an exhaustive description of the phenotypic features of VCFS. Much of the book is dedicated to providing an overview of all possible complications or associations related to VCFS. While it is helpful to the clinician to be aware of the variability and extent of the phenotype, little perspective is given regarding the actual prevalence of various phenotypic findings. Equal attention is given to findings that have been reported in one (e.g. cerebellar ataxia) individual with VCFS as to findings that are reported to occur in a high percentage of individuals with VCFS (e.g. chronic leg pains). The reader may develop a skewed perception of what to typically expect, and thus could have benefited by a classification of common versus less common phenotypic features. More focus on the typical VCFS phenotype and its most commonly associated complications/associations, would have helped the reader to develop a clearer gestalt of the syndrome. This would lead to more sensitive recognition and ultimately a better diagnostic rate amongst speech and language pathologists and other health care providers. Special attention is given to the presentation of speech and language disorders. Many of the statements about the speech and language profiles are based on the authors' extensive experiences in treating this patient population in a communication disorders clinical setting and leaves the reader with the impression that nearly all affected individuals have gross VPI and resultant severe speech issues. The widely variable heterogeneity in this population with regards to speech/resonance disorders, and the resulting wide range of management options are not well outlined which may contribute to under-diagnosis of the condition. We have seen children with VCFS with normal resonance or slight/mild hypernasality. In addition, alternative, non-surgical management options need to be considered for carefully selected young children who may benefit from speech/resonance therapy alone. Some young children with VCFS and compensatory articulation substitutions (glottal stops) are able to achieve correct articulatory placement and avoid the need for surgical management for VPI. While the authors encourage advocating for speech therapy as soon as a need is identified, in this patient population, a more preventative approach should be recommended; specifically to avoid the development of compensatory articulation substitutions and to facilitate expressive language development.

The third chapter reviews the genetic basis of VCFS. The diagrams in this chapter provide a helpful adjunct to the written explanations. The explanatory text appears to be directed toward the lay public and is thus oversimplified for the health professional reader. Some of the analogies used to explain complex genetic mechanisms may be more confusing than clarifying. The advice provided regarding genetic counseling relies on the authors' anectodal experiences, rather than on evidence-based practice published in the genetic counselling literature. This is highlighted in part by a failure to address the principles of counseling individuals with intellectual disabilities.

The fourth chapter provides general recommendations regarding medical management of the various phenotypic features in VCFS. This chapter is the most useful as it provides the reader with a thorough overview of the various investigations that should be considered in the overall care of a child with VCFS. The authors, however, draw upon their extensive anecdotal experience, at the expense of evidence-based medical references to support their recommendations.

The fifth chapter focuses specifically on issues regarding growth and feeding. A valuable contribution to this chapter is the longitudinal information on height and weight which the authors have used to generate VCFS specific growth curves. This may prove to be a helpful tool once validated independently. As in the previous chapter, the use of evidence-based medical references would have served to strengthen the authors' recommendations regarding medical management.

Overall, this publication has many strengths, including clear visual aids and photographs. The variety of images of patients with VCFS helps the clinician recognize the variability of the syndrome dysmorphology. The enclosed CD ROM serves as a good teaching tool, for speech-language pathologists in particular, as the majority of samples are nasopharyngoscopy or multiview videofluroscopy studies. Preceding titles and the use of labels superimposed on images provide additional clarification. The synopses of the relevant video clips at the end of Chapters 1 and 2 are helpful as well. The book is clearly organized and simply written, which may make it useful for families who want more detailed information. Visually clear information boxes attempt to define concepts and terms. Overall, readers, health professionals, and caregivers will benefit from the extensive clinical experience and information that the authors have collated into this comprehensive resource.

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