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Progress in pediatric cardiology (“Genetics of Pediatric Heart Disease”)

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1. Introduction

Pediatric heart disease includes both structural and functional abnormalities ranging from congenital heart defects to cardiomyopathy. Congenital heart defects represent a significant proportion of birth defects and account for the majority of morbidity and mortality related to congenital malformations. Genetic abnormalities underlie the cause of many congenital heart malformations. Recent technological advances such as microarray analysis of gene expression, automated DNA sequencing and gene mutation/polymorphism identification and analysis are providing new insights into deciphering the complex networks of gene expression activity that regulate cardiac development and function. The identification of gene mutations and characterization of gene alterations that produce congenital defects are important in understanding causative genetic mechanisms. The use of efficient and rapid molecular methods for identification of disease causing genes allows for not only an expansion exponentially of genetic knowledge but for potential use in treatment and management of individuals with pediatric heart disease.

Our current understanding of genetic factors leading to pediatric heart disease will be addressed in this journal beginning with the first chapter written by Dr. Benson and his colleagues on genetic causes of congenital heart disease and developmental insights. Next, the genetic epidemiology of cardiovascular malformations is addressed by Drs. Lin and Ardinger in their review of the occurrence, morbidity and mortality, and associated anomalies and malformation syndromes related to congenital heart defects. In the third chapter, my colleagues and I discuss the current status of gene expression and micro array technology in detecting pediatric heart disease with special emphasis on conotruncal defects and the identification of potential candidate genes and networks leading to developmental cardiac problems. Chapter 4, written by Dr. Exil and his colleagues, discusses the metabolic basis of pediatric heart disease with particular emphasis on fatty acid oxidation defects and treatment of affected patients. In Chapter 5, Dr. Hershberger reviews and describes the current understanding of familial dilated cardiomyopathy, candidate genes and treatment. The remaining chapters are written by leading experts in the field of medical genetics, molecular biology and pediatric cardiology. Each chapter concentrates on a separate classical genetic condition whereby the genetic lesions have been identified and characterized, beginning with Dr. Spinner’s contribution on the genetics of Alagille syndrome. This is followed by Dr. Noonan as she discusses the syndrome she first reported in 1963 along with other related disorders. The next chapter includes a clinical and historical overview of velo-cardio-facial syndrome written by Dr. Shprintzen, the syndrome first

described by him in 1978. The final chapter discusses the medical overview and genetics of the Williams-Beuren syndrome written by Dr. Pober and her colleagues.

Although an attempt has been made to provide the most recent information relating to the genetics of pediatric heart disease and to provide examples of classical syndromes and their genetic causation, this review is not meant to be exhaustive. We trust this review of genetic causes of pediatric heart disease, epidemiology, animal models for heart disease, micro array gene expression, metabolic basis of pediatric heart disease and description of genetic disorders with specific genetic lesions and associated heart pathology will provide useful information to pediatric cardiologists, surgeons, pediatricians, geneticists, and other health providers caring for children with pediatric heart disease.