Genetic Basis of Oral Health Conditions

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Preface

Human genetics is the discipline that deals with inheritance as it occurs in human beings. It has components of classical genetics, cytogenetics, molecular genetics, biochemical genetics, genomics, population genetics, developmental genetics, clinical genetics, and genetic counseling, and to the list we can now add epigenetics. My main purpose in writing this book is to document the content of 12 years of developing a *Craniofacial Genetics* course for dental students. The course, as is this book, is designed to use the clinical conditions that interest dentists to present genetic concepts. In that sense, this book brings information that is not typically found in other genetic textbooks or dental textbooks. Similarly to how the course addresses inheritance of dental conditions, the book focuses on inheritance, which is a topic not really explored in publications of the conditions highlighted in the following chapters. This text is the result of more than 20 years of interactions with a number of very talented dental and craniofacial scientists, dentists, and physicians from all continents. I thank Lindsay Carol Brown, Jacob I. Khan, Daryna A. Koval, and Catherine A. Roberts who took my *Craniofacial Genetics* course and then accepted the challenge to carefully revise the text for grammar, style, and flow so it resembles closely the discussions that happen in the classroom.

Chapter 1 is the introduction of all other chapters and although the following chapters are structured to stand alone in their respective subspecialties of dentistry; concepts that can be relevant to all conditions are described throughout in the book. In that sense, this book can be read as a novel. Since multifactorial inheritance is the best explanation for most of the conditions presented in the book, this concept is highlighted in all chapters and the repetition is done purposely. There are also other obviously related topics. Individuals interested in cariology, which is discussed in Chap. 4, will find interesting and relevant content in Chaps. 2, 5, 6, and 10. Craniofacial and dental development disturbances are discussed in Chaps. 2, 3, and 7 and have relevant overlap with Chap. 8 that deals with cancer. Orofacial pain, in Chap. 9, can be complemented by Chaps. 4, 5, 8, and 10.

In writing this volume, I have attempted to produce a comprehensive but at the same time concise, well-referenced text on the genetics of selected dental conditions, each area combined in a separate chapter with depth provided by selecting a few ideas we published for detailed consideration, including a few instances of original data.

Readers who should find this volume of interest include dentists, dental hygienists, craniofacial, oral, and maxillofacial surgeons, craniofacial biologists, psychologists, sociologists, anthropologists, students, and others with special interest in dental, oral, and craniofacial structures.

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2019

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Inheritance in Oral Health Conditions

1

1.1 Introduction

Inheritance of human conditions (both traits and diseases, including those of the craniofacial region) can be divided into three main groups. The vast majority are complex or multifactorial. These are the conditions that are defined by more than one gene, and which can be influenced by the environment (Fig. 1.1; Manolio et al. 2008). Some examples of these conditions include dental caries, periodontitis, dental abnormalities of number and structure, most cases of cleft lip and palate, malocclusion, orofacial pain, oral cancer, and temporomandibular joint dysfunction. The mode of inheritance most people relate to is the one defined by a major single gene, which is called monogenic or Mendelian (Fig. 1.1; Manolio et al. 2008). The reason most know of this type of mode of inheritance is because this is the type typically taught in high school and university curricula. Certain forms of cleft lip, cleft palate, and tooth agenesis are monogenic. The third type refers to conditions that are the result of chromosomal abnormalities. To complicate matters further, there are plenty of exceptions to these three groups.

The etiology and progression of all human diseases likely have a genetic and an environmental component, even if we do not have the tools to identify them. If one lined up all human conditions, three main groups can be identified (Fig. 1.2). At one end of the line, we would see a group of diseases that have a very important genetic component, whereas at the opposite extreme, a group of conditions that have a very important environmental component, and in the vast majority of scenarios, there will be diseases that have both significant genetic and environmental contributions. This chapter will thus aim to describe the main concepts underlying complex inheritance, single gene inheritance, and chromosomal abnormalities. These concepts relate to changes in the DNA sequence and do not include therefore epigenetic changes, which are modifications of gene expression that are not due to alterations of the genetic code itself.

1.2 Complex Inheritance

Complex modes of inheritance explain the large majority of the conditions affecting oral, dental, and craniofacial structures. Most forms of oral cancer, cleft lip and palate, craniosynostosis, craniofacial deformities, malocclusion, periodontitis, dental caries, orofacial pain, temporomandibular joint dysfunction, and developmental dental abnormalities have complex inheritance. Furthermore, various traits such as height (Fig. 1.3), weight, blood pressure, glycemia, response to vaccines, intelligence, behaviors, sexual orientation, and cognition also have a complex mode of inheritance.

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