Cleft Palate and Craniofacial Conditions

A Comprehensive Guide to Clinical Management

Ann W. Kummer
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Ann W. Kummer, PhD, CCC-SLP, FASHA
Senior Director, Division of Speech-Language Pathology (Retired)
Cincinnati Children’s Hospital Medical Center
and
Professor of Clinical Pediatrics and
Professor of Otolaryngology–Head and Neck Surgery
University of Cincinnati College of Medicine
This book is dedicated to the three people who have influenced me most in my life and helped me to be the best that I can be. Without their love and support, I would never have had a career and certainly would not have had the opportunity to write this book . . . now for the fourth time.

The first dedication is to my father, who was a wonderful, caring, and talented otolaryngologist whom I always admired. I always wanted to be like my dad when I was growing up.

The next dedication is to my mother, who was the kindest, most thoughtful, and most caring person I have ever known. Once I grew up, I tried to be more like her. (I’m still trying.)

The final dedication is to my husband, who has loved me, supported me, encouraged me, and helped me to focus and succeed in my career. For that I will be eternally grateful!

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Although current medical technology is not advanced enough to prevent the occurrence of these birth defects, most of the speech and functional impairments associated with craniofacial anomalies can be improved or even corrected with the help of a team of professionals. To provide the type of care that these patients require, this group of professionals must be specialists within their fields. For true quality care, they must have a thorough understanding of the current methods of evaluation and treatment of these patients.

Considering the incidence of clefts and craniofacial anomalies in the general population, however, all healthcare providers should have at least basic knowledge about the management of these patients and appropriate referrals. In particular, speech-language pathologists must be trained in the basic evaluation and treatment and appropriate referrals of individuals with these conditions, especially considering the fact that they often have a significant effect on speech. Certainly, school-based speech-language pathologists are very likely to have children on their caseloads with a history of cleft, craniofacial anomalies, or resonance disorders.

Purpose of This Text

The purpose of this text is to inform, educate, and excite students and professionals in speech-language pathology and the medical and dental professions regarding the management of individuals with clefts or craniofacial anomalies. This text is designed to be a textbook for graduate students and a sourcebook for healthcare professionals who provide services in this area. My goal in writing this text was to provide readers with a great deal of information but in a way that is both interesting and easy to read. As an active
clinician myself, my intent was to make this text a very practical how-to guide as well as a source of didactic and theoretical information.

My ultimate goal with this text is to improve the knowledge of treating professionals who work with individuals who are affected by a cleft or other craniofacial conditions. It is hoped that with this knowledge, they can positively affect the quality of care provided to this population.

Organization
This text was written in a purposeful sequence so that the information from each chapter builds on the information from previous chapters.

Part 1 of this text provides basic information on the normal anatomy of the orofacial structures and the normal physiology of the velopharyngeal valve. Once the normal structures and function are described, information on genetics and patterns of inheritance is covered. The rest of Part 1 consists of information about congenital and acquired craniofacial anomalies and craniofacial syndromes. Once the reader has completed the first section, the reader should have a firm understanding of normal and abnormal facial and velopharyngeal features and the potential causes of congenital and even acquired anomalies.

Part 2 of this text includes chapters on the various functional problems associated with clefts and craniofacial conditions. In particular, this section covers the effects of these anomalies on feeding, speech and language development, psychosocial function, and speech and resonance. After completing the second section, the reader will have an understanding of the number, types, and complexity of the problems that are secondary to clefts and craniofacial conditions. It will then be apparent to the reader that there is a need for multidisciplinary management of these patients in an interdisciplinary setting.

Part 3 of this text covers the various diagnostic methods for assessing speech, resonance, and velopharyngeal function. This section includes

Features
- **Chapter outlines:** The outline of each chapter helps readers navigate through the content and find information quickly.
- **Figures:** This text includes almost 700 figures. These photos and illustrations are meant to enhance comprehension of information and concepts discussed in the chapters.
- **Case studies:** Several chapters include patient case studies to illustrate how chapter information applies to real-life situations.
- **Speech Notes:** Chapters regarding anomalies and surgeries have boxed sections called Speech Notes. These sections highlight how these anomalies or surgeries affect speech and resonance.
For Review and Discussion: A list of questions and topics for discussion is included at the end of each chapter. The purpose of this section is to help the reader synthesize and apply information presented in the chapter. Instructors can also use this section for class discussion, student homework, or essay exams.

Definitions: Selected technical and medical terms are presented in bold and defined within the text and in the glossary.

Glossary: There is a glossary of terms at the end of the text that defines all the medical and technical terms that were bold in the individual chapters. The student may find that studying the glossary is helpful for learning much of the information in the text.

Online Resources
The following resources are available for students and instructors. For more information on how to access these resources, please visit go.jblearning.com/cleftpalate.

Cleft Notes: The Cleft Notes are basic summaries in table format provided for each chapter. There are some compare-and-contrast aspects of these tables to help students assimilate the information. There are two versions of the Cleft Notes—a blank version for students to use when taking notes or studying, and a filled-out version for instructors. By completing the Cleft Notes, the students are engaged in more active learning and have a study guide for test preparation.

Handouts: There are online handouts on a variety of topics that are covered in this text. These handouts are designed primarily for parents but can also be helpful to other professionals who are not familiar with the topic area. The handouts are designed so the user can print them directly from the website.

Videos: There are 295 videos/animations/audio files online. These videos illustrate different types of speech and resonance disorders. There are videos of evaluation techniques, including nasopharyngoscopy, videofluoroscopy, and even nasometry studies. Finally, there are videos of speech therapy techniques that are effective with this population and also with other individuals with speech sound disorders. These videos are designed to help the viewer develop diagnostic and treatment skills by watching and listening to each video as many times as necessary. Because these videos are short and carefully edited, they facilitate better learning than direct observation in a clinic.

PowerPoint Presentations: There are PowerPoint presentations, which include important figures and photos, for each chapter. These presentations can be used by the instructor for classroom teaching.

Testbank: Assessment questions are available in a variety of different formats, including multiple choice, labeling, matching, and true/false.

Image Library: The image library provides access to all the art in the textbook. This resource can be searched using keywords and subject areas.

New to This Edition

Photos: Many new photos have been added, most of which are in color.

Drawings: Anatomy figures have been re-rendered for consistency and improved quality.

Tables: Many chapters have information summarized in tables for easy learning. There are also tables of terms for normal and abnormal craniofacial, oral, dental, and pharyngeal structures and anomalies.

Chapter Text: Chapters have been heavily edited with a focus on making the information clear, concise, and easy to read.

Chapter Order: The chapter order has been reorganized for better flow.

Research Updates: Information within the text and the references have been updated to reflect current research and literature.
Format Notes

Service providers must be sensitive to the emotional and psychological needs of the patient. Sensitivity to the feelings of the patient is often overlooked by well-meaning service providers. It is easy to forget that we deal with real people, not just interesting cases. This lack of sensitivity is sometimes reflected in the terminology that is used in the literature and in daily use. I recall listening to a speech given by an adult who was born with a cleft palate. As he described his childhood, he pointed out that being called a “cleft palate child” evoked very negative feelings. Fortunately, this type of phrase is becoming “politically incorrect,” just as the term “harelip” has in the past. Using the anomaly as an adjective to describe the individual is certainly insensitive to the feelings of the person who was born with this anomaly. Therefore, it is preferable to use “patient-first” terminology as in “child with a cleft.”

The reader will note that the word “child” is frequently used throughout the text for the individual with the anomaly. This is because the speech and resonance disorders secondary to cleft lip/palate and craniofacial anomalies are usually addressed during childhood. However, it should be understood that this information also applies to adults with the same anomalies.

Acknowledgments and Thanks

There are so many people that I would like to acknowledge for their help with this edition of the text. Many thanks go to the members of our VPI/Resonance Team at Cincinnati Children’s, including Jenn Marshall, Shyla Miller, Cara Werner, Margaret (Meg) Wilson, and Sarah Woodhouse. They were very helpful in providing feedback, developing the Cleft Notes, and reviewing videos. Special thanks go to Cara Werner, who proofread the entire manuscript and online content. She also provided very valuable suggestions. I would like to thank the members of the Cleft and Craniofacial Center at Cincinnati Children’s for being such great colleagues, mentors, and friends! I have learned so much through our professional interactions over the years. Finally, I’m very grateful to Laura Pagluica and her entire team at Jones & Bartlett including Rebecca Feeney, Vanessa Richards, Thais Miller, and Troy Liston. It was such a great experience working with them. I have been very impressed with the entire company and the quality of their products.

Final Words

I am very grateful for the opportunity to share with you what I have learned through my clinical practice over the years. I sincerely hope that through this text you will be educated, enlightened, and inspired to provide superior clinical services for individuals with clefts or other craniofacial conditions.
## Vowels

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<thead>
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<th>Symbol</th>
<th>Examples</th>
</tr>
</thead>
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<tr>
<td>/ɪ/</td>
<td>bee, see</td>
</tr>
<tr>
<td>/æ/</td>
<td>hat, cat</td>
</tr>
<tr>
<td>/ɑ/</td>
<td>father, pot</td>
</tr>
<tr>
<td>/ə/</td>
<td>teacher, mother</td>
</tr>
</tbody>
</table>

## Consonants

<table>
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<th>Symbol</th>
<th>Letters</th>
<th>Examples</th>
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</thead>
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<td>glottal stop</td>
<td>button, mitten</td>
</tr>
<tr>
<td>/ʃ, s/</td>
<td>sh</td>
<td>shoe</td>
</tr>
<tr>
<td>/ʒ/</td>
<td>zh</td>
<td>measure</td>
</tr>
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<td>/ð/</td>
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<td>then</td>
</tr>
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<td>/ŋ/</td>
<td>ng</td>
<td>sing</td>
</tr>
</tbody>
</table>

Note: This key includes only the phonetic symbols used in this text.
Ann W. Kummer, PhD, CCC-SLP, FASHA, is the former senior director of the Division of Speech-Language Pathology at Cincinnati Children's. Under her direction of over 35 years, the speech-language pathology program at Cincinnati Children's became the largest pediatric program in the nation and one of the most respected. Dr. Kummer is professor of clinical pediatrics and professor of otolaryngology at the University of Cincinnati (UC), College of Medicine.

Dr. Kummer has done hundreds of national and international lectures and seminars in the areas of cleft palate and craniofacial anomalies, resonance disorders, velopharyngeal dysfunction, and business practices in speech-language pathology. She has taught the craniofacial anomalies course for five universities. She has also written numerous professional articles and 22 book chapters in speech pathology and medical texts. In addition to this text, she is one of the authors of the text Business Practices: A Guide for Speech-Language Pathologists. Dr. Kummer is the co-developer of the Simplified Nasometric Assessment Procedures (SNAP) test (1996) and author of the SNAP-R (2005), which is incorporated in the Nasometer™ equipment (PENTAX Medical). She holds a patent on the nasoscope, which is marketed as the Oral & Nasal Listener™ (Super Duper, Inc.). She was one of the main developers of workflow software that won the 1995 International Beacon Award through IBM/ Lotus. (Derivative software is marketed by Chart Links.)

Dr. Kummer has received numerous honors, including Honors of the Southwestern Ohio Speech-Language-Hearing Association (1995); Honors of the Ohio Speech-Language-Hearing Association (OSLHA) (1997); Distinguished Alumnus Award from the Department of Communication Sciences and Disorders, University of Cincinnati (1999); Fellow of the American Speech-Language-Hearing Association (ASHA) (2002); named one of the top 25 most influential therapists in the United States by Therapy Times (2006); Honors for Distinguished Service, Department of Otolaryngology–Head and Neck Surgery, University of Cincinnati (2007); named one of the 10 Most Inspiring Women in Cincinnati (2007); inducted into the National Academy of Inventors, Cincinnati Chapter (2010); Distinguished Alumnus Award, College of Allied Health, University of Cincinnati (2012), Elwood Chaney Outstanding Clinician Award from the Ohio Speech-Language-Hearing Association (OSHLA) (2012); Annie Glenn National Leadership Award, Ohio School Speech Pathology Educational Audiology Coalition (OSSPEAC) (2014); and the Media Outreach Champion award from ASHA (2014). In 2017, she received Honors of the Association from ASHA, the highest award given by the association.
CONTRIBUTORS

It is with great appreciation that I would like to thank the various contributors to this edition. Their expertise was essential in making the contents of many of the chapters both accurate, current, and clinically relevant. I will be forever in their debt for their contributions.

Haithem Elhadi Babiker, MD, DMD, FAAP, FACS
Assistant Professor
Plastic and Oral-Maxillofacial Surgeon
University of Cincinnati College of Medicine
Division of Plastic Surgery
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 17

David A. Billmire, MD
Emeritus Professor of Clinical Surgery
University of Cincinnati College of Medicine
Director of Plastic Surgery
Shriners Hospitals for Children
Cincinnati, Ohio
Chapter 17

Richard Campbell, DMD, MS
Assistant Professor
University of Cincinnati College of Medicine
Director, Orthodontics
Division of Pediatric Dentistry
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 6

Julia Corcoran, MD
Adjunct Associate Professor of Surgery
Feinberg School of Medicine Northwestern University
Attending Surgeon
Shriners Hospital for Children - Chicago
Chicago, Illinois
Chapter 17

Murray Dock, DDS, MSD
Associate Professor of Clinical Pediatrics
University of Cincinnati College of Medicine
Division of Pediatric Dentistry
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 6

Robert J. Hopkin, MD
Associate Professor of Clinical Pediatrics
University of Cincinnati College of Medicine
Division of Human Genetics
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 2

Deepak Krishnan, DDS, FACS
Associate Professor of Surgery & Residency Program Director
Division of Oral & Maxillofacial Surgery
University of Cincinnati Medical Center
Cincinnati, Ohio
Chapter 17

Patricia K. Marik, PsyD
Pediatric Psychologist
Psychiatry and Behavioral Medicine
Children's Hospital of Wisconsin
Assistant Clinical Professor of Psychiatry
Medical College of Wisconsin
Wauwatosa, Wisconsin
Chapter 9

Claire K. Miller, PhD, MHA
Program Director, Aerodigestive and Esophageal Center
Clinical/Research Speech-Language Pathologist
Division of Speech-Language Pathology
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 7
Contributors

Howard M. Saal, MD
Professor of Pediatrics
University of Cincinnati College of Medicine
Director, Clinical Genetics
Division of Human Genetics
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 4

Janet R. Schultz, PhD
Professor
Psychology Department
Xavier University
Cincinnati, Ohio
Chapter 9

J. Paul Willging, MD
Professor
Department of Otolaryngology–Head and Neck Surgery
University of Cincinnati College of Medicine
Cincinnati Children's Hospital Medical Center
Cincinnati, Ohio
Chapter 5
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Kate Bunton
University of Arizona

Marie E. Byrne
Mississippi University for Women

Ellen R. Cohn
University of Pittsburgh

Karen Copple
Eastern New Mexico University

Ramesh Kaipa
Oklahoma State University

Ciara Leydon
Sacred Heart University

Julie Owen Morris
University of Central Oklahoma

Amy Shollenbarger
Arkansas State University Jonesboro

Daniel Valentine
University of Motevallo
PART 1

Normal and Abnormal Craniofacial Structures

CHAPTER 1  Anatomy and Physiology
CHAPTER 2  Genetics and Patterns of Inheritance
CHAPTER 3  Clefts of the Lip and Palate
CHAPTER 4  Dysmorphology and Craniofacial Syndromes
CHAPTER 5  Facial, Oral, and Pharyngeal Anomalies
CHAPTER 6  Dental Anomalies
CHAPTER 1

Anatomy and Physiology

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Craniofacial Bones and Sutures
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Nose and Nasal Cavity
Lips

Intraoral Structures
Tongue
Faucial Pillars, Tonsils, and Oropharyngeal Isthmus
Hard Palate
Velum
Uvula

Pharyngeal Structures
Pharynx
Eustachian Tube

PHYSIOLOGY

Velopharyngeal Valve
Velar Movement
Lateral Pharyngeal Wall Movement
Posterior Pharyngeal Wall Movement

Muscles of the Velopharyngeal Valve
Velopharyngeal Motor and Sensory Innervation

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Patterns of Velopharyngeal Closure
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Subsystems of Speech: Putting It All Together
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Phonation
Prosody
Resonance and Velopharyngeal Function
Articulation
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Summary
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INTRODUCTION

The nasal, oral, and pharyngeal structures are all very important for normal speech and resonance. Unfortunately, these are the structures that are commonly affected by cleft lip and palate and other craniofacial anomalies. Before the speech-language pathologist can fully understand the effects of oral and craniofacial anomalies on speech and resonance, a thorough understanding of normal structure (anatomy) and normal function (physiology) of the oral structures and the velopharyngeal valve is essential.

This chapter reviews the basic anatomy of the structures of the orofacial and velopharyngeal complex as they relate to speech production. The physiology of the subsystems of speech, including the velopharyngeal mechanism, is also described. For more detailed information on anatomy and physiology of the speech articulators, the interested reader is referred to other sources (Cassell & Elkadi, 1995; Cassell, Moon, & Elkadi, 1990; Dickson, 1972; Dickson, 1975; Dickson & Dickson, 1972; Dickson, Grant, Sicher, Dubrul, & Paltan, 1974; Dickson, Grant, Sicher, Dubrul, & Paltan, 1975; Huang, Lee, & Rajendran, 1998; Kuehn, 1979; Maue-Dickson, 1977; Maue-Dickson, 1979; Maue-Dickson & Dickson, 1980; Maue-Dickson, Dickson, & Rood, 1976; Moon & Kuehn, 1996; Moon & Kuehn, 1997; Moon & Kuehn, 2004; Perry, 2011; Seikel, King, & Drumright, 2005).

ANATOMY

Craniofacial Structures

Although the facial structures are familiar to all, some aspects of the face are important to point out for a thorough understanding of congenital anomalies and clefting. The normal facial landmarks can be seen on FIGURE 1-1. The reader is encouraged to identify the same structures on the photo of the normal infant face shown in Figure 1-1B.

Craniofacial Bones and Sutures

The bones of the cranium include the frontal bones, which cover the anterior portion of the brain; the parietal bones, which cover the top and sides of the cranium; the temporal bones, which form the sides and base of the skull; and finally, the occipital bone, which forms the back of the skull (FIGURE 1-2).

Each bone is bordered by an embryological suture line. The frontal bones are divided in midline by the metopic suture and bordered posteriorly by the coronal suture. The coronal suture is across the top of the skull horizontally (like a crown) and separates the frontal bones and parietal bones. The sagittal suture crosses the skull vertically and, therefore, divides the two parietal bones. Finally, the lambdoid suture is between the parietal, temporal, and occipital bones.

FIGURE 1-1 (A) Normal facial landmarks. Note the structures on the diagram. (B) Normal face. Try to locate the same structures on this infant's face.
The anterior fontanelle ("soft spot" of an infant) is on the top of the skull at the junction of the frontal and the coronal sutures. The metopic suture closes between 3 and 9 months of age. The coronal, sagittal, and lambdoid sutures close between 22 and 39 months of age.

The facial bones include the zygomatic bone (also called malar bone), which forms the cheeks and the lateral walls of the orbits; the maxilla, which forms the upper jaw; and the mandible, which forms the lower jaw.

**Ear**

The ear has three distinct parts—the external ear, the middle ear, and the inner ear (FIGURE 1-3). A description of the anatomy of each part follows.

The external ear consists of the pinna and the external auditory canal. The pinna is the delicate cartilaginous framework of the external ear. It functions to direct sound energy into the external auditory canal, which is a skin-lined canal leading from the opening of the external ear to the eardrum.

The middle ear is a hollow space within the temporal bone. The mastoid cavity connects to the middle ear space posteriorly and consists of a collection of air cells within the temporal bone. Both the middle ear and mastoid cavities are lined with a mucous membrane (also known as mucosa), which consists of stratified squamous epithelium and lamina propria. (This should not be confused with mucus, which is the clear, viscid secretion from the mucous membranes.)

The tympanic membrane, also called the eardrum, is considered part of the middle ear. The tympanic membrane transmits sound energy through the ossicles to the inner ear. The ossicles are tiny bones within the middle ear and are called the malleus, incus, and stapes. The malleus (also known as the hammer) is firmly attached to the tympanic membrane. The incus (also known as the anvil) articulates with both the malleus and the stapes. The stapes acts as a piston to create pressure waves within the fluid-filled cochlea, which is part of the inner ear. The tympanic membrane and ossicles act to amplify the sound energy and efficiently introduce this energy into the liquid environment of the cochlea.

The eustachian tube (also known as the auditory tube) connects the middle ear with...
the nasopharynx. The end of this tube, which terminates in the nasopharynx, is closed at rest but opens during swallowing. When it opens, it provides ventilation for the middle ear and mastoid cavities and results in equalization of air pressure between the middle ear and the environment (Cunsolo, Marchioni, Leo, Incorvaia, & Presutti, 2010; Licameli, 2002; Smith, Scoffings, & Tysome, 2016; Yoshida, Takahashi, Morikawa, & Kobayashi, 2007). It also allows drainage of fluids and debris from the middle ear space. (More information about the eustachian tube is noted in the Pharyngeal Structures section.)

The inner ear consists of the cochlea and semicircular canals. The cochlea is composed of a bony spiral tube that is shaped like a snail’s shell. Within this bony tube are delicate membranes separating the canal into three fluid-filled spaces. The organ of Corti is the site where mechanical energy introduced into the cochlea is converted into electrical stimulation. This electrical impulse is conducted by the auditory nerves to the auditory cortex, which results in an awareness of sound. Inner and outer hair cells (sensory cells with hair-like properties) of the cochlea may be damaged by a variety of mechanisms, leading to sensorineural hearing loss.

In addition to hearing, the inner ear is responsible for balance. The semicircular canals are the loop-shaped tubular parts of the inner ear that provide a sense of spatial orientation. They are oriented in three planes at right angles to one another. The saccule and utricle are additional sensory organs within the inner ear. Hair cells within these organs have small calcium carbonate granules that respond to gravity, motion, and acceleration.

### Nose and Nasal Cavity

The nose begins at the nasal root, which is the most depressed, superior part of the nose and at the level of the eyes. The nasal bridge is the saddle-shaped area that includes the nasal root and the lateral aspects of the nose. Finally, the nasion is a midline point just superior to the nasal root and overlying the nasofrontal suture.