FIFTH EDITION

Cleft Palate and Craniofacial Conditions

A Comprehensive Guide to Clinical Management

Ann W. Kummer, PhD, CCC-SLP, FASHA

Plural Publishing Inc.

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PREFACE

Anticipating the birth of a new baby is usually a very exciting time of life. The expectant couple does many things to prepare for the baby, including setting up a nursery, gathering baby clothes and diapers, and deciding on a name. The parents expect to have a normal baby, with 10 fingers, 10 toes, and an intact face. They are totally unprepared for the possibility of a different outcome. Unfortunately, not all babies are born with perfect structures. When a child is born with cleft lip, cleft palate, or other craniofacial anomalies, the experience is profoundly shocking, especially because the face is involved. What was expected to be a joyful and celebratory moment can quickly become stressful and emotionally overwhelming for parents and family members. In the early hours and days, it may be difficult for parents to look beyond the anomaly and truly appreciate their newborn.

Cleft lip with or without cleft palate is the second most common birth defect. In fact, about 1 in every 700 children born in the United States each year has a cleft of the lip and/or palate. Many of these children, particularly those with cleft palate only, have other associated malformations. In fact, cleft palate is a characteristic of well over 400 recognized syndromes.

In past generations, the treatment goal for individuals with cleft palate was simply acceptable or intelligible speech, as normal speech was rarely achievable. Over the past several decades, however, our understanding of the velopharyngeal mechanism has expanded significantly. Advances in evaluation methods and surgical techniques have also contributed to improved outcomes. As a result, most children born with nonsyndromic cleft lip and/or palate can ultimately achieve normal speech with appropriate care. For this reason, every effort should be made to attain normal speech (without nasal emission) and normal resonance (without hypernasality or hyponasality) whenever possible.

Considering the incidence of clefts and other craniofacial conditions in the general population, all healthcare providers should have at least basic knowledge of their management and appropriate referrals when needed. This is especially true for speech-language pathologists (SLPs), who must be trained in the evaluation, treatment, and referral needs of individuals with these conditions, given their significant impact on speech and resonance. School-based SLPs, in particular, are likely to have children on their caseloads with a history of cleft lip/palate, craniofacial anomalies, or resonance disorders, and therefore need this foundational knowledge. As such, this book is intended to provide essential information for professionals who frequently, or even occasionally, work with children who have structural conditions that affect speech.

Purpose of This Book

The purpose of this book is to inform, educate, and excite students and professionals in speech-language pathology and also those in the medical and dental professions regarding the management of individuals with clefts or other craniofacial conditions.

This book is ideal for a graduate-level course on cleft palate and craniofacial conditions. Drawing on my long-term experience teaching graduate courses on this topic, this book now contains all of the resources needed by instructors and students. Although this is the most comprehensive text currently available, it is structured so that it can be used as a full course or part of another course.

This book also serves as a source book for healthcare professionals who provide clinical services to affected patients. Because of my extensive clinical experience, this textbook includes not only didactic and theoretical information, but also includes practical "how-to" guides for clinicians, including a comprehensive collection of speech therapy strategies. Almost 300 clinical short videos of speech disorders, evaluation techniques, and therapy strategies are also provided.

My ultimate goal with this book is to improve the knowledge of treating professionals who work with individuals that are affected by clefts or other craniofacial conditions. It is hoped that with this knowledge, they can positively impact the quality of care provided to affected children.

New to this Edition

The fifth edition has been extensively updated to reflect current clinical practice and research. It includes expanded coverage of speech, resonance, and velopharyngeal dysfunction; updated discussions of genetics, craniofacial syndromes, development, and surgical management; and additional evidence-based strategies for speech therapy and carryover. The text has also been reorganized and streamlined for greater clarity, with new summary tables, clinical photographs, illustrations, diagnostic examples, and enhanced online learning resources.

Organization

This book 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 I consists of information about congenital and acquired craniofacial anomalies and craniofacial syndromes. Once the first section is completed, the reader should have a firm understanding of the types of facial, oral and velopharyngeal conditions, as well as their potential causes.

Part 2 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 finally, 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 covers the various diagnostic methods for assessing speech, resonance, and velopharyngeal function. This section includes the perceptual examination of speech and resonance, and the physical examination of the oral cavity and other orofacial structures. There is an overview chapter on instrumentation that is sufficient for graduate students. There are also individual chapters on the vari-

ous types of instrumental procedures. These chapters are very detailed and are written to provide specific information for practicing clinicians who will be using these procedures.

Part 4 covers the treatment of speech and resonance disorders secondary to clefts, craniofacial anomalies, and velopharyngeal dysfunction. This section includes surgical management, prosthetic management, and speech therapy. The speech therapy chapter includes specific therapy strategies for achieving placement, regardless of the misarticulation or cause. In addition, there is a section on achieving carryover using motor learning and motor memory principles.

Part 5 of this text is short, but important, because it emphasizes the fact that many disciplines are needed to provide care for patients affected by clefts or craniofacial conditions. The reader will complete this section with an understanding that quality patient care requires interdisciplinary interaction and collaboration in the assessment and treatment of these patients.

Features

Features within the text:

- **Chapter outlines:** The outlines of each chapter help readers navigate through the content and find information quickly.
- **Figures:** This text includes close to 700 photographs and illustrations that are meant to enhance comprehension of information and concepts discussed in the chapters.
- **Tables:** Each chapter has one or more tables that summarize important information that is described in the text.
- **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. Professors can also use this section for class discussion, student homework, or for essay exams.
- **Definitions:** Selected technical and medical terms are presented in italics and defined in the text and in the glossary.
- **Glossary:** There are more than 750 medical and technical terms that are defined in the glossary of the book. All of these terms are also italicized and defined in the individual chapters. The student may find that studying the glossary is helpful in learning much of the information in this book.

Online Resources

Online resources for instructors include:

- **PowerPoint® Presentations:** Each chapter includes a PowerPoint® presentation, which contains key information from the chapter and relevant figures, photos, and clinical videos. These PowerPoints can be used by instructors for classroom teaching.
- **Recorded Lectures:** Each PowerPoint® presentation also includes a recorded lecture given by the author. For convenience, instructors may choose to use these recorded lectures for their students, rather than present the lecture themselves.

- **Test Bank:** There is a quiz with many multiple-choice questions for each chapter. These can be used by the instructor for exams or can be given to the students to use for studying the material.
- **Image Bank:** The images from the text are made available in PowerPoint slides so instructors can use them as needed for instruction.
- **Syllabus:** A syllabus is provided for instructors of a graduate-level course on cleft palate and craniofacial conditions. This can be modified based on the number of credit hours earned for the course.
- ***Cleft Notes*:** *Cleft Notes* are structured tables that students can complete while reading each chapter. The tables require active learning and help the student to remember, organize, and internalize complex information efficiently. Each chapter has two formats: a blank version for students to complete during independent study, and a fully completed version for instructors. Completion of the *Cleft Notes* may be required as a homework assignment, allowing students to earn points toward their grade with each submission. Alternatively, instructors may choose to provide the completed *Cleft Notes* to students so that they can use them as study guides.
- Instructors also have access to the following resources.

Online resources for students and clinicians:

- **Videos:** There are more than 280 short, carefully edited patient videos available online. Many of these clips illustrate a wide range of speech and resonance disorders. Others demonstrate evaluation procedures, including nasopharyngoscopy, videofluoroscopy, and nasometry. In addition, there are videos showing effective speech therapy techniques for this population and for individuals with other speech sound disorders. These videos are designed to help viewers build diagnostic and treatment skills by allowing them to watch and listen as many times as needed. Because the videos are brief, focused, and well edited, they often provide a more efficient learning experience than direct clinic observation.
- **Quizzes:** There is a quiz with many multiple-choice questions for each chapter. These can be used by the instructor for exams or can be given to the students to use for studying the material.
- **Handouts:** There are online handouts on a variety of topics that are covered in this book. 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, as long as the heading, logo, and content are not altered.
- ***Cleft Notes:*** Students can download blank versions to complete during independent study.

Final Words

Speaking for both myself and for all the contributors, we are grateful for the opportunity to present this information to you. We are hopeful that you will become educated, enlightened, and inspired to provide superior clinical services for individuals with clefts or other craniofacial conditions.

ABOUT THE AUTHOR



Ann W. Kummer, PhD, CCC-SLP, FASHA, retired as Senior Director of the Division of Speech-Language Pathology at Cincinnati Children's Hospital Medical Center after more than four decades of service. During her tenure, the program became the nation's largest and one of its most respected. She also served as Professor of Clinical Pediatrics and Professor of Otolaryngology at the University of Cincinnati College of Medicine. She is now Professor Emerita.

During her career, Dr. Kummer has delivered over 400 national and international lectures on cleft palate, craniofacial anomalies, resonance disorders, velopharyngeal dysfunction, and professional practices. She taught a graduate-level course on craniofacial conditions at five universities for many years. In addition to this textbook, now in its 5th edition, Dr. Kummer has published over 30 book chapters and over 70 peer-reviewed articles. Additional contributions include development of the SNAP-R Test, which is incorporated into the Pentax Medical Nasometer software, and co-development of professional workflow software, which received the International IBM Beacon Award in 1996.

Dr. Kummer has received numerous professional honors, including state and national association awards, distinguished alumni recognitions, and ASHA's Honors of the Association, the organization's highest award.

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Anatomy and Physiology

With Contributions from Jamie Perry, PhD

CHAPTER 1

CHAPTER OUTLINE

Introduction

ANATOMY

Craniofacial Structures
- Craniofacial Bones and Sutures
- Eyes
- Ears
- Nose and Nasal Cavity
- Lips

Intraoral Structures
- Tongue
- Tonsils and Adenoids
- Hard Palate
- Velum and 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

Variations in Velopharyngeal Closure
- Patterns of Velopharyngeal Closure
- Pneumatic Versus Nonpneumatic Activities
- Timing of Closure
- Height of Closure
- Firmness of Closure
- Effect of Rate and Fatigue
- Changes with Growth and Age

Physiological Subsystems of Speech
- Respiration
- Phonation
- Prosody
- Resonance and Velopharyngeal Function
- Articulation
- Subsystems as "Team Players"

Summary

For Review and Discussion

References

INTRODUCTION

The nasal, oral, and pharyngeal structures play a critical role in normal speech production 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 et al., 1990; Dickson, 1972, 1975; Dickson & Dickson, 1972; Dickson et al., 1974, 1975; Huang et al., 1998; Kuehn, 1979; Maue-Dickson, 1977, 1979; Maue-Dickson & Dickson, 1980; Maue-Dickson et al., 1976; Moon & Kuehn, 1997, 2004; Perry, 2011; Seikel et al., 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 clefts. Normal facial landmarks can be seen in Figure 1–1A and Figure 1–1B. The reader is encouraged to identify the same structures in the photo of the normal infant face shown in Figure 1–1B.

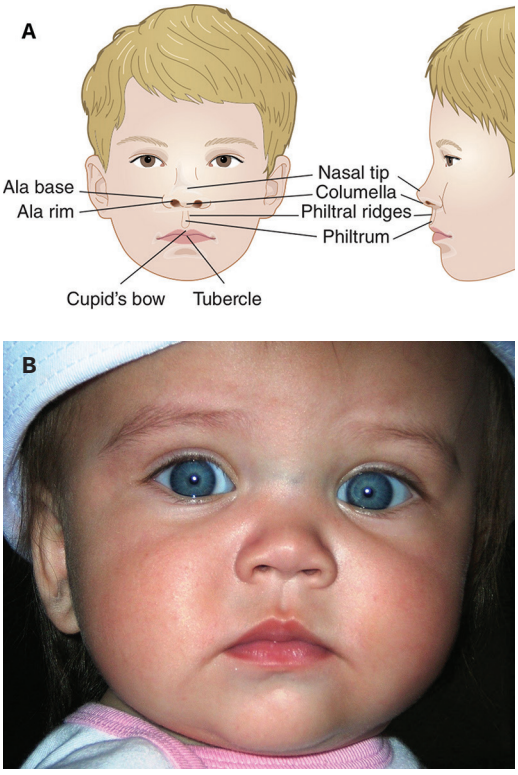


FIGURE 1–1. Normal facial landmarks. **A.** Note the structures on the diagram. **B.** Normal face. Try to locate the same structures on this infant's face.

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).

Cranial bones meet at fibrous joints known as sutures. The frontal bones are sepa-

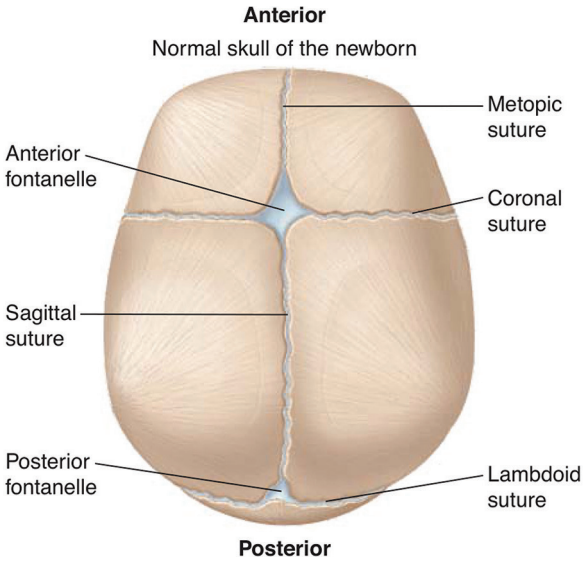


FIGURE 1–2. Cranial suture lines.

rated at the midline by the *metopic suture* and are bordered posteriorly by the coronal suture. The *coronal suture* extends across the top of the skull horizontally (like a crown) and separates the frontal bones and parietal bones. The *sagittal suture* runs along the midline in an anteroposterior direction and divides the two parietal bones. Finally, the *lambdoid suture* separates the parietal bones from the occipital bones.

The *anterior fontanelle* (commonly referred to as the "soft spot" of an infant) is on the top of the skull at the junction of the sagittal, frontal, and coronal sutures. The metopic suture (fibrous joint in the midline of the frontal bone) typically closes between three and nine months of age. In contrast, the coronal, sagittal, and lambdoid sutures generally remain open throughout childhood and adolescence and may not fully fuse until adulthood.

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

Eyes

The external anatomy of the eyes includes the eyelids and eyelashes. The eyelids consist of movable folds of skin and muscle. Their purpose is to shield the eye from debris and help distribute the tear film with each blink. The eyelashes act as tactile sensors that trigger a protective blink reflex. The opening between the upper and lower lids is called the *palpebral fissure*. It varies in width and slant and can offer important clinical clues in the craniofacial assessment. A *canthus* (plural: *canthi*) is the corner of the eye where the upper and lower eyelids meet. There are two canthi in each eye: the medial (inner) canthus near the nose, which connects to the tear-bearing lacrimal glands, and the lateral (outer) canthus near the temple, which aids in eyelid movement. Surrounding each eye is the bony orbit, which provides the eye with external structural support.

Ears

The ear has three distinct parts—the external ear, the middle ear, and the inner ear (Figure 1–3). The anatomy of each part is described below.

The *external ear* consists of two main structures: 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 an air-filled space within the temporal bone. The *mastoid cavity* connects to the middle ear space posteriorly and contains a collection of air cells within the temporal bone. Both the middle ear and mastoid cavities are lined with a *mucous membrane* (*mucosa*), which is composed primarily of simple cuboidal or columnar epithelium with an underlying lamina propria, whereas stratified squamous epithelium is limited to the external auditory canal and the lateral surface of the tympanic membrane. (This should not be confused with *mucus*, which is the clear, viscid secretion from the mucous membranes.)

The *tympanic membrane*, also called the *eardrum,* forms the lateral boundary of the middle ear and separates it from the external auditory canal. 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* (hammer) is firmly attached to the tympanic membrane. The *incus* (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. Together, the tympanic membrane and ossicles amplify sound energy and efficiently transmit it into the cochlea.

The *eustachian tube* connects the middle ear with the nasopharynx. The end of this

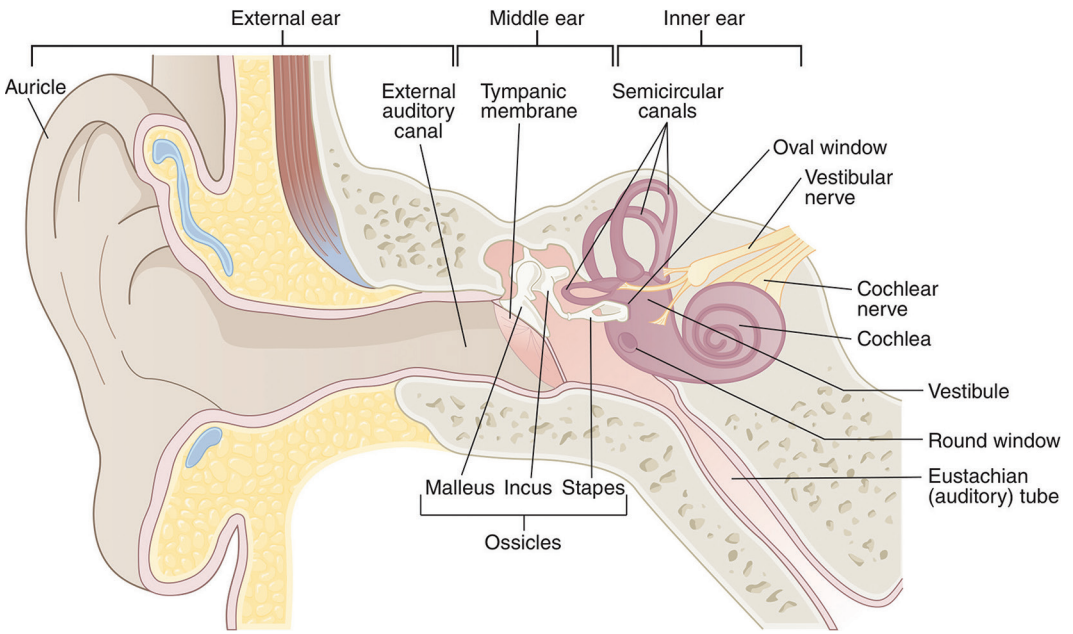


FIGURE 1–3. Ear showing external, middle, and inner ear structures and the eustachian tube.

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 et al., 2010; Licameli, 2002; Yoshida et al., 2007). It also allows drainage of fluids and debris from the middle ear space. (More information about the eustachian tube is noted below under Pharyngeal Structures.)

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 and contains membranous structures that divide it into three separate fluid-filled spaces. The *organ of Corti*, located within the cochlea, is the sensory structure responsible for converting mechanical sound energy into electrical signals that are transmitted via the cochlear nerve to the auditory cortex, resulting in the perception 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 portion of the external nose and lies at the level of the eyes. The nasal root overlies the *nasion,* a midline craniometric point at the junction of the frontal bone and the paired nasal bones at the nasofrontal suture. The *nasal bridge* is the saddle-shaped area that includes the nasal root and the lateral aspects of the nose.

The nostrils are separated externally by the *columella* (little column). The anterior nasal spine of the maxilla forms a base for the columella. The columella is like a supporting column in that it provides support for the nasal tip. The columella must be long enough so that the nasal tip has an appropriate degree of projection. Ideally, the columella is straight and backed by a straight nasal septum.

The nostrils are frequently referred to as *nares*, although an individual nostril is a *naris*. The *ala nasi* (*ala* is Latin for "wing") is the outside curved side of the nostril. The *alae* (plural version of *ala*) are the two curved sides of each nostril. The *alar rim* is the outside curved edge that surrounds the opening to the nostril on either side, and the *alar base* is the area where the ala meets the upper lip. The *nasal sill* forms the base of the nostril opening. The *nasal vestibule* is the most anterior part of the nasal cavity and is enclosed by the cartilages of the nose.

The opening to the bony inside of the nose is called the *piriform* (meaning "pear-shaped") *aperture*. This pear-shaped opening (thus the name) is bordered by the nasal and maxillary bones (Figure 1–4).

The *nasal septum* is located in the midline of the nose and serves to separate the right and left nasal cavities (Figure 1–5). It consists of both cartilage in the anterior portion of the nose and bone in the posterior portion. The *quadrangular cartilage* forms the anterior nasal septum and projects anteriorly to the columella. The bones of the septum include the maxillary crest, the vomer, and the perpendicular plate of the ethmoid. The *vomer* is a trapezoidal-shaped bone in the nasal septum which is positioned perpendicular to the palate. Its lower border fits within a groove on the nasal aspect of the maxilla created by the *median (intermaxillary) palatine suture*, which is the midline joint where the right and left