

# 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

## CHAPTER OUTLINE

#### **INTRODUCTION**

#### ANATOMY

#### **Craniofacial Structures**

Craniofacial Bones and Sutures Ear 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

#### 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

#### Subsystems of Speech: Putting It All Together

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 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, 1972; Dickson, 2008, 200

## ANATOMY

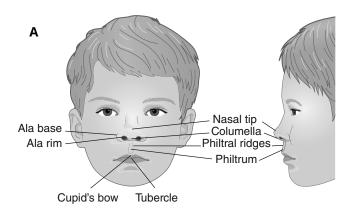
## **Craniofacial Structures**

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.

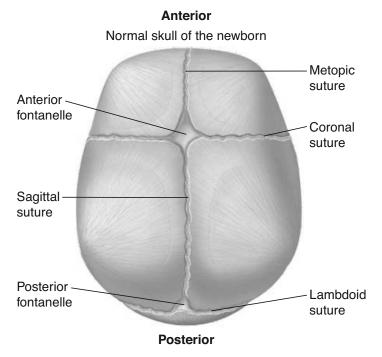


FIGURE 1-2 Cranial suture lines.

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

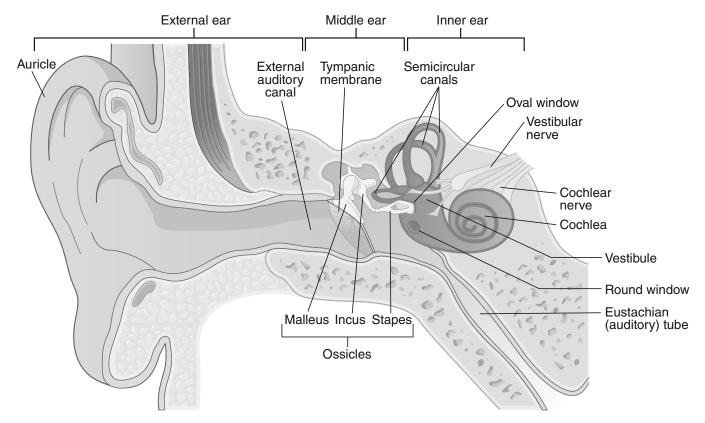


FIGURE 1-3 Ear showing external, middle, and inner ear structures and the eustachian tube.

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. 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** is 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 **pyriform aperture** (also spelled as "piriform," means "pear shaped"). This pearshaped 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 nasal cavity into two nostrils (**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

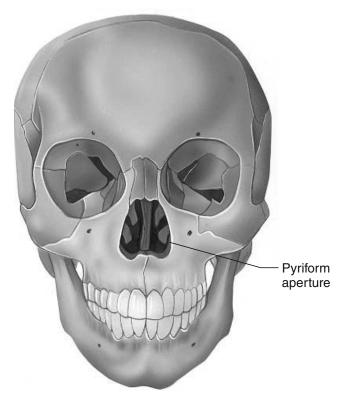


FIGURE 1-4 Pyriform aperture.

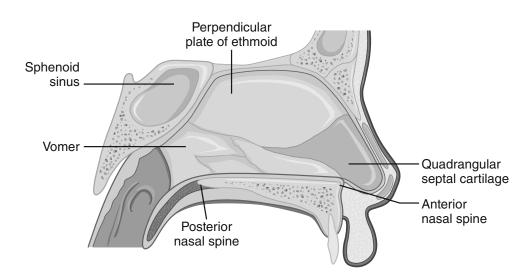


FIGURE 1-5 The nasal septum and related structures.

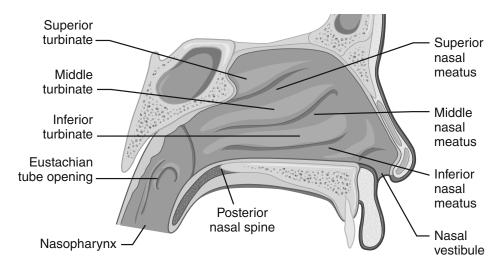


FIGURE 1-6 The lateral wall of the nose showing the turbinates.

**vomer** is a trapezoidal-shaped bone in the nasal septum. It is positioned perpendicular to the palate, and as such, the lower portion of the vomer fits in a groove formed by the median palatine suture line on the nasal aspect of the maxilla. The **perpendicular plate of the ethmoid** projects downward to join the vomer. It is not uncommon for the nasal septum to be less than perfectly straight, particularly in adults. The nasal septum is covered with mucous membrane, which is the lining tissue of the nasal cavity, oral cavity, and the pharynx.

The nasal turbinates, also called nasal conchae (concha, singular), are paired bony structures within the nose that are covered with mucosa (FIGURE 1-6). They are attached to the lateral walls of the nose and protrude medially into the nasal cavity. They are long, narrow, shelf-like, and curled in shape. As air flows underneath them, the curled shape helps to create turbulent airflow (thus the name "turbinate") to maximize contact of the inspired air with the nasal mucosa.

The nasal turbinates within the nose have three distinct functions. First, the mucus that covers the nasal mucosa filters inspired air of gross contaminants by trapping particulate contaminants. Second, the turbinates warm and humidify the inspired air. Finally, the turbinates deflect air superiorly in the nose in order to enhance the sense of smell. Directly under the turbinates are the superior, middle, and inferior **nasal meatuses** (meatus, singular), which are the openings or passageways through which the air flows. At the back of the nasal cavity, on each side of the posterior part of the vomer, is a **choana** (choanae, plural), which is a funnel-shaped opening that leads to the nasopharynx.

Finally, the **paranasal sinuses** are air-filled spaces in the bones of the face and skull. These structures are each about the size of a walnut. There are four pairs of paranasal sinuses: frontal sinuses (in the forehead area), ethmoid sinuses (between the eyes), maxillary sinuses (under the cheeks), and sphenoid sinuses (deep in the skull). These sinuses are connected to the nose by a small opening called an **ostium** (ostia, plural). **FIGURE 1-7** shows the sinuses through computed tomography.

### Lips

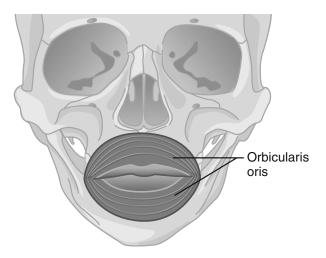
The features of the upper lip can be seen in Figure 1-1A. An examination of the upper lip reveals the **philtrum**, which is a long dimple or indentation that courses from the columella down to the upper lip. The philtrum is bordered by the **philtral ridges** on each side. These ridges are actually embryological suture lines that are formed as the segments of the upper lip fuse. The philtrum and philtral ridges course downward from the nose and terminate at the edge of the upper lip.



FIGURE 1-7 Radiograph of the nasal sinuses.

The top of the upper lip is called the **Cupid's bow** because of its characteristic shape of bilateral rounded peaks with a midline indentation. On the upper lip, the inferior border of the midsection of the vermilion is referred to as the **labial tubercle** because it comes to a slight point and can be somewhat prominent. The lips are surrounded by border tissue, called the **white roll**. The skin of the lips is called the **vermilion** because it is redder (and darker) than the skin of the rest of the face.

In its naturally closed position, the upper lip rests over and slightly in front of the lower lip, although the inferior border of the upper lip is inverted. Movement of the lips is primarily because of the orbicularis oris muscle. The **orbicularis oris** muscle is actually a complex of four independent quadrant muscles in the lips that encircle the mouth (**FIGURE 1-8**). This group of muscles is responsible for pursing and puckering of the lips for kissing and whistling.



**FIGURE 1-8** Orbicularis oris muscles, which circle the mouth.

## **Intraoral Structures**

The intraoral structures include the tongue, faucial pillars, tonsils, hard palate, soft palate, uvula, and oropharyngeal isthmus (**FIGURE 1-9**). These structures are discussed in detail as follows.

## Tongue

The tongue resides within the arch of the mandible and fills the oral cavity when the mouth is closed. With the mouth closed, the slight negative pressure within the oral cavity ensures that the tongue adheres to the palate and the tip rests against the alveolar ridge. The **dorsum** (dorsal surface) is the superior surface of the tongue and the **ventrum** (ventral surface) is the inferior surface of the tongue.

## Faucial Pillars, Tonsils, and Oropharyngeal Isthmus

At the back of the oral cavity on both sides are the paired curtain-like structures called the **faucial pillars** (Figure 1-9). Both the anterior and posterior faucial pillars contain muscles that assist with velopharyngeal movement. (See section called *Muscles of the Velopharyngeal Valve.*)

Most people think of the **tonsils** as the tissue in the oral cavity that can become infected,

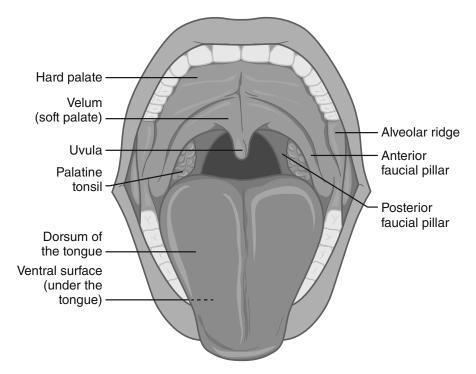


FIGURE 1-9 The structures of the oral cavity.

causing **tonsillitis**. Actually, there are three sets of tonsils, which surround the opening to the oro-pharynx, collectively known as **Waldeyer's ring**.

The **palatine tonsils** (usually known as just the tonsils) are located at the back of the mouth and between the anterior and posterior faucial pillars on both sides. Although the palatine tonsils are bilateral, differences in size are common, so it is not unusual for one tonsil to be larger than the other. The **lingual tonsil** is located at the base of the tongue and extends to the epiglottis (**FIGURE 1-10**). Finally, the pharyngeal tonsil, also known as the adenoids, is located in the nasopharynx. All tonsils consist of tissue similar to lymph nodes. They are covered by mucosa with various pits, called **crypts**, throughout.

Tonsillar tissue serves as part of the body's immune system by developing antibodies against infections, and therefore, this tissue is especially important during the child's first 2 years of life (Brodsky, Moore, Stanievich, & Ogra, 1988). Over time, the tonsil and adenoid tissue tends to atrophy, particularly with puberty, so that by around the age of 16, only small remnants of this tissue remain. Fortunately, atrophy (and even surgical removal) of tonsil and/or adenoid tissue has little effect on immunity because of the redundancy in the immune system. In fact, the entire gastrointestinal tract is lined with the same type of tissue as found in the tonsils so that it also supports immunity.

The **oropharyngeal isthmus** is the opening between the oral cavity and the pharynx. It is bordered superiorly by the velum, laterally by the faucial pillars, and inferiorly by the base of the tongue.

## **Hard Palate**

The **hard palate** is a bony structure that separates the oral cavity from the nasal cavity. It serves as both the roof of the mouth and the floor of the nose. The anterior portion of the hard palate is called the **alveolar ridge** (Figure 1-9). This ridge forms the bony support for the teeth. The rest of the hard palate forms a rounded dome on the upper part of the oral cavity, called the **palatal vault**.