ANATOMY AND EMBRYOLOGY OF THE PEDIATRIC AIRWAY

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The differences between the airway of the adult and child have been described eloquently and extensively.[2-7] Without repeating the specifics of the differences in size, shape, and location of the airway and their well-recognized changes from the neonatal period through childhood and adulthood, and with some poetic license, one can imagine that evolutionary events that required 500 million years to take place are recapitulated during embryogenesis. These precisely choreographed processes, when gone awry, may provide challenges even for the experienced clinician or pediatric specialist. Surgical progress has improved outcomes for these children because of advances in technique as well as earlier intervention; indeed, such progress makes it incumbent upon anesthesiologists to understand the embryology of the craniofacial complex and the details of the surgical repair so that appropriate evaluations, plans, and interventions can be made and counseling given to patients and parents. This article reviews the development of normal and abnormal anatomy of the head and neck and will proceed in a cranio-caudad fashion, much like embryogenesis itself.

FORMATION OF THE CRANIAL VAULT AND BASE

The *skull* develops from a membranous and cartilaginous *neurocranium*. The *membranous* neurocranium gives rise to the flat bones of the skull (the *cranial vault*) and the cartilaginous neurocranium (chondrocranium) forms the skull base (Fig. 1). The flat bones of the neurocranium, which edge-to-edge form sutures, also form *fontanelles* where more than two bones meet (Fig. 2). The frontal bones meet at the metopic suture, whereas the frontal and parietal bones meet at the coronal suture. The anterior fontanelle is the junction of the frontal and parietal bones. The sagittal suture is where the parietal bones meet. Posteri-

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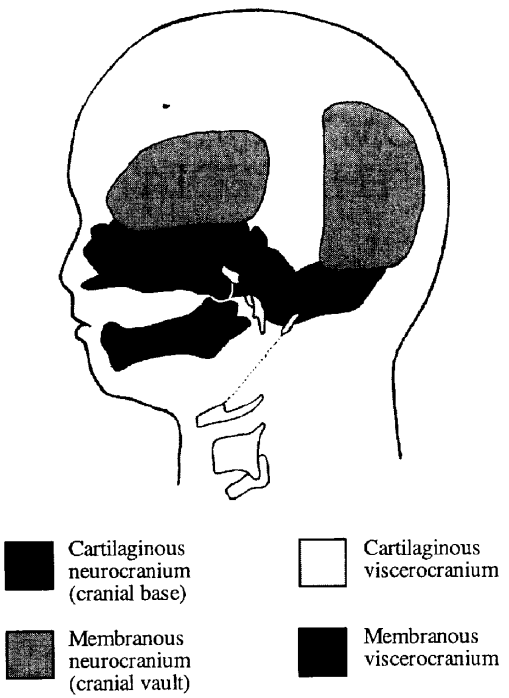

Figure 1. The membranous neurocranium develops into the flat bones of the cranial vault while the cartilaginous neurocranium develops into the skull base, the roof of the face, and the floor of the calvarium. The branchial arch mesoderm develops into membranous and cartilaginous viscerocranium. (*From* Moore K: Essentials of Human Embryology. Toronto, BC Decker, 1988, p 143; with permission.)

orly, the parietal bones meet the occipital bone at the lambdoid suture, and the posterior fontanelle is the junction of parietal and occipital bones.

The base of the skull is formed from the cartilaginous neurocranium that then becomes the base of the occipital bone, the sphenoid, ethmoid, and petrous bone, and portions of the temporal bone. Where the base of the skull (primarily the squamous temporal bone) opposes the parietal bone, it becomes the *squamous suture*. Where the two bones meet the frontal and occipital bone, the anterolateral and posterolateral fontanelles are formed, respectively.

The cranial base, phylogenetically the oldest skeletal component, provides a floor for the calvarium and a roof for the face. The shaping of the skull base and contiguous structures is a dynamic process involving reciprocal influences between the cranial base, the pharynx, the face, and primary and secondary palates. During fetal life and early childhood, *neural influences* predominate because of the rapid growth of the brain. During postnatal development of the airway, *nasal influences* play a major role, and because of speech and nutritional requirements, the *pharynx* also influences the development of the skull base. The anterior portion of the skull base is the roof of the nasomaxillary complex, whereas the posterior portion of the cranial base is the roof of the nasopharynx. During development, the depth of the nasopharynx increases due to remodeling

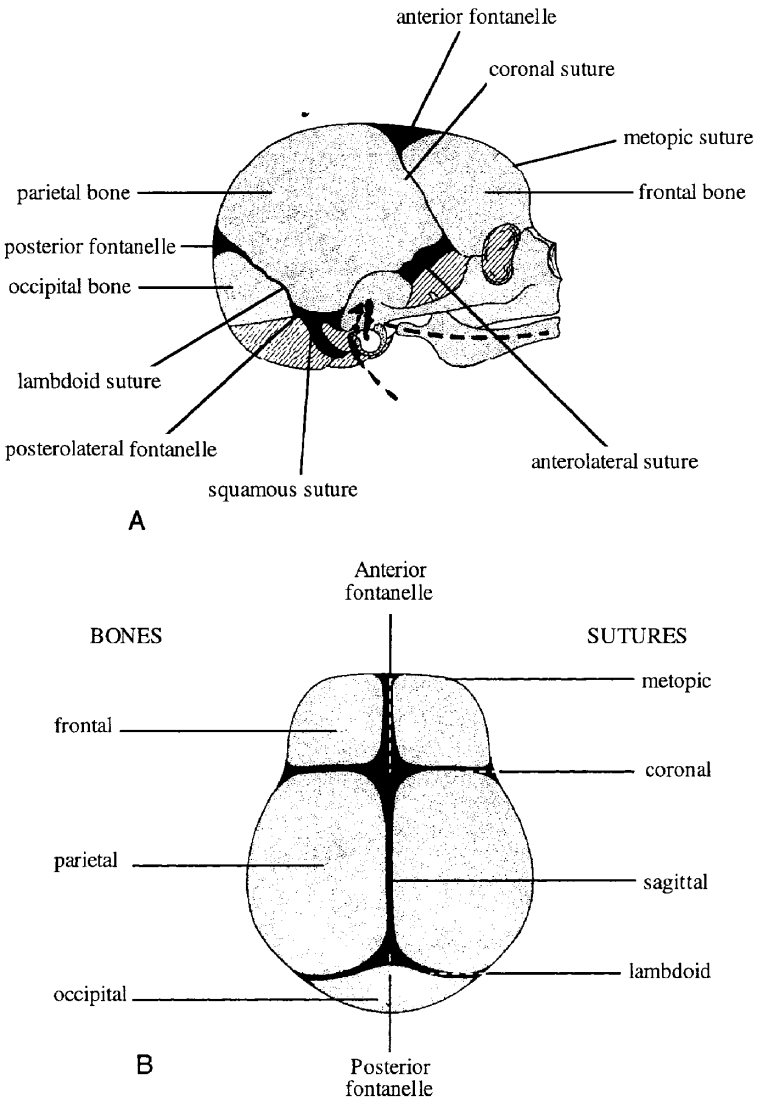

Figure 2. Nomenclature of cranial structures in infants. *A*, Lateral view. *B*, Superior view. (*From* Snell R: Clinical Embryology for Medical Students. Boston, Little, Brown, 1972, p 303; with permission.)

of the palate as well as changes in the angulation of the skull base, providing an enlarged nasal airway for the adult.

Cranial Deformities

A variety of terms are essential when describing cranial deformities. A *synostosis* is a union between adjacent bones, whereas a *dysostosis* is a defect in

ossification. *Craniosynostosis* typically refers to a premature closure of a cranial suture, with primary craniosynostosis resulting from closure of one or more sutures and secondary craniosynostosis resulting from failure of brain growth and expansion. Altered skull morphology results in either event, with growth arrested in a direction perpendicular to the affected suture. Scaphocephaly (boat-shaped skull) is the result of closure of the sagittal suture, brachycephaly (broadened skull, especially of the forehead) is the result of closure of the coronal suture, and turricephaly (tower skull) is the result of closure of the sagittal and coronal sutures. Trigonocephaly (triangular skull) is the result of closure of the metopic suture, with a mid-forehead keel. Multiple suture closure with inferolateral expansion leads to cloverleaf dysostosis or kleeblattschädel. Asymmetrical suture closure may result in a tilted cranium—plagiocephaly (slanting skull).

That the facial bones are hypoplastic in craniosynostosis should not be surprising, as the current hypothesis about the cause of the disorder is that it is actually a skull base defect. The osteoblasts and osteoclasts of the sutures actually are normal rather than defective. The skull base dysmorphism yields a maxilla that is small, a high palate, and poorly developed paranasal sinuses. Dental crowding, with deformed and malocclusion teeth, is common. The orbits are shallow, with a very steep orbital roof, whereas the optic foramina are typically normal in size. The sphenoid bones are widened in a transverse direction, and the interorbital distance is increased. The ears may appear to be low-set and are occasionally deformed. The nasal bridge is broad with narrow nares. Most children are mouth breathers, with very noisy respirations through a small mouth. Table 1 lists genetic disorders associated with craniosynostosis.

CRANIOVERTEBRAL DEVELOPMENT

Embryonic mesoderm differentiates into three distinct regions early in development—paraxial, intermediate, and lateral mesoderm. The paraxial mesoderm is a column of tissue on either side of the midline of the embryo, and at about the fourth week of development it becomes divided into blocks of tissue called *somites* (Fig. 3). Whereas most of the muscles of the head are derived from mesenchyme of the branchial arches, three pairs of occipital myotomes migrate ventrally and cranially to form the muscles of the tongue. Caudal to the occipital myotomes, each cervical somite differentiates into a ventromedial sclerotome and a dorsolateral dermomyotome. The sclerotome consists of mesenchymal cells that migrate medially during the fourth week of development and surround the notochord. The caudal half of each sclerotome fuses with the cephalic half of the immediately succeeding sclerotome to form the mesenchymal vertebral body. Each vertebral body is therefore an intersegmental structure, with the notochord degenerating completely in the region of the vertebral body but enlarging in the intervertebral region to form the nucleus pulposus and intervertebral disc. The annulus fibrosus is derived from sclerotomic mesenchyme between adjacent vertebral bodies.

Craniovertebral Anomalies

Faulty segmentation in the occipitovertebral area can occur either contemporaneously with branchial arch or nonbranchial arch anomalies, or in isolation. Vertebral anomalies are due to failure of normal segmentation, which can be traced back to the third embryonic week, when segmentation of mesodermal

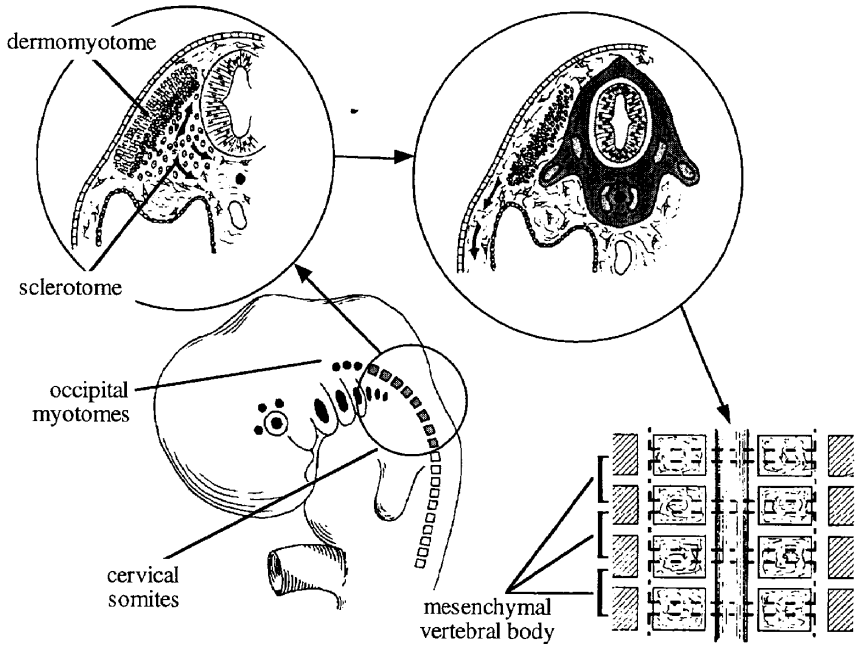

Figure 3. Development of normal vertebral structures from cervical somites, illustrating mesodermal differentiation into muscular and skeletal elements. Note the formation of vertebral bodies from the caudal and cranial portions of adjacent somites. (*From* Snell R: Clinical Embryology for Medical Students. Boston, Little, Brown, 1972, 294; with permission.)

somites takes place. Signs of cranial nerve, root, and spinal cord impairment are variable but not surprising, given the intersegmental formation noted previously. Structures near the posterior part of the foramen magnum may be displaced cranialward, and atlanto-occipital fusion, atlantoaxial dislocation, stenosis, deformity of the foramen magnum, failure of fusion of the posterior arch of the atlas, anterior and posterior clefts of the lower cervical vertebrae, and fusion of cervical vertebrae (Klippel-Feil syndrome) may all occur because of such defects. Clinical symptoms may vary from pain; limitation of movement; shortening of the neck; raised intracranial pressure; hydrocephalus; cerebellar, bulbar, or cranial nerve symptoms; or degenerative changes in lower cervical segments. Faulty segmentation of two or more cervical vertebrae may result in the Klippel-Feil syndrome. Several or all of the cervical vertebrae may be fused into a single column; the number of vertebrae may be reduced and the vertebral bodies flattened. The spinous processes are often small or bifid, and intervertebral foramina may be smaller than normal. Hemivertebrae and other anomalies may be additional expressions of abnormal segmentation; there is an association with hemifacial microsomia. A short neck with limited mobility, giving the impression that the head sits on the shoulders, with a low hairline and neck webbing is often seen.

Table 1. GENETIC DISORDERS OF CRANIOSYNOSTOSIS

Syndrome	Inheritance	M:F Ratio	Principal Craniofacial Disorder	Associated Features
Apert syndrome	Sporadic defect	1 to 1	Primarily coronal synostosis, although multiple sutures may also be involved	Sphenoethmoidomaxillary hypoplasia
	Autosomal dominant			Prominent mandible Hypertelorism Proptosis Downward-slanting palpebral fissures Small nasal bridge Hydrocephalus Progressive cervical spine calcification Finger and toe syndactyly are extremely common
Carpenter syndrome	Autosomal recessive	1 to 1	Wide, towering skull (premature closure of all cranial sutures)	Severe mental retardation Hypoplastic mandible Short neck Omphalocele Variety of cardiac anomalies (atrial septal defect, ventricular septal defect, pulmonic stenosis, Tetralogy of Fallot) Short upper extremities Syndactyly of upper- and lower-extremity digits

Crouzon syndrome	Autosomal dominant	1 to 1	Wide, towering skull Coronal and sagittal sutures most commonly affected	Proptosis Orbital and maxillary hypoplasia Orbital hypertelorism Intelligence is usually normal High-arched, short, and occasionally cleft palate
Pfeiffer syndrome	Sporadic defect Autosomal dominant	1 to 1	Turricephaly Widely spaced eyes	Intelligence is unaffected Thumbs and great toes are short and broad Finger and toe syndactyly is common
Saethre-Chotzen syndrome	Autosomal dominant	1 to 1	Asymmetric craniosynostosis Plagiocephaly	Facial asymmetry Ptosis Short fingers Soft-tissue syndactyly of the second and third fingers

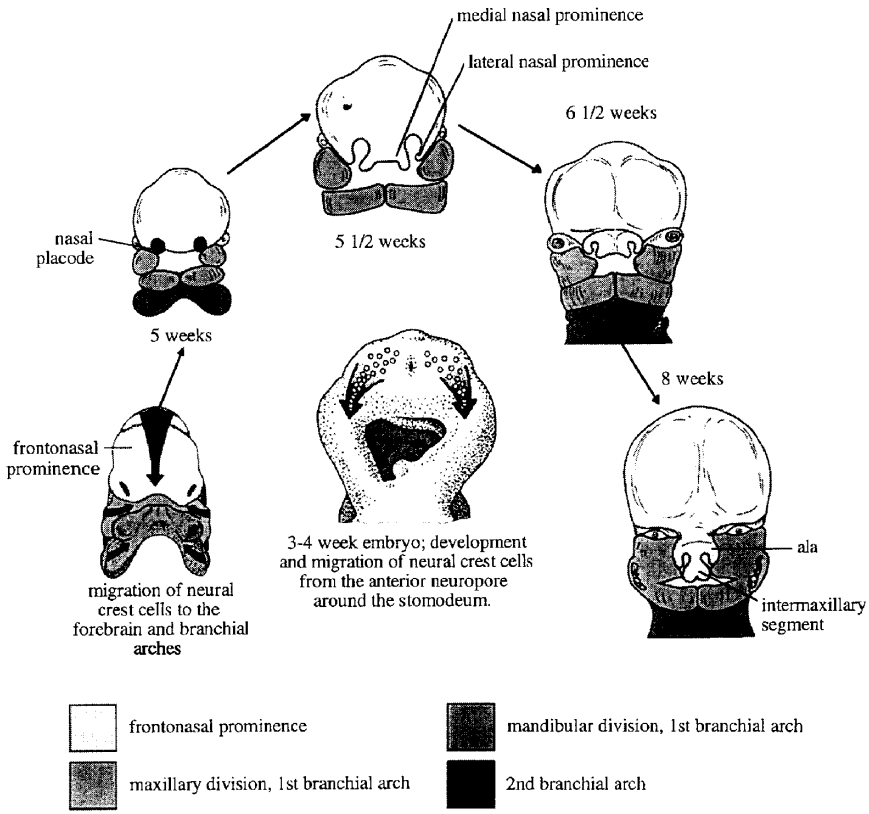

Figure 4. Formation of the face from the confluence of the developing frontonasal prominence and elements of the first and second branchial arch apparatus. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 8.)

THE FACE

Just as the neurocranium forms the cranial vault and base, the viscerocranium forms the face and is derived mainly from cartilage of the first two branchial arches. Neural crest cells of the developing 3- to 4-week embryo, composed of ectoderm, are found at the junction of the neural plate and surface ectoderm. These neural crest cells then migrate to branchial arch mesoderm, and the face develops as a result of these massive cell migrations and their interactions with differentiating cells of mesodermal origin (Fig. 4). Those cells forming the frontonasal process are derived from the forebrain fold and migrate a relatively short distance as they pass into the nasal region. Those cells that form the mesenchyme of the maxillary and mandibular processes have a considerably longer distance to migrate, as they must move into the branchial arches, where they surround the mesodermal muscle plates and contribute to the formation of the face.

At 28 days, the face barely shows its eventual relationship to the five

primordia from which it is derived: the frontonasal prominence, which is the cranial boundary of the primitive mouth (stomodeum); the paired maxillary prominences (first branchial arch); and the paired mandibular prominences (also first branchial arch). The mandibular prominences grow medially and begin to merge with each other by the end of the fourth week, forming the lower lip, the chin, and the mandible.

The frontonasal prominence develops in a cranio-caudad fashion from mesoderm, under the influence of neural crest tissue. Ultimately, the keystone of the midface, the sides of the frontonasal prominence, gives rise to the nasal placodes. Small horseshoe-shaped ridges develop around the nasal placodes, called the medial and lateral nasal prominences, at about 33 days. Between the fifth and eighth weeks, the nasal pits form in the floor of the nasal placodes and the maxillary prominences increase in size and grow medially. This moves the medial nasal prominences toward the median plane, and the groove between the lateral nasal prominence and the maxillary prominence disappears during the merger, thus completing the "keystone."

The primitive mouth (stomodeum) appears as a slight depression in the surface ectoderm separated by the oropharyngeal membrane. This membrane ruptures at about 24 to 26 days' gestation, and the primitive gut then communicates with the amniotic cavity. The involved germ layers are endoderm internally and ectoderm externally.

The tongue surface arises primarily from first arch mesenchyme, with significant contributions from third and fourth arches, hence its complex innervation by the facial nerve in the anterior two-thirds (that portion formed by the mandibular division of the first branchial arch) and the hypoglossal nerve (that portion formed by the third branchial arch) (Fig. 5). The foramen cecum is located posterior to the first branchial arch derivatives. The muscle bulk of the tongue arises primarily from occipital somites, explaining the hypoglossal nerve (XII) innervation of tongue musculature.

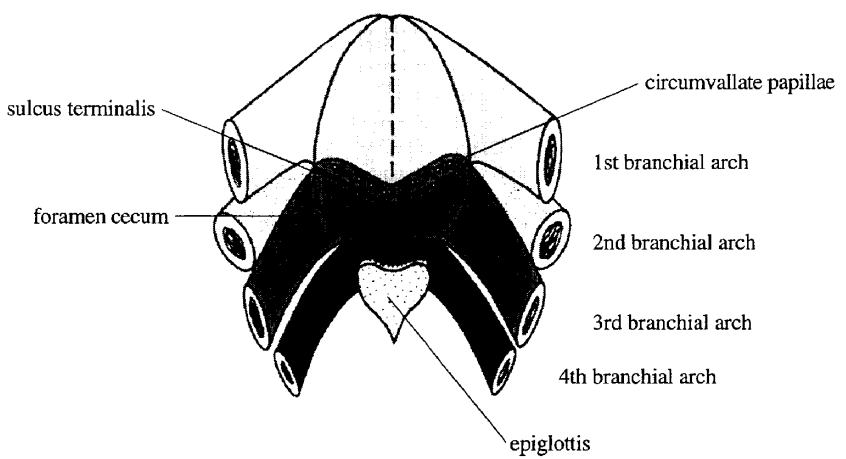

Figure 5. The complexity of the tongue is explained by its diverse embryogenesis; muscle bulk from occipital myotomes (innervated by cranial nerve XII), and compound sensation by the nerves of the first (cranial nerve VII) and third (cranial nerve IX) branchial arches. (*From* Snell R: Clinical Embryology for Medical Students. Boston, Little, Brown, 1972, p 106; with permission.)

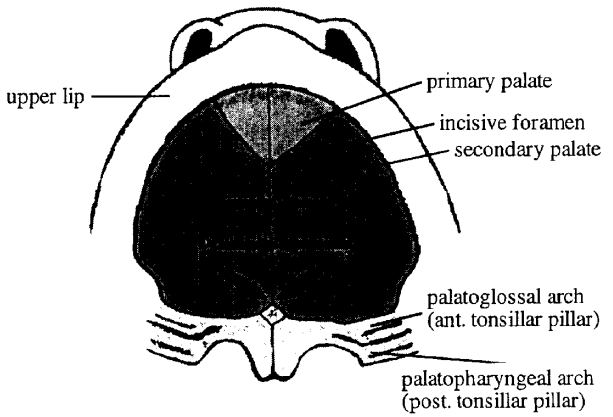

Figure 6. Formation of the primary and secondary plate. Typical cleft lip formation occurs along a line joining the primary and secondary palates through the middle of the ipsilateral ala, at the junction of the anlage of the nostril, the lateral and medial nasal processes. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 867.)

The upper lip is formed by the merging of the maxillary prominences with the medial nasal prominences. The lateral nasal prominences do not form part of the upper lip but rather form the alae of the nose. The intermaxillary segment in the central portion of the upper lip area consists of three parts: the labial component, forming the philtrum, a maxillary component associated with the four incisor teeth, and a palatal component, which becomes the primary palate.

The development of the palate serves to divide the nasomaxillary complex from the oral cavity (Fig. 6). The premaxilla (intermaxillary segment), containing the incisor teeth, forms from the fusion of the globular processes of the median nasal process. The palatal processes advance in a medial direction from the maxillary processes of the first branchial arch, fusing in the midline in an anterior-to-posterior sequence, and unite with the premaxilla and the developing nasal septum. The soft palate forms from continued growth of the posterior edge of these palatal processes, ending with the formation and fusion of the two halves of the uvula.

The nose originates in the cranial ectoderm, which subsequently develops into the frontonasal prominence, with the paired nasal placodes becoming recognizable during the third week of development. The superior portion of the nose is formed from the lateral nasal processes, whereas the inferior portion of the nasal cavity is incomplete until the paired maxillary processes of the first branchial arch grow anteriorly and medially to fuse with the median nasal processes. The nasal cavities extend posteriorly during development under the influence of the posteriorly directed fusion of the palatal processes, thinning out the membrane which separates them from the oral cavity. By the 38th day of development, the two-layer membrane consisting of nasal and oral epithelia ruptures, and forms the choanae (posterior nares). Failure of such rupture results in choanal atresia, although these choanae are not in the same location as the definitive choanae, which will eventually be located more posteriorly; however, it does explain the unexpectedly anterior extent of choanal atresia given the eventual location of the choanae. The cranial base and nasal septum are in

cartilaginous continuity, and fetal facial bones and teeth organize themselves around the cartilaginous nasal capsule. The normal nasomaxillary complex grows both downward and forward.

THE BRANCHIAL APPARATUS

Branchial Arches

The branchial apparatus (Fig. 7) consists of four numbered branchial arches that are visible on the surface of the embryo, as well as a fifth and sixth arch that cannot be seen on the surface. Branchial pouches and clefts are likewise numbered craniocaudally. The first branchial arch cartilage, known as Meckel's cartilage, is the position of the future mandible, as well as the eventual middle ear bones, the malleus and incus, which develop by endochondral ossification. The sphenomandibular ligament remains from this embryological anlage, whereas the balance of the first arch cartilage disappears almost completely, the mandible subsequently developing from intramembranous ossification around Meckel's cartilage.

The second branchial arch cartilage produces the stapes, the styloid process, the stylohyoid ligament, and the superior portion of the body of the hyoid. The

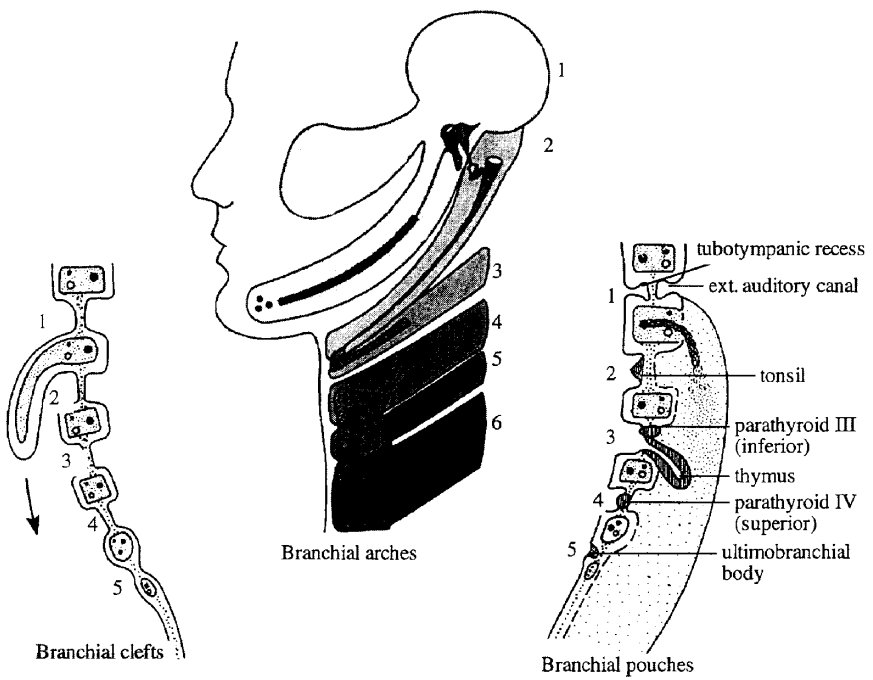

Figure 7. The branchial arch apparatus and its various fates. (*From* Snell R: Clinical Embryology for Medical Students. Boston, Little, Brown, 1972, pp 102, 104; with permission.)

other branchial arch cartilages contribute to the inferior portion of the hyoid as well as the thyroid cartilage.

Striated muscles also are formed in the respective branchial arch mesenchyme. Myoblasts differentiate and migrate to various parts of the head and neck, where they form the muscles of mastication and facial expression, each retaining their own original nerve supply. Although we think of muscular action in the head and neck as far removed from the origin and course of the cranial nerves, fetal nerves that supply the branchial arch derivatives have only a short distance to travel from the brain. The trigeminal nerve (V) supplies the skin covering the part of the face derived from the first branchial arch via its maxillary and mandibular divisions (the ophthalmic division does not make a contribution). The facial nerve (VII) supplies the muscles derived from the first arch. The nerve of the third branchial arch is the glossopharyngeal (IX) cranial nerve. Two branches of the vagus nerve (X) supply the remaining branchial arches. The superior laryngeal nerve innervates derivatives of the fourth branchial arch, whereas the recurrent laryngeal nerve supplies derivatives of the sixth branchial arch.

Branchial Pouches

The first branchial pouch develops into the tubotympanic recess, becoming the auditory tube and the middle ear cavity (Fig. 8). The cavity of the second branchial pouch is largely obliterated as the palatine tonsil develops, but part of it remains as the tonsillar fossa (intratonsillar cleft). The endoderm of the second branchial pouch becomes the surface epithelium of the tonsil and the lining of its crypts, with the mesenchyme around the pouch differentiating into lymphoid tissue. The endoderm of the dorsal part of the third branchial pouches differentiates into the inferior parathyroids, and the ventral parts unite to become the thymus. The endoderm of the fourth branchial pouches differentiates into the superior parathyroid glands, and the ventral parts develop into the ultimobranchial bodies (the calcitonin-secreting portion of the thyroid).

Branchial Clefts

The first branchial cleft (between the first and second arches) forms the external ear, which is the only normal structure to arise from a branchial cleft;

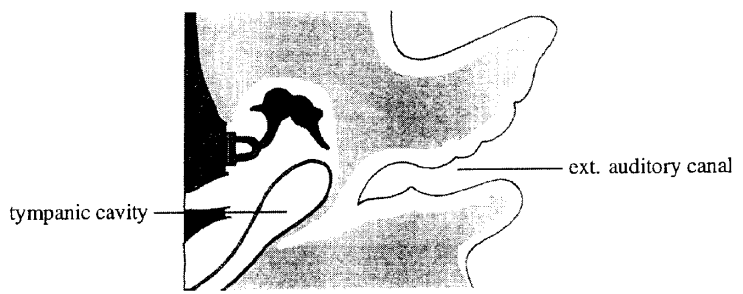

Figure 8. Development of the tympanic membrane from the approximation of the tubotympanic recess (tympanic cavity) and the external auditory canal. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 90.)

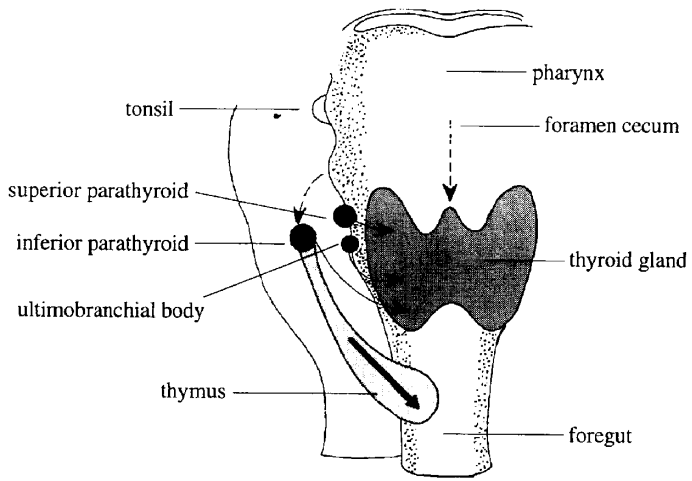

Figure 9. Migration of neck structure derivatives of the branchial arches and oropharyngeal mesoderm. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 1359.)

however, if the second arch does not grow caudally over the third and fourth arches (as it normally should), then the second, third, and fourth clefts can remain as branchial fistulas, in contact with the skin surface between the clavicle and the mandible.

The thyroid begins as a thickening of the endoderm of the floor of the pharynx, in the midline between the first and second pouches, at the foramen cecum (Figs. 5 and 9). A thin connection, the thyroglossal duct, remains attached to the oral cavity, and its point of attachment marks the origin of the thyroid gland. The thyroid descends along the thyroglossal duct and reaches the level of the first tracheal ring at about the seventh week of gestation. The thyroglossal duct is then normally obliterated. Accessory thyroid tissue may be deposited anywhere along this cranio-caudal path; on the other hand, failure of the thyroid to descend may result in a lingual thyroid (Fig. 10). The paired parathyroid glands develop from separate pouches, with one pair derived from the third and the other from the fourth branchial arch. At the seventh week of development, the parathyroid glands move caudally from their respective branchial pouches with the parathyroids derived from the third arch moving more caudally than the parathyroids of the fourth arch. Accessory parathyroid tissue again may be left along the line of migration (Fig. 9).

Facial Defects

Cleft lip with or without a cleft palate occurs in approximately 1:1000 live births. Cleft lip occurs with greater frequency in boys (63%) and cleft palate with a greater frequency in girls (70%); 50% of affected patients have both. Associated congenital defects occur in 13% to 50% of patients with cleft palates and 7% to 13% of those with cleft lips. There is a tendency toward repairs at younger ages for cleft palate because speech development has been found to be

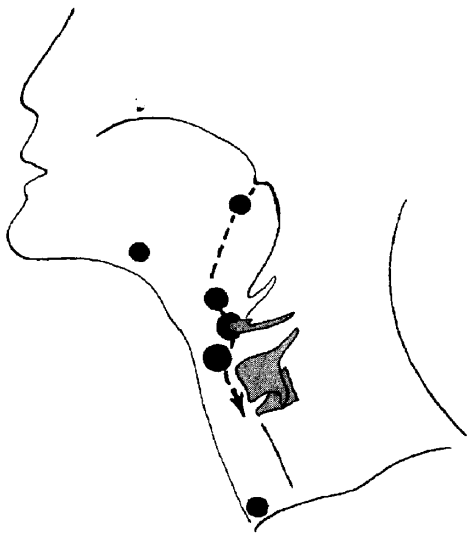

Figure 10. Migration path and typical locations for ectopic thyroid tissue as well as thyroglossal duct cysts. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 1400.)

more normal, the incidence of ear infections less, and orthodontic treatment can be instituted more effectively. Initial repairs may be followed by additional procedures to correct physiologic or cosmetic deformities during subsequent growth and development.

In choanal atresia, lack of communication exists between the posterior nares and nasopharynx. Although the obstruction may occasionally be membranous, over 90% of obstructions involve bone. Choanal atresia may be unilateral or bilateral and, in severe forms, accompanied by apnea, respiratory obstruction, and cyanosis. Whereas membranous atresia may be perforated therapeutically and tubes passed from the nose into the nasopharynx, bony atresia requires transnasal or transpalatal repair. A special subset of patients with choanal atresia also has the CHARGE association:

Colobomatous malformation (iris or retinal)	80%
*H*eart defect	58%
*A*tresia choanae	100%
Mental *R*etardation	94%
*G*rowth deficiency	87%
*G*enital hypoplasia (males)	75%
*E*ar anomalies	88%

The Pierre Robin syndrome is characterized by micrognathia and glossoptosis (tongue falling backward), often in association with a cleft palate thought to be secondary to tongue interference with palatal fusion. With mandibular hypoplasia as the initial malformation, a relatively normal tongue occupies a small, developing oral cavity. Chronic upper airway obstruction represents a constant threat to the infant. Symptoms usually improve, and by 2 to 3 years of age, most children outgrow airway or feeding difficulties.

Branchial Arch Defects

In hemifacial microsomia (otomandibular dysostosis), the embryological structures involved are the first and second branchial arches, including the intervening first cleft; this involvement may cause bilateral deformity as well as the more common unilateral deformity. Hemifacial microsomia is characterized by unilateral ear abnormalities and hypoplasia of the mandibular condyle and ramus. Various gradations exist, depending on the severity of the hypoplasia, and are categorized according to the association of orbital, mandibular, ear, facial nerve, and soft-tissue defects (Table 2). The chin usually is deviated toward the affected side, and mandibular mobility may be impaired not only due to the bony abnormalities, but also because of abnormal development of the muscles of mastication and developmental effects on innervation (Table 2).

The Goldenhar syndrome (oculoauriculovertebral dysplasia) is characterized by preauricular appendages and fistulas associated with mandibulofacial dysostosis. It probably is the same entity as hemifacial microsomia. Micrognathia, unilateral mandibular hypoplasia, and cleft palate are found, along with a 40% incidence of vertebral abnormalities such as the Klippel-Feil syndrome and a 35% incidence of congenital heart disease. The defect may originate as

Table 2. THE OMENS CLASSIFICATION FOR HEMIFACIAL MICROSOMIA[8]

*O*rbit	O_0	Normal orbital size and position
	O_1	Abnormal orbital size
	O_2	Abnormal orbital position; arrow denotes relative position of affected size (i.e., $O_2\uparrow$ denotes superior position)
	O_3	Abnormal orbital size and position
*M*andible	M_0	Normal mandible
	M_1	Mandible and glenoid fossa small; short ramus
	M_2	Mandibular ramus short/abnormally shaped
	2_A	Glenoid fossa in anatomically acceptable position with reference to opposite temporomandibular joint (TMJ)
	2_B	TMJ is inferiorly, medially, and anteriorly displaced; severely hypoplastic condyle
	M_3	Complete absence of ramus, glenoid fossa, and TMJ
*E*ar	E_0	Normal ear
	E_1	Mild hypoplasia and cupping with all structures present
	E_2	Absence of external auditory canal with variable hypoplasia of the concha
	E_3	Malpositioned lobule with absent auricle. Lobular remnant usually inferiorly and anteriorly displaced
Facial *N*erve	$N7_0$	No facial nerve involvement
	$N7_1$	Upper facial nerve involvement (temporal and zygomatic branches)
	$N7_2$	Lower facial nerve involvement (buccal, mandibular, and cervical branches)
	$N7_3$	All branches of the facial nerve affected. Other nerve involvement may be designated (i.e., trigeminal N5, hypoglossal N12)
*S*oft tissue	S_0	No obvious soft-tissue or muscle deficiency
	S_1	Minimal subcutaneous/muscle deficiency
	S_2	Moderate—between the two extremes, S1 and S3
	S_3	Severe soft-tissue deficiency due to subcutaneous and muscular hypoplasia

early as the seventh to eighth week of development, and hence is frequently associated with other anomalies of early development.

Malar hypoplasia and lower lid coloboma with associated ear and mandibular deformities are characteristic of the Treacher Collins syndrome (mandibulofacial dysostosis). The syndrome is classically subdivided into five categories: complete, incomplete, unilateral, abortive, and atypical. The unilateral form probably is really hemifacial microsomia. The complete form includes downward-slanting palpebral fissures, notching or coloboma of the lower eyelid, hypoplasia of the facial bones (especially the zygoma and mandible), malformation of the ear (external, middle, and occasionally inner), macrostomia with high palate and abnormal dentition, blind fistulas between the ear and the angle of the mouth, and patchy projections of hair from the sideburns onto the cheeks. Very commonly, the palate may be cleft as well. The incomplete form usually presents with lesser degrees of deformity, frequently with associated deafness, and is probably the original Treacher Collins syndrome. Patients with Treacher Collins syndrome have marked cross-sectional narrowing of the pharynx at the level of the base of the tongue, resulting in significant problems with airway obstruction and obstructive sleep apnea.

Prominent features of the Hallermann-Streiff syndrome (oculomandibulofacial syndrome) include dwarfism along with diminished scalp and body hair. The skull is wide and occasionally macrocephalic. Ophthalmologic defects include microphthalmia, downward-slanting palpebral fissures, blue sclerae, congenital cataracts, and decreased visual acuity. Typically, the patients have "bird-like" facies. They have malar hypoplasia, micrognathia, mandibular hypoplasia, and a high-arched palate. The entire temporomandibular joint may be displaced forward, up to 2 cm.

Branchial Pouch Defects

First branchial pouch defects (Fig. 11) are rare but may occur in association with abnormalities of the first branchial arch. Such defects may include commu-

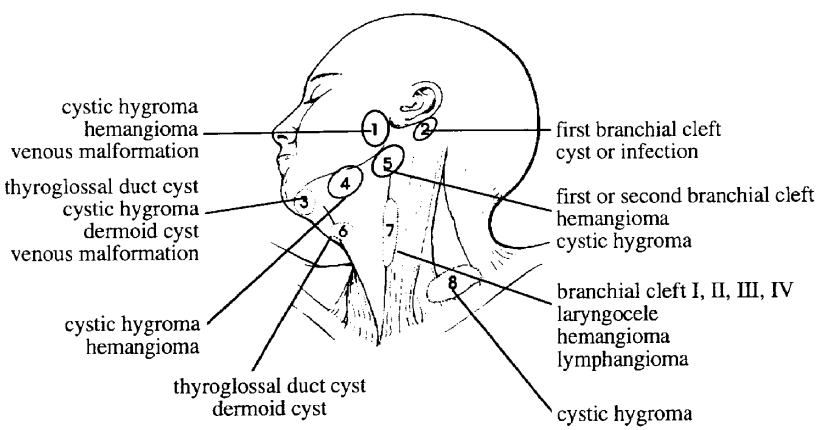

Figure 11. Congenital abnormalities may present in various locations in the neck. (*From* May M: Neck masses in children: Diagnosis and treatment. Pediatric Annals 5:519, 1976; with permission.)

nication between the first branchial cleft and first pouch presenting as an external sinus in continuity with the Eustachian tube. The second branchial pouch is the precursor of the palatine tonsil; persistence of the second branchial pouch may result in an internal sinus at the level of the palatine tonsil, which may present with recurrent infection. The third branchial pouch gives rise to the thymus and the inferior parathyroid gland; agenesis of the third and fourth branchial pouches results in the absence of the thymus and parathyroids (DiGeorge syndrome), which may present with neonatal tetany and impaired cellular immunity. A persistent third pouch may result in a sinus tract from the skin of the anterior neck to the pyriform sinus.

Branchial Cleft Defects

Isolated branchial cleft defects (Fig. 11) result from failure of normal regression. Branchial cleft defects generally present unilaterally, with cysts outnumbering fistulas 3:1. First branchial cleft cysts are found entirely above the hyoid bone and can communicate with the external auditory canal. Their clinical significance is their proximity to the facial nerve. First branchial cleft cysts need to be included in the differential diagnosis of parotid gland masses. Second branchial cleft cysts may appear externally anywhere along the anterior border of the lower two thirds of the sternocleidomastoid. Approximately 50% fistulize to the tonsillar fossa, with a path between the internal and external carotid arteries; they usually present as a dimple in the skin. Third branchial cleft cysts open externally at about the same location as the second, but present internally at the pyriform sinus, after traveling down the sheath of the common carotid artery. Fourth branchial cleft cysts are rare and pass from the upper esophagus to the mediastinum.

THE LARYNX

Development of the respiratory system begins at approximately 3 weeks of gestational age with formation of the laryngotracheal tube from the ventral wall of the foregut. The normal laryngotracheal tube grows faster than the esophagus, which forms from the caudal growth of the foregut. The laryngotracheal tube then grows caudally into the splanchnic mesoderm on the ventral surface of the foregut, dividing into the right and left lung buds. The epiglottis begins to form from the hypobranchial eminence of the third and fourth arches at approximately 30 to 32 days of gestation (Fig. 12). The anlage of the arytenoid cartilages can be identified on both sides of the laryngeal slit at this time and continue to grow during the fifth week of development. The aryepiglottic folds develop from the lateral boundaries of the fourth arch along a line from the hypobranchial eminence (epiglottis) to the arytenoid eminence of the sixth arch. Incomplete development at this stage may produce variable degrees of persistent laryngeal cleft. A definite larynx may be seen by 41 days of gestation.

The cricoid and thyroid cartilages begin to develop prior to the arytenoid cartilages and begin chondrification at about 7 weeks of gestation. As the thyroid cartilages develop, the glottis deepens and the true vocal cords are aligned within the thyroid laminae. Failure of the true vocal cords to split at 10 weeks of gestation to form the primitive glottis results in congenital atresia of the larynx, or more often, a complete or partial congenital laryngeal web. Although

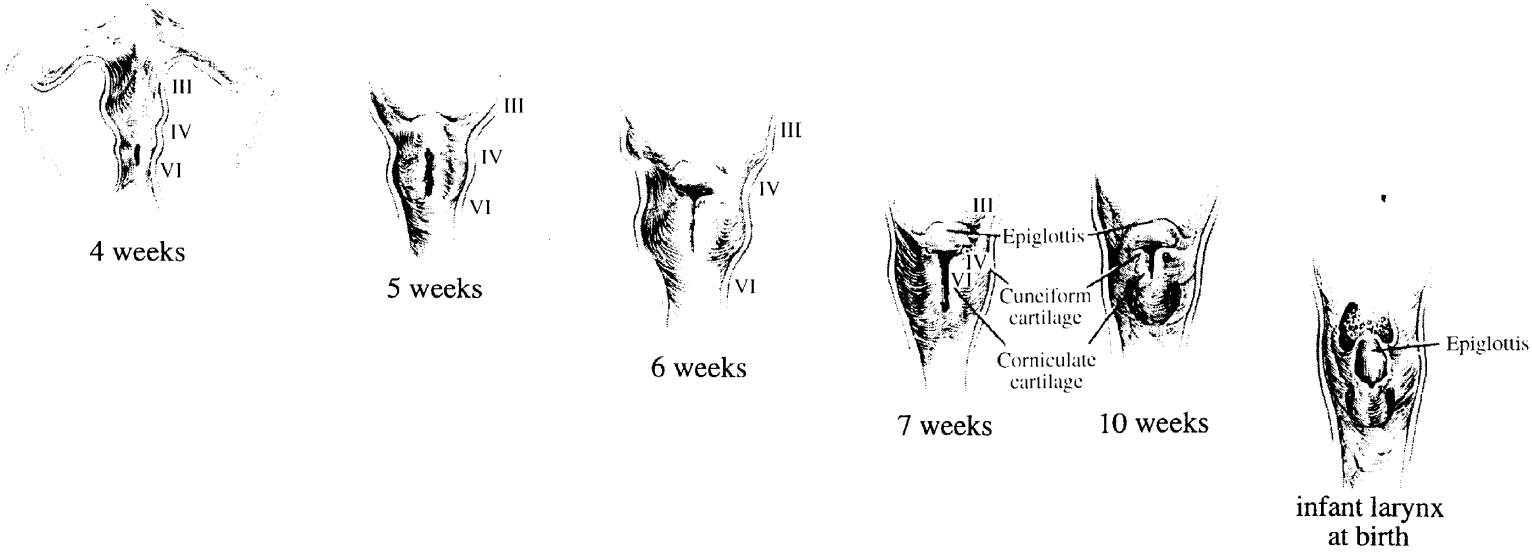

Figure 12. Development of the larynx. (*From* Bluestone C, Stool S: Pediatric Otolaryngology. Philadelphia, WB Saunders, 1983, p 1136.)

webs may be supraglottic or subglottic, most occur at the level of the glottis. Congenital cysts of the supraglottic region are possibly remnants of the third branchial pouch and lie superior to the derivations of the fourth arch. By the 10th to 11th weeks of gestation, the major structures of the larynx have developed and the cartilages are chondrifying.

If the separation of the esophagus and trachea is slightly delayed and the margins of the laryngotracheal groove fail to fuse adequately, the rapidly growing trachea separates the proximal and distal esophagus, resulting in the most common form of esophageal atresia and distal tracheoesophageal fistula.

Laryngeal Defects

Developmental abnormalities of the larynx presenting in the newborn period may be related to either structural or functional impairment. The principal functional impairment is cartilaginous incompetence—laryngo, tracheo, or bronchomalacia.

Laryngomalacia (congenital flaccid larynx) is responsible for about 75% of laryngeal problems in infancy. Characteristic findings on awake laryngoscopy or fiberoptic nasopharyngoscopy are infolding of an abnormally flaccid epiglottis and aryepiglottic folds into the glottis during inspiration. Tracheomalacia with or without bronchomalacia may present with expiratory stridor, signifying an intrathoracic obstruction, and the evaluation includes diagnostic bronchoscopy to rule out extrinsic compression from vascular structures or solid masses.

Structural defects may occur anywhere from the supraglottic to infraglottic larynx (Fig. 13). Supraglottic atresia has a high frequency of associated anomalies, which may include esophageal atresia, tracheoesophageal fistula, and nonforegut anomalies. Survival may be enhanced if a tracheoesophageal fistula is present. Laryngeal webs occur from the supraglottic to the subglottic areas; two thirds occur at the level of the vocal cords and vary from thin to dense membranes, producing variable degrees of airway obstruction. Supraglottic locations represent less than 2% of congenital laryngeal webs, whereas over 75% of laryngeal webs are located at the level of the glottis. Treatment is directed to sharp or laser resection of the obstruction. A thick membranous web often requires a tracheotomy.

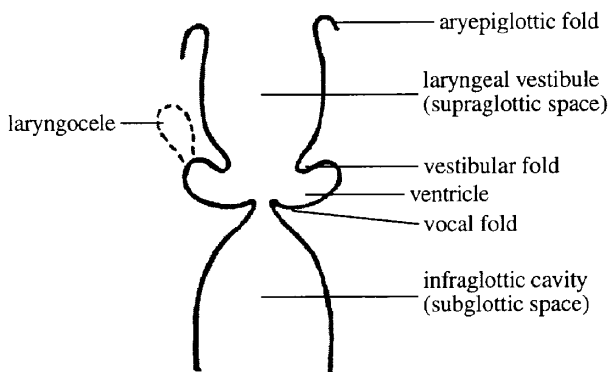

Figure 13. Laryngeal topography for localizing congenital laryngeal abnormalities.

Laryngeal cysts contain fluid, do not communicate with the airway, and are located in the supraglottic larynx. They must be distinguished from laryngoceles, which contain both air and fluid, as they arise from the airway. Laryngoceles also are located in the supraglottic larynx and represent a dilatation of the ventricular sinus beyond the limits of the normal laryngeal cartilage. Laryngoceles cause intermittent hoarseness and dyspnea that increase with crying. Clinical presentation is characterized by stridor and airway obstruction in both disorders. Endoscopy includes the risk of cyst rupture, with release of fluid and blood into the airway.

Stridor is the presenting symptom of subglottic stenosis, which may be congenital or iatrogenic, often following airway support for respiratory distress. Laser resection, tracheal reconstruction in older age groups, or tracheostomy (for severe cases) is the usual treatment.

Vocal cord paralysis can have a variety of causes. Bilateral vocal cord paralysis may be caused by CNS malformations such as cerebral agenesis, cervical meningocele, and Arnold-Chiari malformation, or by intracranial hemorrhage. In unilateral vocal cord paralysis, the left cord is affected more commonly than the right because of the longer intrathoracic course of the recurrent laryngeal nerve, making it more susceptible to the influence of any intrathoracic anomaly such as vascular malformations, abnormalities of the great vessels, malformations of the conducting airways, or mediastinal masses. Transient vocal cord paralysis may be caused by trauma to the recurrent laryngeal nerve during birth.

Hemangiomas and lymphangiomas are the most common benign tumors of infancy and are caused by excesses of embryonic mesodermal tissue which gives rise to arteries, veins, and lymphatics. Lymphangiomas are the most common cause of macroglossia in infancy. Large lymphangiomas are called hygromas because of their fluid content. Cystic or cavernous, they are usually evident by ultrasound shortly after birth or prior to delivery. Approximately 70% arise from the posterior triangle of the neck, 20% from the axilla, and 10% in other areas of the body; they may be very large. Anterior triangle lymphangiomas are associated with intraoral lymphangiomas, often with airway compromise, and are occasionally accompanied by mediastinal extension. Mediastinal compression of the airway may require support with an endotracheal tube.

CONCLUSION

Understanding the embryology of the head, face, and neck can help anesthesiologists to better predict and assess patients with difficult airways and plan appropriately for their care. The craniosynostoses, for example, rarely will provide a challenge for direct laryngoscopy and intubation of the airway. Because of the mid-face dysmorphism due to the base of skull malformation, however, the mask airway often will be a challenge. By dividing the face into three horizontal and three vertical planes, the contribution of the maxillary and mandibular divisions of the first branchial arch are better appreciated, particularly in relation to congenital abnormalities of jaw development that may severely limit temporomandibular joint excursion and direct laryngoscopy. Impaired growth of the nasomaxillary complex, which should grow both downward and outward, helps explain the mid-face dysmorphism of the CHARGE syndrome. Such understanding, rooted in the embryology of the head and neck, is critical for consultant-level care of patients with congenital airway abnormalities.

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