DEVELOPMENT OF THE FACE

Prof Malek

Face

The main development of the face occurs between the 4th and 8th weeks.

Facial primordium

Early in the 4th week, face is represented by an area around the mouth bounded cranially by the neural plate, caudally by the pericardial bulge and laterally by the mandibular precess of 1st pharyngeal arches.

Facial development is induced by brain vesicles.

Facial prominences

In week 4 five facial prominences appear around stomodeum:

-Single ***frontonasal prominence*** (FNP) above the stomodeum.

-Paired ***maxillary prominences*** (Mx P) lateral to stomodeum.

-Paired ***mandibular prominences*** (Md P) below stomodeum.

The paired prominences are derivatives of the first pair of pharyngeal arches.

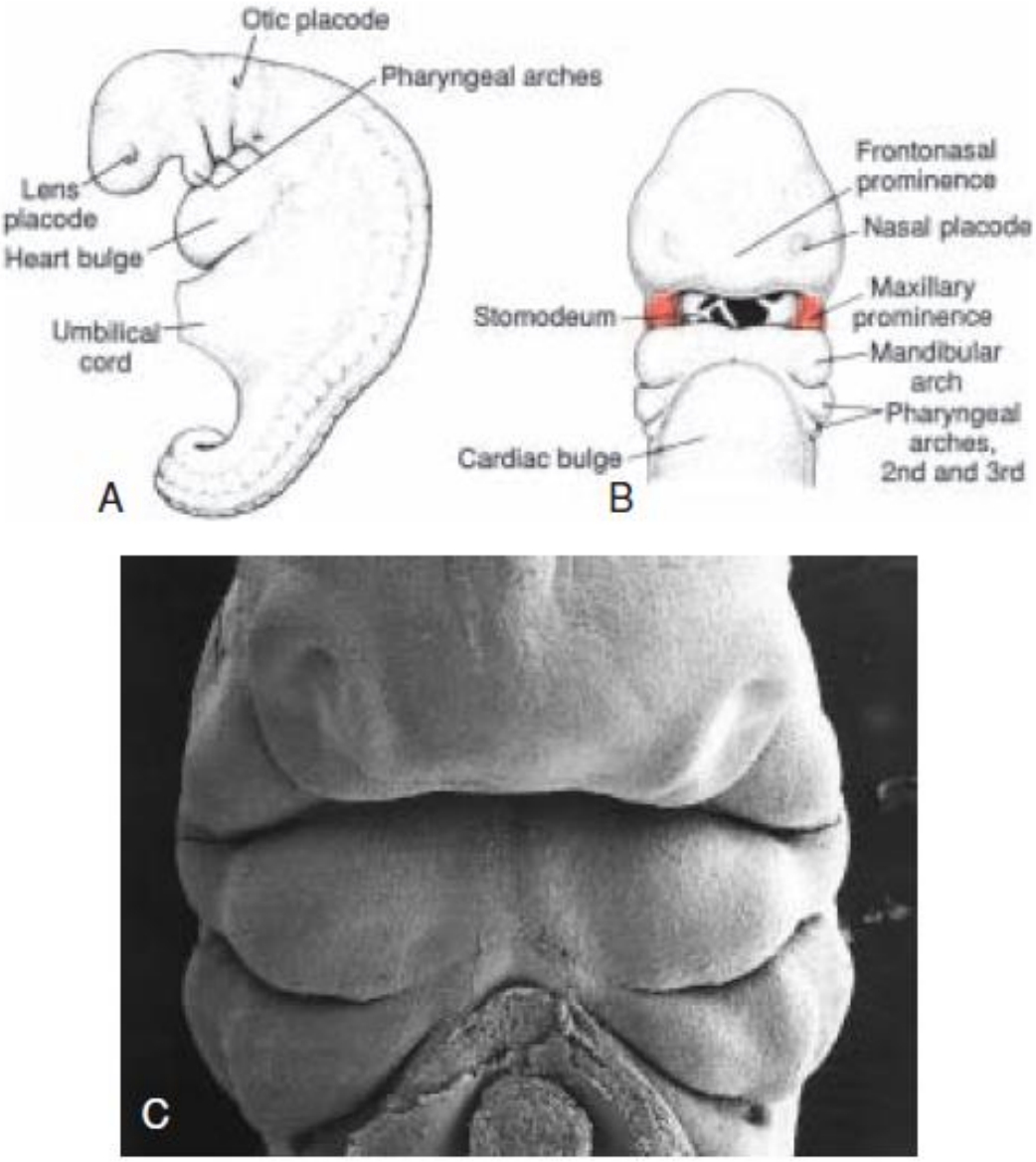


Figure 15.21 A. Lateral view of an embryo at the end of the fourth week, showing position of the pharyngeal arches. **B.** Frontal view of a 4.5-week embryo showing the mandibular and maxillary prominences. The nasal placodes are visible on either side of the frontonasal prominence. **C.** Scanning electron micrograph of a human embryo at a stage similar to that of **B.**

Facial prominences are active centers of growth in the mesenchyme (underlying ectoderm) and this connective tissue is continuous from one prominence to the other. Facial prominences are produced by proliferation and migration of ***neural crest cells***.

By the end of the 4th week bilateral oval thickenings of ectoderm – ***nasal (olfactory) placodes*** appear in frontonasal prominence. Mesenchyme in the margins of the nasal placodes proliferate producing horseshoe elevations – the ***medial and lateral nasal processes***.

The lower lip and jaw are the first parts of the face to form. They result from merging of the medial ends of the mandibular prominences in the midline.

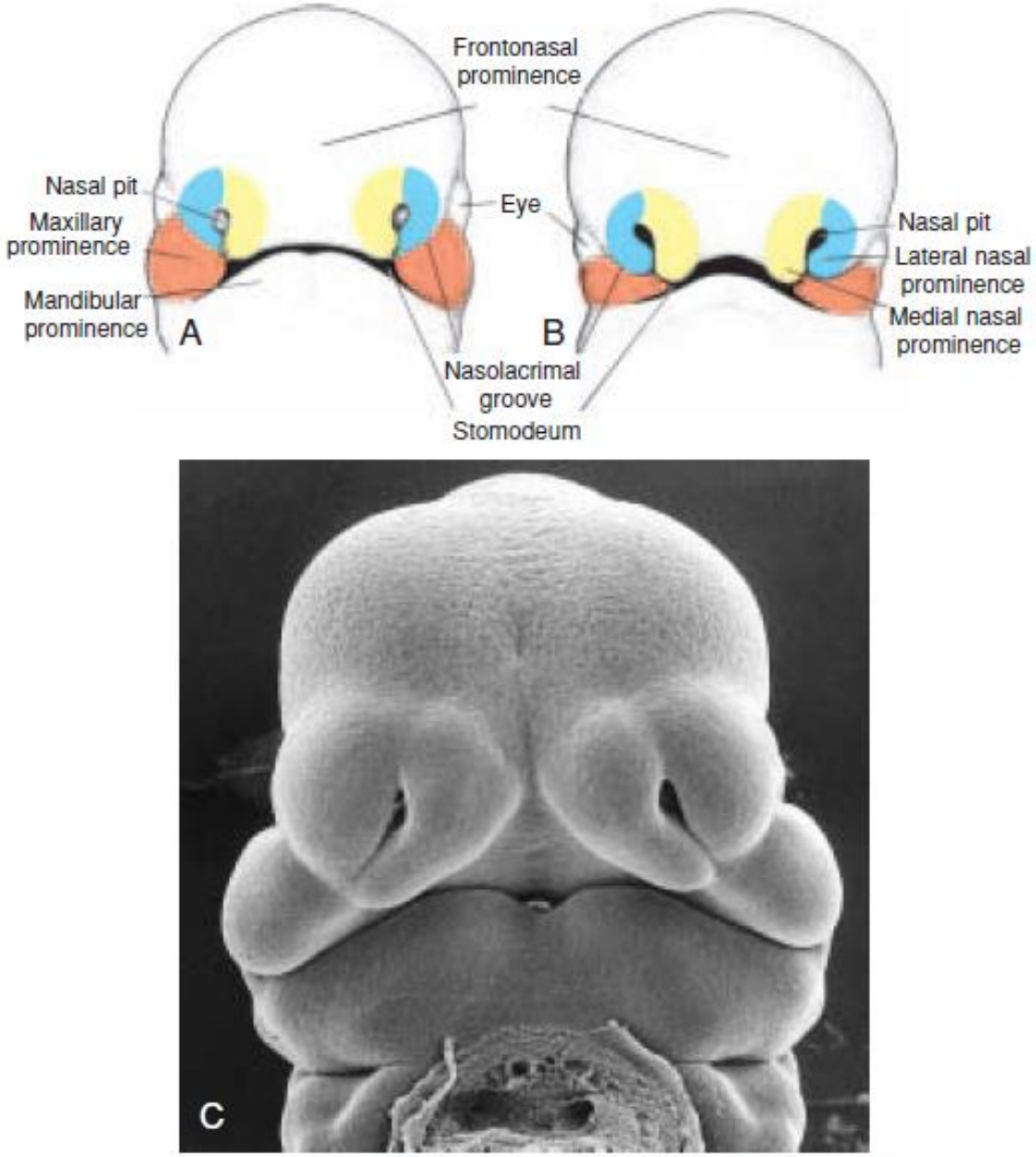


Figure 15.22 Frontal aspect of the face. **A.** 5-week embryo. **B.** 6-week embryo. The nasal prominences are gradually separated from the maxillary prominence by deep furrows. **C.** Scanning electron micrograph of a mouse embryo at a stage similar to that of **B.**

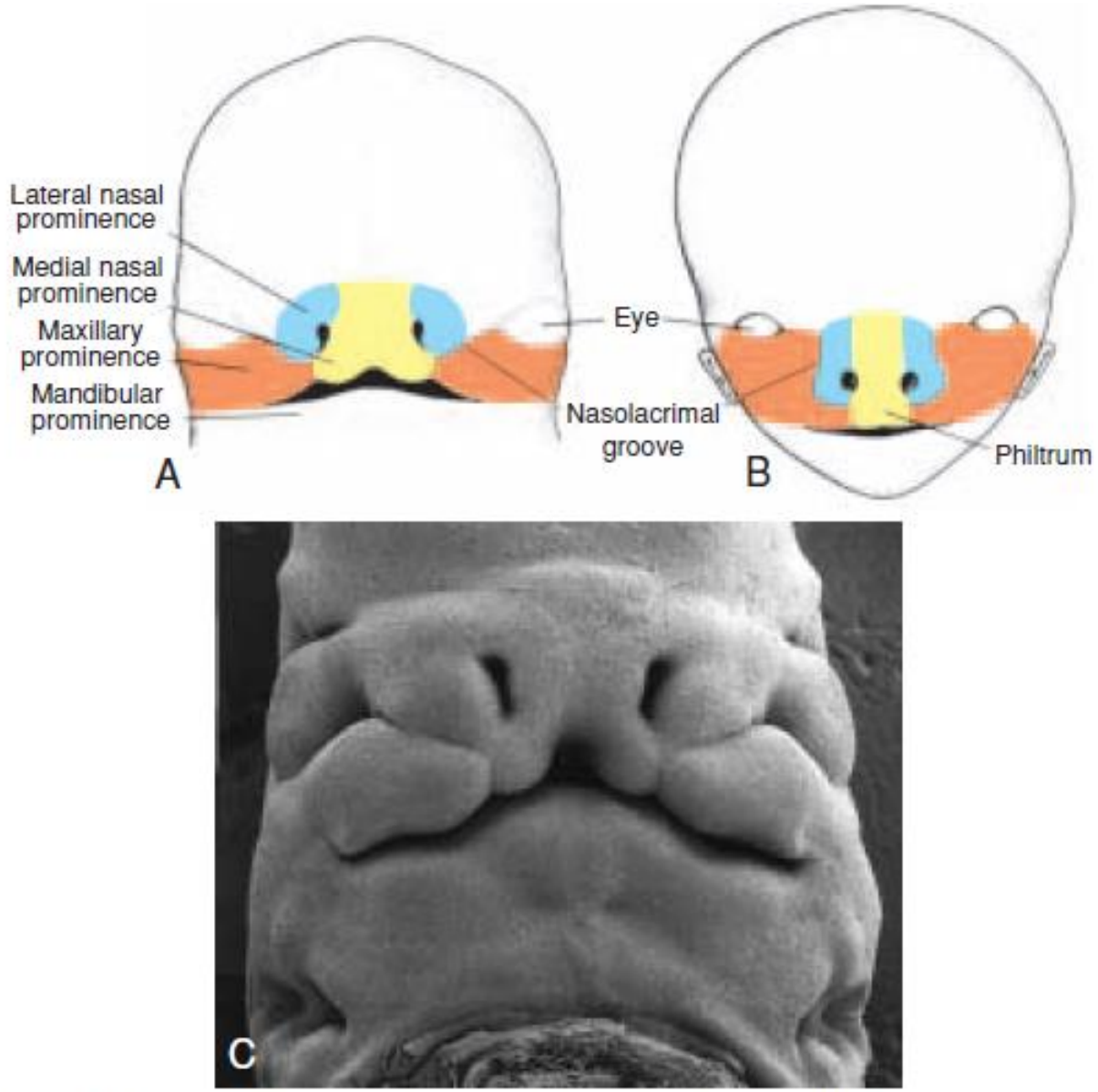


Figure 15.23 Frontal aspect of the face. **A.** 7-week embryo. Maxillary prominences have fused with the medial nasal prominences. **B.** 10-week embryo. **C.** Scanning electron micrograph of a human embryo at a stage similar to that of **A.**

During the 5th week, the nasal placodes invaginate to form the ***nasal pit*** surrounded laterally by the lateral nasal process and medially by the medial nasal process. The nasal pits are the primordium of the anterior **nares and nasal cavities**.

By the 6th week the maxillary prominences grow medially toward the nasal processes. MxP is separated from the lateral nasal process by a cleft called ***naso-optic furrow***. It fuses with the lateral nasal process at the naso-optic furrow to form the lower border of the developing orbit. Ectoderm in this groove forms a solid epithelial cord that detaches from surface ectoderm and canalizes to form the **nasolacrimal duct**. The maxillary prominences compress the medial nasal processes and push them medially.

By the 7th week, the compressed medial nasal processes merge and fuse to form **philtrum of the upper lip**. Fusion of the maxillary prominences with the medial nasal processes results in formation of the **upper lip**.

The lips are separated from the gums by linear thickening of ectoderm – the ***labiogigival laminae*** which grow into the underlying mesoderm and later degenerate creating the **labiogingival groove** between the lips and gums. A small area persists in the median plane to form **frenulum of the upper lip**.

Intermaxillary segment

The structure formed by the two merged medial nasal processes is known as the intermaxillary segment. It is composed of:

A ***labial component***, which forms the philtrum of the upper lip.

An ***upper jaw component*** which carries the four incisor teeth.

A ***palatal component***, which forms the triangular primary palate.

Palate

The palate develops from two sources; the ***primary palate*** and the ***secondary palate***.

The primary palate develops from the deep part of the ***intermaxillary segment***.

During the 6th week the secondary palate develops by two shelf-like ingrowths from the inner aspects of the ***maxillary prominences*** called ***palatal shelves.*** Palatal shelves are directed obliquely on each side of the tongue. In week 7, palatal shelves ascend above the tongue and fuse together forming the secondary palate. Anteriorly, fusion with the triangular primary palate and the incisive foramen is the midline landmark between primary and secondary palate. Bone forms and extends from maxillae into palatal shelves to form the hard palate. The posterior part remains unossified to form the soft palate and uvula.

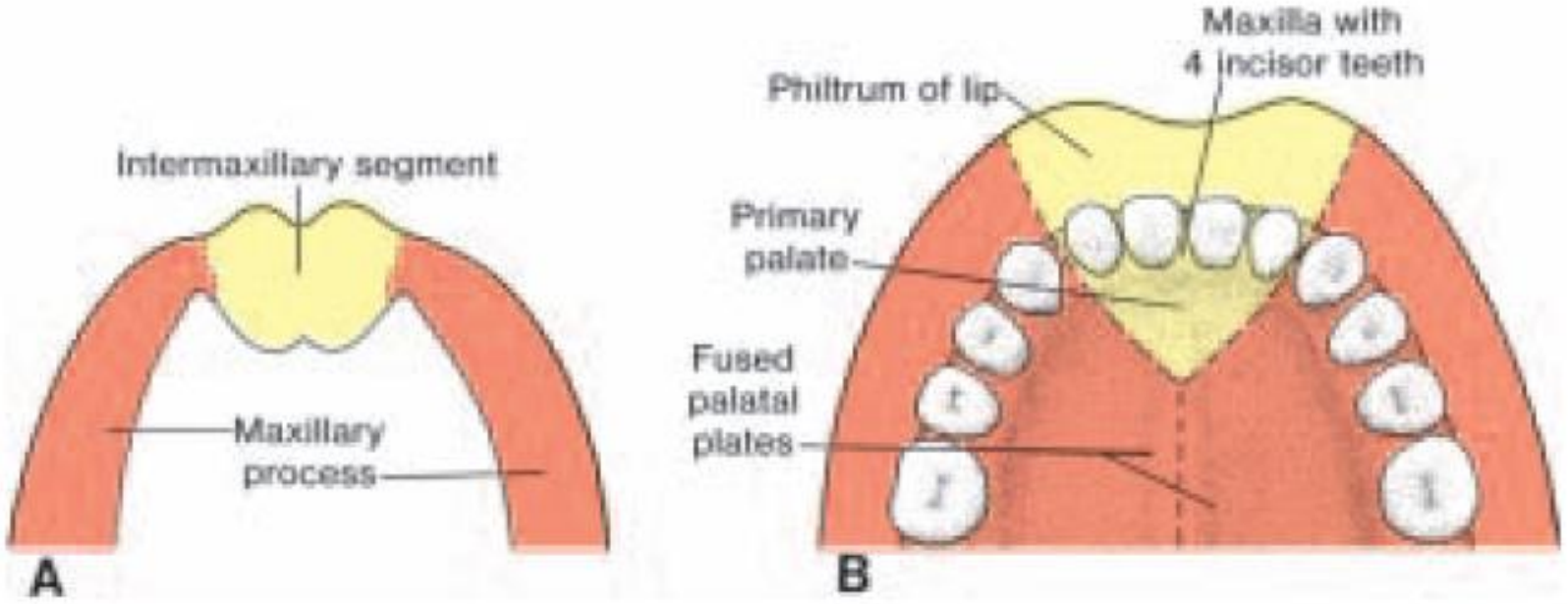


Figure 15.24 A. Intermaxillary segment and maxillary processes. **B.** The intermaxillary segment giving rise to the philtrum of the upper lip, the median part of the maxillary bone with its four incisor teeth, and the triangular primary palate.

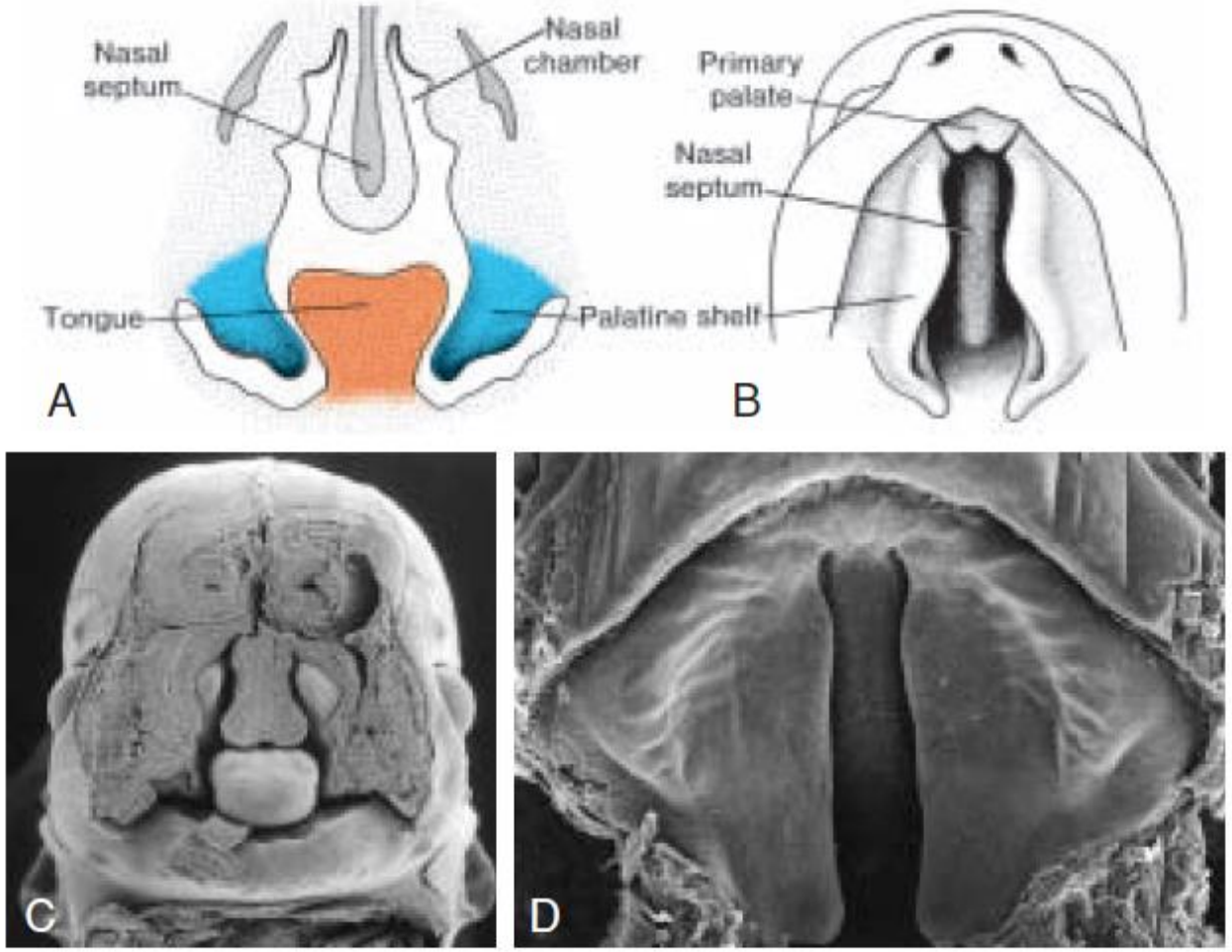


Figure 15.25 A. Frontal section through the head of a 6.5-week-old embryo. The palatine shelves are in the vertical position on each side of the tongue. **B.** Ventral view of the palatine shelves after removal of the lower jaw and the tongue. Note the clefts between the primary triangular palate and the palatine shelves, which are still vertical. **C.** Scanning electron micrograph of a mouse embryo at a stage similar to that of **A.** **D.** Palatal shelves at a stage slightly older than those in **B.** The shelves have elevated, but they are widely separated. The primary palate has fused with the secondary palatal shelves.

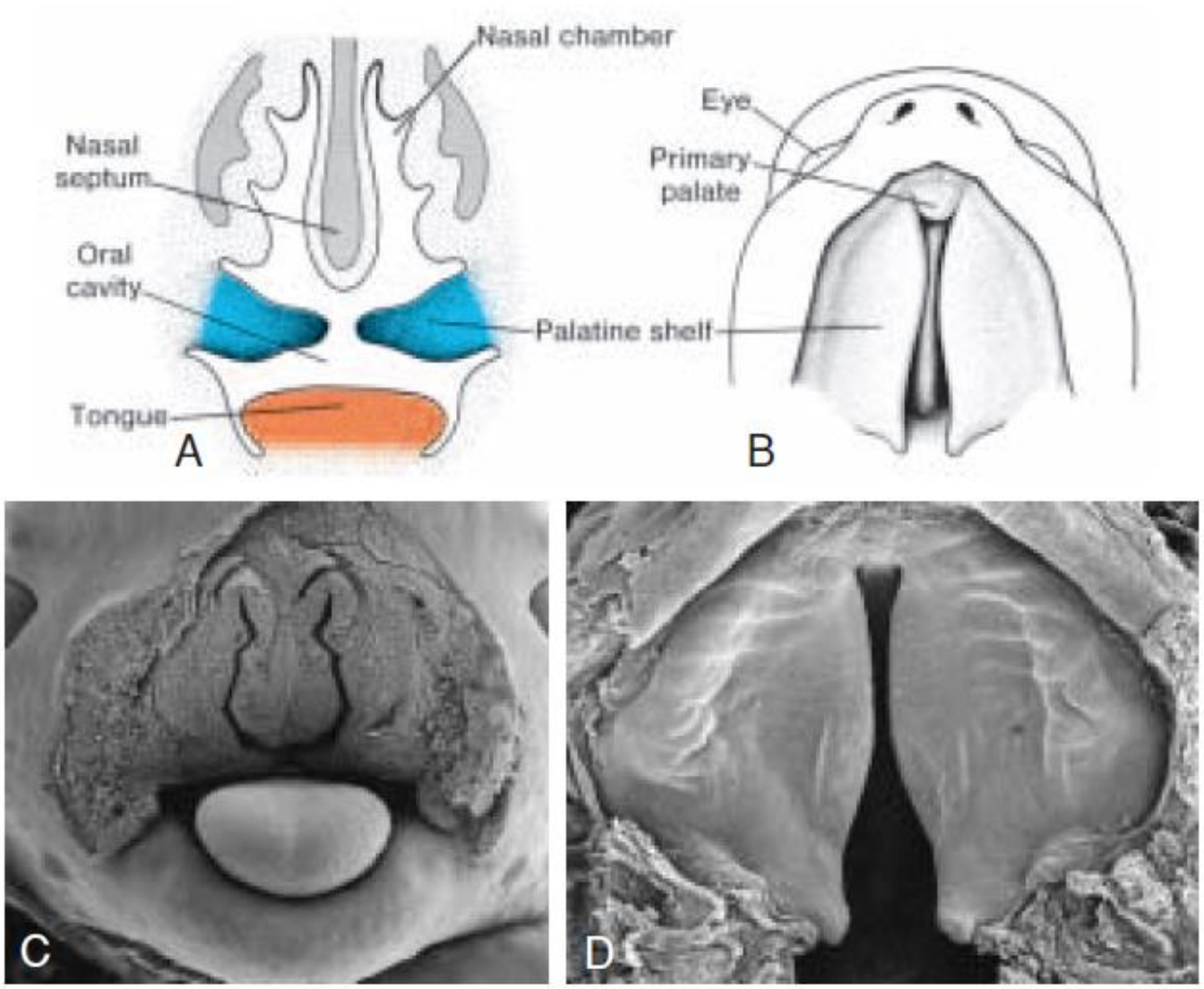


Figure 15.26 A. Frontal section through the head of a 7.5-week embryo. The tongue has moved downward, and the palatine shelves have reached a horizontal position. **B.** Ventral view of the palatine shelves after removal of the lower jaw and tongue. The shelves are horizontal. Note the nasal septum. **C.** Scanning electron micrograph of a mouse embryo at a stage similar to that of **A. D.** Palatal shelves at a stage similar to that of **B.**

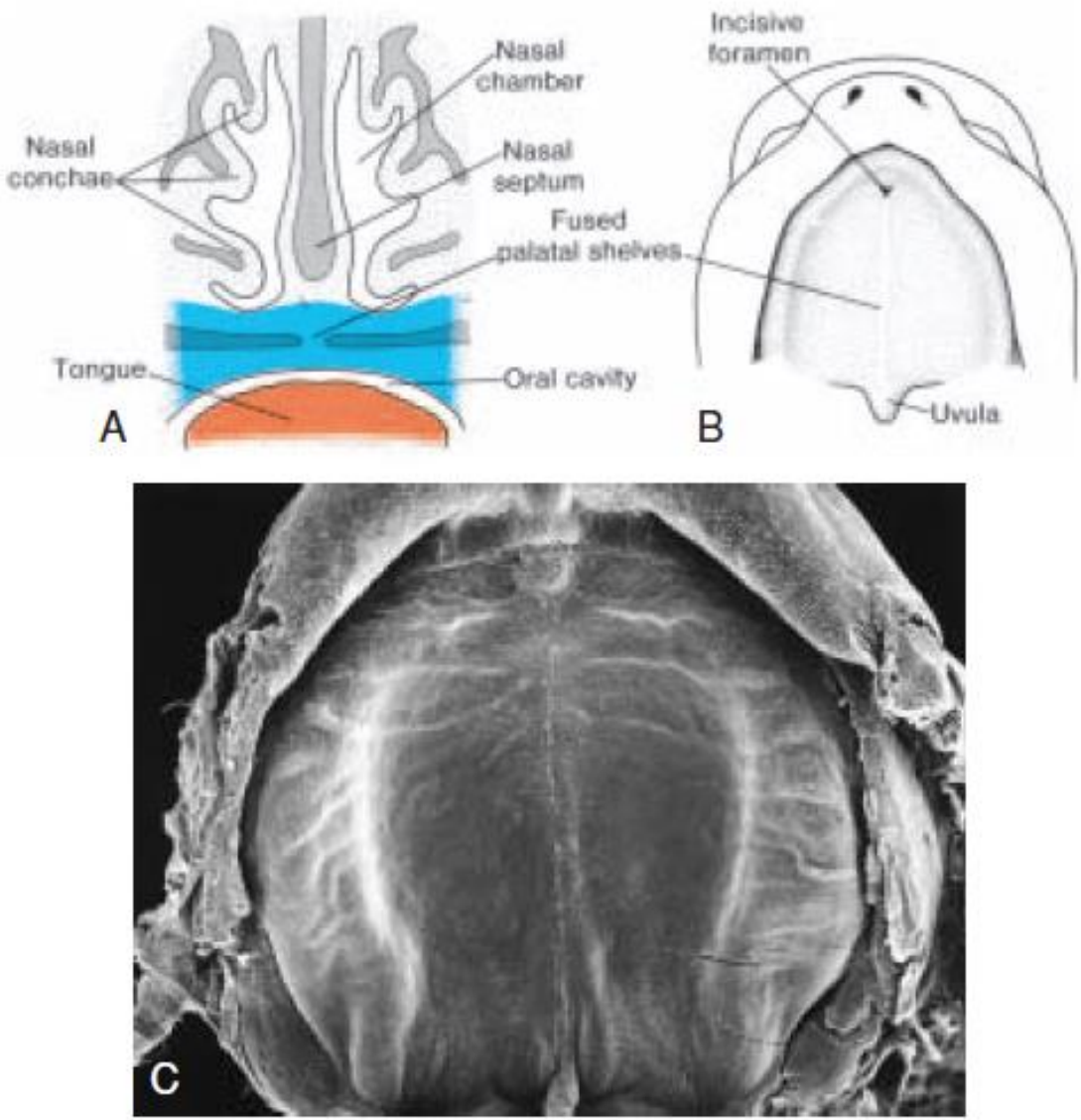


Figure 15.27 A. Frontal section through the head of a 10-week embryo. The two palatine shelves have fused with each other and with the nasal septum. **B.** Ventral view of the palate. The incisive foramen forms the midline between the primary and secondary palate. **C.** Scanning electron micrograph of the palatal shelves of a mouse embryo at a stage similar to that of **B.**

Nasal septum

The nasal septum develops as down growth from the internal parts of the merged ***medial nasal processes.*** Fusion of the nasal septum with the cephalic aspect of the palate is completed during the 12^{th} week.

Nasal cavities

Nasal pits deepen to form the nasal cavities. After the palate develops the **choanae** are located at the junction of the nasal cavities and the pharynx.

The nasal pits form the **nostrils**. The lateral nasal process forms the roof and lateral wall of the nasal cavity. Ectoderm in the roof becomes specialized to form olfactory epithelium. The floor of the nasal cavity is formed as a result of fusion of the nasal septum with the palate. Nasal **conchae** develop as inward projections from the lateral nasal wall. **Paranasal sinuses** develop during the fetal life as diverticulae from the lateral nasal process.. The frontal sinus develops from the anterior ethmoidal cells, and the sphenoidal sinus develops from the posterior ethmoidal cells. During early childhood air sinuses grow slowly and cells inside degenerate and are replaced by air. Growth of sinuses reaches maximum size at puberty.

During early fetal period the nose is flat and the mandible is underdeveloped. As the brain enlarges a prominent forehead is created and the eyes move medially. As the head and mandible enlarge the auricles rise to the level of the eyes. Prenatal smallness of the face results from:

-Rudimentary upper and lower jaws.

-Small size of the nose and paranasal sinuses.

-Unerupted teeth.

Anomalies of the face

1-Cleft lip and / or palate

-Cleft lip (1/1000 more common in males) and cleft palate (1/2500 more common in females) are mainly due to multifactorial inheritance (genetic and non-genetic factors). Some cases of cleft lip and palate result from use of anticonvulsant drugs. Few cases are associated with syndromes, eg.trisomy 13.

-Cleft lip and or palate result in facial disfigurement and defective speech.

-Cleft lip and palate are mainly due to partial or complete lack of fusion of the maxillary prominence and the medial nasal process on one or both sides.

Cleft lip

a)**Unilateral cleft upper lip** is due to failure of maxillary prominence to fuse with the medial nasal process in one side.

b)**Bilateral cleft upper lip**, is usually associated with anterior cleft palate.

It is due to failure of maxillary prominences to fuse with the medial nasal processes.

c)**Median cleft upper lip** is very rare.

It is due to failure of medial nasal processes to merge.

d)**Cleft lower lip,** very rare and is due to failure of mandibular prominences to fuse.

Cleft palate

Incidence is 1/2500, more often in females than males and this could be due to the fact that in females palatal shelves fuse one week later than males.

Using the incisive fossa as a reference landmark, cleft palate is classified into anterior and posterior clefts.

a)**Cleft anterior (primary) palate**, is due to failure of fusion of primary palate with palatine shelves.

b)**Cleft of posterior (secondary) palate** and uvula, is due to failure of palatine shelves to fuse together. It can be partial or complete. Failure of fusion may include the nasal septum.

c)**Cleft of anterior and posterior palate**, is due to failure of palatine shelves to fuse together and with the primary palate.. It can be total unilateral or total bilateral. It is usually associated with cleft upper lip and jaw.

2-**Oblique facial cleft** is due to failure of maxillary prominence to fuse with the lateral nasal process. Nasolacrimal duct is exposed.

3-**Microstomia** is small mouth resulting from excessive merging of mesenchymal masses in the maxillary and mandibular prominences.

4-**Macrostomia** is excessively large mouth due to excessive furrows between maxillary and mandibular prominences.

Anomalies of the nose

a)**Absent nose** is due to failure of nasal placodes to develop.

b)**Single nostril** occurs when only one nasal placode develops.

c)**Bifid nose** is due to failure of fusion of medial nasal processes.

d)**Median nasal furrow** is due to split in the nasal septum.

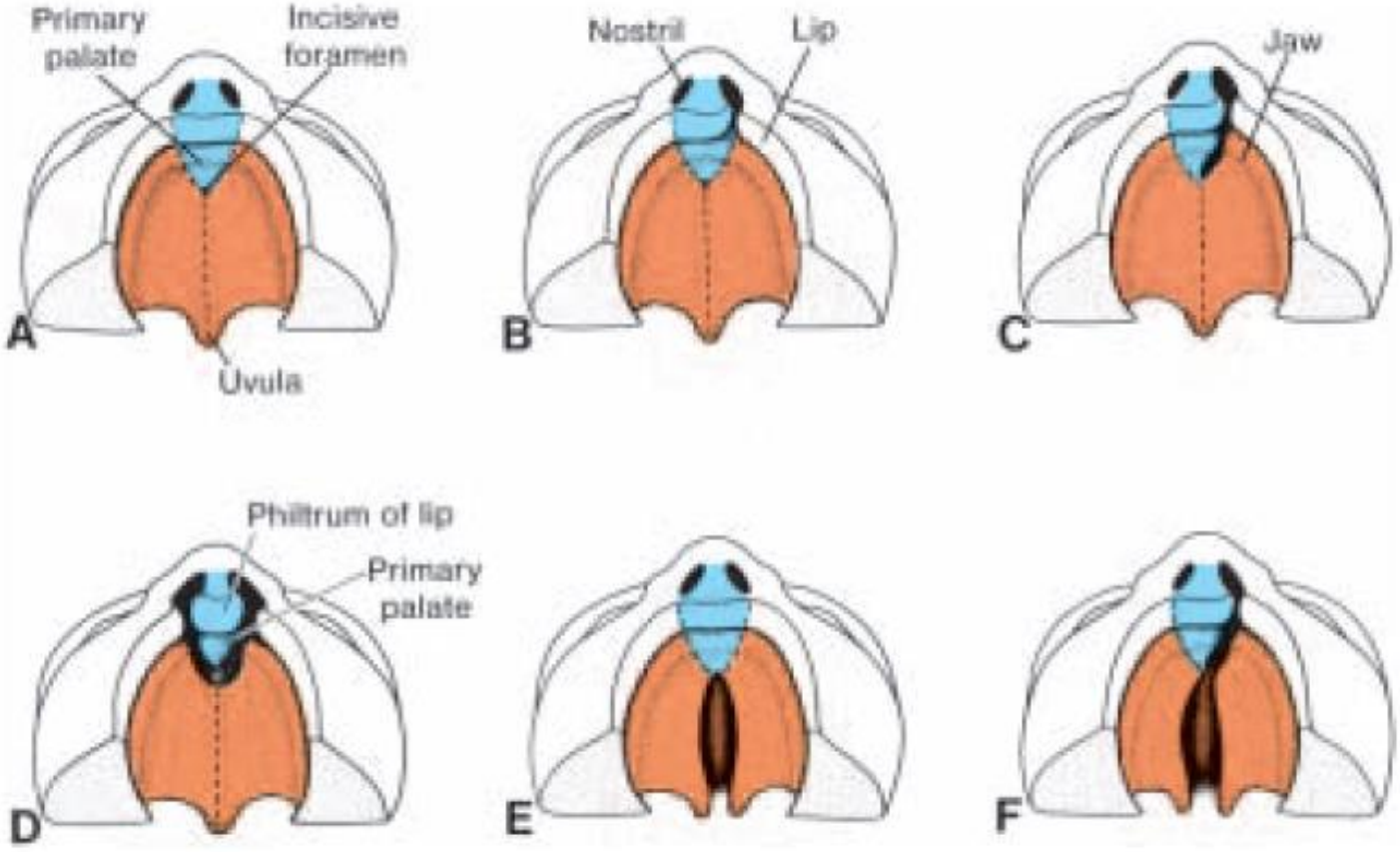


Figure 15.28 Ventral view of the palate, gum, lip, and nose. **A.** Normal. **B.** Unilateral cleft lip extending into the nose. **C.** Unilateral cleft involving the lip and jaw and extending to the incisive foramen. **D.** Bilateral cleft involving the lip and jaw. **E.** Isolated cleft palate. **F.** Cleft palate combined with unilateral anterior cleft lip.

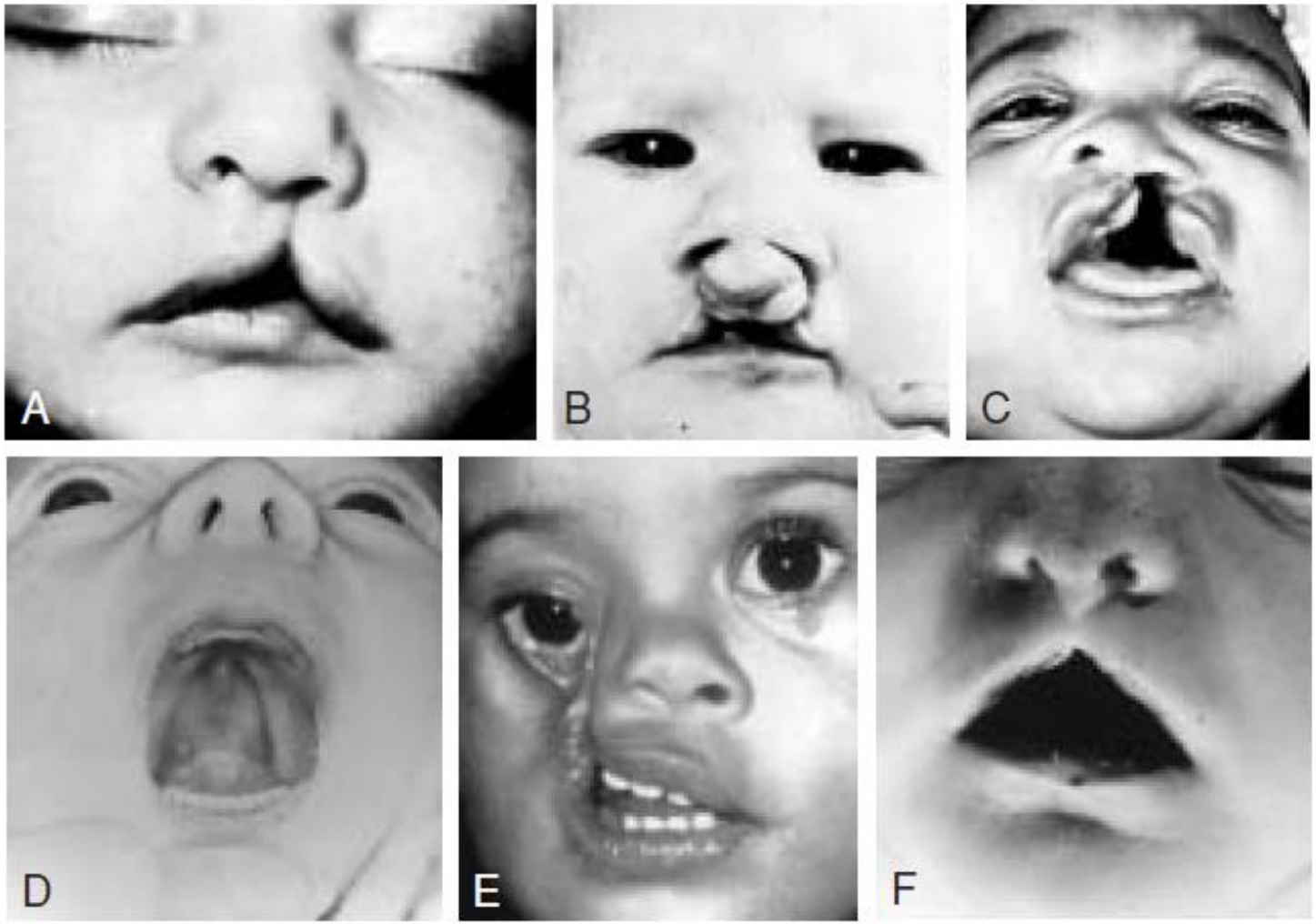


Figure 15.29 A. Incomplete cleft lip. **B.** Bilateral cleft lip. **C.** Cleft lip, cleft jaw, and cleft palate. **D.** Isolated cleft palate. **E.** Oblique facial cleft. **F.** Midline cleft lip.

Development of teeth

Teeth are derived from ectoderm and mesoderm. Neural crest cells play an important role in embryogenesis of teeth. Many factors control and affect teeth development involving genetic and environmental factors.

-By the 6th week, ectodermal epithelial lining of the oral cavity forms a C-shaped ***dental lamina*** along the upper and lower jaws. Dental lamina gives 10 ***dental buds*** in each jaw.

-**Cap stage of tooth development**

The deep surface of the dental bud is invaginated resulting in cap stage of tooth development. This cap consists of an outer and inner dental epithelium, and a central core of loosely woven tissue – the ***stellate reticulum***. Mesenchyme of neural crest origin forms the **dental papilla.**

-**Bell stage of tooth development**

Indentation of the dental cap deepens and the tooth takes the shape of a bell.

Mesenchyme cells of the papilla differentiate into **odontoblasts,** which produce **dentin. With** thickening of dentin layer odontoblasts retreat into the dental papilla, leaving cytoplasmic (dental) processes in the dentin. Odontoblasts persist throughout the life of the tooth and continuously providing prodentin which becomes dentin.

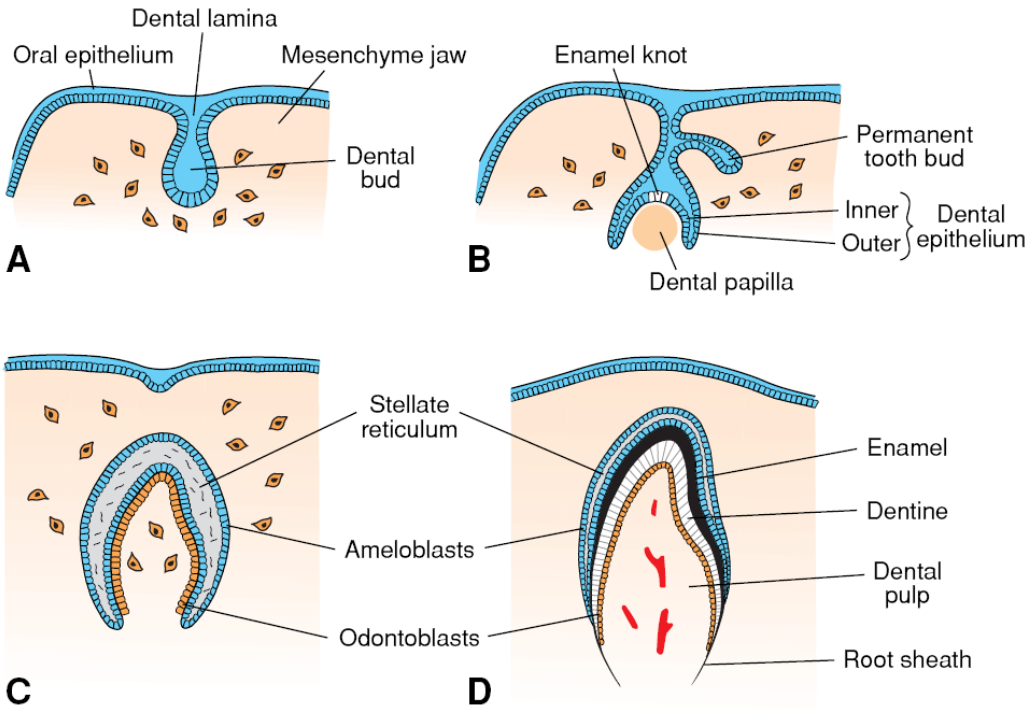


Figure 15.31 Formation of the tooth at successive stages of development. **A.** Bud stage; 8 weeks. **B.** Cap stage; 10 weeks. **C.** Bell stage; 3 months. **D.** 6 months.

The remaining cells of the dental papilla form the **pulp of tooth**. Meanwhile cells of the outer dental epithelium differentiate into **ameloblasts.** These cells produce e**namel** that is deposited over the dentin. The contact layer between enamel and dentin is called enamel dentin junction. Enamel is first laid down at the apex and then spreads toward the neck of the tooth. When enamel thickens, ameloblasts retreat into the stellate reticulum. Here they regress temporarily leaving a thin membrane (***dental cuticle***) on the surface of enamel. After tooth erupts this membrane sloughs off.

Formation of the tooth root

Formation of the root begins when dental epithelium penetrates into underlying mesenchyme to form the epithelial root sheath. Cells of the dental papilla lay down a layer of dentin continous with that of the crown. As more dentin is deposited, the pulp chamber narrows and finally forms a canal containing blood vessels and nerves of the tooth (root canal).

Formation of cementum

Mesenchymal cells outside the tooth and in contact with dentin of the root differentiate into **cementoblasts**. These cells produce a thin layer of specialized bone – **cementum.** Outside the cement layer, mesenchyme gives the **periodontal ligament** which holds tooth firmly in position and function as shock absorber.

Tooth eruption

With further lengthening of the root the crown is pushed through the overlying tissue into the oral cavity. Eruption of deciduous (milk) teeth occurs 6-24 months after birth. Buds for permanent teeth are located on the lingual aspect of milk teeth. They are formed during the 3rd month. These buds remain dormant until the 6th year when they begin to grow pushing against the underside of the corresponding milk teeth and aiding in their shedding.

Anomalies of teeth

1-**Natal teeth** are erupted at birth. Usually involve mandibular incisors that may be abnormally formed and have little enamel.

2-**Enamel hypoplasia** is defective enamel formation. It causes pits and fissures in enamel. Various factors may injure ameloblasts, e.g.measles, vitamen D deficiency and tetracyclines.

3-**Abnormal shape of teeth**

-Enamel pearls which are attached to the tooth and they are formed by aberrant groups of ameloblasts.

-Slender tapering (peg-shaped) incisors.

-Screwdriver incisors with central notches (congenital syphilis).

4-**Abnormal number of teeth**

-Supernumerrary teeth are usually in the maxillary incisors. They erupt behind normal teeth.

-Partial anodontia is a familial anomaly with missing one or more of the teeth.

-Complete anodontia is very rare.

5-**Abnormal size**

Macordontia or microdontia.

6-**Fused teeth**

Sometimes permanent and deciduous teeth fuse together.

7-**Dentigerous cyst** containing unerupted tooth.

8-**Amelogenesis imperfecta** is abnormal enamel formation. Enamel is soft and friable because of hypocalcification and the teeth are yellow or brown in color.

9-**Dentinogenesis imperfecta** Is autosomal dominant trait affecting chromosome 4. Teeth are brown to gray-blue. Odontoblasts fail to differentiate normally, and poorly calcified dentin results in enamel wear rapidly exposing dentin.

10-**Discolored teeth**

Foreign substances incorporated in enamel or dentin result in discoloring teeth. Hemolytic diseases may produce black discoloration of teeth. Tetracyclines produce yellow-brownish coloration of teeth.

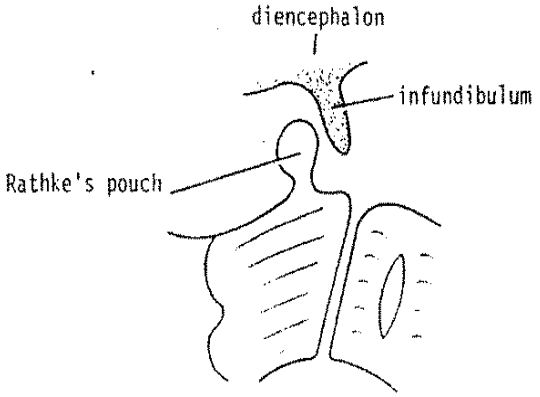
PITUITARY GLAND

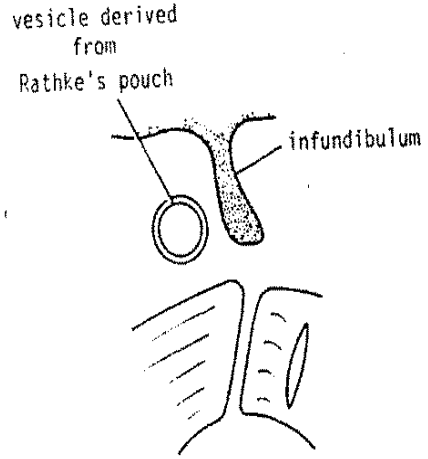
The pituitary gland is ectodermal in origin and is derived from two sources:

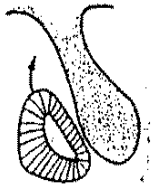
1-From the roof of the mouth a diverticulum grows superiorly called **Rathke's pouch.**

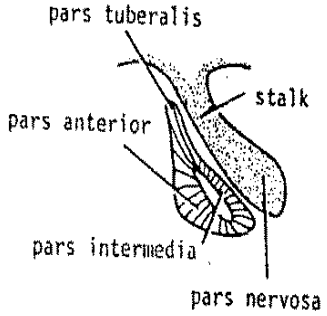
2-From the floor of the diencephalon a divirticulum grows inferiorly called the **Infundibulum.**

Rathke’s pouch comes into contact with the anterior surface of the infundibulum, and its connection with the oral epithelium and disappears . Rathke's pouch is now a vesicle that flattens itself around the anterior and lateral surfaces of the infundibulum . Cells of the anterior wall of the vesicle proliferate and form **pars distalis** , and from its upper part there is a cellular extension that grows superiorly around the stalk of the infundibulum, forming **pars tuberalis** . Cells of the posterior wall of the vesicle develop slowly and form **pars intermedia**. The cavity of the vesicle is reduced to a **residual cleft** . Meanwhile the infundibulum has differentiated into the stalk and the posterior **lobe(pars nervosa**). Neuroglia of the pars nervosa differentiate into pituicytes . Nerve cells in the hypothalamus give rise to nerve fibers that grow inferiorly into the pars nervosa, and neurosecretory activity begins in late fetal life. ‘During the third month cells of the pars distalis differentiate into chromophil and chromophobe cells, and the cells become arranged in columns around blood sinusoids. The gland becomes vascularized, and a portal system of blood vessels is established .


diencephalon
infundibulum
Rathke's pouch


vesicle derived
from
Rathke's pouch
infundibulum




pars tuberalis
stalk
pars anterior
pars intermedia
pars nervosa

Congenital anomalies of the pituitary gland

1-Absence of the pituitary: congenital absence of the pituitary is incompatible with life. Infants born with no anterior lobe have maldeveloped adrenals, thyroid and testes.

Congenital absence of cells secreting specific hormone is recognized; for example hereditary growth hormone deficiency.

2-Pharyngeal pituitary: if part of Rathke's pouch remain attached to the roof of the stomodeum .

3-Craniopharyngioma is development of a pituitary tumour in the pharynx from remnants of pitutary pouch.

Development of the thyroid gland

Thyroid is endodermal in origin and appears as a **thyroid diverticulum** at a midpoint between tuberculum impar and copula. It descends as a bilobed structure in front of hyoid bone. It remains connected to tongue by a narrow canal, the **thyroglossal duct**. The thyroid reaches its final position in front of trachea by the 7th week. By then it has acquired a small median isthmus and two lateral lobes. The thyroglossal duct obliterates and disappeas. Aggregations of cells are invaded by blood vessels and change into clusters. These clusters will form the *thyroid follicles* which start to function by the 3rd month when follicles containing colloid become visible. The colloid is the source of thyroxine and triiodothyronine. *Parafollicular (C) cells* are derived from the **ultimobranchial body** of the 5th pharyngeal pouch.

Anomalies of the thyroid

1-**Agenesis** of the thyroid which results in **congenital cretinism**.

2-Failure of descent may result in:

a)**Lingual thyroid** is found just behind foramen cecum.

b)**Sublingual thyroid** is found just below the hyoid bone.

3-Failure of obliteration of the thyroglossal duct may result in:

a)Complete failure of obliteration results in **thyroglossal fistula**. The fistula may be connected to the tongue or to the outside.

b)Incomplete failure of obliteration may result in **thyroglossal sinus** or **thyroglossal cyst**. The thyroglossal cyst is located along the path of decent but usually in the midline of the neck.

4-Congenital **hyperfunction** occurs in babies born to thyrotoxicosis mothers.

5-Congenital **hypofunction** is associated with congenital cretinism.

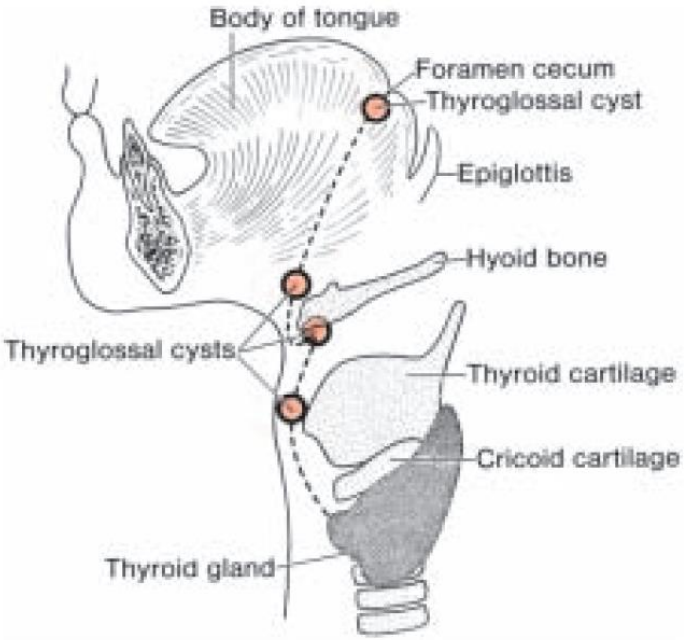


Figure 15.19 Thyroglossal cysts. These cysts, most frequently found in the hyoid region, are always close to the midline.

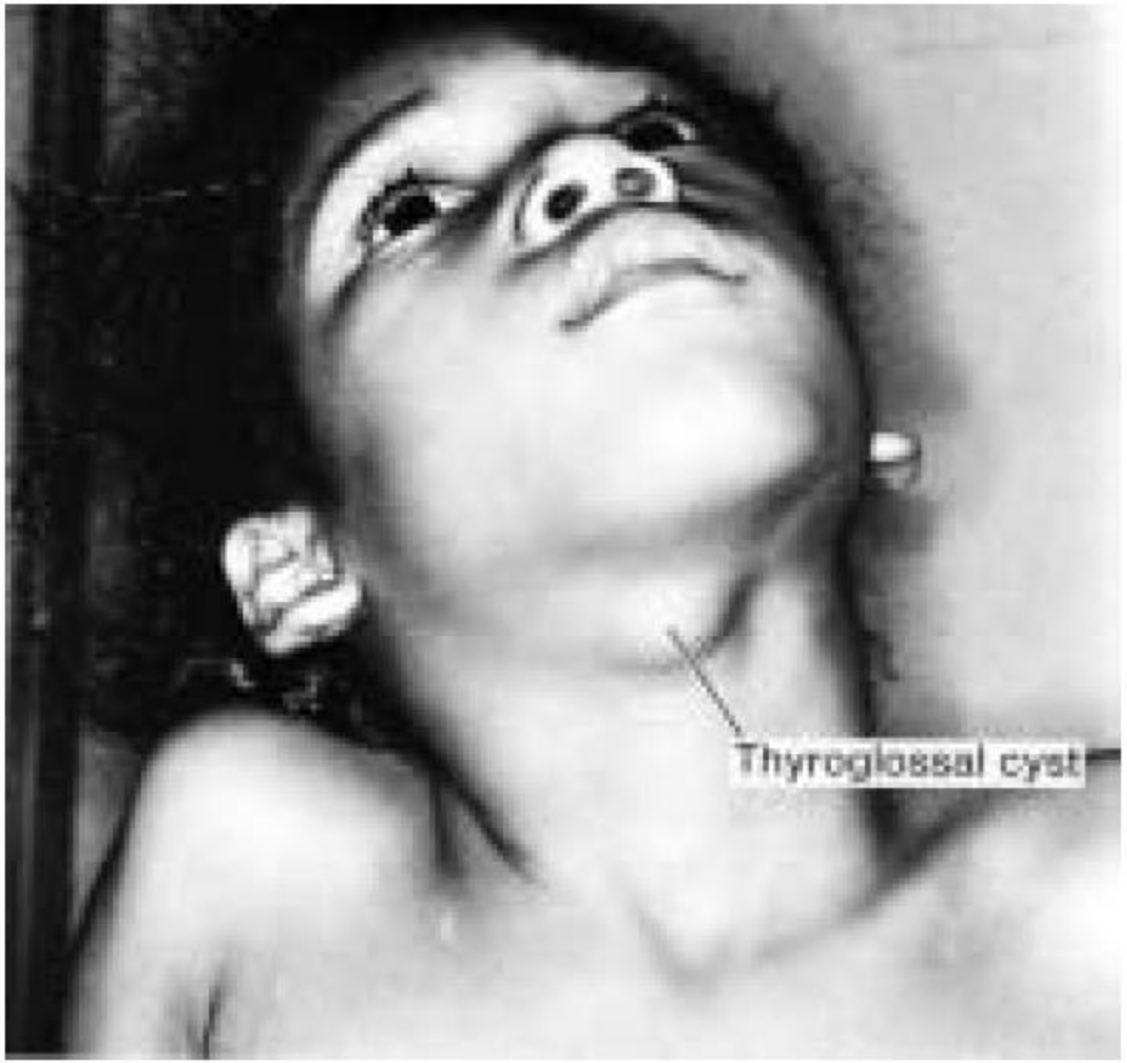
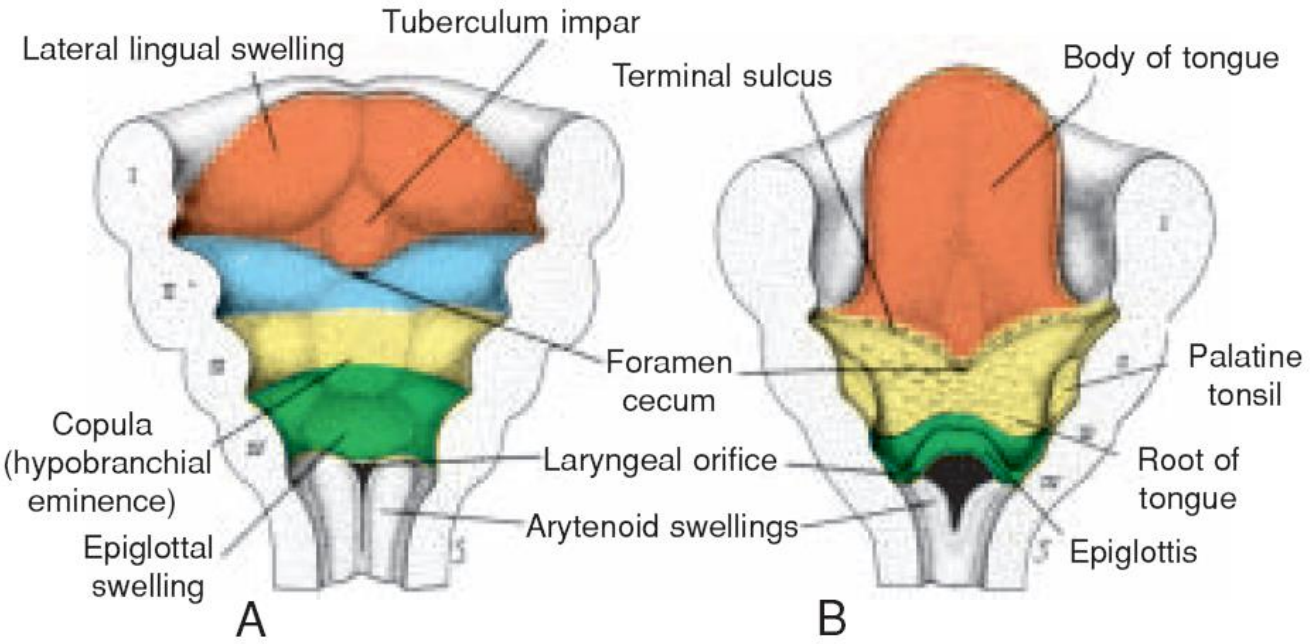


Figure 15.20 Thyroglossal cyst. These cysts, which are remnants of the
duct, may be anywhere along the migration pathway of the thyroid gland. T
monly found behind the arch of the hyoid bone. An important diagnostic c
is their midline location.

Development of the tongue

Anterior two thirds of the tongue

By the end of the 4th week mesenchymal proliferations of the ventromedial parts of the first pair of pharyngeal arches result in appearance of three elevations of endoderm of the floor of the pharynx; a median swelling called **tuberculum impar** followed by two lateral **lingual swellings**. The two lingual swellings increase in size, overgrow the tuberculum impar and merge forming the anterior two-thirds of the tongue. Fusion of the lingual swellings is indicated by the *median sulcus* of the tongue and internally by the fibrous *lingual septum*. Sensory innervation of the mucosa of the anterior 2/3 is by lingual nerve, branch of Mandibular, the nerve of the first pharyngeal arch. Chorda tympani of facial nerve supplies taste of the anterior 2/3 of the tongue.


Lateral lingual swelling
Tuberculum impar
Terminal sulcus
Body of tongue
Foramen cecum
Palatine tonsil
Copula (hypobranchial eminence)
Laryngeal orifice
Root of tongue
Arytenoid swellings
Epiglottal swelling
Epiglottis
A
B

Posterior one-third of the tongue

Two median swellings are formed by proliferation of the ventromedial mesoderm of the 2nd (**copula**), 3rd and 4th (**hypobranchial eminence**) pharyngeal arches. The copula is overgrown by the hypobranchial eminence and disappears. The cranial part of the hypobranchial eminence forms the posterior 1/3 (pharyngeal part) of the tongue. Line of fusion of the anterior 2/3 and posterior 1/3 is indicated by a V-shaped groove – the *sulcus terminalis*. The caudal part of the hypobranchial eminence forms the epiglottis. Immediately behind this eminence is the laryngeal orifice. The glossopharyngeal nerve (nerve of 3rd arch) supplies sensations of the posterior 1/3 of the tongue and the vallate papillae. The superior laryngeal nerve of vagus (nerve of 4th arch) innervates the epiglottis. The facial nerve (nerve of 2nd arch) does not supply the posterior 1/3 of the tongue because the second arch component, copula, is overgrown by the hypobranchial eminence and disappears.

Pharyngeal arch mesenchyme forms the connective tissue and vasculature of the tongue. Muscles of the tongue are derived from myoblasts that migrate from 3 occipital (pos-otic) myotomes and pull with them their nerve supply which is the hypoglossal nerve.

Papillae and taste buds

-Lingual papillae appear towards the end of the 8^{th} week.

-Taste buds develop during the 11^{th} to the 13th week and most of them are on the dorsum of the tongue.

-Fetal response to taste takes place at the 28^{th} week.

-Cellular degeneration occurs to free the tongue from the floor of the mouth except the frenulum.

The entire tongue is within the mouth at birth, its posterior 1/3 descends into the oropharynx by the age of 4 years.

Anomalies of the tongue

1-Tongue-tie (**ankyloglossia)**,where the tongue is not freed from the floor of the mouth and is due to short frenulum. This anomaly disturbs speech and interferes with feeding.

2-**Macroglossia** is excessively enlarged tongue and is due to angioma or muscular hypertrophy.

3-**Microglossia** is small tongue and usually associated with small mandible (micrognathia).

4-**Bifid tongue** is due to failure of fusion of the two lingual sweelings.

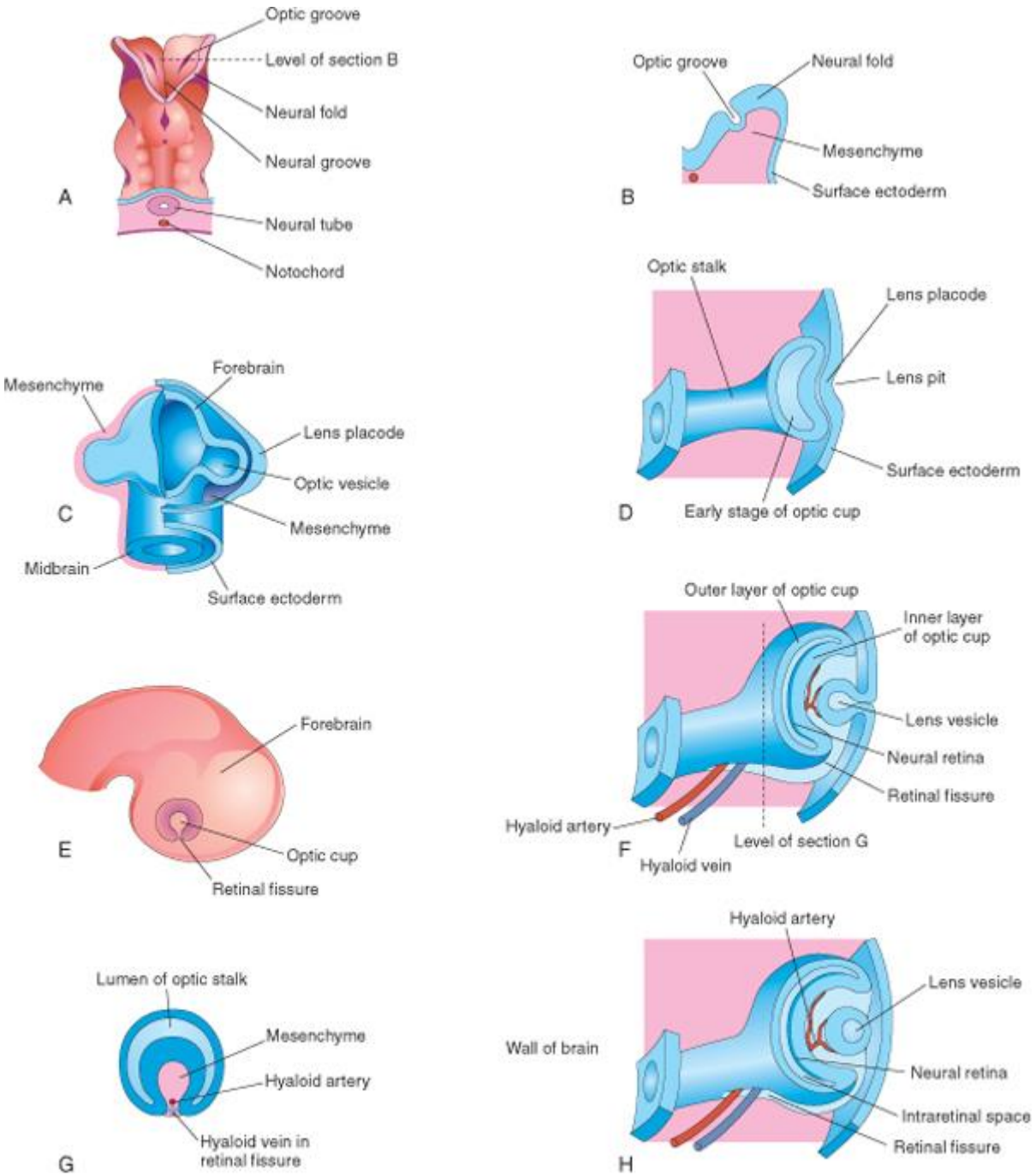
5-**Lingual cyst** is due to persistence of the thyroglossal duct.

DEVELOPMENT OF HUMAN EYE AND ASSOCIATED STRUCTURES

Embryonic sources

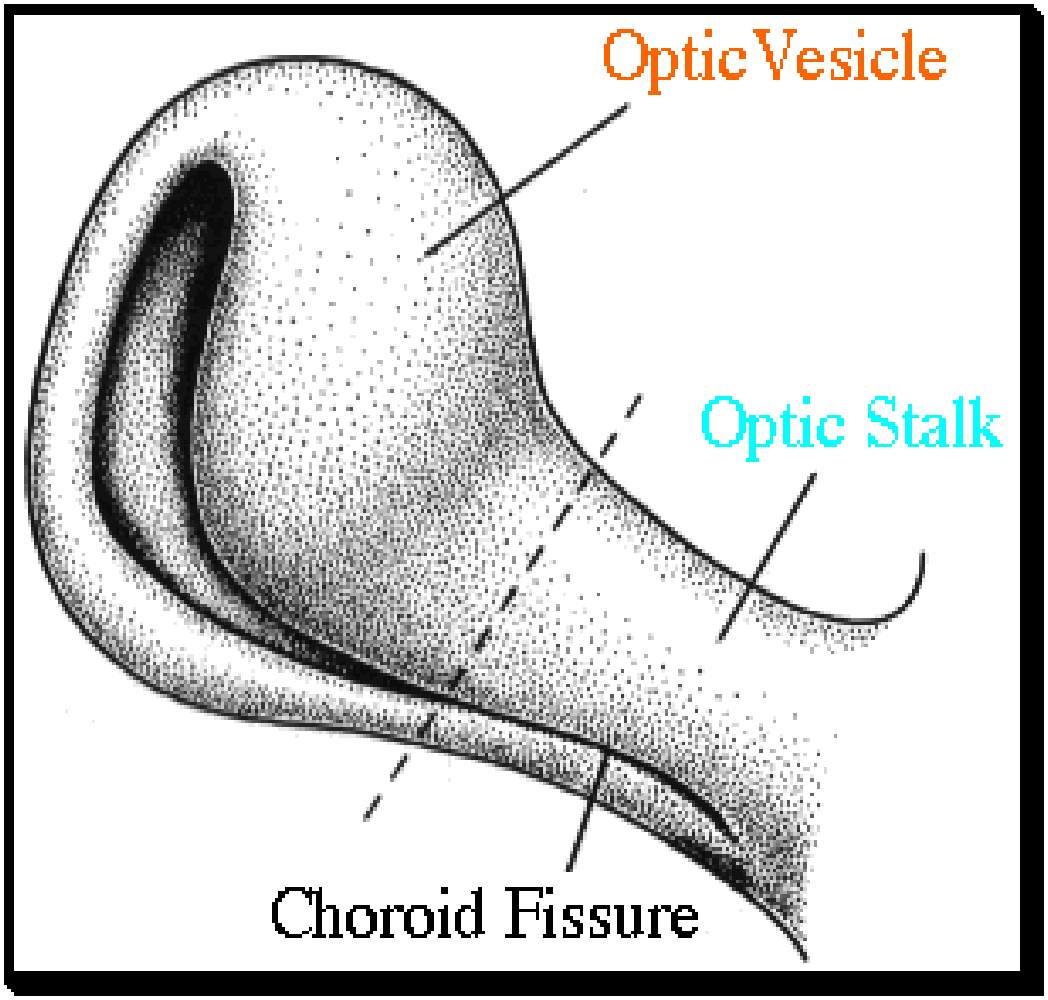
The eyes are derived from four sources:

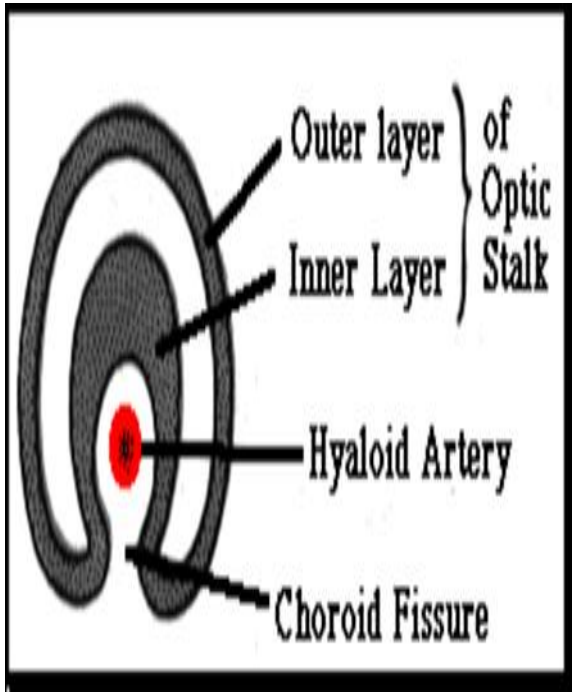
- The **neuroectoderm** of the forebrain
- The **surface ectoderm** of the head
- The **mesenchyme** between the above layers
- **Neural crest cells.**



- The optic vesicles as it grows makes contact with the surface ectoderm leading to its thickening to form **lens placodes**, the **lens primordia**.
- Formation of lens placodes is induced by the optic vesicles.
- The lens placodes invaginate to form **lens pits**.
- The edges of the pits approach each other and fuse to form spherical **lens vesicles**, which soon lose their connection with the surface ectoderm.

- As the lens vesicles are developing, the optic vesicles invaginate to form double-walled **optic cups**.
- The opening of each cup is large at first, but its rim infolds around the lens.
- The lens vesicles lose their connection with the surface ectoderm and enter the cavities of the optic cups.
- **Retinal fissures** (optic fissures) develop on the ventral surface of the optic cups and along the optic stalk.
  - The fissures contain vascular mesenchyme from which the hyaloid blood vessels develop.
- The **hyaloid artery**, a branch of the ophthalmic artery, supplies the inner layer of the optic cup, the lens vesicle, and the mesenchyme in the **cavity of the optic cup**.
- The **hyaloid vein** returns blood from these structures.
- As the edges of the retinal fissure fuse, the hyaloid vessels are enclosed within the **primordial optic nerve**.
- Distal parts of the hyaloid vessels eventually degenerate, but proximal parts persist as the **central artery** and **vein of the retina**.



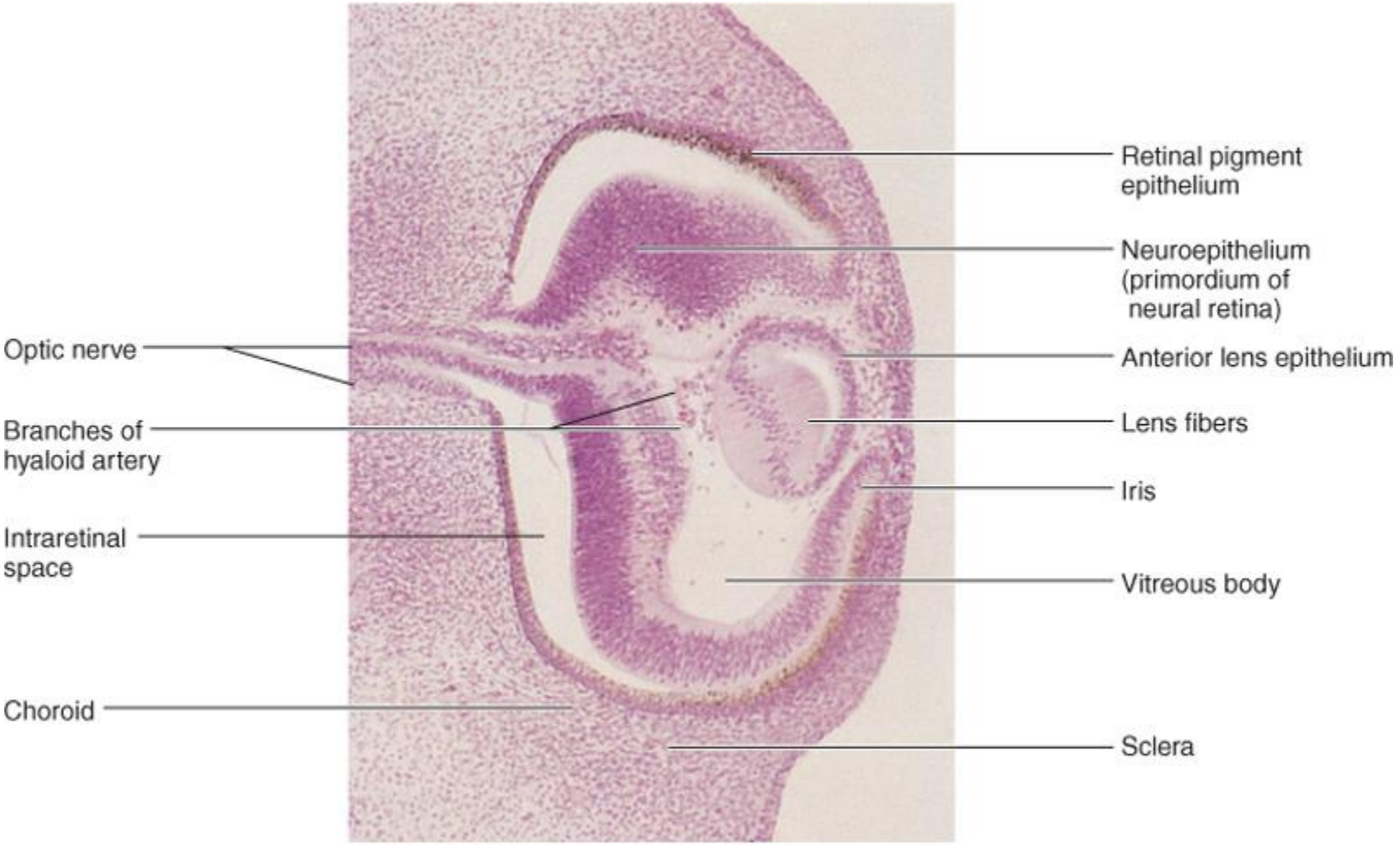


The optic vesicle and the optic stalk invaginate, forming the choroid fissure inferiorly.

DEVELOPMENT OF THE RETINA

- The retina develops from the walls of the *optic cup*, an outgrowth of the forebrain.
  - The outer, thinner layer of the optic cup becomes the retinal pigment epithelium (**pigmented layer of retina**),
  - The inner, thicker layer differentiates into the neural retina (**neural layer of retina**).
- During the embryonic and early fetal periods, the two retinal layers are separated by an **intraretinal space**, which is the original cavity of the optic cup.

- Before birth, intraretinal space gradually disappears as the two layers of the retina fuse, but this fusion is not firm - can separate neural layer from the pigment layer.
- Layers of the optic cup are continuous with the wall of the brain-out growth of forebrain.
- Under the influence of the developing lens, the inner layer of the optic cup proliferates to form a thick **neuroepithelium**.
- **Neuroepithelium** cells differentiate into the **neural retina**.



Photomicrograph of a sagittal section of the eye of an embryo (×100) at Carnegie stage 18, approximately 44 days. Observe that it is the posterior wall of the lens vesicle that forms the lens fibers. The anterior wall does not change appreciably as it becomes the anterior lens epithelium.

- Neural retina contains photoreceptors (rods and cones) and the cell bodies of neurons (e.g., bipolar and ganglion cells).
  - Retinal ganglion cell differentiation is signaled/regulated by Fibroblast growth factor.
- Because the optic vesicle invaginates as it forms the optic cup, the neural retina is "inverted," that is, light-sensitive parts of the photoreceptor cells are adjacent to the retinal pigment epithelium.
- Light must pass through the thickest part of the retina before reaching the receptors; however, because the retina overall is thin and transparent, it does not form a barrier to light.
- The axons of ganglion cells in the superficial layer of the neural retina grow proximally in the wall of the **optic stalk** to the brain.
- As a result, the cavity of the optic stalk is gradually obliterated as the axons of the many ganglion cells form the **optic nerve**.

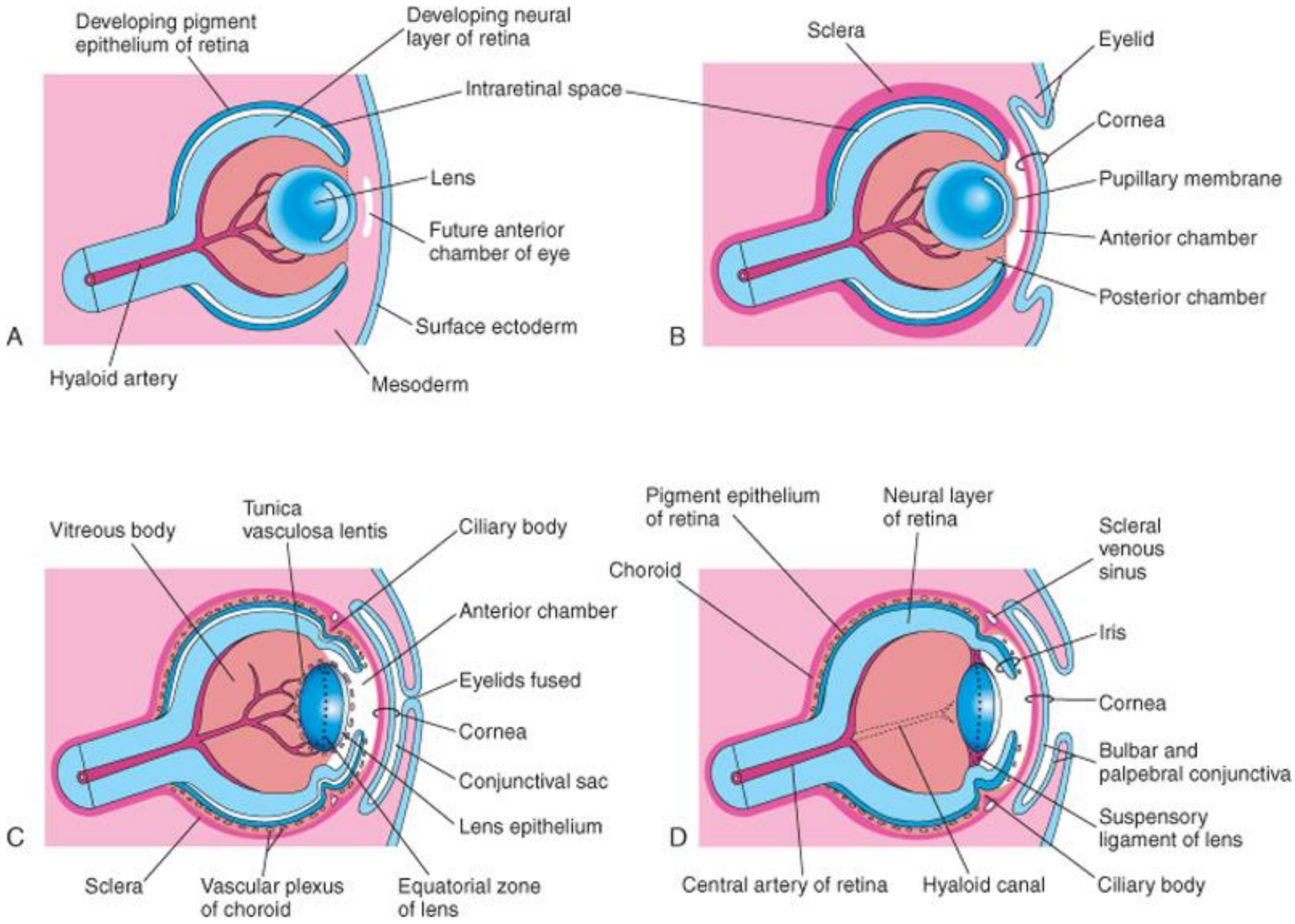
- Myelination of optic nerve fibers is incomplete at birth.
- After the eyes have been exposed to light for approximately 10 weeks, myelination is complete, but the process normally stops short of the optic disc, where the optic nerve enters the eyeball.
- Normal newborn infants can see, but not too well; they respond to changes in illumination and are able to fixate points of contrast.
  - Visual acuity has been estimated to be in the range of 20/400.
- At 2 weeks of age, the infants show a more sustained interest in large objects.

CONGENITAL DETACHMENT OF THE RETINA

- Congenital detachment of the retina occurs when the inner and outer layers of the optic cup fail to fuse during the fetal period to obliterate the intraretinal space - primary detachment.
- Detachment may be partial or complete.
- Retinal detachment may result from unequal rates of growth of the two retinal layers; as a result, the layers of the optic cup are not in perfect apposition.
- Secondary detachments usually occur in association with other anomalies of the eye and head.

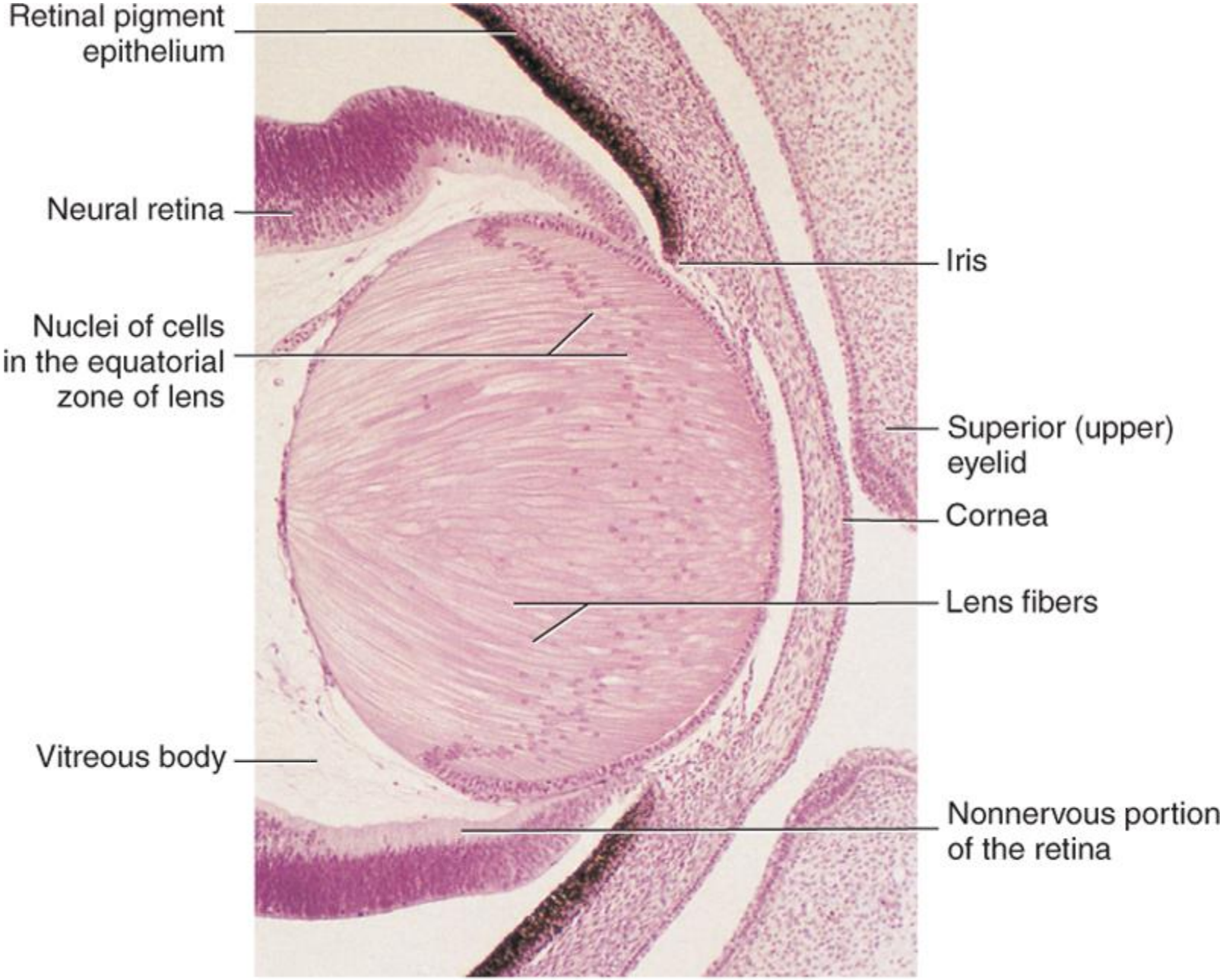
DEVELOPMENT OF THE CILIARY BODY

- The ciliary body is a wedge-shaped extension of the choroid.
- Its medial surface projects toward the lens, forming **ciliary processes**.
- The **pigmented** portion of the ciliary epithelium is derived from the outer layer of the optic cup and is continuous with the retinal pigment epithelium.
- The **nonpigmented** portion of the ciliary epithelium represents the anterior prolongation of the neural retina in which **no neural elements develop.**
- The **ciliary muscle**-the smooth muscle of the ciliary body that is responsible for focusing the lens-and the connective tissue in the ciliary body develop from mesenchyme located at the edge of the optic cup in the region between the anterior scleral condensation and the ciliary pigment epithelium.



DEVELOPMENT OF THE IRIS

- The iris develops from the rim of the optic cup, which grows inward and partially covers the lens.
- The two layers of the optic cup remain thin in this area.
- The epithelium of the iris represents both layers of the optic cup; it is continuous with the double-layered epithelium of the ciliary body and with the retinal pigment epithelium and neural retina.
- The connective tissue framework (stroma) of the iris is derived from **neural crest cells** that migrate into the iris.
- The **dilator pupillae** and **sphincter pupillae** muscles of the iris are derived from **neuroectoderm of the optic cup**.
- They appear to arise from the anterior epithelial cells of the iris.
- **These smooth muscles result from a transformation of epithelial cells into smooth muscle cells.**



COLOR OF THE IRIS

- The iris is typically light blue or gray in most newborn infants.
- The iris acquires its definitive color as pigmentation occurs during the first 6 to 10 months.
- The concentration and distribution of pigment-containing cells-**chromatophores**-in the loose vascular connective tissue of the iris determine eye color.
  - If the melanin pigment is confined to the pigmented epithelium on the posterior surface of the iris, the iris appears blue.
  - If melanin is also distributed throughout the stroma (supporting tissue) of the iris, the eye appears brown.

CONGENITAL ANIRIDIA

- In this rare anomaly, there is almost complete absence of the iris.
- This defect results from an arrest of development at the rim of the optic cup during the eighth week.
- Aniridia may be familial, the transmission being dominant or sporadic.

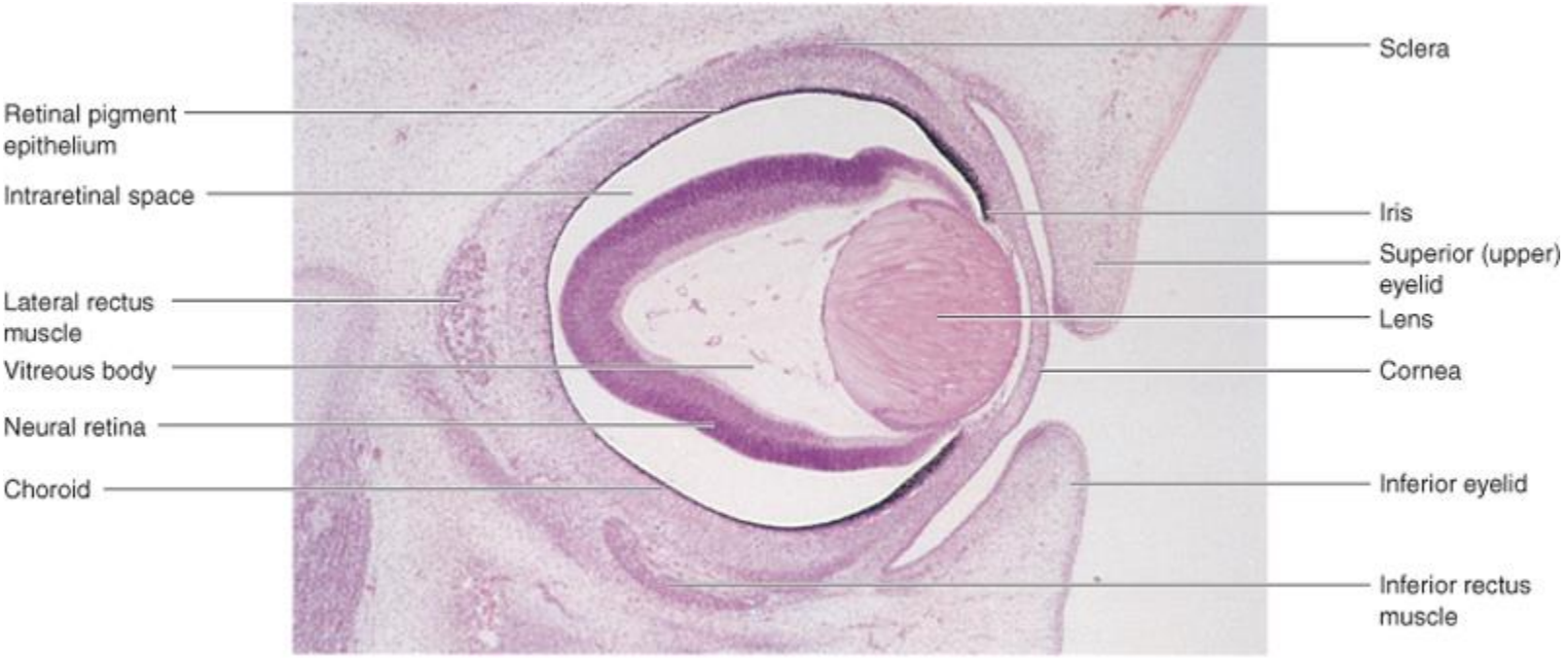
Heterochromia (also known as a heterochromia iridis or heterochromia iridium) is an ocular condition in which one iris is a different color from the other iris. The subject on the photo above has a brown and hazel eye.

Coloboma of the Iris

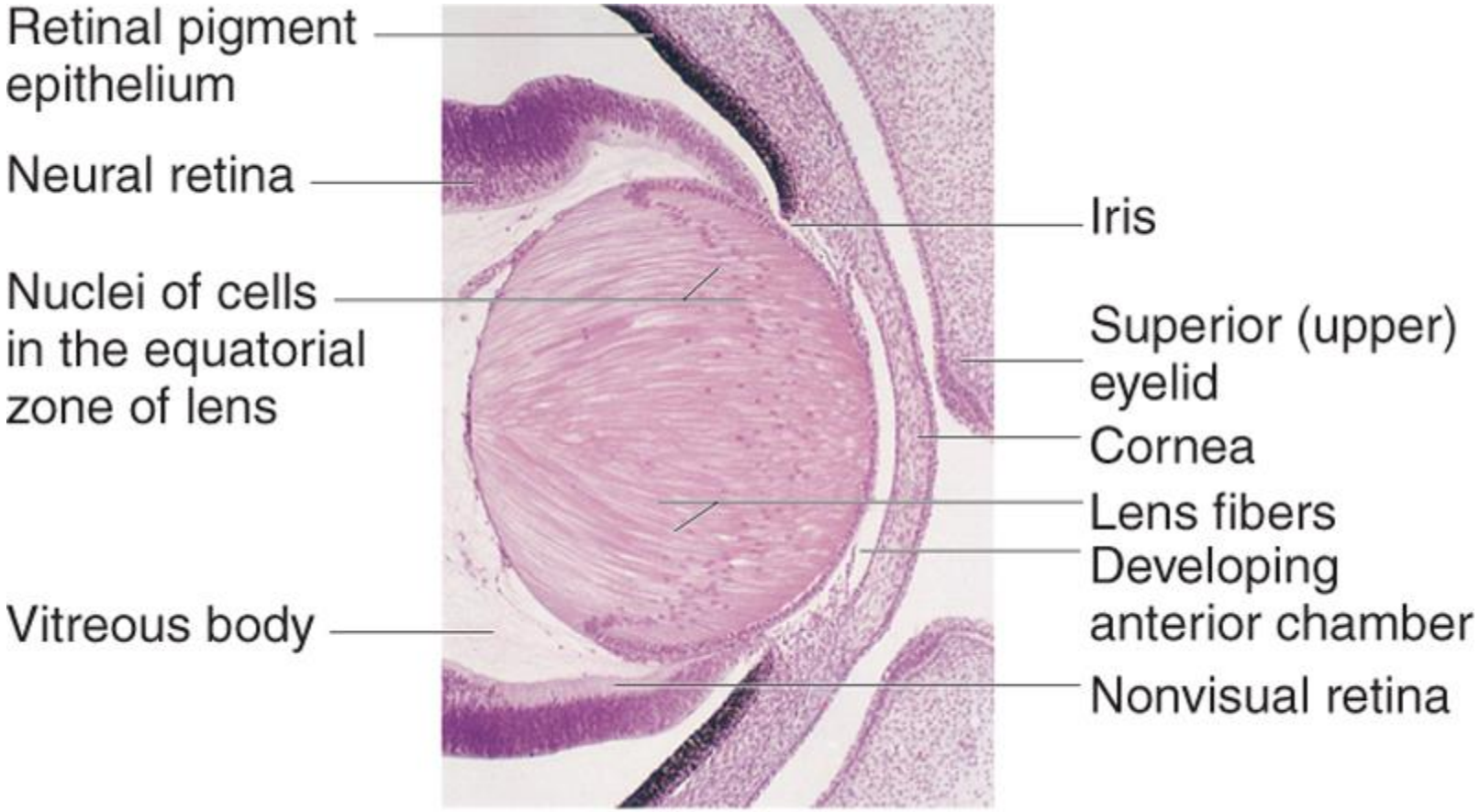
- Coloboma is a defect in the inferior sector of the iris or a notch in the pupillary margin, giving the pupil a keyhole appearance.
- The defect may be limited to the iris or it may extend deeper and involve the ciliary body and retina.
- *A typical coloboma of the iris results from failure of closure of the retinal fissure during the sixth week*.

DEVELOPMENT OF THE LENS

- The lens develops from the **lens vesicle**, a derivative of the surface ectoderm.
- The anterior wall of this vesicle, composed of cuboidal epithelium, becomes the subcapsular **lens epithelium.**
- The nuclei of the tall columnar cells forming the posterior wall of the lens vesicle undergo dissolution.
  - These cells lengthen considerably to form highly transparent epithelial cells, the **primary lens fibers**.
- As these fibers grow, they gradually obliterate the cavity of the lens vesicle.



- The rim of the lens is known as the **equatorial zone** because it is located midway between the anterior and posterior poles of the lens.
- The cells in the equatorial zone are cuboidal; as they elongate, they lose their nuclei and become **secondary lens fibers**.
- These new lens fibers are added to the external sides of the primary lens fibers.
- Although secondary lens fibers continue to form during adulthood and the lens increases in diameter, the primary lens fibers must last a lifetime.



Photomicrograph of a portion of the developing eye. Observe that the lens fibers have elongated and obliterated the cavity of the lens vesicle. Note that the inner layer of the optic cup has thickened greatly to form the neural retina and that the outer layer is heavily pigmented (retinal pigment epithelium).

- Lens nutrition
- During development: distal part of the **hyaloid artery**;.
- Later nutrition when avascular: from the aqueous humor in the **posterior and anterior chamber of the eye**, and from the vitreous humor.
- The developing lens is invested by a vascular mesenchymal layer, the **tunica vasculosa lentis**.
- The anterior part of this capsule is the **pupillary membrane**.
- When hyaloid artery degenerate, the tunica vasculosa lentis and pupillary membrane degenerate.
- The **lens capsule** produced by the anterior lens epithelium and the lens fibers persists.
- The lens capsule represents a **greatly thickened basement membrane**.

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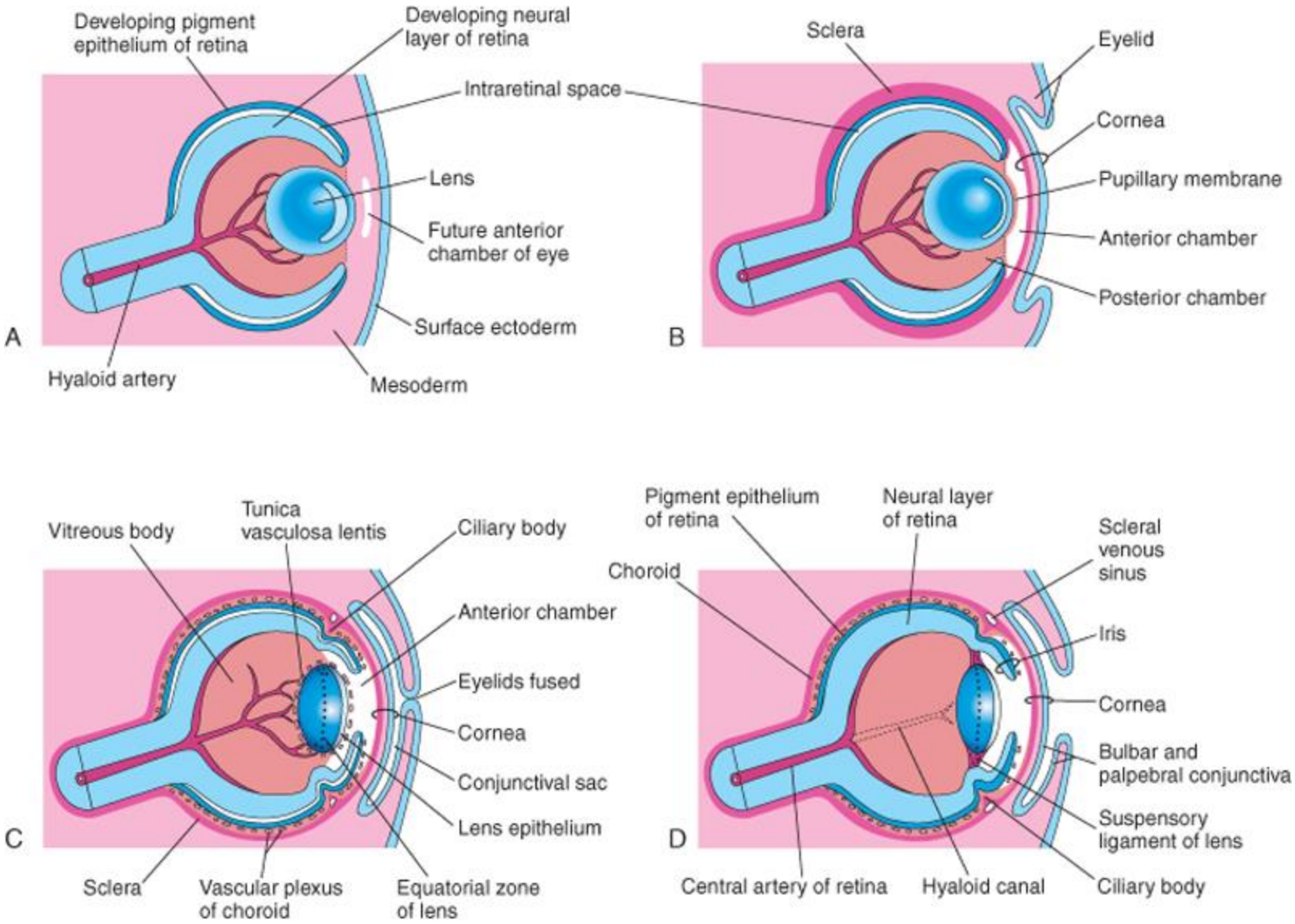


- The **vitreous body** forms within the cavity of the optic cup.
  - It is composed of **vitreous humor**, an avascular mass of transparent, gel-like, intercellular substance.
- The **primary vitreous humor** is derived from mesenchymal cells of **neural crest origin**.
- The primary vitreous humor does not increase but it is surrounded by a gelatinous **secondary vitreous humor**, generally believed to arise from the inner layer of the optic cup.
  - The secondary vitreous humor consists of primitive hyalocytes (vitreous cells), collagenous material, and traces of hyaluronic acid.

PERSISTENT PUPILLARY MEMBRANE

- Remnants of the pupillary membrane, which covers the anterior surface of the lens during the embryonic period, may persist as weblike strands of connective tissue or vascular arcades over the pupil in newborns, especially in premature infants.
- This tissue seldom interferes with vision and tends to atrophy.
- Very rarely the entire pupillary membrane persists, giving rise to congenital atresia of the pupil.

PERSISTENCE OF THE HYALOID ARTERY

- The distal part of the hyaloid artery normally degenerates as its proximal part becomes the central artery of the retina.
- If the distal part of the hyaloid artery persists, it may appear as a freely moving, nonfunctional vessel or as a wormlike structure projecting from the optic disc.
- Sometimes the hyaloid artery remnant may appear as a fine strand traversing the vitreous body.

CONGENITAL APHAKIA

- Absence of the lens is extremely rare and results from failure of the lens placode to form during the fourth week.
- Congenital aphakia could also result from failure of lens induction by the optic vesicle.

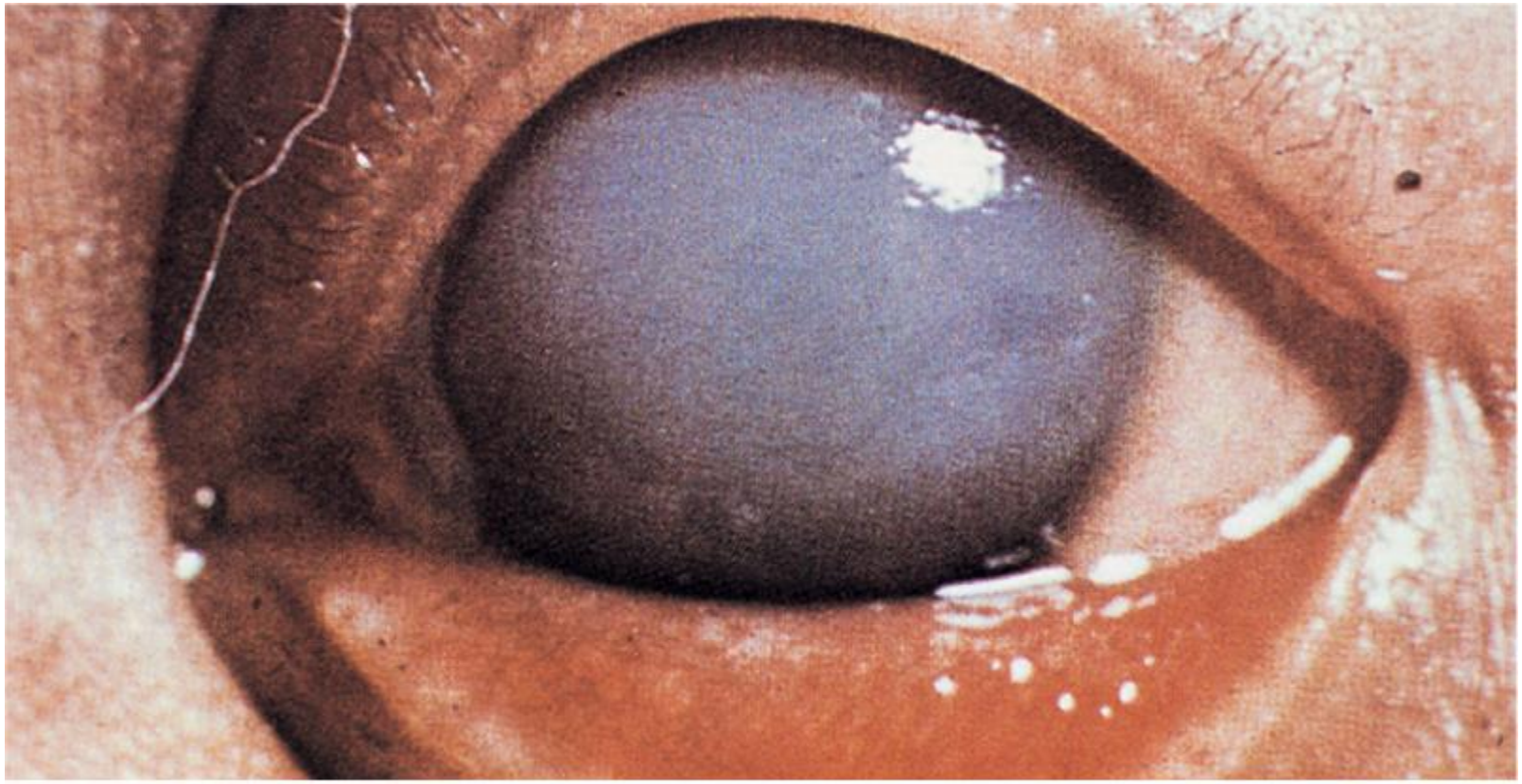
DEVELOPMENT OF THE AQUEOUS CHAMBERS

- The anterior chamber of the eye develops from a cleftlike space that forms in the mesenchyme located between the developing lens and cornea.
- The mesenchyme superficial to this space forms the substantia propria of the cornea and the mesothelium of the anterior chamber.
- After the lens is established, it induces the surface ectoderm to develop into the epithelium of the cornea and conjunctiva.

- The **posterior chamber of the eye** develops from a space that forms in the mesenchyme posterior to the developing iris and anterior to the developing lens.
- When the pupillary membrane disappears and the pupil forms, the anterior and posterior chambers of the eye are able to communicate with each other through a circumferential **scleral venous sinus**.
- This vascular structure encircling the anterior chamber is the outflow site of aqueous humor from the anterior chamber of the eye to the venous system.

CONGENITAL GLAUCOMA

- Abnormal elevation of intraocular pressure in newborn infants usually results from abnormal development of the drainage mechanism of the aqueous humor during the fetal period.
- Intraocular tension rises because of an imbalance between the production of aqueous humor and its outflow.
- This imbalance may result from abnormal development of the scleral venous sinus.
- Congenital glaucoma is genetically heterogeneous, but the condition may result from a rubella infection during early pregnancy.



CONGENITAL CATARACTS

- In this condition, the lens is opaque and frequently appears grayish white.
- Without treatment, blindness results.
- Many lens opacities are inherited, dominant transmission being more common than recessive or sex-linked transmission.
- Some congenital cataracts are caused by teratogenic agents, particularly the rubella virus, that affect early development of the lenses.
- The lenses are vulnerable to rubella virus between the fourth and seventh weeks, when primary lens fibers are forming.

- Physical agents, such as **radiation**, can also damage the lens and produce cataracts.
- Another cause of cataract is an enzymatic deficiency-**congenital galactosemia**.
  - These cataracts are not present at birth, but may appear as early as the second week after birth.
- Because of the enzyme deficiency, large amounts of galactose from milk accumulate in the infant's blood and tissues, causing injury to the lens and resulting in cataract formation.

- Treatment of cataracts requires surgery, typically at a very early age (younger than 4 months), to remove the damaged lens.
- In most cases, corrective eyewear is required, although some studies have shown that artificial intraocular lenses may be safely implanted.
- More than 70% of patients with bilateral congenital cataracts can attain reasonable visual acuity.
- Extended treatment with refractive correction and additional surgery may be required.

DEVELOPMENT OF THE CORNEA

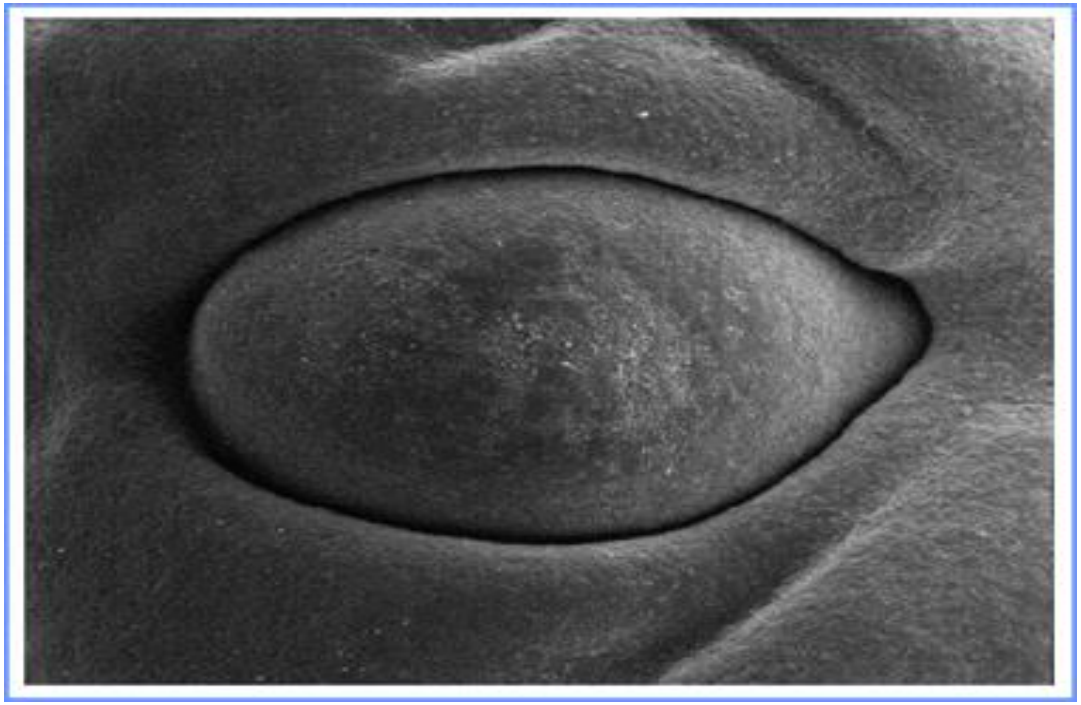
- The cornea is induced by the lens vesicle.
- The inductive influence results in transformation of the surface ectoderm into the transparent, multilayered avascular cornea, the part of the fibrous tunic of the eye that bulges out of the orbit.
- The cornea is formed from three sources:
  - The external corneal epithelium, derived from surface ectoderm
  - The mesenchyme, derived from mesoderm, which is continuous with the developing sclera
  - Neural crest cells that migrate from the lip of the optic cup and differentiate into the corneal endothelium

DEVELOPMENT OF THE CHOROID AND SCLERA

- The mesenchyme surrounding the optic cup (largely of neural crest origin) reacts to the inductive influence of the retinal pigment epithelium by differentiating into:
  - an inner vascular layer, the choroid,
  - an outer fibrous layer, the sclera.
- The sclera develops from a condensation of mesenchyme external to the choroid and is continuous with the stroma of the cornea.
- Toward the rim of the optic cup, the choroid becomes modified to form the cores of the ciliary processes, consisting chiefly of capillaries supported by delicate connective tissue.
- The first choroidal blood vessels appear during the 15th week; by the 23rd week, arteries and veins can be easily distinguished.

DEVELOPMENT OF THE EYELIDS

- The eyelids develop during the sixth week from:
  - neural crest cell mesenchyme
  - two cutaneous folds of ectoderm that grow over the cornea.
- The eyelids adhere to one another by the beginning of the 10^{th} week and remain adherent until the 26th to the 28th week.
- While the eyelids are adherent, there is a closed conjunctival sac anterior to the cornea.
- As the eyelids open, the bulbar conjunctiva is reflected over the anterior part of the sclera and the surface epithelium of the cornea.
- The palpebral conjunctiva lines the inner surface of the eyelids.

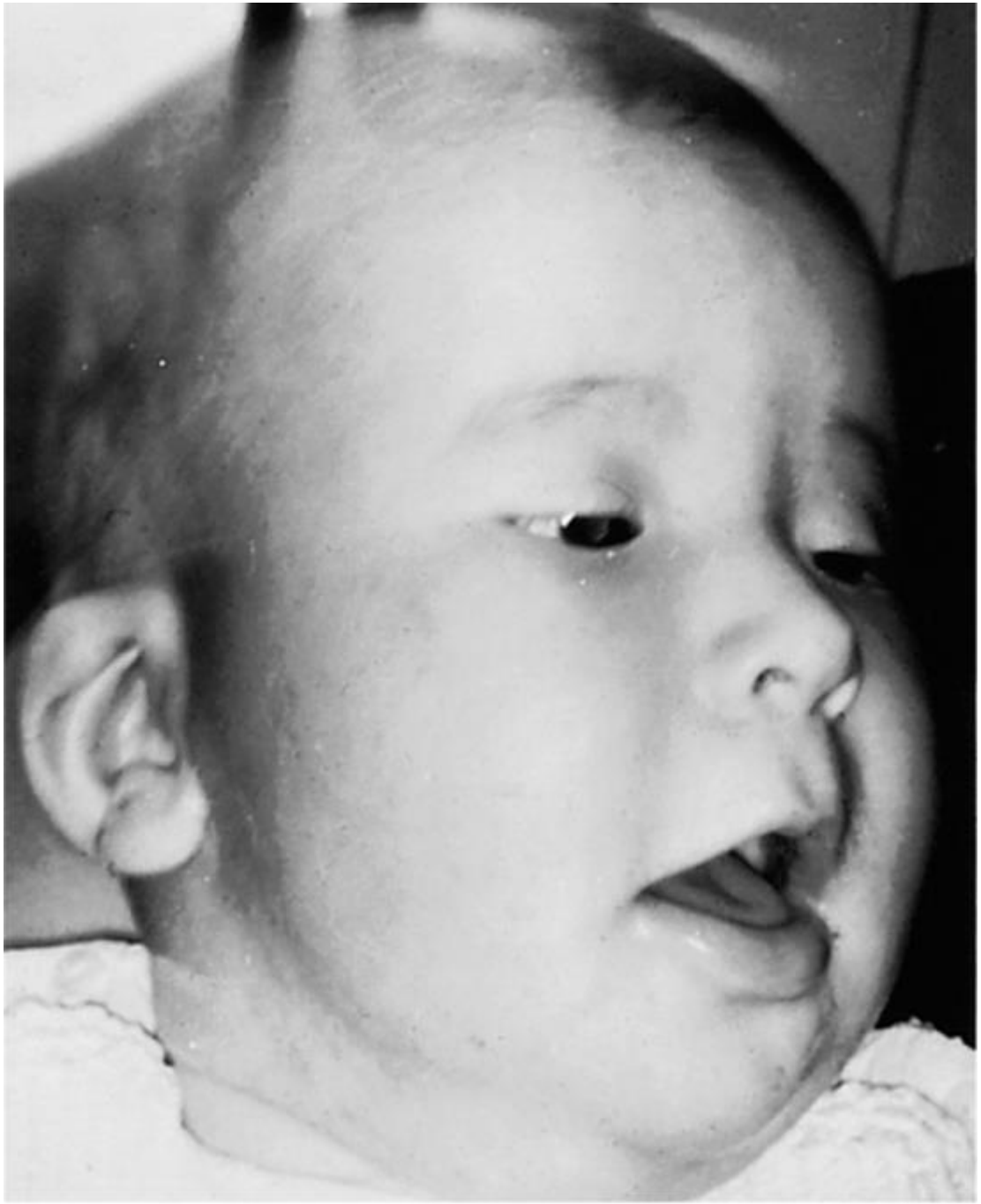


By the end of the embryonic period, eyelids begin to form.

- The **eyelashes** and **glands** in the eyelids are derived from the surface ectoderm in a manner similar to that described for other parts of the integument.
- The connective tissue and **tarsal plates** develop from mesenchyme in the developing eyelids.
- The orbicularis oculi muscle is derived from mesenchyme in the second pharyngeal arch and is supplied by its nerve (CN VII).

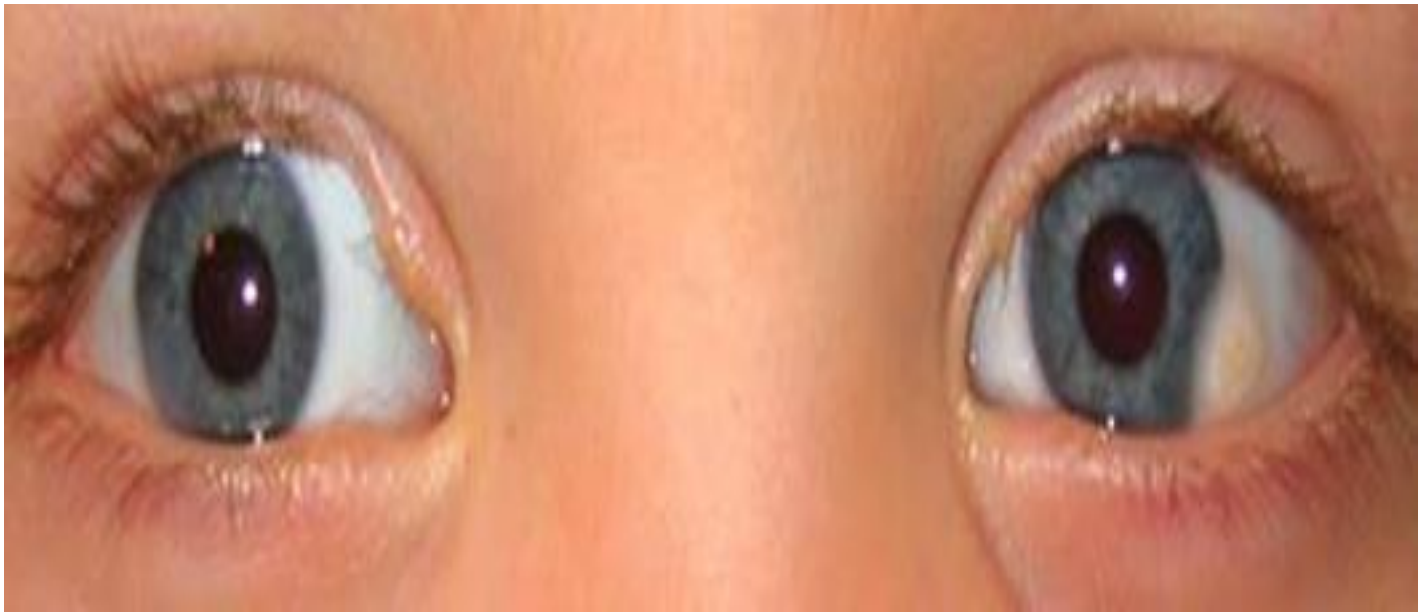
CONGENITAL PTOSIS OF THE EYELID

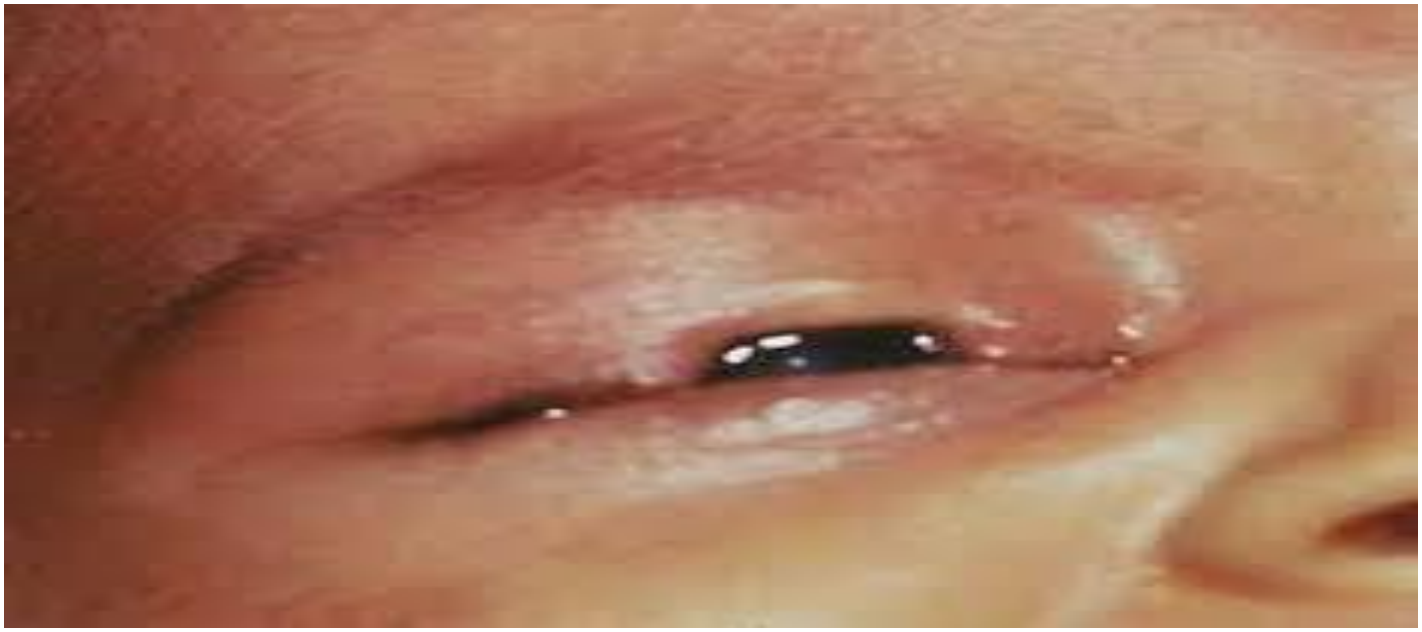
- Drooping of the superior (upper) eyelids at birth is relatively common.
- Ptosis (blepharoptosis) may result from failure of normal development of the levator palpebrae superioris muscle.
- Congenital ptosis may more rarely result from prenatal injury or dystrophy of the superior division of the oculomotor nerve (CN III), which supplies this muscle.
- If ptosis is associated with inability to move the eyeball superiorly, there is also failure of the superior rectus muscle of the eyeball to develop normally.
- Congenital ptosis may be transmitted as an autosomal dominant trait.
- Congenital ptosis is also associated with several syndromes.



Coloboma of the Eyelid

- Large defects of the eyelid (palpebral colobomas) are uncommon.
- A coloboma is usually characterized by a small notch in the superior (upper) eyelid, but the defect may involve almost the entire lid.
- A coloboma of the inferior (lower) eyelid is rare.
- Palpebral colobomas appear to result from local developmental disturbances in the formation and growth of the eyelids.





Cryptophthalmos

- Cryptophthalmos results from congenital absence of the eyelids; as a result, skin covers the eye.
- The eyeball is small and defective, and the cornea and conjunctiva usually do not develop.
- Fundamentally, the defect means absence of the palpebral fissure (slit) between eyelids; usually there is varying absence of eyelashes and eyebrows and other eye defects.
- Cryptophthalmos is an autosomal recessive condition that is usually part of the cryptophthalmos syndrome.

DEVELOPMENT OF THE LACRIMAL GLANDS

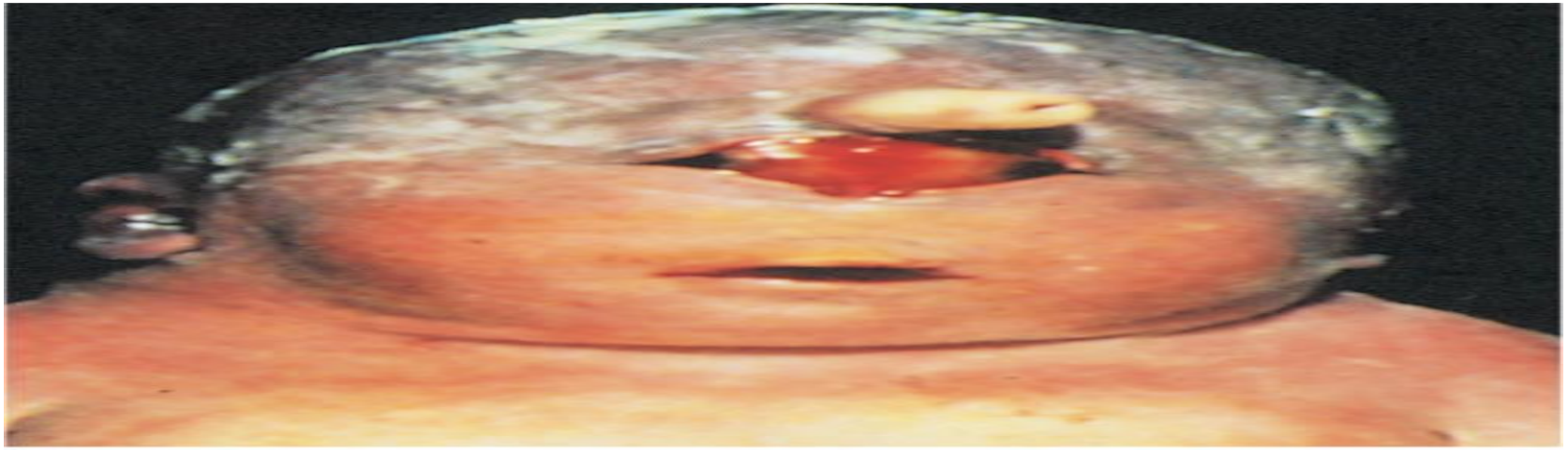
- At the superolateral angles of the orbits, the lacrimal glands develop from a number of solid buds from the surface ectoderm.
- The buds branch and become canalized to form the nasolacrimal ducts.
- The lacrimal glands are small at birth and do not function fully until approximately 6 weeks; hence, the newborn infant does not produce tears when it cries.
- Tears are often not present with crying until 1 to 3 months.

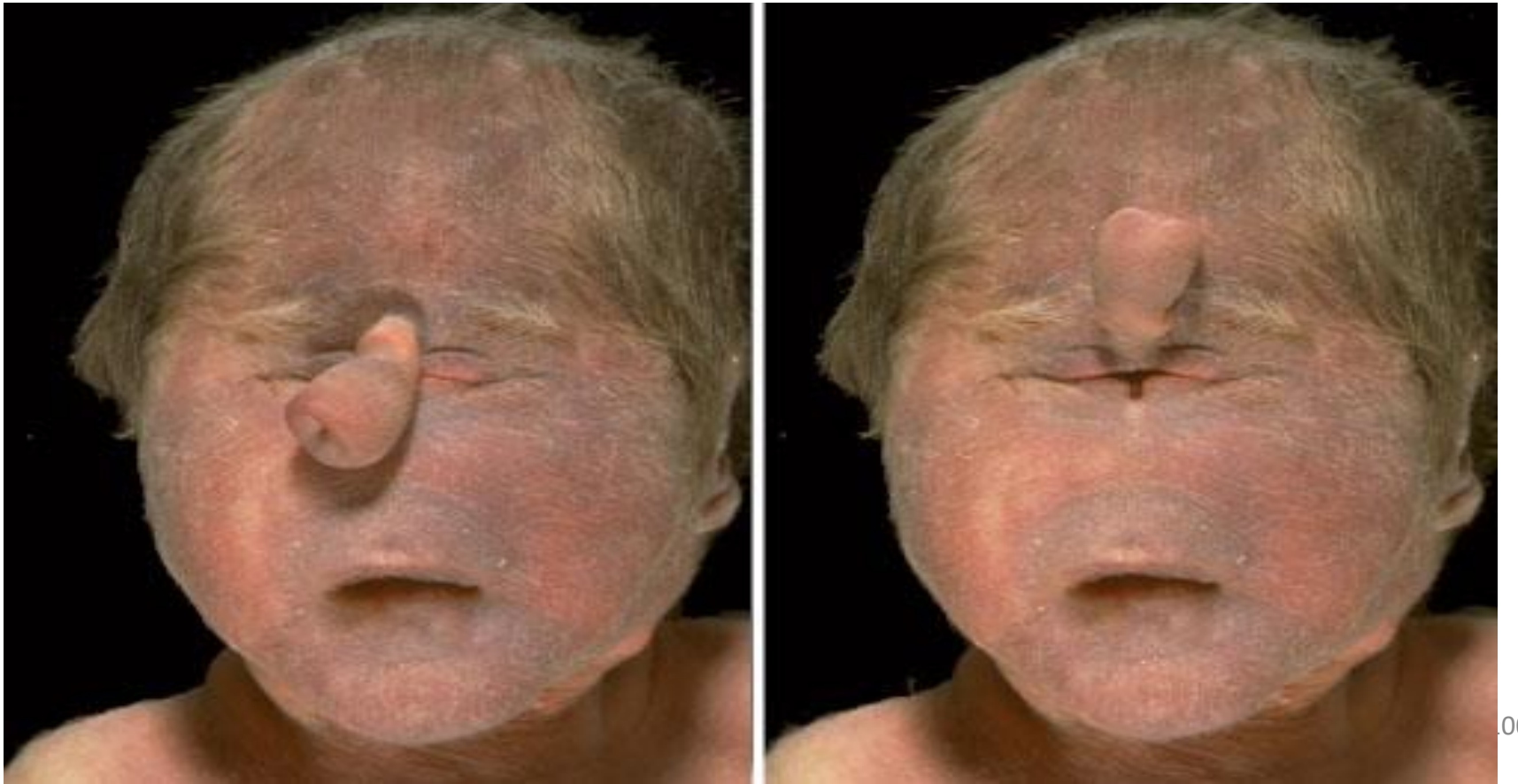
CONGENITAL ANOMALIES OF THE EYE

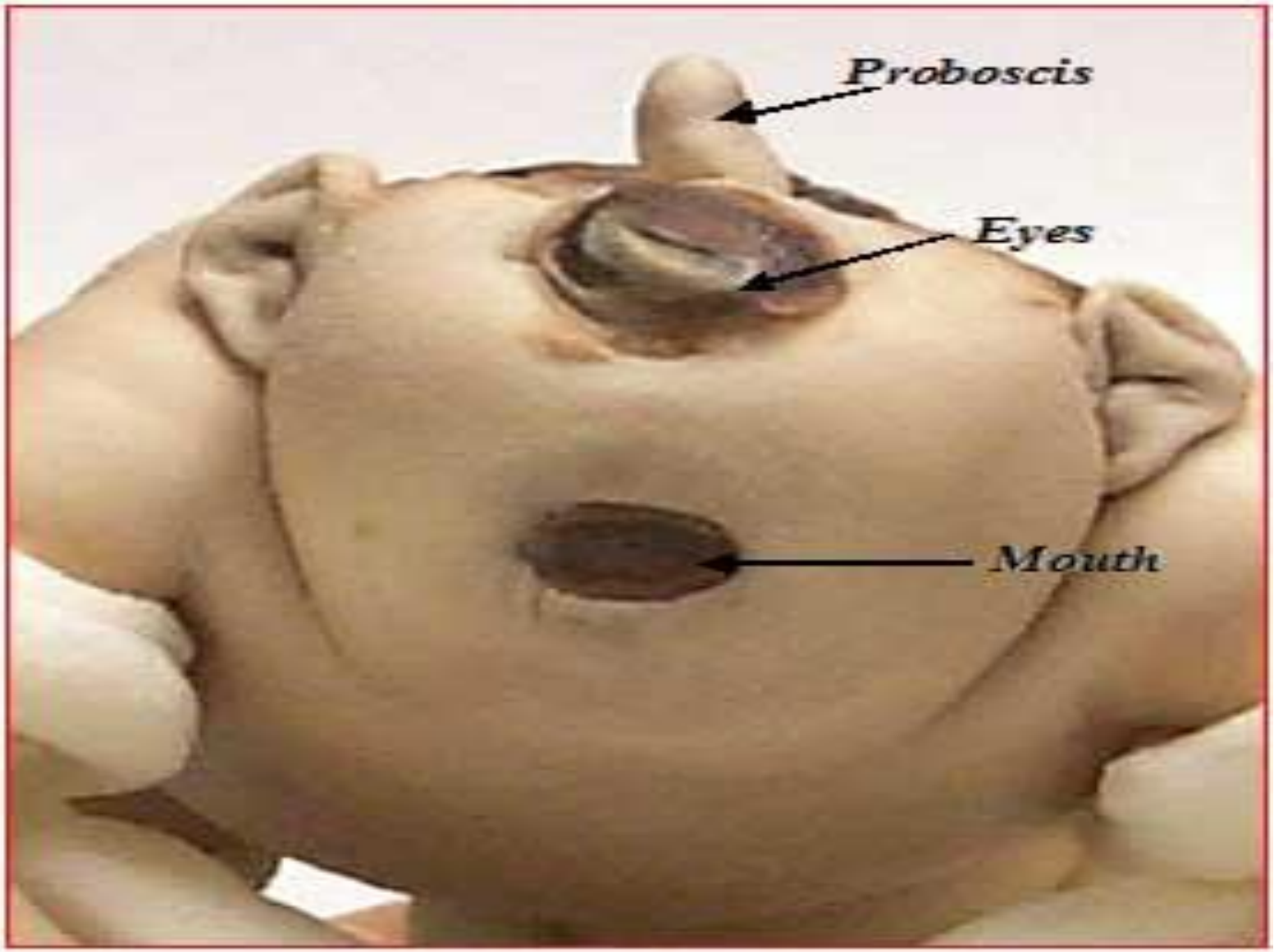
- The type and severity of congenital eye anomalies depend on the embryonic stage during which development is disrupted.
- Several environmental teratogens cause congenital eye defects.
- Most common eye anomalies result from defects in closure of the retinal/choroid fissure.

CYCLOPIA

- In this very rare anomaly, the eyes are partially or completely fused, forming a single **median eye** enclosed in a single orbit.
- There is usually a tubular nose (**proboscis**) superior to the eye.
- **Cyclopia** (single eye) and **synophthalmia** (fusion of eyes) represent a spectrum of ocular defects in which the eyes are partially or completely fused.
- These severe eye anomalies are associated with other craniocerebral defects that are incompatible with life.
- Cyclopia appears to result from severe suppression of midline cerebral structures-**holoprosencephaly**-that develop from the cranial part of the neural plate.
- Cyclopia is transmitted by recessive inheritance.






Proboscis
Eyes
Mouth

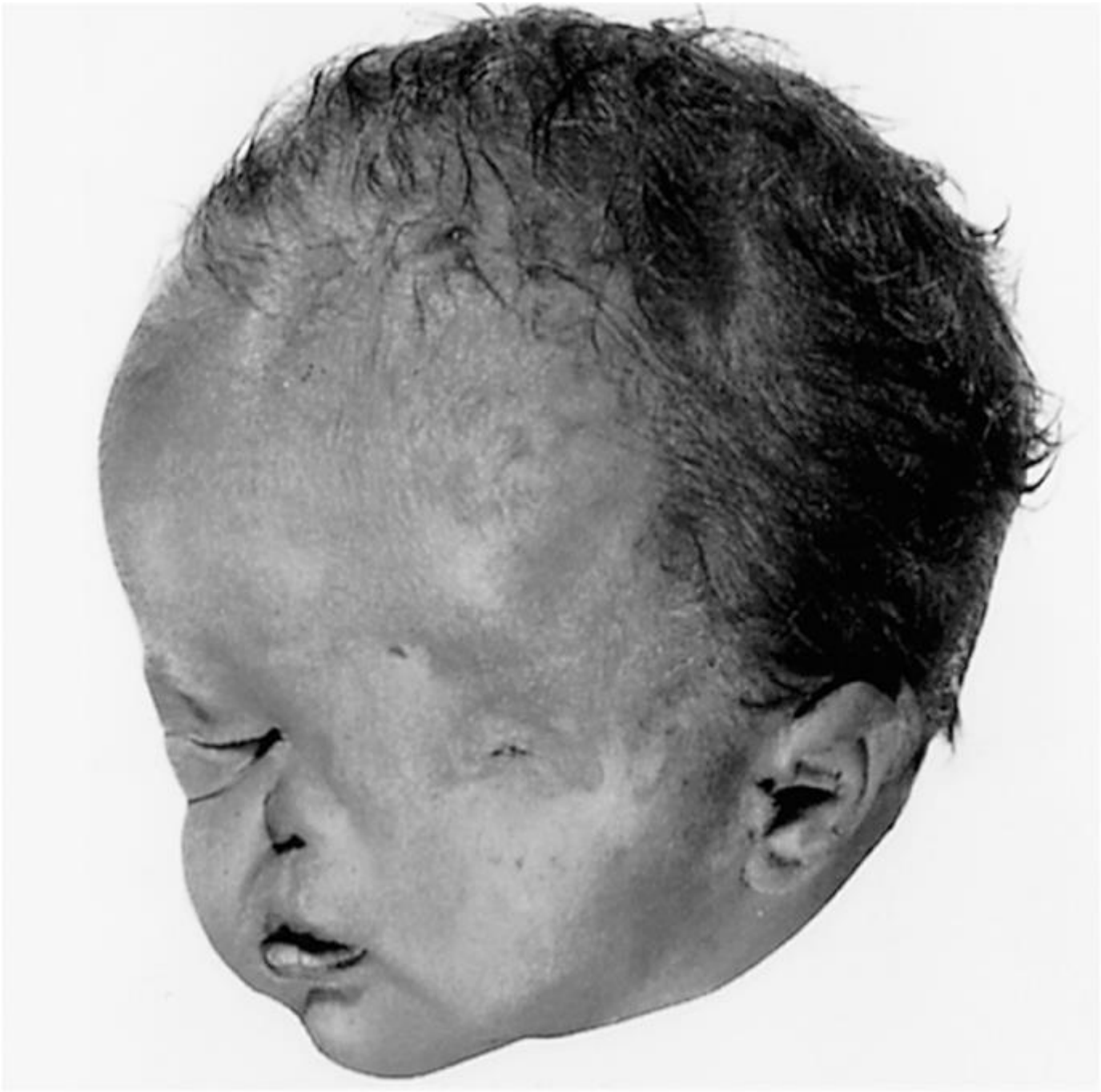
Microphthalmia

- Congenital microphthalmia is a heterogeneous group of eye anomalies.
- The eye may be very small with other ocular defects or it may be a normal-appearing rudimentary eye.
- The affected side of the face is underdeveloped and the orbit is small.
- Microphthalmia may be associated with other congenital anomalies (e.g., a facial cleft) and be part of a syndrome (e.g., trisomy 13).
- Severe microphthalmia results from arrested development of the eye before or shortly after the optic vesicle has formed in the fourth week.

- The eye is essentially underdeveloped and the lens does not form.
- Development arrested in the 6th week - the eye is larger, but the microphthalmos is associated with gross ocular defects.
- Development is arrested in the 8th week or during the early fetal period, simple microphthalmos results (small eye with minor ocular abnormalities).
- Some cases of microphthalmos are inherited: autosomal dominant, autosomal recessive, or X linked.
- Most cases of simple microphthalmia are caused by **infectious agents** (e.g., rubella virus, *Toxoplasma gondii*, and herpes simplex virus) that cross the placental membrane during the late embryonic and early fetal periods.

Anophthalmia

- Congenital absence of the eye - a rare condition.
- The eyelids form, but no eyeball develops.
- This severe defect is usually accompanied by other severe craniocerebral anomalies.
- In **primary anophthalmos**, eye development is arrested early in the fourth week and results from failure of the optic vesicle formation.
- In **secondary anophthalmos**, development of the forebrain is suppressed and absence of the eye or eyes is one of several associated anomalies.



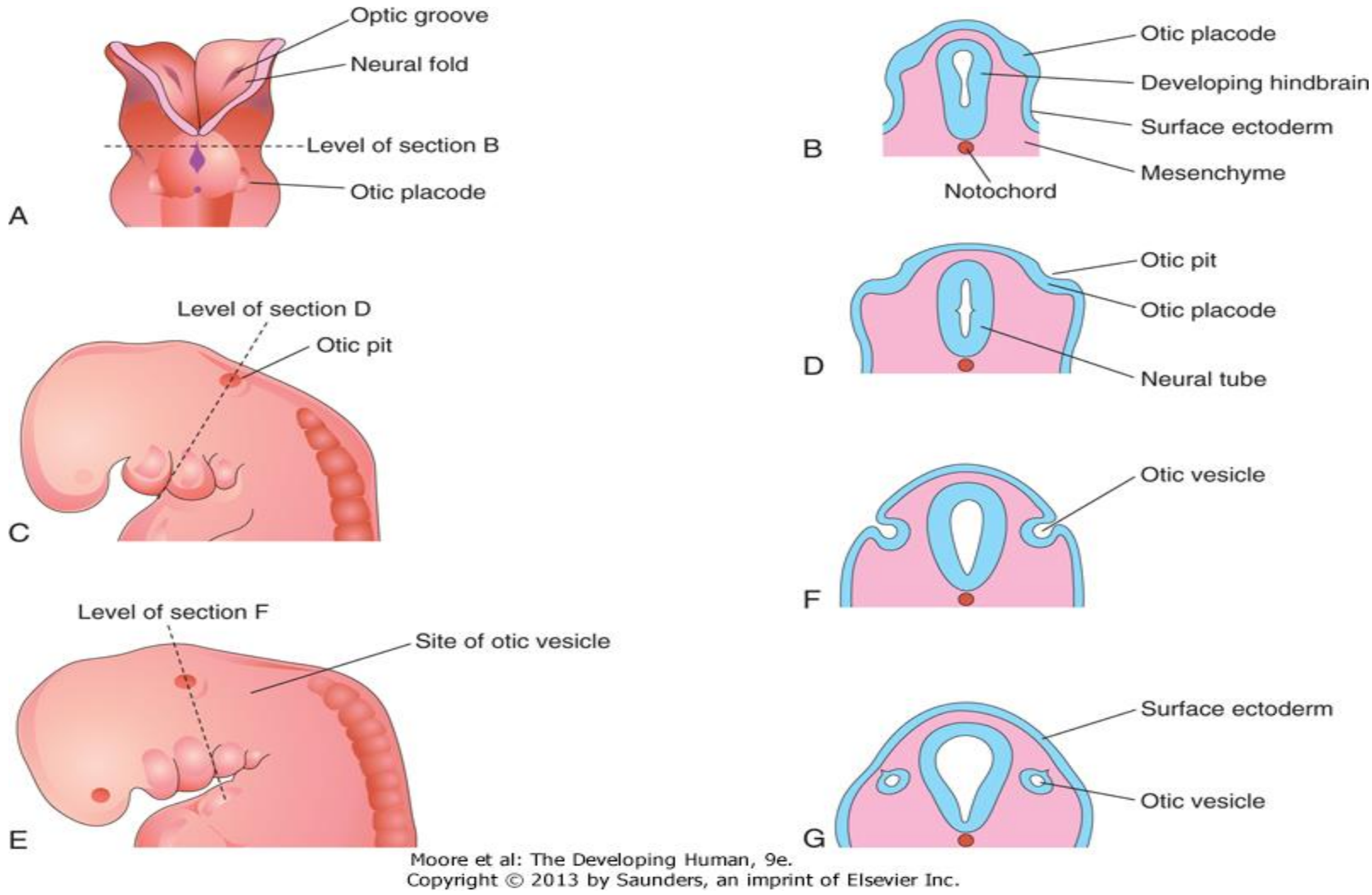
DEVELOPMENT OF EAR

Anatomic parts

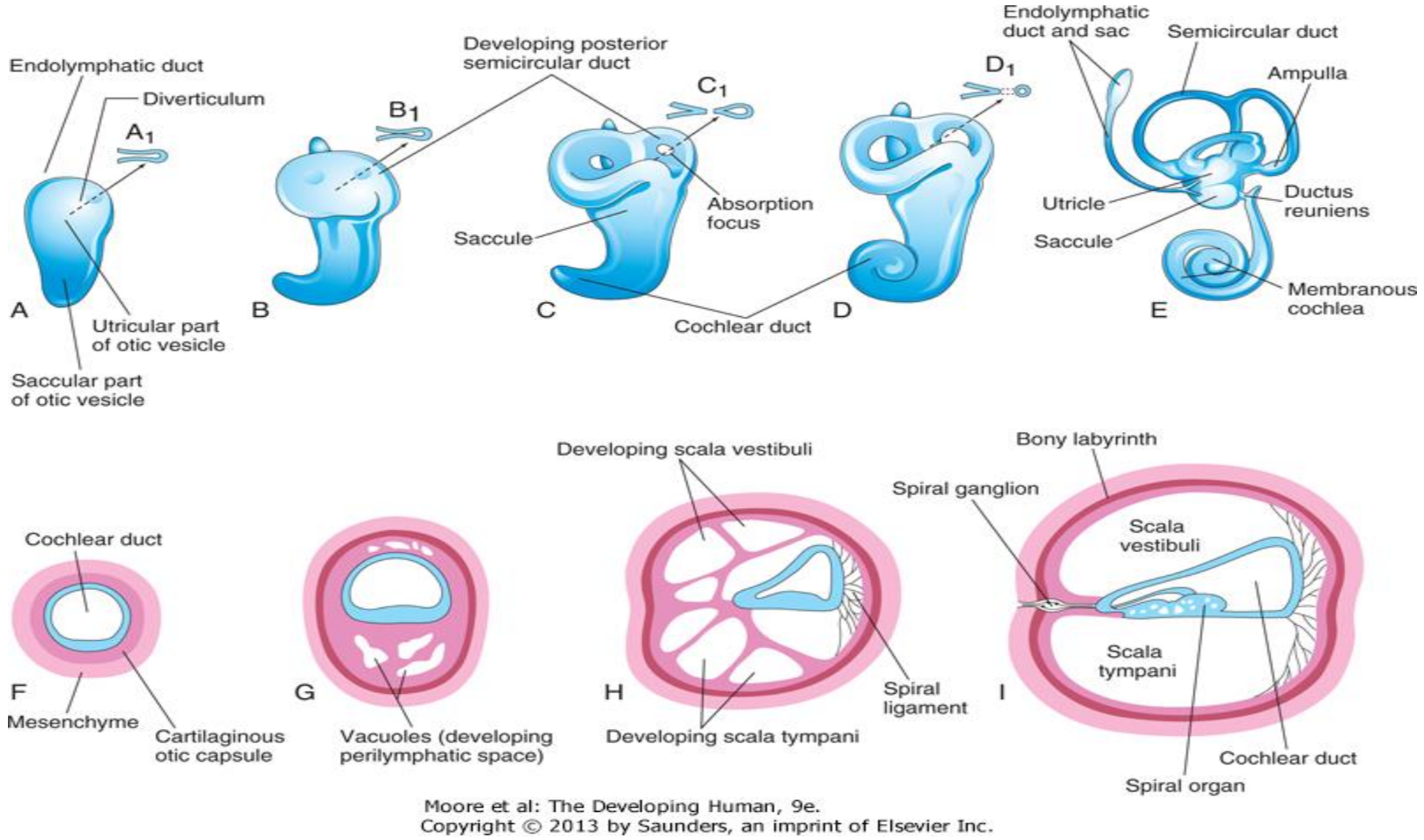
- *External ear*, consisting of the:
  - auricle (pinnae),
  - external acoustic meatus (passage),
  - the external layer of the tympanic membrane (eardrum)
- *Middle ear*, consisting of:
  - three small auditory ossicles (ear bones)
  - the internal layer of the tympanic membranes
- *Internal ear*, consisting of:
  - the vestibulocochlear organ

Development of Internal Ear

- First part of the ears to develop.
- Early in the 4th week, a thickening of surface ectoderm, the **otic placode**, appears on each side of the hindbrain.
- Each otic placode soon invaginates forming **otic pit** and sinks deep to the surface ectoderm into the underlying mesenchyme.
- The edges of the pit soon come together and fuse to form an **otic vesicle**-the *primordium of the membranous labyrinth*.
- The otic vesicle loses its connection with the surface ectoderm.
- A diverticulum grows from the vesicle and elongates to form the **endolymphatic duct** and **sac**.
- The otic vesicles has two parts:
  - Dorsal **utricular parts**, from which the small endolymphatic ducts, utricles, and semicircular ducts arise.
  - Ventral **saccular parts**, which give rise to the saccules and cochlear ducts.



Drawings illustrating early development of the internal ear. **A,** Dorsal view of an embryo at approximately 22 days, showing the otic placodes. **B, D, F,** and **G,** Schematic coronal sections illustrating successive stages in the development of otic vesicles. **C** and **E,** Lateral views of the cranial region of embryos, at approximately 24 and 28 days, respectively.



Drawings of the otic vesicles showing the development of the membranous and bony labyrinths of the internal ear. **A** to **E,** Lateral views showing successive stages in the development of the otic vesicle into the membranous labyrinth from the fifth to eighth weeks. **A** to **D,** Diagrammatic sketches illustrating the development of a semicircular duct. **F** to **I,** Sections through the cochlear duct showing successive stages in the development of the spiral organ and the perilymphatic space from the 8th to the 20th weeks.

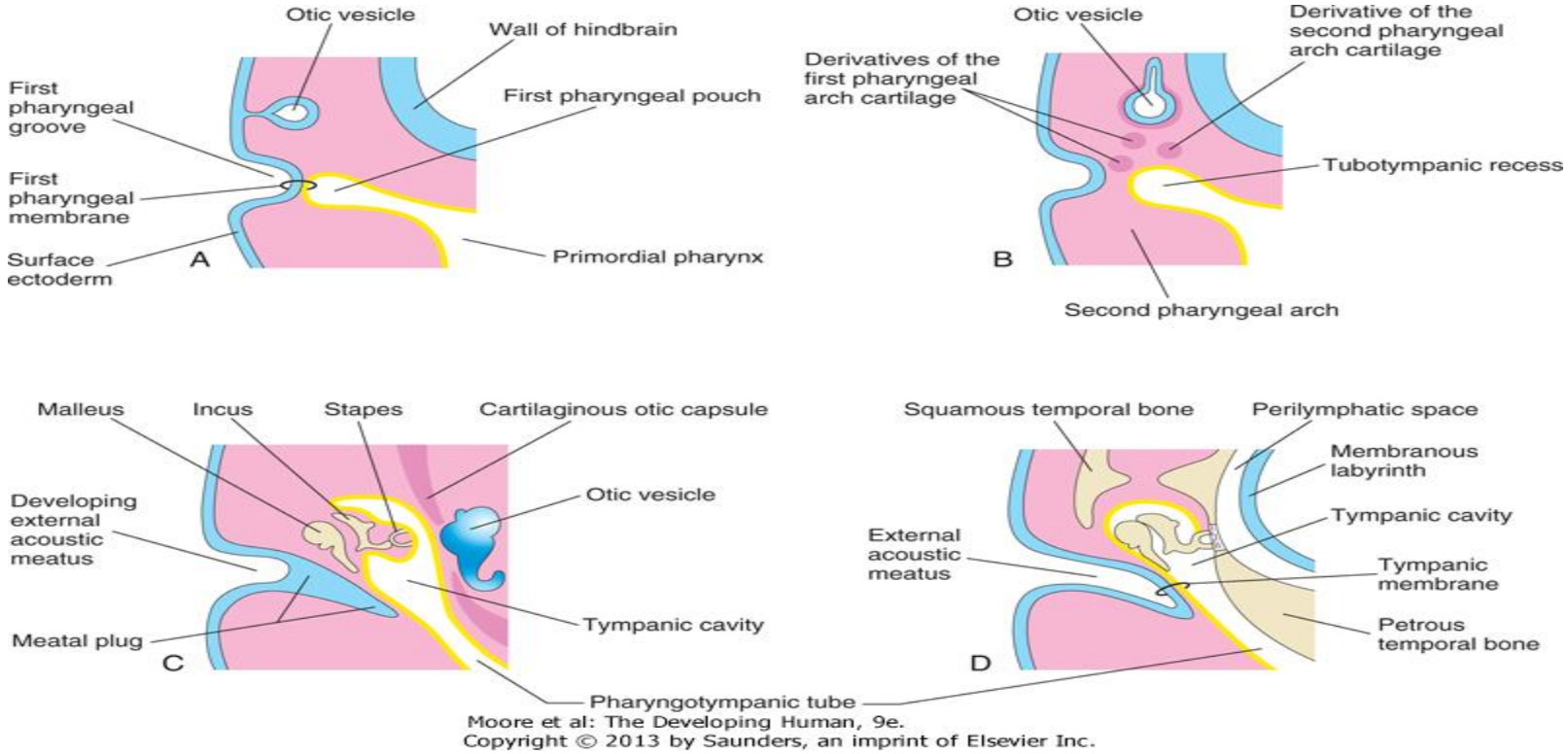
- Three disc-like diverticula grow out from the utricular parts of the **primordial membranous labyrinths**.
- Soon the central parts of these diverticula fuse and disappear.
- The peripheral unfused parts of the diverticula become the **semicircular ducts**, which are attached to the utricle and are later enclosed in the semicircular canals of the bony labyrinth.
- Localized dilatations, the **ampullae**, develop at one end of each semicircular duct.
- Specialized receptor areas-**cristae ampullares**-differentiate in the ampullae and the utricle and saccule (maculae utriculi and sacculi).
- From the **ventral saccular** part of the otic vesicle, a tubular diverticulum-the **cochlear duct**-grows and coils to form the **membranous cochlea**.

- A connection of the cochlea with the saccule, the **ductus reuniens**, soon forms.
- The **organ of Corti** differentiates from cells in the wall of the cochlear duct.
- Ganglion cells of the **vestibulocochlear nerve** (cranial nerve VIII) migrate along the coils of the membranous cochlea and form the **spiral ganglion** (of cochlea).
- Nerve processes extend from this ganglion to the spiral organ, where they terminate on the **hair cells**.
- The cells in the spiral ganglion retain their embryonic bipolar condition.
- Inductive influences from the otic vesicle stimulate the mesenchyme (mesoderm) around the otic vesicle to condense and differentiate into a **cartilaginous otic capsule**.

- With the enlargement of the **membranous labyrinth**, vacuoles appear in the cartilaginous otic capsule and soon coalesce to form the **perilymphatic space**.
- The membranous labyrinth is now suspended in **perilymph** (fluid in the perilymphatic space).
- The perilymphatic space, related to the cochlear duct, develops two divisions:
  - **scala tympani**
  - **scala vestibuli**.
- The cartilaginous otic capsule later ossifies (25 weeks) to form the **bony labyrinth** of the internal ear.
- The internal ear reaches its adult size and shape by the middle of the fetal period - 20-22 weeks.

Development of Middle Ear

- T**ubotympanic recess** develops from the first pharyngeal pouch.
- The proximal part of the tubotympanic recess forms the **pharyngotympanic tube** (auditory tube).
- The distal part of the recess expands and becomes the **tympanic cavity**, which gradually envelops the small bones of the middle ear-**auditory ossicles** (malleus, incus, and stapes), their tendons and ligaments, and the chorda tympani nerve.
- These structures receive a more or less complete epithelial investment.
  - It has been suggested that, in addition to apoptosis in the middle ear, an epithelium-type organizer located at the tip of the tubotympanic recess probably plays a role in the early development of the middle ear and tympanic membrane.

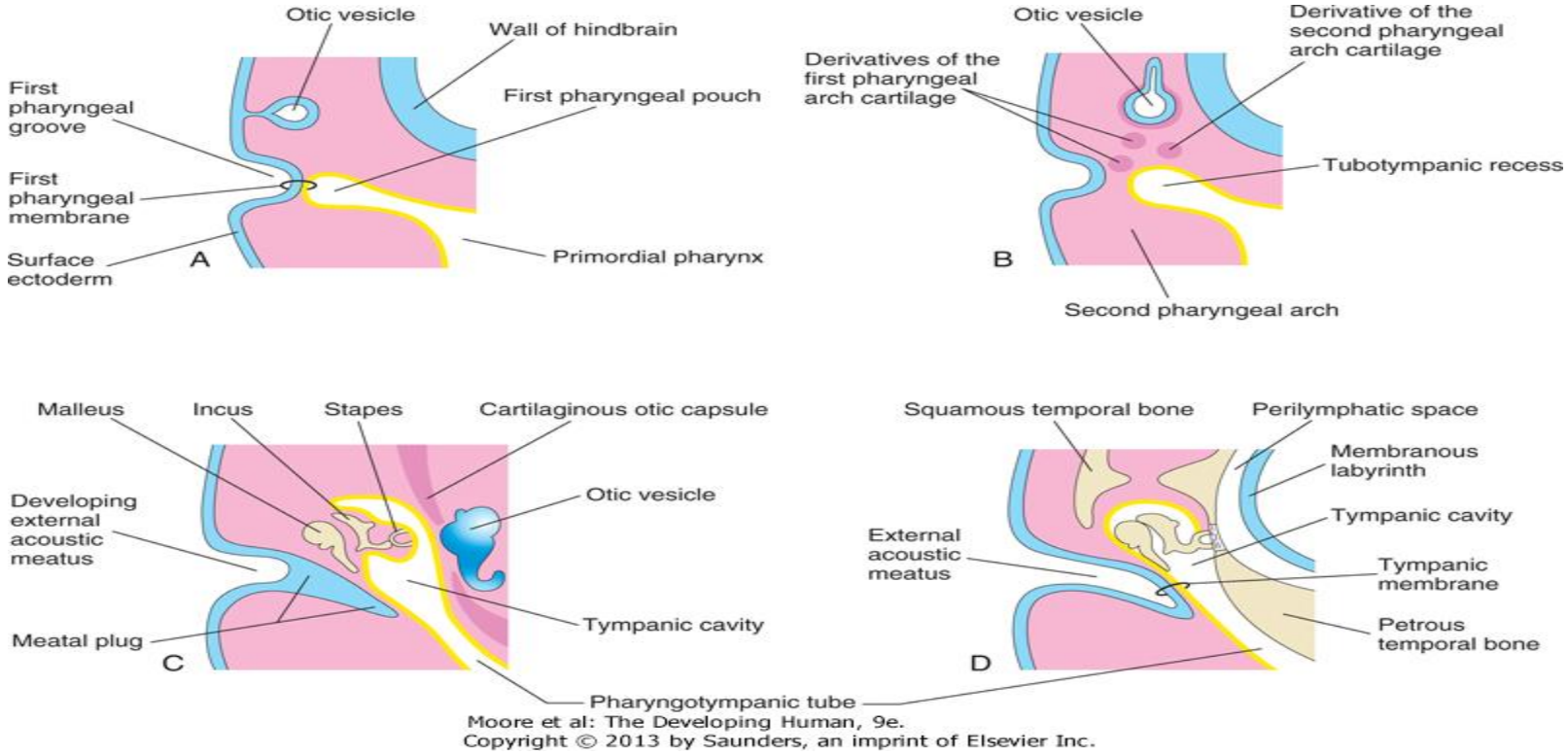


Schematic drawings illustrating development of the external and middle parts of the ear. Observe the relationship of these parts of the ear to the otic vesicle, the primordium of the internal ear. **A,** At 4 weeks, illustrating the relation of the otic vesicle to the pharyngeal apparatus. **B,** At 5 weeks, showing the tubotympanic recess and pharyngeal arch cartilages. **C,** Later stage, showing the tubotympanic recess (future tympanic cavity and mastoid antrum) beginning to envelop the ossicles. **D,** Final stage of ear development showing the relationship of the middle ear to the perilymphatic space and the external acoustic meatus. **Note that the tympanic membrane develops from three germ layers: surface ectoderm, mesenchyme, and endoderm of the tubotympanic recess.**

- Expansion of the **tympanic cavity** gives rise to the **mastoid antrum**, located in the petromastoid part of the temporal bone.
- The mastoid antrum is almost adult size at birth; however, no mastoid cells are present in neonates.
- By 2 years of age, the mastoid cells are well developed and produce conical projections of the temporal bones, the **mastoid processes**.
- The middle ear continues to grow through puberty.
- The **tensor tympani muscle**, attached to the malleus, is derived from mesenchyme in the first pharyngeal arch and is innervated by trigeminal nerve (CN V), the nerve of this arch.
- The **stapedius muscle** is derived from the second pharyngeal arch and is supplied by facial nerve (CN VII), the nerve of this arch.
  - *The signaling molecules FGF-8, endothelin-1, and Tbx1 are involved in middle ear development.*

Development of External Ear

- The **external acoustic meatus**, develops from the dorsal part of the first pharyngeal groove.
- The ectodermal cells at the bottom of this funnel-shaped tube proliferate to form a solid epithelial plate, the **meatal plug**.
- Degeneration of the central cells of this plug forms a cavity that becomes the internal part of the external acoustic meatus.
- The meatus, relatively short at birth, attains its adult length in approximately the ninth year.
- Tympanic membrane develops from three sources:
  - Ectoderm of the first pharyngeal groove
  - Endoderm of the tubotympanic recess, a derivative of the first pharyngeal pouch
  - Mesenchyme of the first and second pharyngeal arches which forms collagen in the mebrane



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- The **auricle** (pinna), develops from 6 mesenchymal proliferations in the first and second pharyngeal arches – 6 **auricular hillocks** -surrounding the first pharyngeal groove .
- The **earlobule** (earlobe) of the auricle is the last part of the auricle to develop.
- The auricles begin to develop at the base of the neck.
- As the mandible develops, the auricles assume their normal position at the side of the head.

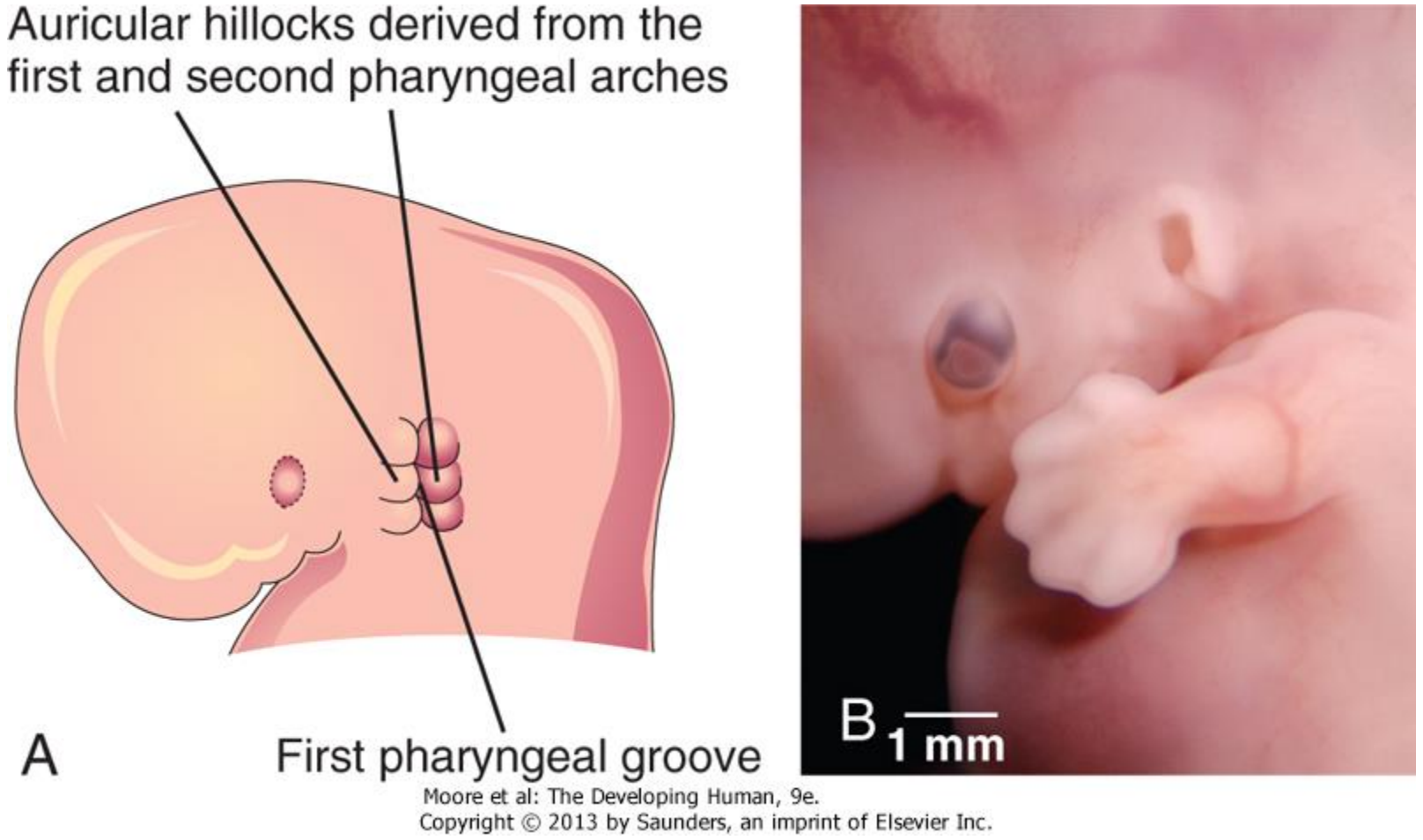
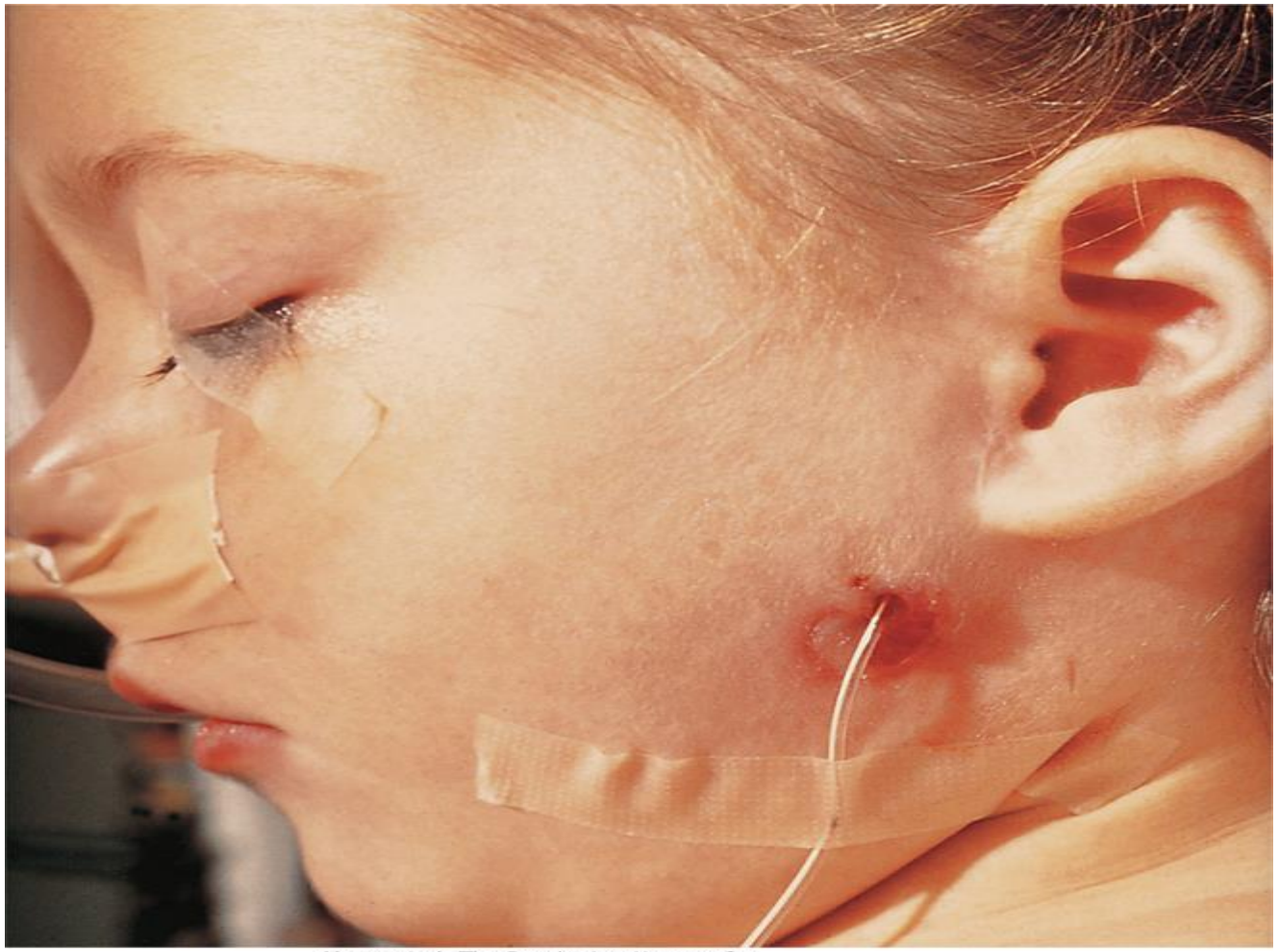


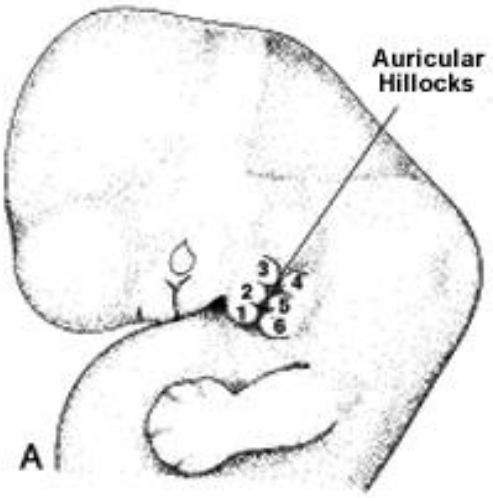
Illustration of the development of the auricle, the part of the external ear that is not within the head. **A,** At 6 weeks. Note that three auricular hillocks are located on the first pharyngeal arch and three on the second arch. **B,** Photograph of a 7-week embryo. Note the developing external ear.

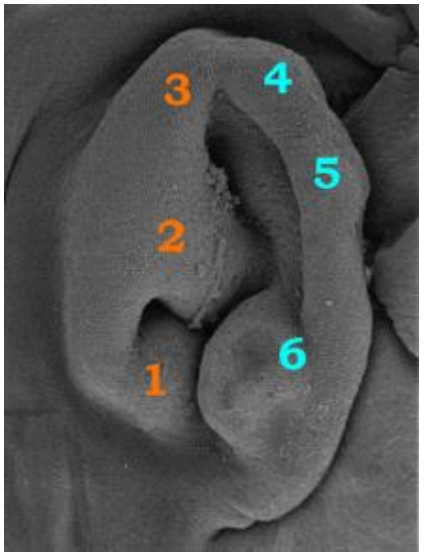


A child with an auricular fistula relating to the first pharyngeal arch. Note the external orifice of the fistula below the auricle and the upward direction of the catheter (in sinus tract) toward the external acoustic meatus.

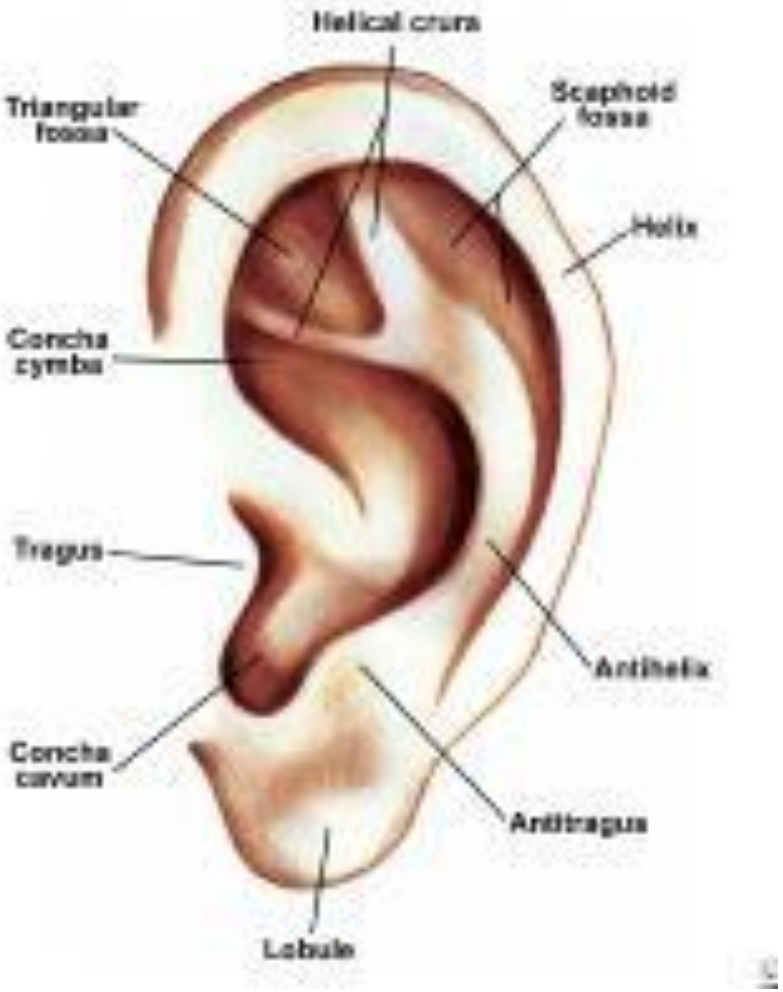
Auricle

- 6 Hillocks of His (mesoderm)
- 1st pharyngeal arch
  - 1: Tragus
  - 2: Helical crus
  - 3: Helix
- 2nd pharyngeal arch
  - 4: antihelix
  - 5: antihelix
  - 6: antitragus





Auricle

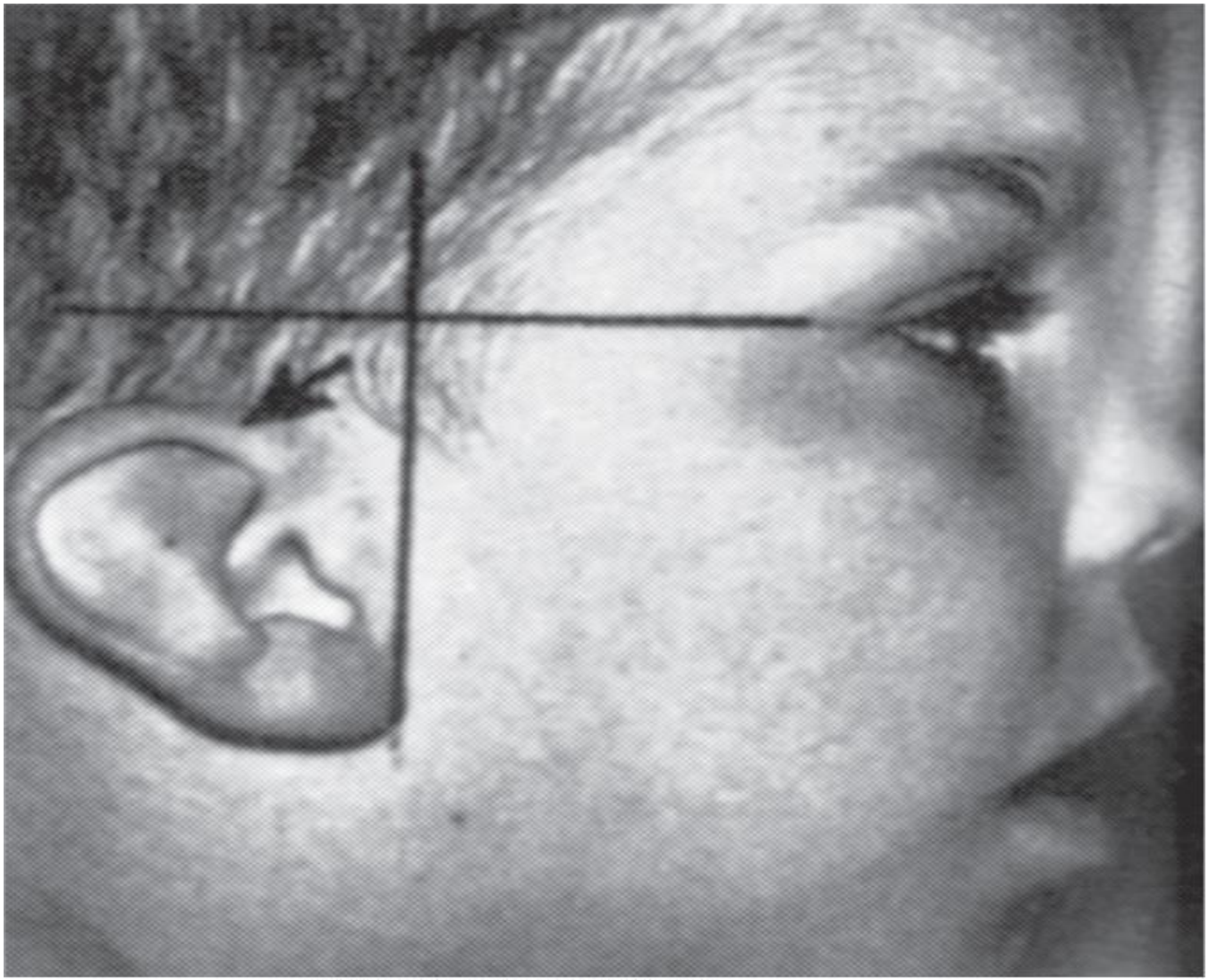

Helical crura
Triangular fossa
Scaphoid fossa
Helix
Concha cymba
Tragus
Antihelix
Concha cavum
Antitragus
Lobule

CONGENITAL DEAFNESS

- Approximately 3 in every 1000 newborns have significant hearing loss, of which there are many subtypes.
- Most types of congenital deafness are caused by genetic factors and many of the genes responsible have been identified.
- Mutations in the *GJB2* gene are responsible for approximately 50% of nonsyndromic recessive hearing loss.
- Congenital deafness may be associated with several other head and neck defects as a part of the first arch syndrome.
- Abnormalities of the malleus and incus are often associated with this syndrome.
- A **rubella infection** during the seventh and eighth weeks, can cause defects of the spiral organ and deafness.
- **Congenital fixation of the stapes** results in conductive deafness in an otherwise normal ear.
  - Failure of differentiation of the annular ligament, which attaches the base of the stapes to the oval window (*fenestra vestibuli*), results in fixation of the stapes to the bony labyrinth.

AURICULAR ABNORMALITIES

- Severe defects of the external ear are rare, but minor deformities are common.
- There is a wide variation in the shape of the auricle.
- Almost any minor auricular defect may occasionally be found as a usual feature in a particular family.
- Minor defects of the auricles may serve as indicators of a specific pattern of congenital anomalies.
- For example, the auricles are often abnormal in shape and low-set in infants with chromosomal syndromes such as trisomy 18 and in infants affected by maternal ingestion of certain drugs (e.g., trimethadione).



Low-set slanted ear. This designation is made when the margin of the auricle or helix (*arrow*) meets the cranium at a level inferior to the horizontal plane through the corner of the eye.

Absence of the Auricle

- Anotia (absence of the auricle) is rare but is commonly associated with the first pharyngeal arch syndrome.
- **Anotia** results from failure of mesenchymal proliferation.

Auricular Appendages

- Auricular appendages (skin tags) are common and may result from the development of accessory auricular hillocks.
- The appendages usually appear anterior to the auricle, more often unilaterally than bilaterally.
- The appendages, often with narrow pedicles, consist of skin but may contain some cartilage.

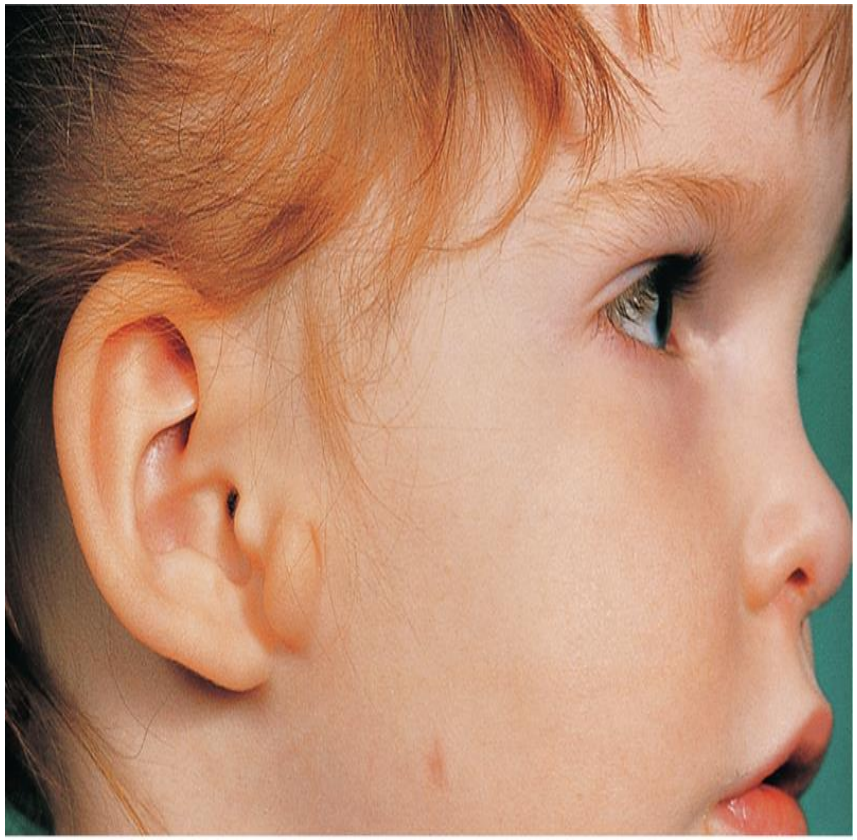
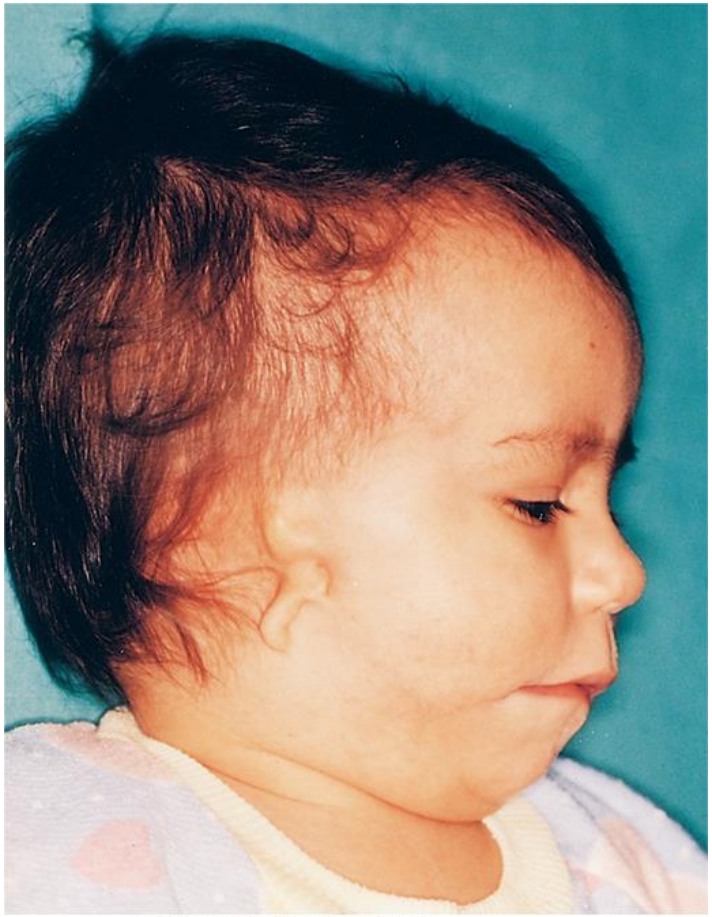


FIGURE 18–21 A child with a preauricular tag or

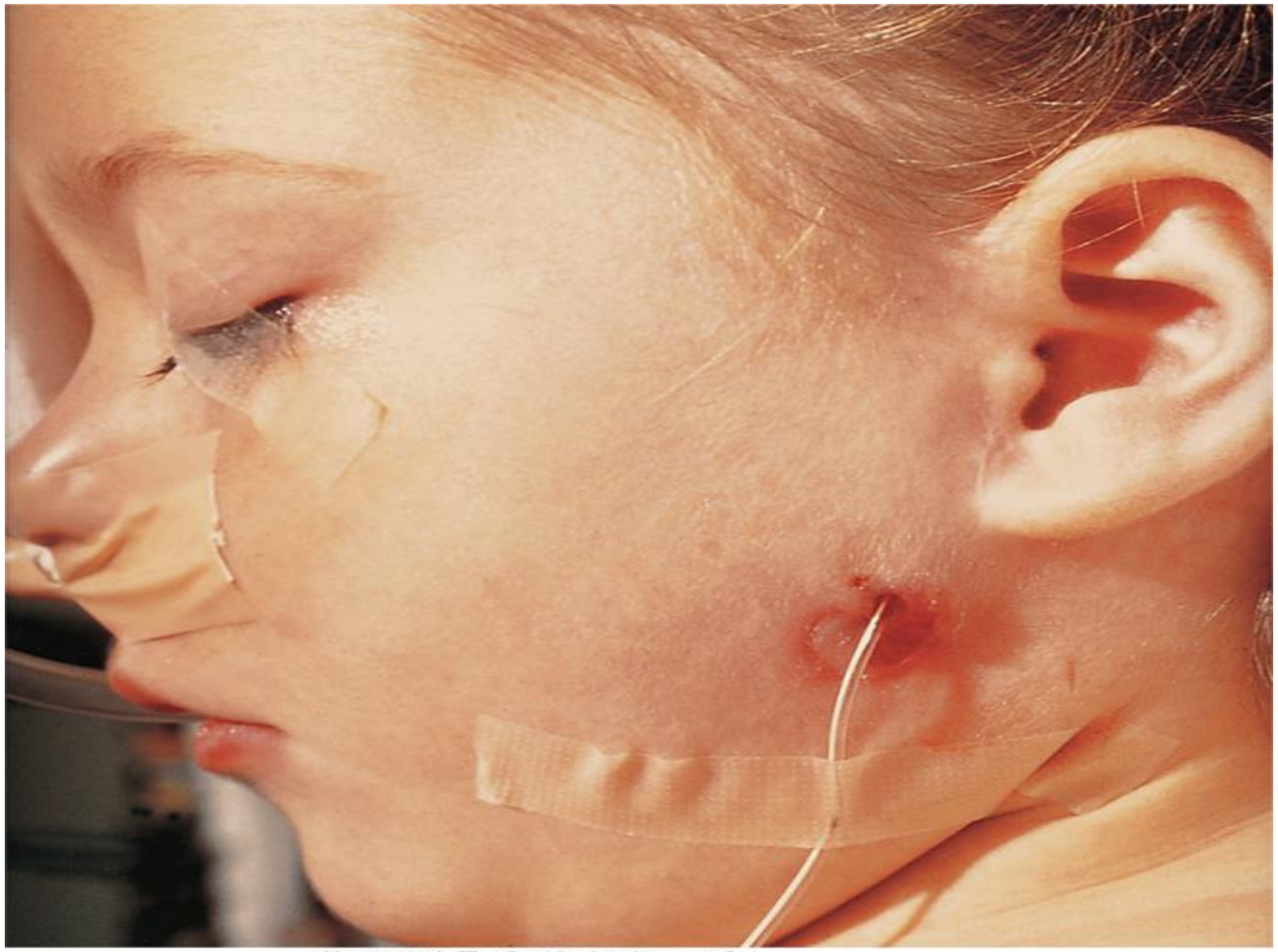
Microtia

- Microtia (a small or rudimentary auricle) results from suppressed mesenchymal proliferation
- This defect often serves as an indicator of associated birth defects, such as an atresia of the external acoustic meatus (80% of cases) and middle ear anomalies.
- The cause can be both genetic and environmental.



- **Preauricular Sinuses and Fistulas**
- Pit-like cutaneous depressions or shallow sinuses are occasionally located in a triangular area anterior to the auricle.
- The sinuses are usually narrow tubes or shallow pits that have pinpoint external openings.
- Some sinuses contain a vestigial cartilaginous mass.
- **Preauricular sinuses** may be associated with internal anomalies, such as deafness and kidney malformations.
- The embryologic basis of auricular sinuses is uncertain but it may relate to incomplete fusion of the auricular hillocks or to abnormal mesenchymal proliferation and defective closure of the dorsal part of the first pharyngeal groove.

- Most of this pharyngeal groove normally disappears as the external acoustic meatus forms.
- Other auricular sinuses appear to represent ectodermal folds that are sequestered during formation of the auricle.
- Bilateral preauricular sinuses are typically familial.
- The majority of sinuses are asymptomatic and have only minor cosmetic importance; however, they can become infected.
- **Auricular fistulas** (narrow canals) connecting the preauricular skin with the tympanic cavity or the tonsillar fossa are extremely rare.

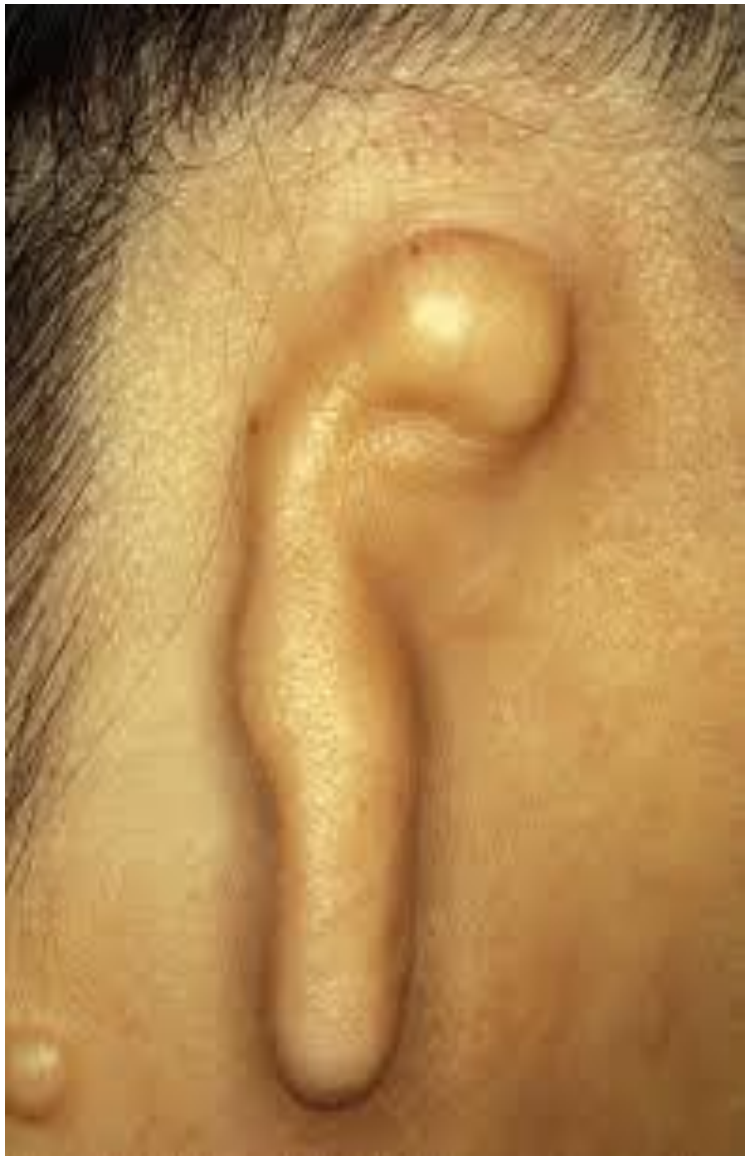


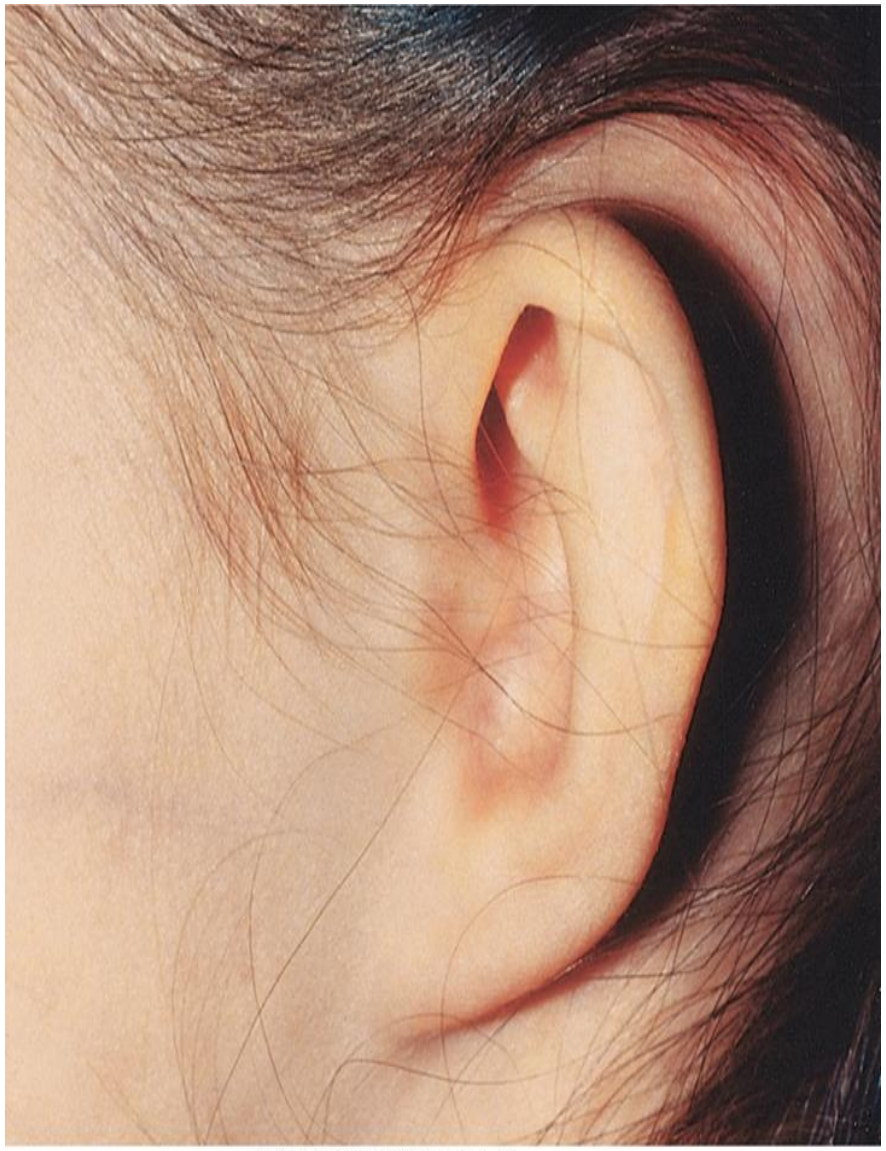
A child with an auricular fistula relating to the first pharyngeal arch. Note the external orifice of the fistula below the auricle and the upward direction of the catheter (in sinus tract) toward the external acoustic meatus.

Atresia of External Acoustic Meatus

- Atresia (blockage) of this canal results from failure of the meatal plug to canalize .
- Usually the deep part of the meatus is open, but the superficial part is blocked by bone or fibrous tissue.
- Most cases are associated with the *first arch syndrome*.
- Often abnormal development of both the first and second pharyngeal arches is involved.
- The auricle is also severely affected and defects of the middle and/or internal ear are sometimes present.
- Atresia of the external acoustic meatus can occur bilaterally or unilaterally and usually results from autosomal dominant inheritance.

- **Absence of External Acoustic Meatus**
- Absence of the external acoustic meatus is rare; usually the auricle is normal.
- This defect results from failure of inward expansion of the first pharyngeal groove and failure of the meatal plug to disappear.
- **Congenital Cholesteatoma**
- This is a fragment of keratinized epithelial cells that is retained after birth.
- The "rest" appears as a white cyst-like structure medial to and behind the tympanic membrane.
- It may be that the rest consists of cells from the meatal plug that was displaced during its canalization.
- It has been suggested that congenital cholesteatoma may originate from an epidermoid formation that normally involutes by 33 weeks gestation.
- Choleseatomas can exhibit growth and invasion of neighboring bone.





a- Large ears.
b- Protruding ears.
c- Lop ear, Cupped ear or Constricted ear (when the seems to fold down and forward).
d- Shell ear (when the curve in the outer rim, as well as the natural folds and creases, are missing).
e- Stahl ear or Spock's ear (pointed outer rim).
f- Cryptotia (Absence of depression between ear and head in back of the ear).
g- Micotia or anotia (Small or Absence of ear with or without external ear opening) 3 grades.
h- Ear lobes with large creases and wrinkles (old age).
i- Accessory lobe (Skin tags in cheeks with or without cartilage).

THANK YOU