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Fetal Face and Neck: Anatomy and Anomalies

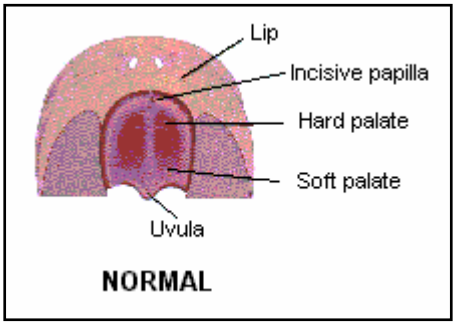
Normal Anatomy

1. **Face:** The upper lip and nares may be visualized in an oblique coronal plane and is useful in searching for facial clefts and some types of proboscis.
2. **Eyes:** The eyes may be imaged in either a true coronal or a transverse plane. Measurement of the outer orbital distance is valuable in diagnosing hypertelorism or hypotelorism. Inner orbital distance measurements may also be used.
3. **Neck**: Soft tissue structures of the neck may be evaluated in both sagittal and transverse planes. Special attention should be paid to surface contours since soft tissue masses may cause protrusion. Transverse sections allow the measurement of the nuchal fold. Studies have shown an association with Down's syndrome when this measurement exceeds 5mm.

Facial Abnormalities

1. **Cleft Palate**: The second most common congenital malformation (13% of all fetal anomalies), most congenital malformations of head and neck originate during transformation of the branchial apparatus into adult derivatives. Occurs in approximately 1: 2500 births. May be unilateral or bilateral.
   a. **Etiology**: Cleft palate and cleft lip are two separate anomalous entities, which evolve at different points in differentiation.

   b. **Embryology**: Results from a failure of mesenchymal masses of the lateral palatine processes to fuse with each other, with the nasal septum and/or median palatine process. Great majority are determined by multiple factors, genetic and non-genetic each causing only a minor developmental defect.



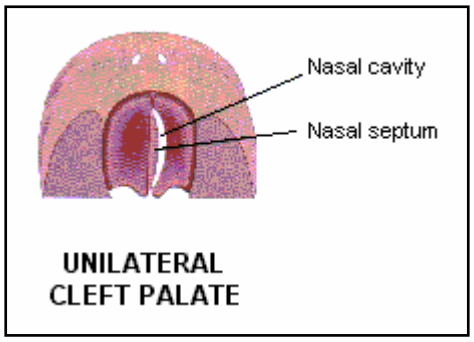
   c. **Pathology**: Clefts of the upper lip and palate are common. The defects are usually classified according to developmental criteria, with the incisive fossa as a reference landmark. Cleft lip and palate are especially

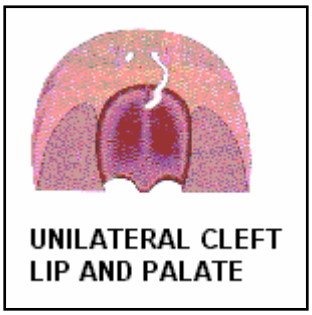
conspicuous because they result in an abnormal facial appearance and defective speech. There are **two major groups** of cleft lip and palate:

i. Clefts involving the upper lip and anterior part of the maxilla, with or without involvement of parts of the remaining hard and soft region of the palate.
ii. Clefts involving the hard and soft regions of the palate.

Specific types of facial clefting include:

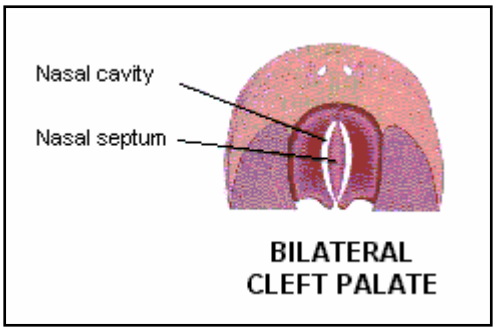
iii. **Anterior cleft anomalies** include cleft lip, with or without cleft of the alveolar part of the maxilla. A complete anterior deft anomaly is one in which the cleft extends through the lip and the alveolar part of the maxilla to the incisive fossa, separating the anterior and posterior parts of the palate.
iv. **Posterior cleft anomalies** include clefts of the secondary or posterior palate that extend through the soft and hard regions of the palate to the incisive fossa, separating the anterior and posterior parts of the palate.
v. **Clefts involving the upper lip**, with or without cleft palate, occur about once in 1000 births, however, their frequency varies widely among ethnic groups**;** 60 - 80% of affected Infants are males. The clefts vary from small notches of the vermilion border of the lip to larger ones that extend into the floor of the nostril and through the alveolar part of the maxilla. Cleft lip can he unilateral or bilateral.
vi. **Unilateral cleft lip** results from failure of the maxillary prominence on the affected side to unite with the merged medial nasal prominences. This results in a persistent labial groove. In addition, the epithelium in the labial groove becomes stretched and the tissues in the floor of the persistent groove break down. As a result, the lip **is** divided into medial and lateral parts. Sometimes a bridge of tissue; a Simonart band, joins the parts of the incomplete cleft.

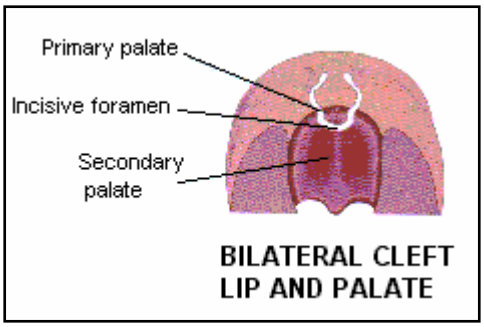




vii. **Bilateral cleft lip** results from failure of the mesenchymal masses in the maxillary prominences to meet and unite with the merged medial nasal prominences. The epithelium in both labial grooves becomes stretched and breaks down. In bilateral cases the defects may be dissimilar, with varying degrees of defect on each side. When there is a complete bilateral cleft of the lip and alveolar part of the maxilla,

the intermaxillary segment hangs free and projects anteriorly. These defects are especially deforming because of the loss of continuity of the orbicularis oris muscle, which closes the mouth and purses the lips as occurs when whistling.





viii. **Median cleft** of the upper lip is an extremely rare defect (see below). It results from a mesenchymal deficiency, which causes partial or complete failure of the medial nasal prominences to merge and form the intermaxillary segment.

ix. A **complete cleft palate** indicates the maximum degree of clefting of any particular type; for example, a complete cleft of the posterior palate is an anomaly in which the cleft extends through the soft palate and anteriorly to the incisive fossa. The landmark for distinguishing anterior from posterior cleft anomalies is the *incisive fossa.* Anterior and posterior cleft anomalies are embryologically distinct.

d. **Sonographic findings**:
   i. Sonographic demonstration of the defect as a groove extending from one nostril through the lip and possible the alveolar ridge.
   ii. Usually best demonstrated in coronal section
   iii. Possible associated polyhydramnios resulting from swallowing difficulties.

2. Median cleft lip

a. **Definition**: Malformation of upper lip with/without cleft palate. The embryologic development of this anomaly is related to the differentiation process of the forebrain and is frequently associated with other midline defects of the face and brain, such as holoprosencephaly.

b. **Pathology**
   i. Vary from small notch to complete division of lip and alveolar part of maxilla.
   ii. Unilateral results from failure of maxillary prominence to fuse with medial nasal prominence. Bilateral results from failure of maxillary processes to meet and merge with medial prominences

c. **Sonographic findings**:
   i. Similar to the diagnosis of cleft lip/cleft palate.

ii. Visualization of the tongue in a higher than usual position in the mouth.
iii. Search for other anomalies and measure orbital distance to detect hyper/hypotelorism.

3. **Epignathus**
   a. **Definition:** A teratoma arising from the oral cavity or pharynx. May arise from the sphenoid bone, hard or soft palate, pharynx, tongue or jaw.
   b. **Pathology**: Most tumors arise from the sphenoid bone. Some arise from the hard and soft palate, the pharynx, the tongue, and jaw. From their sites of origin, the tumors grow into the oral or nasal cavity or intracranially. Most tumors are benign. Histologically, they consist of tissues derived from any of the three germinal layers. Most often, they contain adipose tissue, cartilage, bone, and nervous tissue. These tumors can fill the mouth and airways and lead to acute asphyxia immediately after birth. Obstruction of the mouth is responsible for polyhydramnios.
   c. **Associated anomalies**: Six percent of these tumors have associated anomalies. They include cleft palate, multiple facial hemangiomas, branchial cysts, hypertelorism, umbilical hernia, and congenital heart defects. Facial anomalies have been attributed to the mechanical effects of the tumor on developing structures.
   d. **Sonographic findings**:
      i. Solid, complex tumor seen extruding from the fetal mouth
      ii. Calcifications may be present within mass

Neck Abnormalities

1. **Teratoma of the neck**
   a. **Definition:** Similar to epignathus except the tumor arises from the neck.
   b. **Pathology:** Tumors are generally unilateral and encapsulated. They vary in size and generally consist of a mixture of cystic and solid components. Malignant transformation is extremely rare and there are nor reports of recurrence after complete surgical resection. Obstruction of the airway by the tumor may lead to respiratory failure during the neonatal period.
   c. **Sonographic findings**:
      i. Complex, cystic/solid tumor seen near fetal neck
      ii. Identification of the origin of the mass is essential to differentiate from epignathus.
      iii. Polyhydramnios (30%)

2. **Nuchal thickening**
   a. **Definition**: Increased soft tissue thickness (>5mm) over the posterior aspect of the neck. Infrequently associated with chromosomal syndromes such as Down's syndrome. The measurement is taken in an axial plane of section at the level of the thalami.

b. **Sonographic findings**:
   i. Cross section taken behind the occipital bone
   ii. > 5mm raises suspicion of Down's (15-20 weeks)
   iii. Fetal head must NOT be hyperextended

3. **Cystic hygromas**
   a. **Definition**: Benign developmental anomaly of lymphatic origin characterized by single or multiple cystic areas within soft tissues surrounding the neck. The term “hygroma” means moist tumor. Cystic hygromas are anomalies of the lymphatic system characterized by single or multiple cysts within the soft tissues, usually involving the neck.
   b. **Etiology**: Cystic hygromas are frequently found in association with chromosomal aberrations (mainly Turner’s syndrome). When isolated, this anomaly can be inherited as an autosomal recessive trait. Webbed necks or redundant skin are found in genetic and nongenetic syndromes, such as Noonan’s syndrome, familial pterygium colli, and fetal alcohol syndrome. In the embryo, the lymphatic system drains into the jugular lymphatic sac. A communication between this primitive structure and the jugular vein is formed at 40 days of gestation (conceptional age). Failure of development of this communication results in lymphatic stasis. Dilatation of the lymphatic channels.
   c. **Pathology:** Overdistention of the jugular lymphatic sacs that are located in both sides of the neck results in the formation of a cystic structure that is usually partitioned by a thick fibrous band corresponding to the nuchal ligament. Within the cystic structure, thinner septa are seen and are thought to derive from either fibrous structures of the neck or deposits of fibrin. The size of the lesions may vary greatly from small collections of fluid to enormous cysts that may be larger than the fetus. In cases of generalized hydrops, pleural effusions, ascites, and severe skin edema are present.
   d. **Associated anomalies**: Cystic hygromas are very frequently associated with chromosomal aberrations and, consequently, with a wide variety of anatomic defects.
   e. **Sonographic findings**:
      i. Fluid-filled structure presenting as a cystic mass contiguous with chest wall.
      ii. Thin walled, multiseptated cyst usually located posterior to fetal head/neck.
      iii. Associated with fetal ascites, fetal edema, enlarged edematous placenta, and intradermal fluid collections (cystic cutaneous lymphangiectasia)
      iv. Differential includes teratoma, neural tube defect