Fetal Face and Neck

**NORMAL SONOGRAPHIC ANATOMY**

**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.

**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.

**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 6mm between 15 and 21 weeks.

**FACIAL CLEFTING**

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.

Cleft palate and cleft lip are two separate anomalous entities which evolve at different points in differentiation.

- 1 : 2500 births
- May be unilateral or bilateral.
- 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.
SONOGRAPHIC FINDINGS:
- Sonographic demonstration of the defect as a groove extending from one nostril through the lip and possible the alveolar ridge.
- Usually best demonstrated in coronal section
- Possible associated polyhydramnios
- Cannot detect isolated soft palate clefts

**MEDIAN CLEFT LIP**
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.

PATHOLOGY:
- Vary from small notch to complete division of lip and alveolar part of maxilla.
- 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.

SONOGRAPHIC FINDINGS:
- Similar to the diagnosis of cleft lip/cleft palate.
- Visualization of the tongue in a higher than usual position in the mouth.
- Search for other anomalies and measure orbital distance to detect hyper/hypotelorism.

**EPIGNATHUS**
A teratoma arising from the oral cavity or pharynx. May arise from the sphenoid bone, hard or soft palate, pharynx, tongue or jaw.

SONOGRAPHIC FINDINGS:
- Solid, complex tumor seen extruding from the fetal mouth
- Calcifications may be present within mass
**Teratoma of the neck (Cervical teratoma)**
Similar to epignathus except the tumor arises from the neck.

**Sonographic Findings:**
- Complex, cystic/solid tumor seen near fetal neck
- Identification of the origin of the mass is essential to differentiate from epignathus.

**Nuchal Thickening**
Increased soft tissue thickness over the posterior aspect of the neck. Increased nuchal thickening is reported to be associated with a slightly higher risk of chromosomal syndromes, especially Trisomy 21.

**Sonographic Findings:**
- Oblique axial cross section showing CSP, 3rd ventricle, cerebellum and CM
- > 6mm considered abnormal between 15 and 21 weeks
- Fetal head must NOT be hyperextended

**Cystic Hygroma**
Benign developmental anomaly of lymphatic origin characterized by single or multiple cystic areas within soft tissues surrounding the neck.

**Sonographic Findings:**
- Fluid-filled structure presenting as a cystic mass contiguous with chest wall.
- Thin walled, multiseptated cyst usually located posterior to fetal head/neck but also may be anterior.
- Can be associated with fetal ascites, fetal edema, enlarged edematous placenta, and intradermal fluid collections (cystic cutaneous lymphangiectasia)
- May mimic cervical teratoma, neural tube defect

Coronal section through fetus with cystic hygromas, bilateral pleural effusions, severe soft tissue edema.