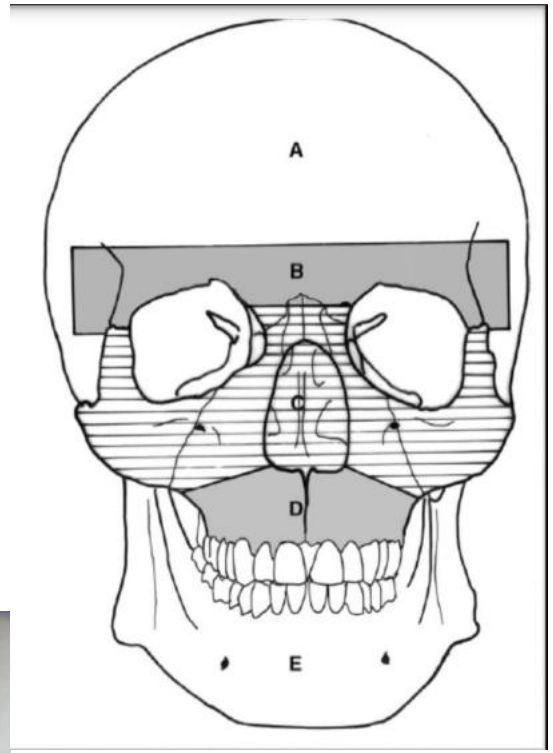
Craniofacial Anomalies



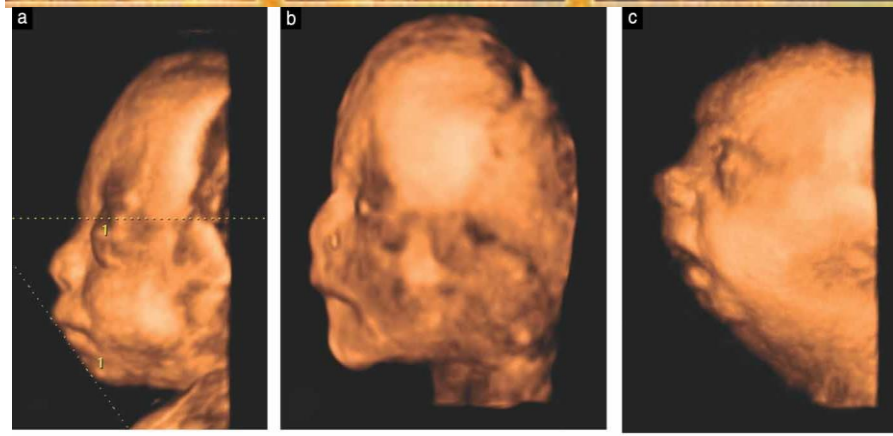
Prof. Abbas AY Taher

Craniofacial Anomalies :

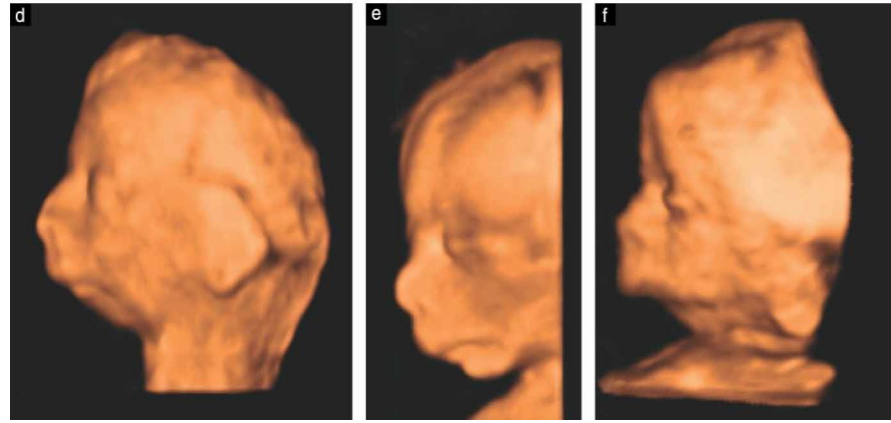
1-Craniofacial anomalies are **deformities that affect a child's head and facial bones**. **These disorders are typically present at birth (congenital)** and **can range from mild to severe**. Common craniofacial anomalies include: **cleft lip and palate**: a separation in the lip and the palate

2-Craniofacial anomalies include a wide spectrum of malformations. They may **be clinically relevant as such**, but they may also be associated with other **congenital anomalies or be part of a syndrome**.

Evaluation of the face is indeed an important part of the clinical genetic examinations performed postnatally. Therefore, any time a fetal anomaly is identified during an **antenatal US scan**, the diagnostic workup should include a detailed examination of the fetal face. Apart from obvious malformations, a prenatal sonogram may also identify subtle dysmorphisms that may be crucial for a definitive diagnosis.

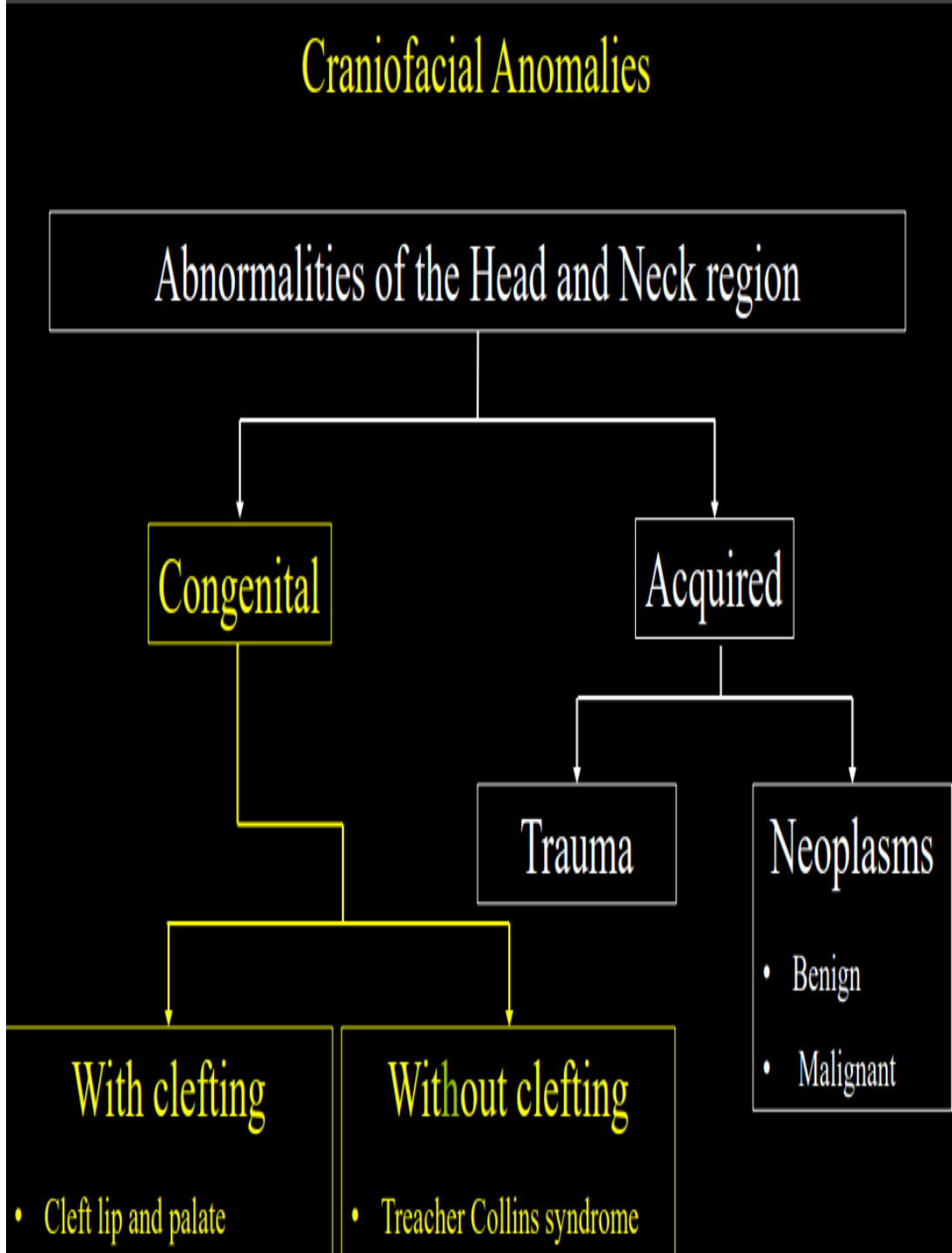


Prenatal sonographic diagnosis of craniofacial anomalies is possible in early gestation.The diagnostic accuracy of referral centers in the investigation of selected patients at increased risk is quite high,whereas the sensitivity of standard examinations of low-risk patients is low, in the range of **20% to -40%,** with a general tendency to recognize facial malformations associated with other anomalies and to miss the isolated ones.



However, until a few years ago, most national guidelines for the standard examination of fetal anatomy suggested only demonstration of the orbits and the eyes. More recently, the recommendations include visualization of the nose, lips, and chin, with an expected positive impact on the detection of craniofacial anomalies

• Craniosynostosis
Plagiocephaly/Trigonocephaly/Scaphocephaly/Brachycephaly
• Craniofacial Dysostosis
Crouzon Syndrome/Apert Syndrome/Pfeiffer Syndrome/Cloverleaf skull
• Craniofacial Syndromes and Anomalies
Treacher Collins Syndrome/Hemifacial Microsomia/Binder Syndrome/Pierre Robin Sequence/Craniofrontonasal Dysplasia/Craniofacial Clefts • Cleft Lip and Palate
• Benign and Malignant Head and Neck Tumors
Encephaloceles/Dermoid cysts/Hemangiomas/Vascular Malformations/Fibrous Dysplasia/Sarcoma and other malignancies
• Craniomaxillofacial Trauma
• Non-syndromic Orthognathic Deformities



Developmental disturbances can affect the normal growth and differentiation of craniofacial structures, resulting in observable craniofacial anomalies.
Thesedevelopmental craniofacial anomalies are usually first discovered in infancy or childhood.
Some are caused by known and recently discovered genetic mutations, others result from environmental factors, and a third group are multifactorial.
These conditions are manifested by a **variety of abnormalities of the face and jaws**, including abnormalities of structure, shape, **organization**, and **function of hard and soft tissues**. A multitude of conditions affect the morphogenesis of the face and jaws, many of which are rare syndromes.

Cleft lip/palate results in defects in the alveolar ridge and abnormalities of the dentition. (A) Bilateral clefts of the maxilla in the lateral incisor regions and defects of the dentition. (B) Lateral cephalometric skull image shows underdevelopment of the maxilla.

Cleft Lip and Palate

Facial clefts of various types are caused by a failure of fusion of the developmental processes of the face during fetal development. Cleft lip and cleft palate (CL/P), cleft lip (CL), and cleft palate (CP) together are the most common developmental craniofacial anomalies worldwide. Their incidence varies with geographic location, ethnic background, and socioeconomic status. Worldwide, the average birth prevalence of all orofacial clefts is 9.92 per 10,000. In Caucasian populations, the incidence of CL is 1 : 800 to 1 : 1000 live births, and the incidence of CP is approximately 1 : 1000. CL with or without CL/P and CP are two different conditions with different etiologies.

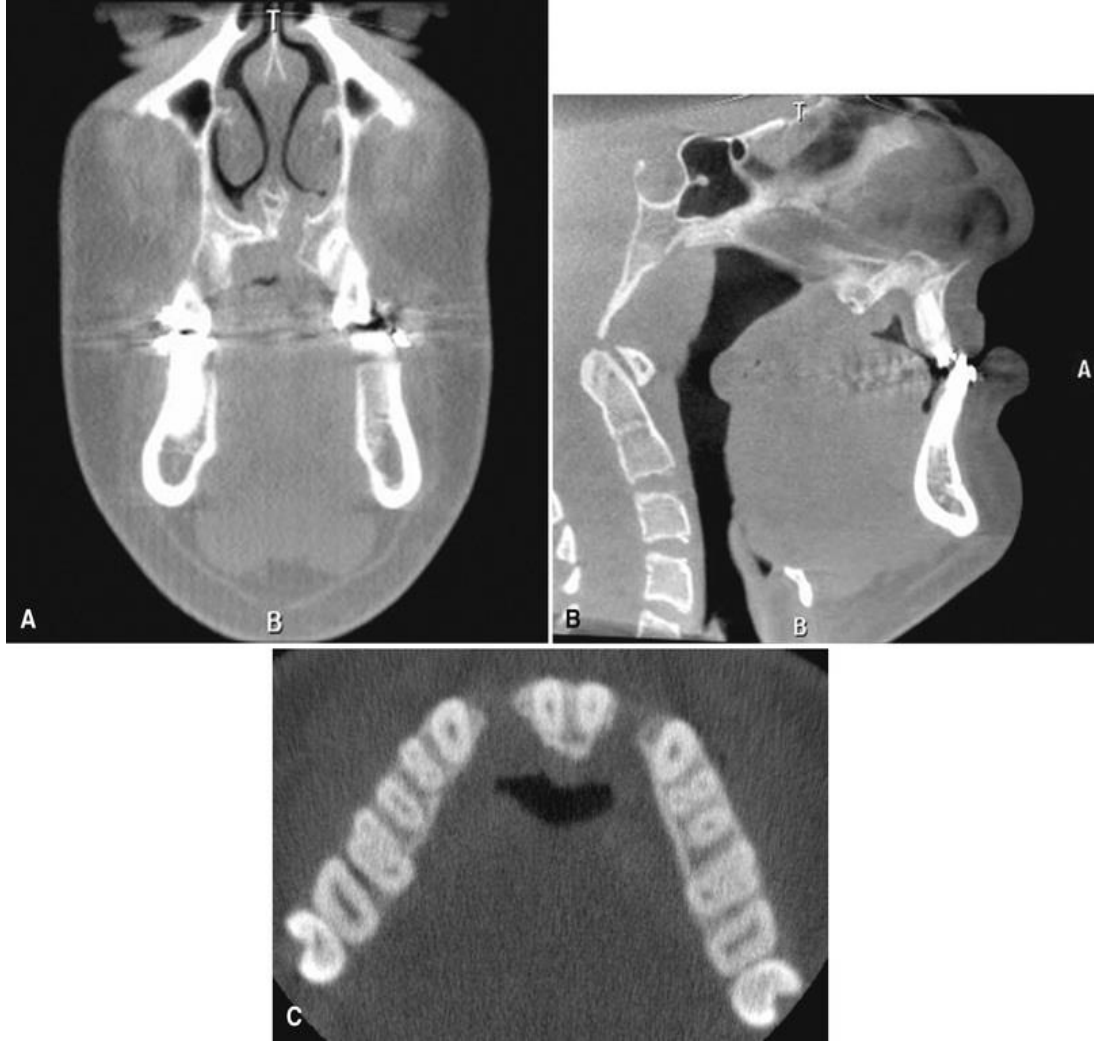
Left lip and palate :

CL/P results from a failure of fusion of the medial nasal process with the maxillary process, and can range in severity from a unilateral CL to bilateral complete clefting through the lip, maxillary alveolar processes, and hard and soft palate in the most severe cases. CP develops from a failure of fusion of the lateral palatal shelves.

The minimal manifestation of CP is a submucous cleft in which the palate appears to be intact except for notching of the uvula (bifid uvula) or notching in the posterior border of the hard palate, detectable by palpation. The most severe presentation of CP is complete clefting of the hard and soft palate.

The precise etiology of orofacial clefting is not yet completely understood. However, most cases of non-syndromic CL/P and CP are considered to follow a multifactorial threshold model, with environmental risk factors and a strong genetic component.

Several genes are associated with orofacial clefting. CL/P and CP each can be associated with other abnormalities, as part of a genetic malformation syndrome, **such as 22q.11 deletion syndrome (velocardiofacial syndrome—CP and facial** and cardiac abnormalities) or van der Woude syndrome (CL or CP or both and lip pits).



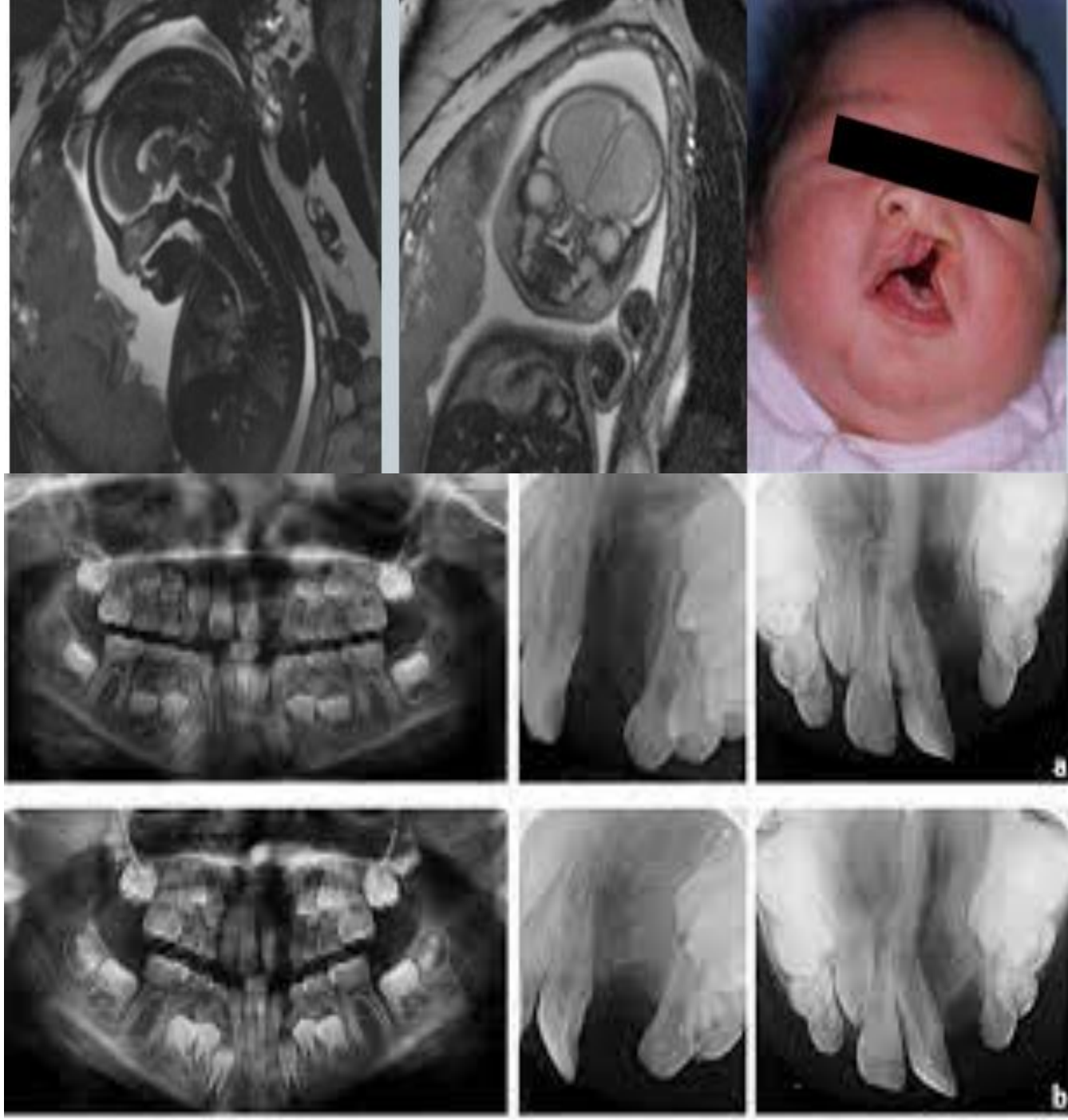
Cone beam computed tomography (CBCT) images of a patient with left unilateral cleft lip/palate. (A) Coronal view. Note the discontinuity in the nasal floor visible on the patient's left side. (B) Sagittal view of the same patient shows maxillary hypoplasia and deficient palatal anatomy. (C) Axial
CBCT image of a different patient with bilateral clefts shows bilateral defects in the maxillary alveolar processes. ([A and B],

A vertical hypoechoic region through the fetal upper lip usually represents the defect in cleft lip. This finding may be corroborated by a similar defect of the soft tissues of the upper lip overlying the maxilla in the axial plane. The palate can be examined in the transverse (axial) plane.

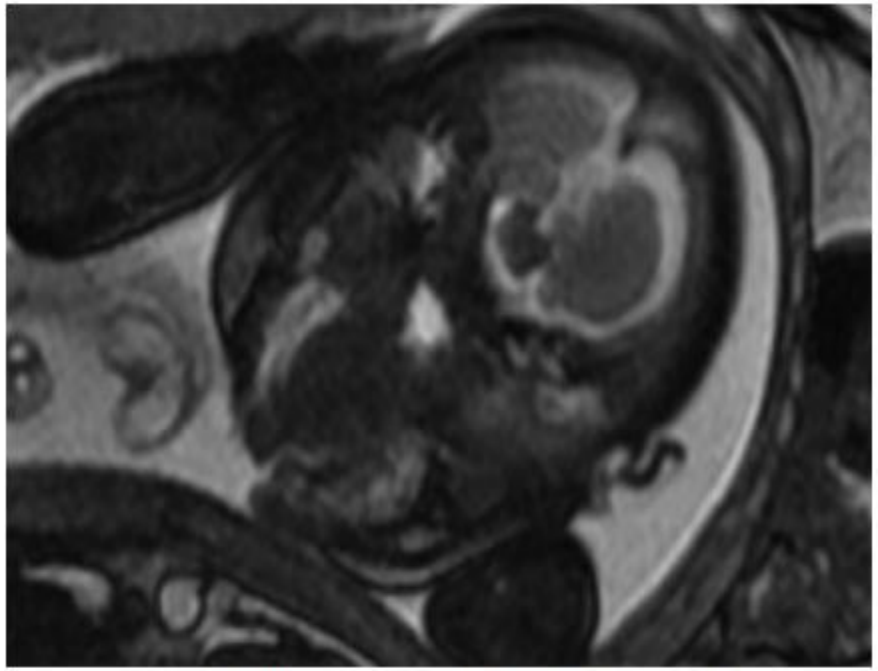
Imaging Modalities and Protocols Conventional and Three-dimensional US Conventional US can depict cleft lip and palate with use of standard orthogonal views.

The coronal view allows visualization of the nares and upper lip .

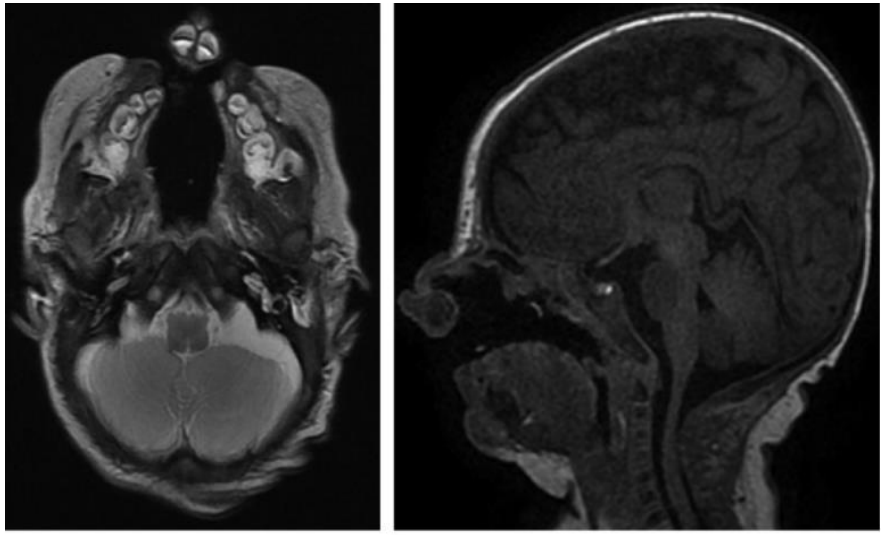
The midsagittal view depicts the hard and soft palate, and the axial view demonstrates the maxillary alveolar ridge . Three dimensional (3D) US allows better diagnosis of cleft lip and palate, and surface renderings are useful to help prepare parents psychologically and can also be used by the medical teams for postnatal management



Radiography and Computed Tomography Radiography is not typically helpful for diagnosis of cleft lip and palate; however, panoramic and dental radiographs are used to detect associated dental anomalies.
Computed tomographic (CT) imaging of CLP is particularly useful for demonstrating bone and dental anatomy and is often used before repair of dentofacial deformities; multiplanar reformations of helical CT images with use of bone and soft-tissue algorithms can help depict anatomic abnormalities, and 3D reconstructions can aid in surgical planning as well as patient and family education. Intravenous contrast material is not typically required. MR Imaging MR imaging is the criterion standard for evaluation of possible associated intracranial abnormalities in the setting of CLP or cleft palate. Prenatal (fetal) MR imaging can aid in confirmation and characterization of the cleft itself as well as associated intracranial and extracranial findings .
Postnatal MR imaging is not typically required for characterization of the cleft itself.
Althoughcleft palate can be seen on sagittal MR images,coronal MR images best depict cleft palate. Axial MR images may help confirm the integrity of the maxillary arch. **T2-weighted MR imaging with or without fat saturation can be helpful**. Intravenous contrast material is not typically required for evaluation **of CLP or cleft palate**.



Axial MR image in a fetus at 32 weeks of gestation shows a cleft of the maxillary alveolus.

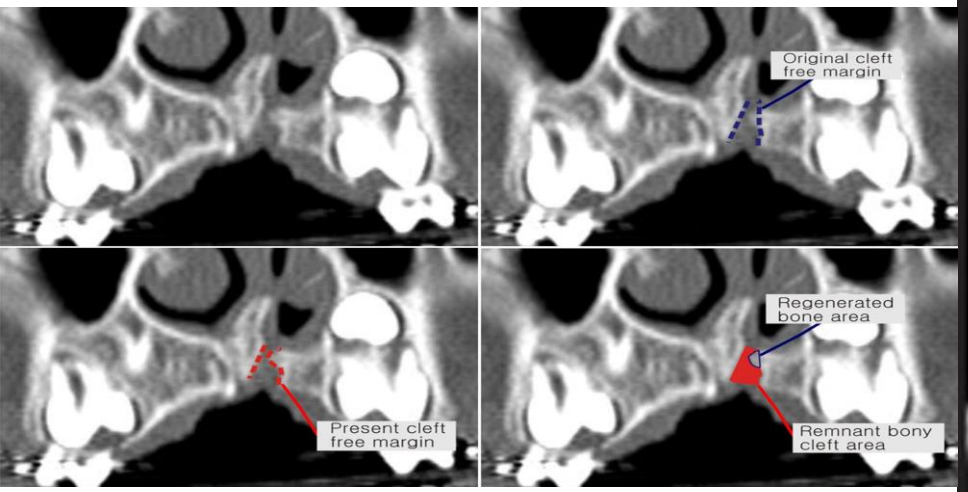


b.

Axial **(a)** and sagittal **(b)** MR images in a 3-week-old boy show bilateral complete cleft lip and pal-
ate with a protrusive premaxillary segment.

Other factors that are implicated in the development of orofacial clefts include nutritional disturbances (prenatal folate deficiency), environmental teratogenic agents (maternal smoking, *in utero* exposure to anticonvulsants),stress (which results in increased secretion of hydrocortisone), defects of vascular supply to the involved region, and mechanical interference with the fusion of the embryonic processes (CP in Pierre Robin sequence). **Cleft involving the lower lip and mandible are extremely rare**

Clefts of the palate are usually surgically repaired within the first year of life, whereas clefts of the lip are usually **repaired within the first 3 months** to aid in feeding and maternal-infant bonding. The bone in the cleft site is often augmented with bone graft material before replacement of missing teeth with either fixed or removable prosthodontics or dental implants. Orthodontic treatment, without or with orthognathic surgery, is usually necessary to re-create a normal arch form and functional occlusion



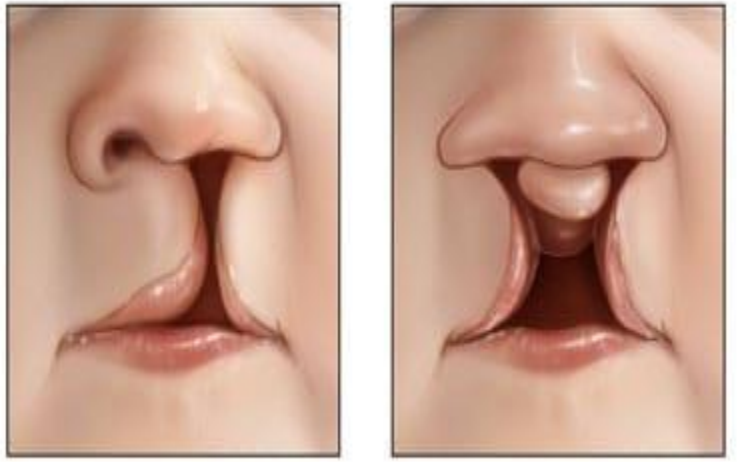
Cleft Lip

Incomplete unilateral cleft lip

Complete Unilateral cleft lip

Incomplete bilateral cleft lip

Complete bilateral cleft lip



Unilateral cleft lip

Bilateral cleft lip

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unilateral cleft palate

Thank You!