Diploma in Fetal Medicine & ISUOG Educational Series

The 18 - 23 weeks scan

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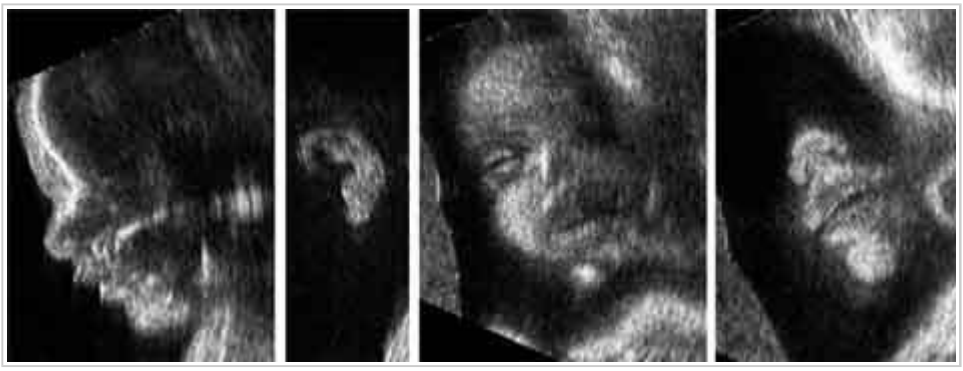
Chapter 3

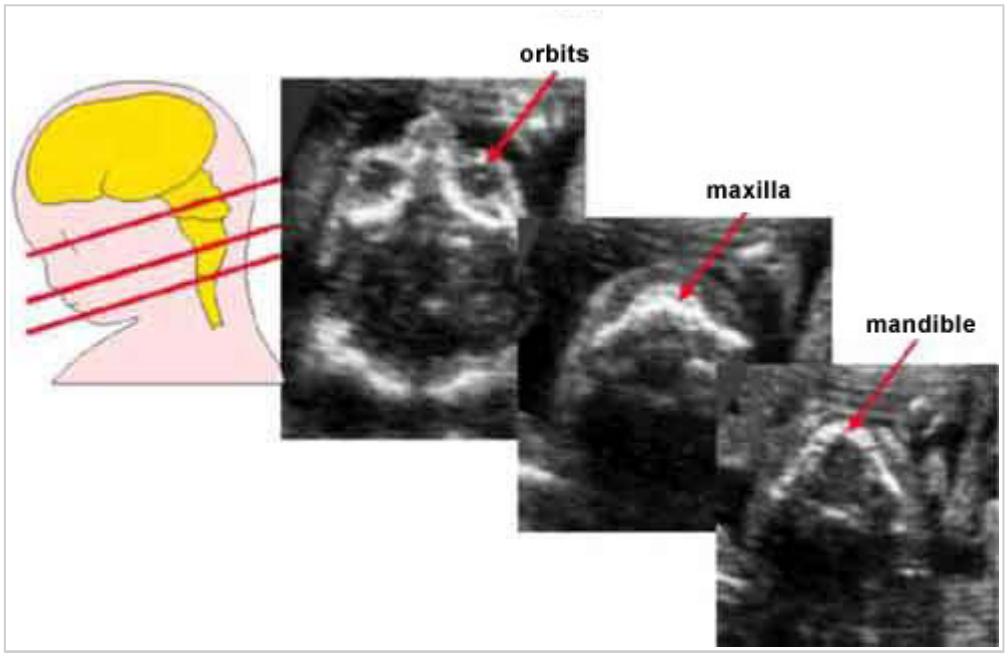
Face

NORMAL SONOGRAPHIC ANATOMY

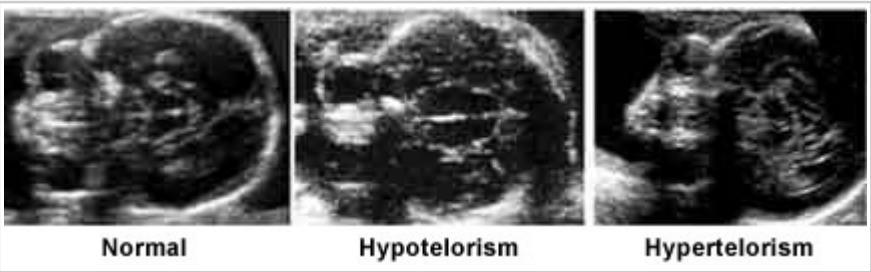
The forehead, orbits, nose, lips and ears can be consistently identified from 12 weeks of gestation. Sagittal, transverse and coronal planes are all useful for the evaluation of normal and abnormal anatomy. A mid-sagittal plane allows visualization of the fetal profile, whereas the ears are visualized in parasagittal scans tangential to the calvarium. The coronal planes are probably the most important ones in the evaluation of the integrity of facial anatomy. Orbits, eyelids, nose, and lips are well visualized. The tip of the nose, the alae nasi, and the columna are seen above the upper lip. The nostrils typically appear as two little anechoic areas.



A series of transverse scans from the top of the head moving caudally allows examination of the forehead, nasal bridge, orbits, nose, upper lip and anterior palate, the tongue within the oral cavity, lower lip and mandible. The presence and size of the eyes are assessed subjectively. As a rule of thumb, each orbital diameter is equal in size to the interorbital diameter. In cases of suspected defects measurement of the internal and external orbital diameters may be necessary.



ORBITAL DEFECTS



Hypertelorism (Euryopia)

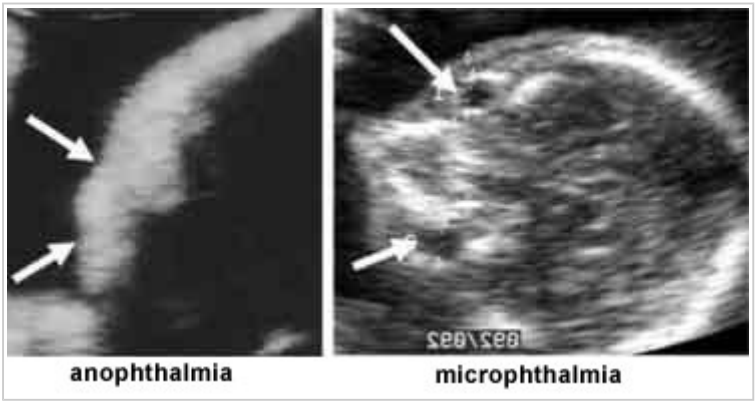
In early development the eyes are placed laterally in the primitive face in a fashion similar to that of lower animals with panoramic vision. As gestation progresses, they migrate toward the mid-line, creating favorable conditions for the development of stereoscopic vision. Hypertelorism is an increased interorbital distance and this can be either an isolated finding or associated with many clinical syndromes or malformations. The most common syndromes with hypertelorism are the median cleft syndrome (hypertelorism, median cleft lip with or without a median cleft of the hard palate and nose, and cranium bifidum occultum), craniosynostoses (including Apert, Crouzon, and Carpenter syndromes), agenesis of the corpus callosum and anterior encephaloceles. Hypertelorism per se results only in cosmetic problems and possible impairment of stereoscopic binocular vision. For severe cases, a number of operative procedures, such as canthoplasty, orbitoplasty, surgical positioning of the eyebrows, and rhinoplasty, have been proposed. The median cleft face syndrome is usually associated with normal intelligence and life span. However, there is a high likelihood of mental retardation when either extracephalic anomalies or an extreme degree of hypertelorism is found. The severity of the cosmetic disturbance should not be underestimated, because this syndrome may be associated with extremely grotesque features.

Hypotelorism (stenopia)

Hypotelorism (decreased interorbital distance) is almost always found in association with other severe anomalies, such as holoprosencephaly, trigonocephaly, microcephaly, Meckel syndrome, and chromosomal abnormalities. The prognosis, which depends on the associated anomalies, is usually very poor.

Microphthalmia / anophthalmia

Microphthalmia is defined as a decreased size of the eyeball and anophthalmia refers to the absence of the eye; however, the term anophthalmia should be reserved for the pathologist, who must demonstrate not only absence of the eye but also of optic nerves, chiasma, and tracts. Microphthalmia / anophthalmia, which is either unilateral or bilateral, is usually associated with with one of about 25 genetic syndromes. In Goldenhar syndrome (found in about 1 per 5,000 births) there is unilateral anophthalmia, together with ear and facial abnormalities. Prenatal diagnosis is based on the demonstration of decreased ocular diameter and careful examination of the intraorbital anatomy is indicated to identify lens, pupil, and optic nerve. Congenital microphthalmia is frequently associated with visual disorders and with other anomalies.

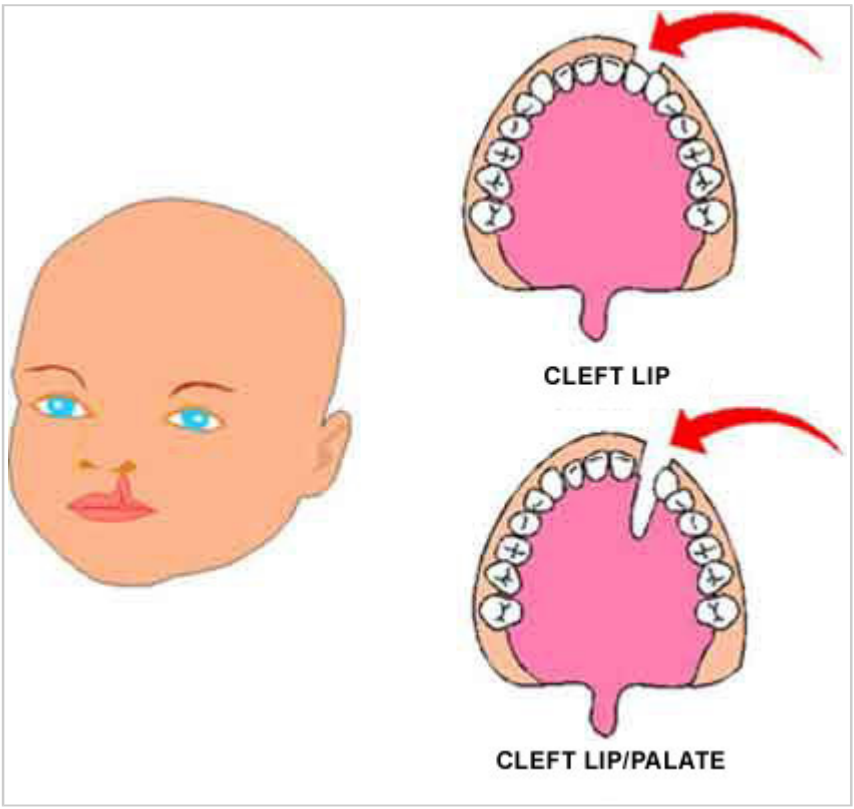


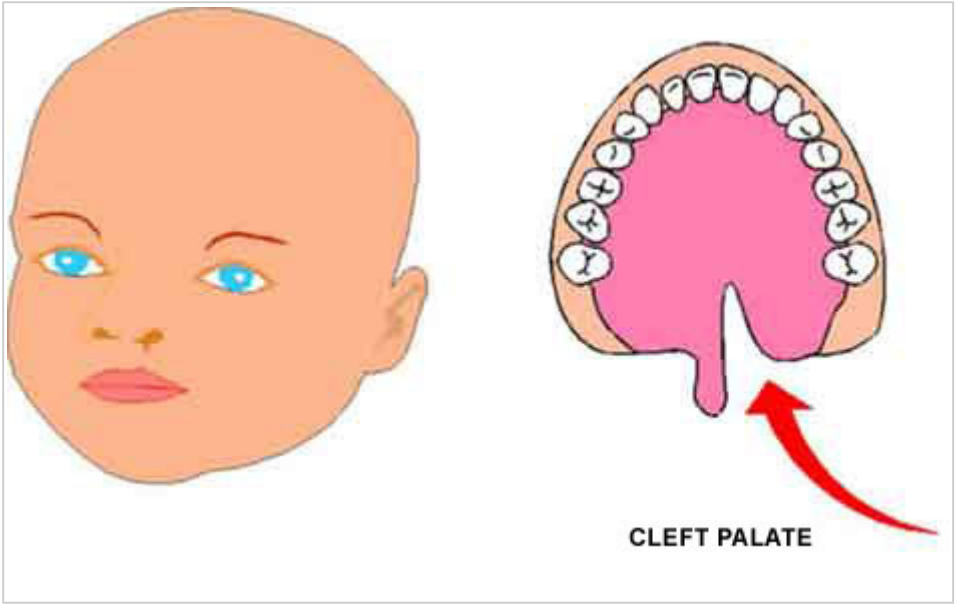
FACIAL CLEFT

This term refers to a wide spectrum of clefting defects (unilateral, bilateral and less commonly mid-line) usually involving the upper lip, the palate, or both. Cleft palate without cleft lip is a distinct disorder. Facial clefts encompass a broad spectrum of severity, ranging from minimal defects, such as a bifid uvula, linear indentation of the lip, or submucous cleft of the soft palate, to large deep defects of the facial bones and soft tissues. The typical cleft lip will appear as a linear defect extending from one side of the lip into the nostril. Cleft palate associated with cleft lip may extend through the alveolar ridge and hard palate, reaching the floor of the nasal cavity or even the floor of the orbit. Isolated cleft palate may include defects of the hard palate, the soft palate, or both. Both cleft lip and palate are unilateral in about 75% of cases and the left side is more often involved than the right side.

Prevalence

Facial clefting is found in about 1 per 800 births. In about 50% of cases both the lip and palate are defective, in 25% only the lip and in 25% only the palate is involved.



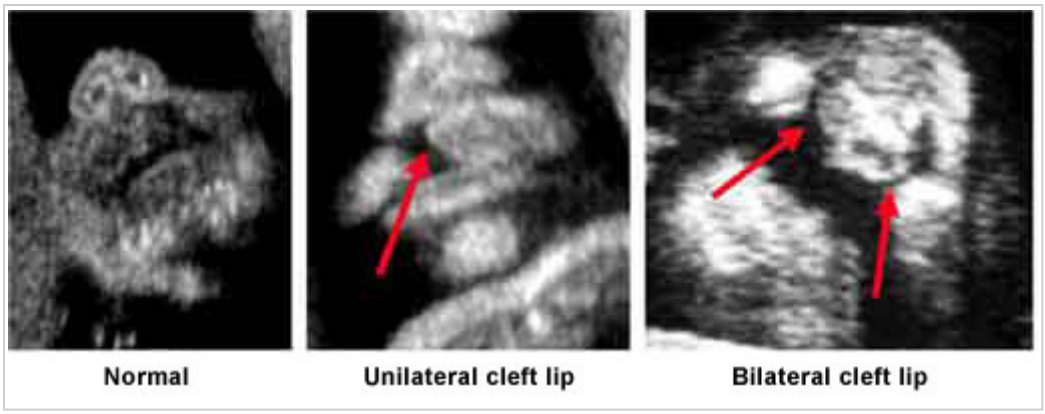


Etiology

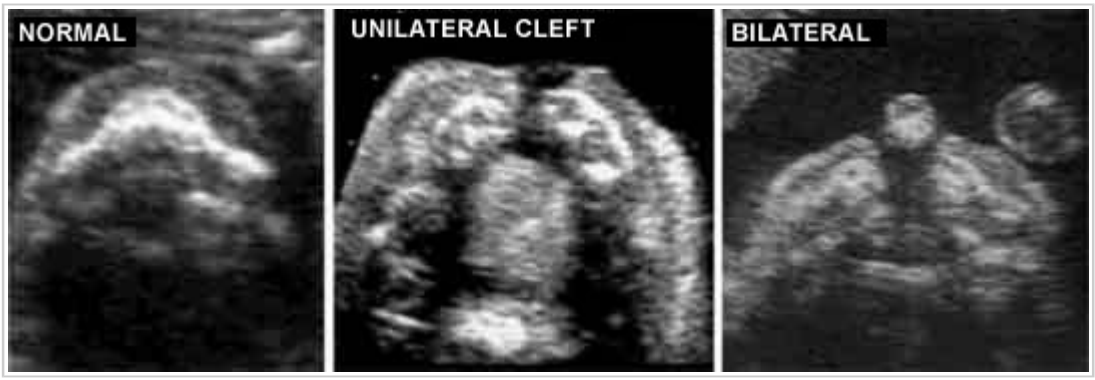
The face is formed by the fusion of four outgrowths of mesenchyme (frontonasal, mandibular and paired maxillary swellings) and facial clefting is caused by failure of fusion of these swellings. Cleft lip with or without cleft palate is usually (more than 80% of cases) an isolated condition, but in 20% of cases it is associated with one of more than 100 genetic syndromes. Isolated cleft palate is a different condition and it is more commonly associated with any one of more than 200 genetic syndromes. All forms of inheritance have been described, including autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive. Associated anomalies are found in about 50% of patients with isolated cleft palate and in about 15% of those with cleft lip and palate. Chromosomal abnormalities (mainly trisomy 13 and 18) are found in 1-2% of cases and exposure to teratogens (such as antiepileptic drugs) in about 5% of cases. Recurrences are type specific; if the index case has cleft lip and palate there is no increased risk for isolated cleft palate, and vice versa. Median cleft lip, which accounts for about 0.5% of all cases of cleft lip, is is usually associated with holoprosencephaly or the oral-facial-digital syndrome.

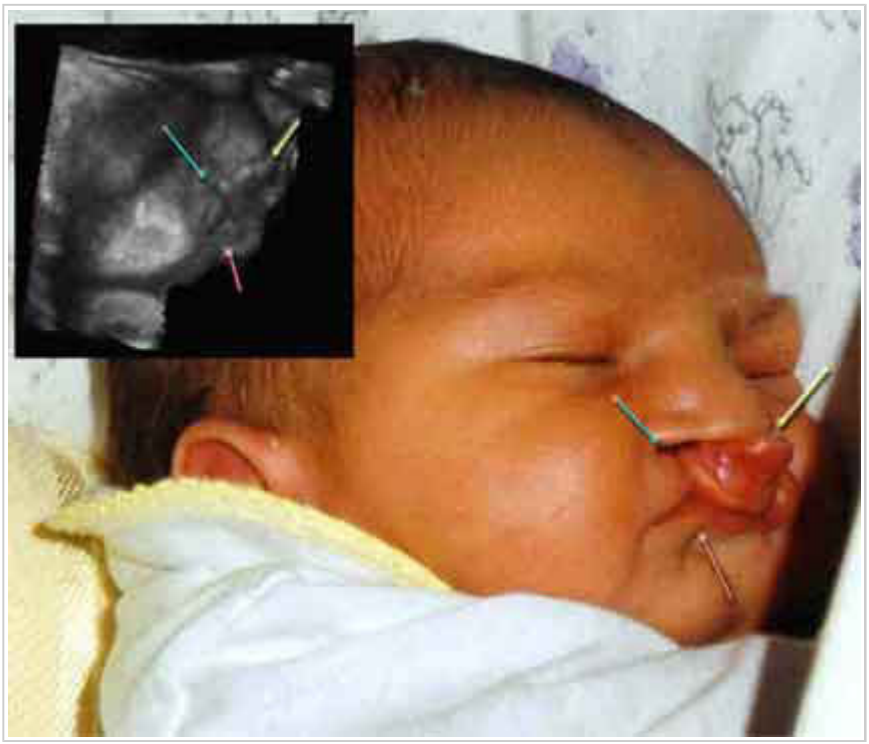
Diagnosis

The sonographic diagnosis of cleft and palate depends on demonstration of a groove extending from one of the nostrils inside the lip and possibly the alveolar ridge. Both transverse and coronal planes can be used.



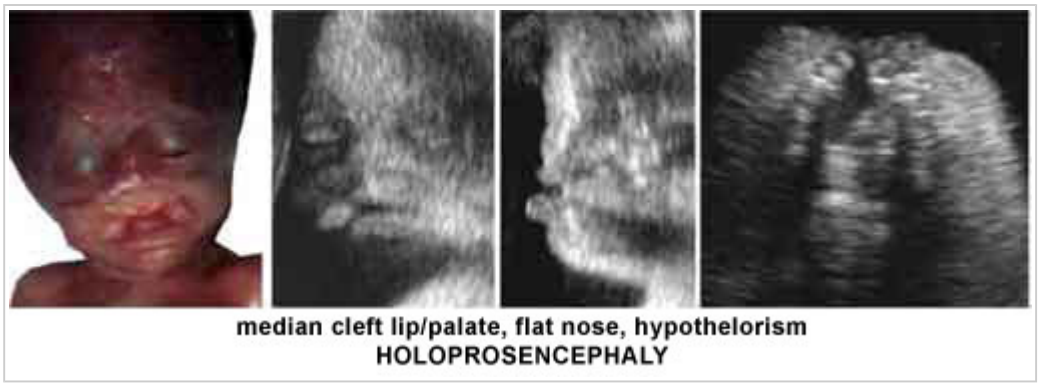
A transverse scan is required to distinguish isolated cleft lip from cleft lip/palate.





Bilateral Cleft Lip and Palate - 3D view (yellow arrow "flap")

Median cleft lip is usually associated with other facial anomalies (hypertelorism with median cleft face syndrome, hypotelorism with holoprosencephaly).



The diagnosis of isolated cleft palate is difficult and in cases at risk for Mendelian syndromes fetoscopy may be necessary.

Prognosis

Minimal defects, such as linear indentations of the lips or submucosal cleft of the soft palate, may not require surgical correction. Larger defects cause cosmetic, swallowing, and respiratory problems. Recent advances in surgical technique have produced good cosmetic and functional results. However, prognosis depends primarily on the presence and type of associated anomalies.

MICROGNATHIA

Micrognathia is characterized by mandibular hypoplasia causing a receding chin.

Prevalence

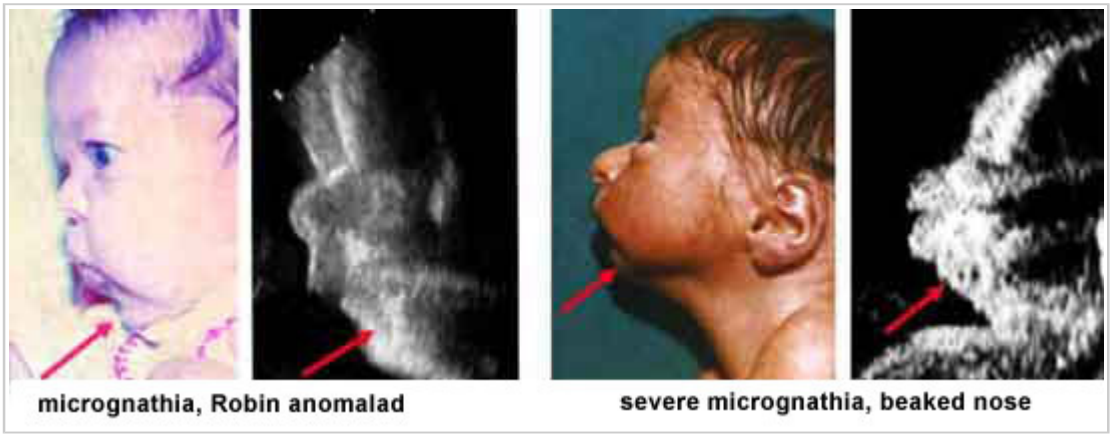
Micrognathia is found in about 1 per 1,000 births.

Etiology

Micrognathia is usually associated with genetic syndromes (such as Treacher-Collins, Robin and Robert syndromes), chromosomal abnormalities (mainly trisomy 18 and triploidy) and teratogenic drugs (such as methotrexate). The Robin anomalad (severe micrognathia, glossoptosis and a posterior cleft palate or an arched palate) may be a sporadic isolated finding (in about 40% of cases) or it may be associated with other anomalies or with recognized genetic and non-genetic syndromes. Otocephaly is a rare, lethal, sporadic abnormality characterized by severe hypoplasia of the mandible (agnathia) and severe midline defects, including holoprosencephaly, anterior encephalocele, cyclopia, aglossia, microstomia, and mid-facial location of the ears ('ear-head').

Diagnosis

Micrognathia is a subjective finding in the midsagittal view of the face and is characterized by a prominent upper lip and receding chin. The diagnosis can be confirmed by the demonstration of a short mandible. Severe micrognathia is associated with polyhydramnios possibly because of the glossoptosis preventing swallowing.



micrognathia, Robin anomalad

severe micrognathia, beaked nose

Prognosis

This depends on the presence of associated anomalies. Severe micrognathia can be a neonatal emergency due to airway obstruction by the tongue in the small oral cavity. If prenatal diagnosis is made a pediatrician should be present in the delivery room and be prepared to intubate the infant. Otocephaly is lethal.

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