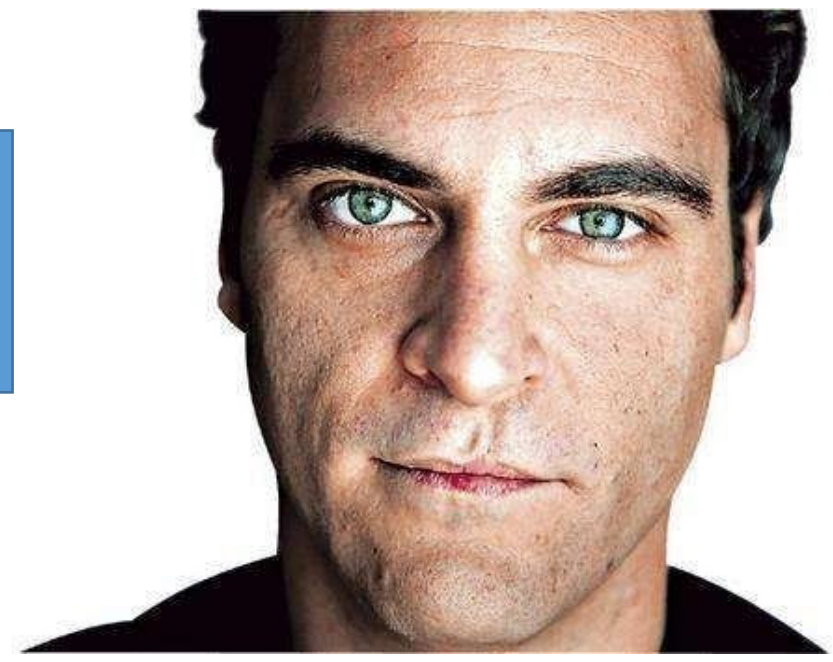
Cleft lip, cleft palate and craniofacial syndromes

Department of Burns and Plastic Surgery

Cleft lip and palate



Epidemiology

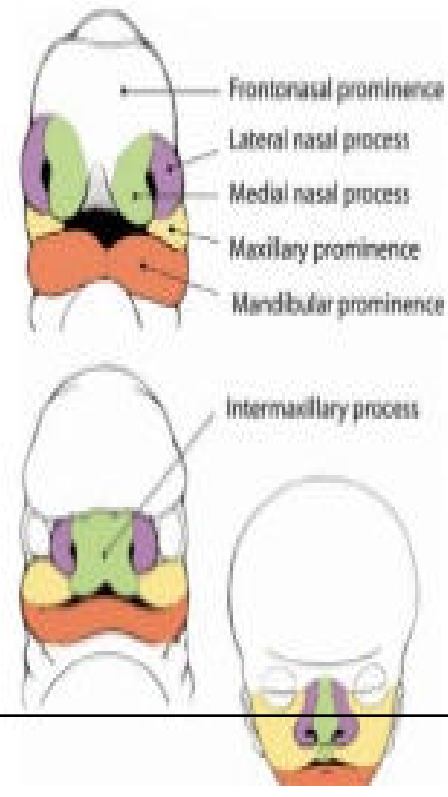
Incidence

- The approximate incidence is 1 in 700 live births, among them 25% are bilateral and 85% are associated with cleft palate. Isolated cleft palate occurs in 1 in 2000 live births .
- Negroids having least incidence (0.4/1000) and mongoloid and afghans(4.9/1000) having the highest incidence.
- Cleft lip is more common among males and cleft palate is more commoly among females.
- Unilateral clefts accounting for 80% of incidence and bilateral for remaining 20%.

Embryology

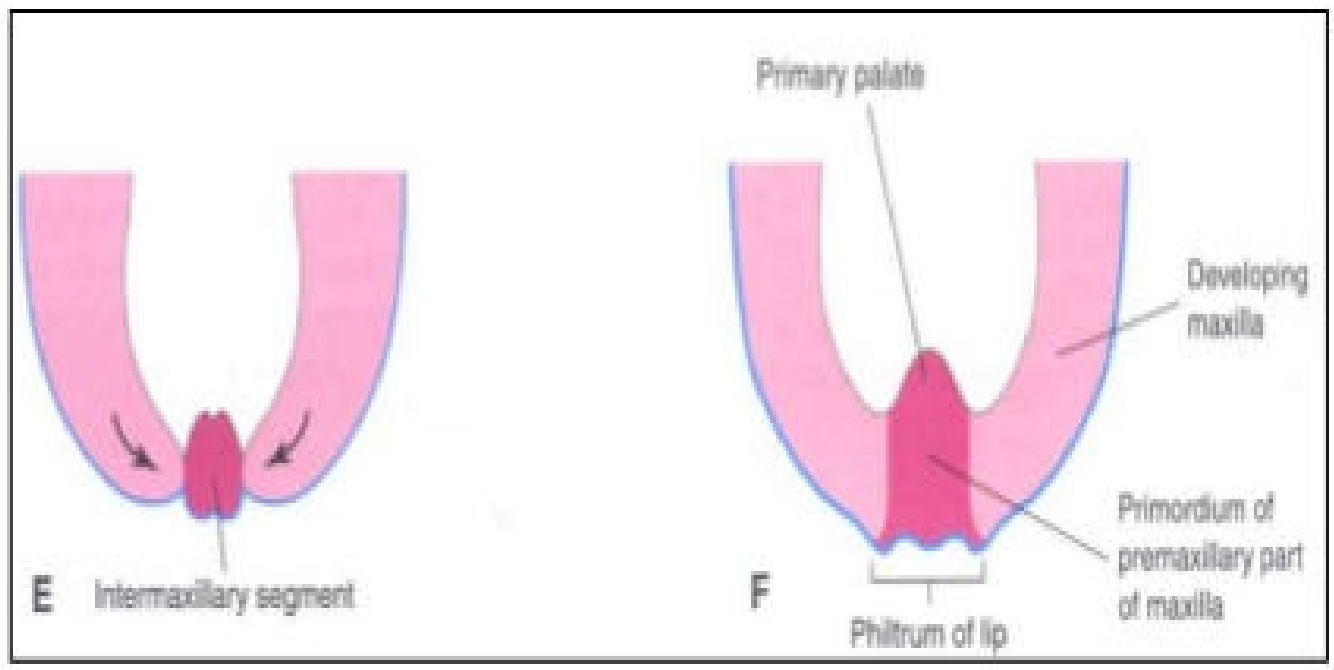
- Face starts forming by 4 rth week and completed by 8 th week
- Palate formation is completed by 10 th week

- Development of facial structures starts at the end of 4th week
- 5 facial prominences around stomatodeum
  1. Unpaired frontonasal process
  2. Paireed maxillary prominences
  3. Paired mandibular prominences

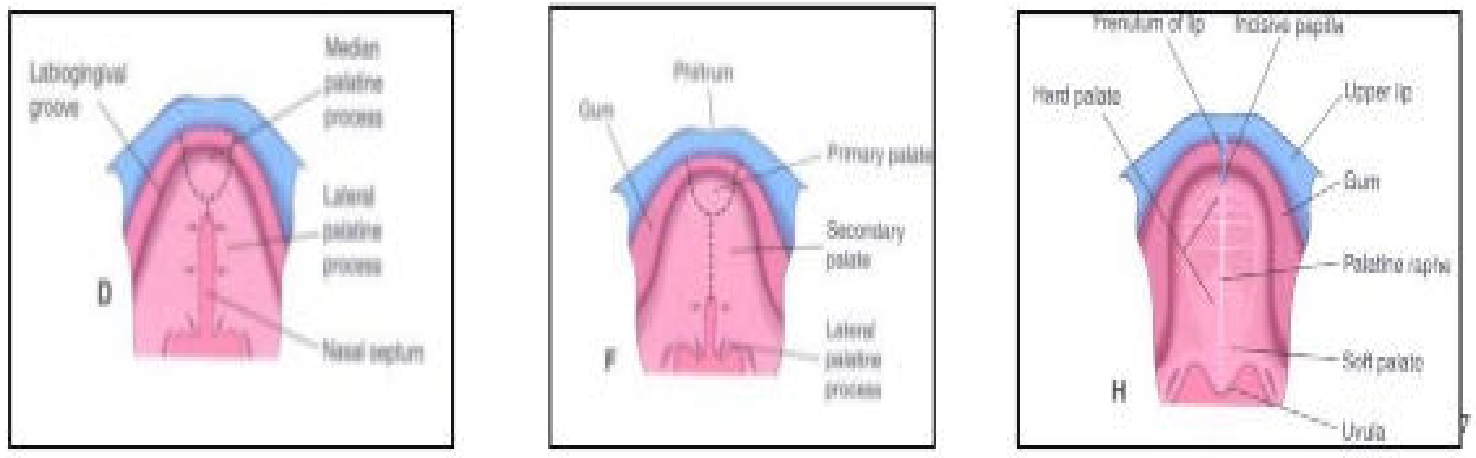


- In following 2 weeks –
  - The 2 medial nasal processes fuse in midline – **upper lip**
  - Mandibular processes fuse in midline – **lower lip**

- **Primary palate** – maxillary and medial nasal process merge
- Formation of **intermaxillary segment** from merged medial nasal prominences

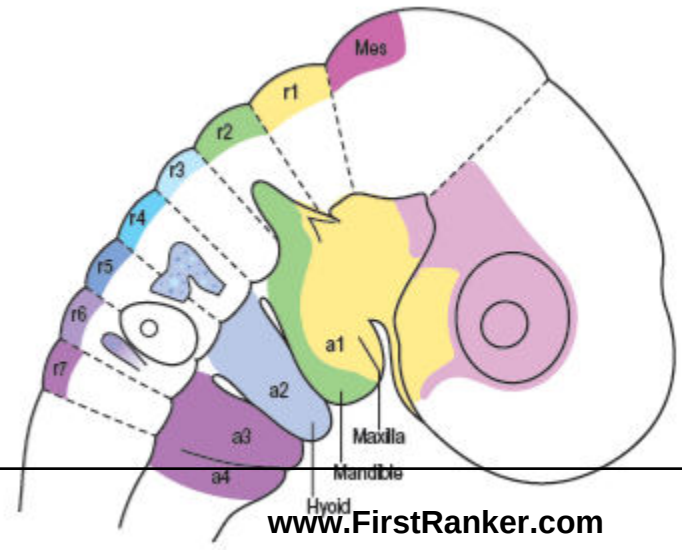


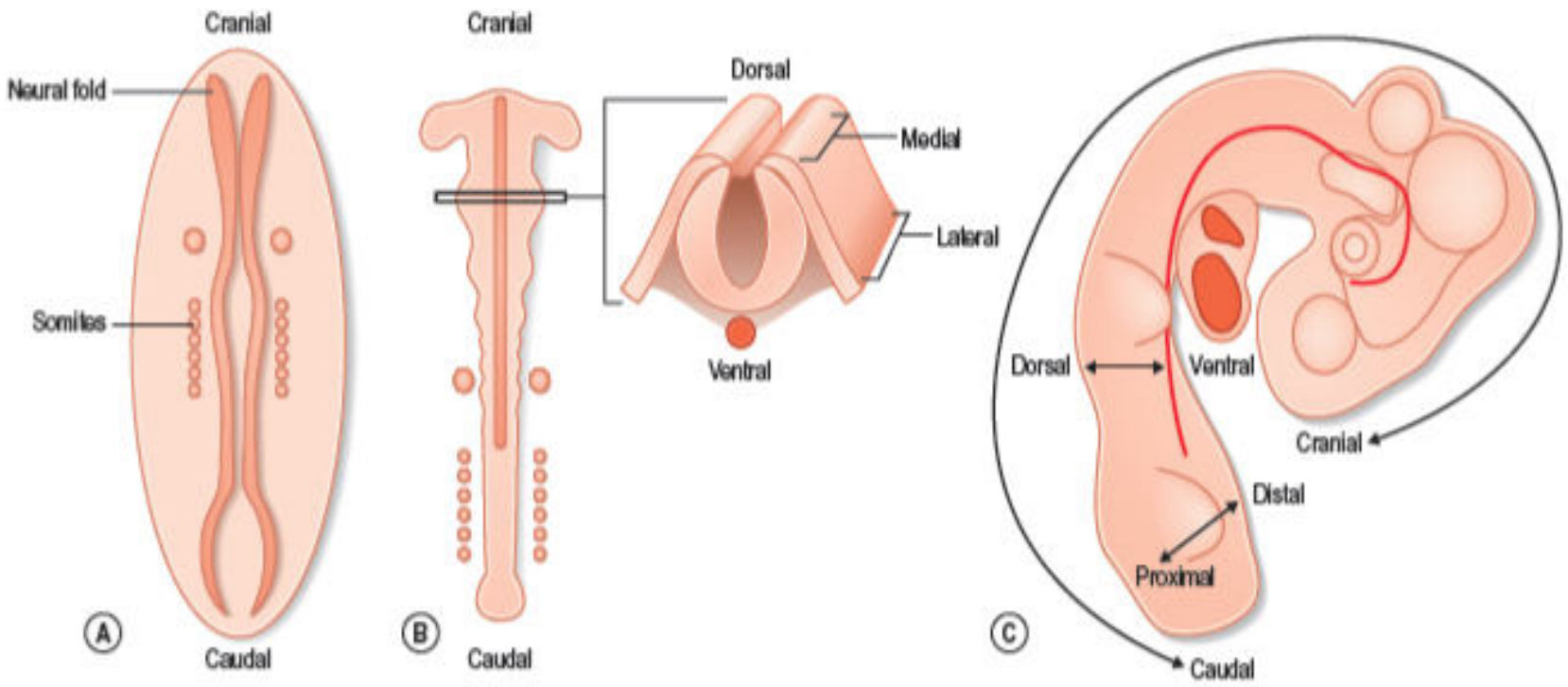
- Secondary palate – formed from 2 outgrowths from maxillary prominences – palatine shelves
- Fuse in midline at 7th week
- Incisive foramen – midline landmark between primary and secondary palate

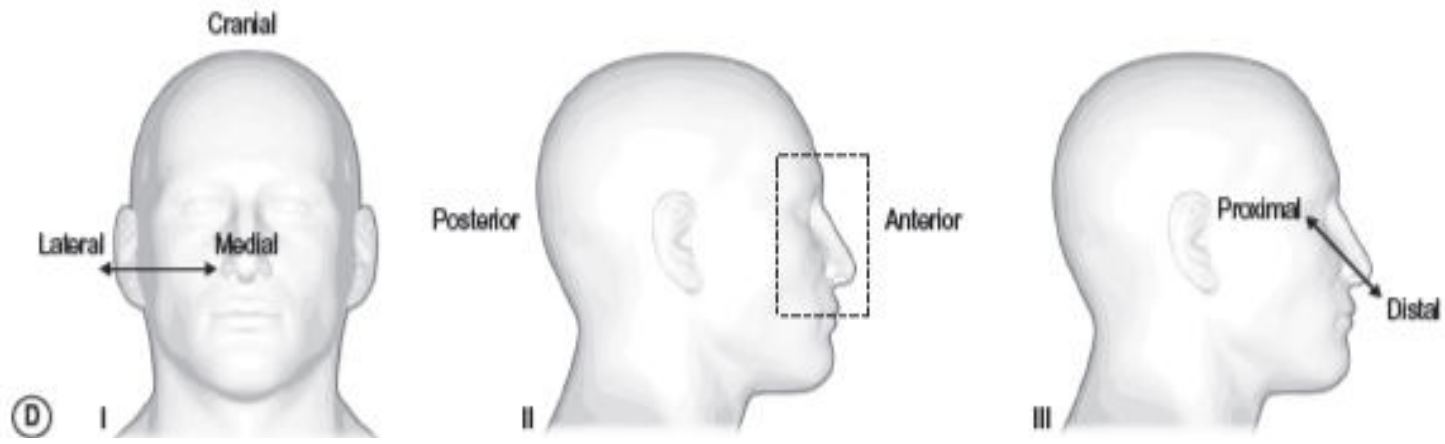


What is unique about craniofacial development?

- Dual origin
- Tissue interactions
- Elaborately choreographed morphogenic movements

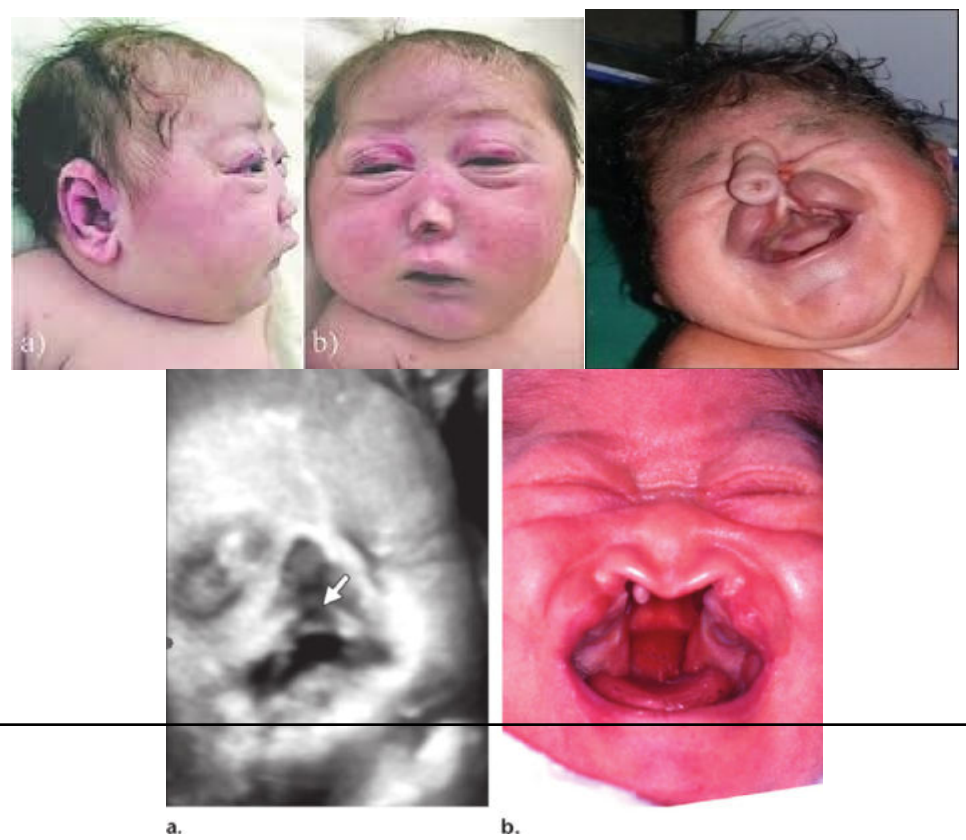


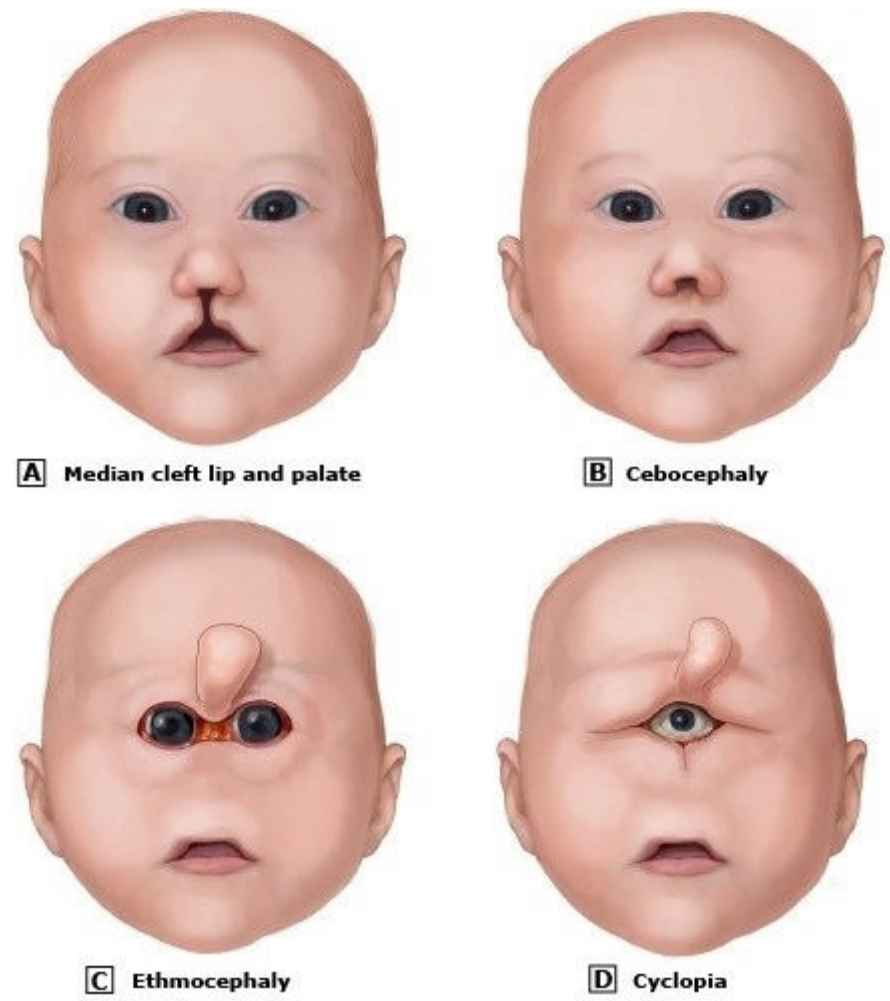




Cyclopia

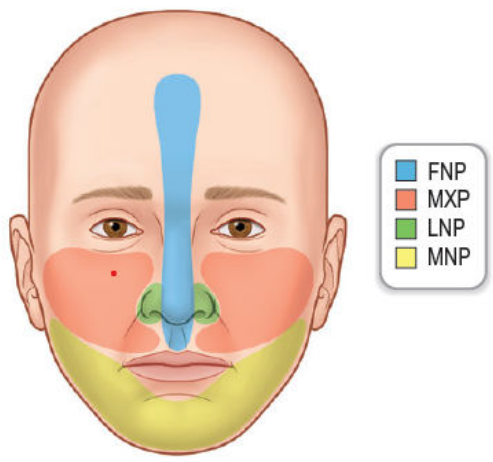
- Part of holoprosencephaly
- Holoprosencephaly also associated with-

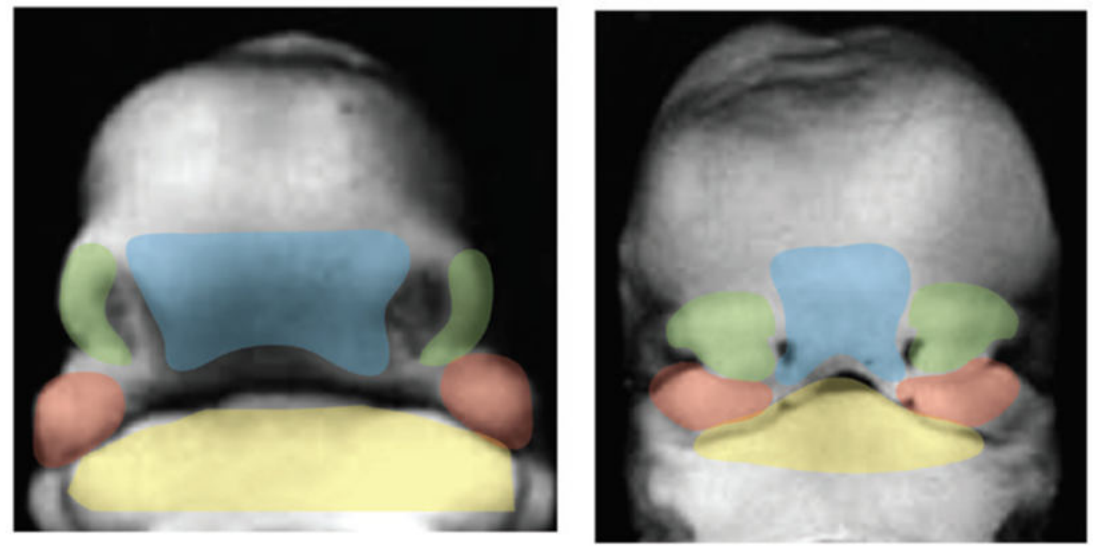




Establishment and fusion of the facial prominences

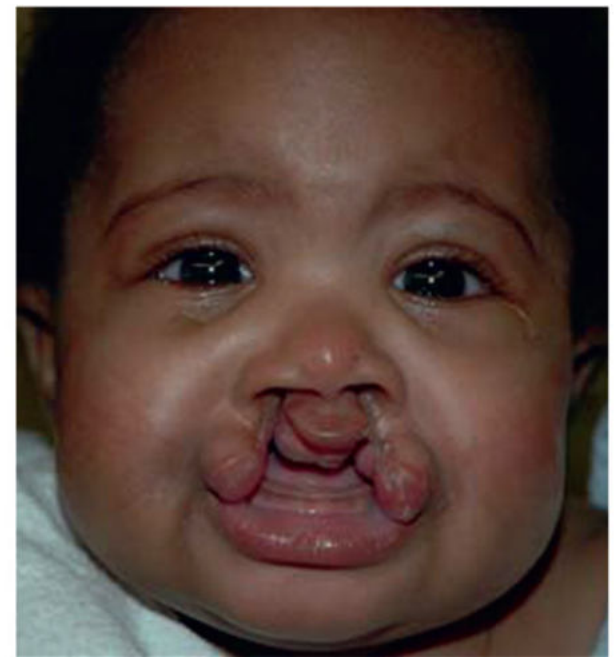
- basic morphology of the face is established between the 4th and 10th weeks
- midline frontonasal prominence,
- 3 paired prominences, the maxillary, lateral nasal, and mandibular prominences





The frontonasal prominence

- forehead, midline of the nose, the philtrum, the middle portion of the upper lip, and the primary palate.
- b/l cleft lip

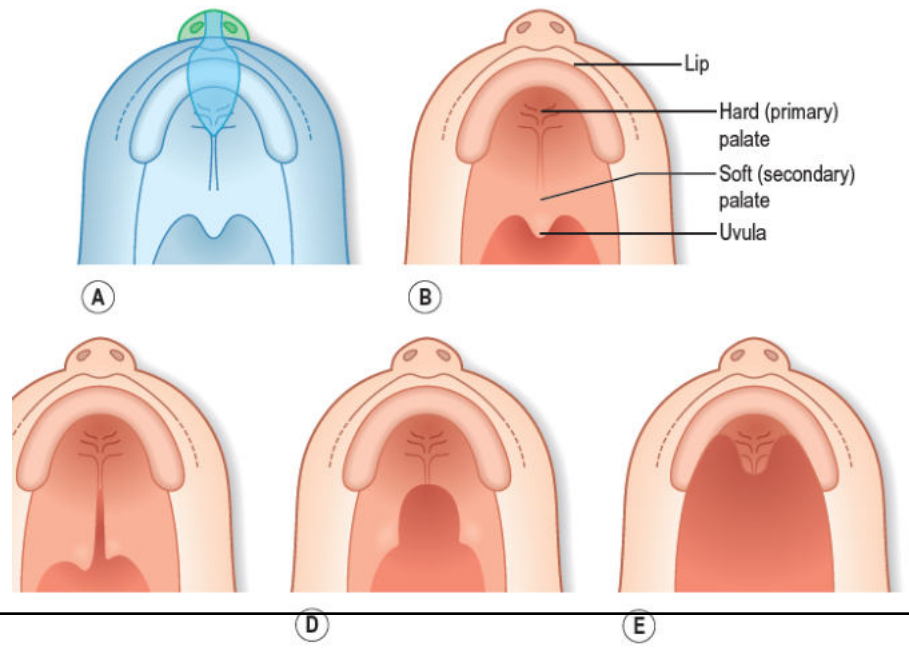


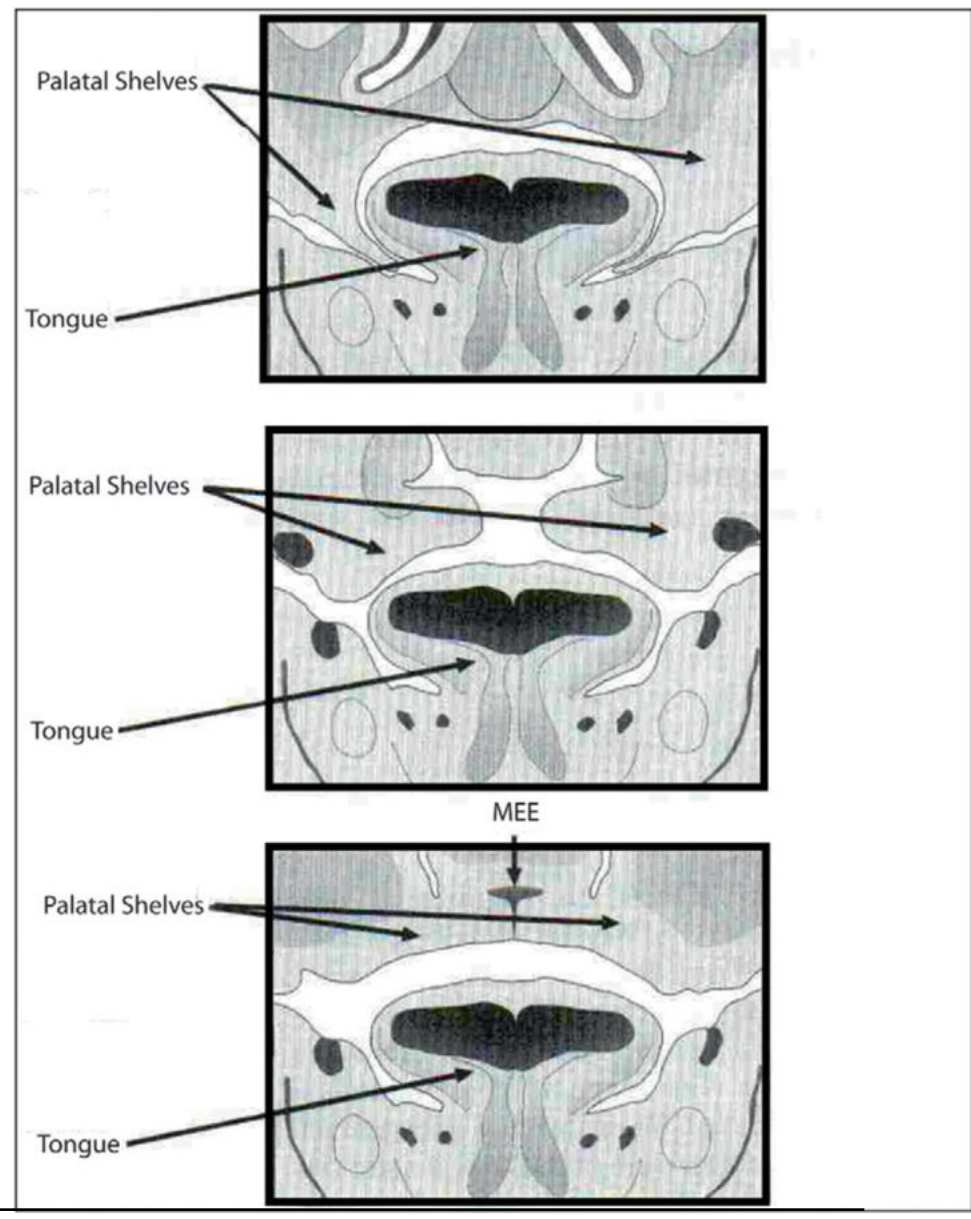
The lateral nasal prominences

- alae of the nose
- failure in the fusion between the lateral nasal prominences and either the frontonasal or the maxillary processes

The maxillary prominences

- upper jaw and the sides of the face
- the sides of the upper lip
- the secondary palate.

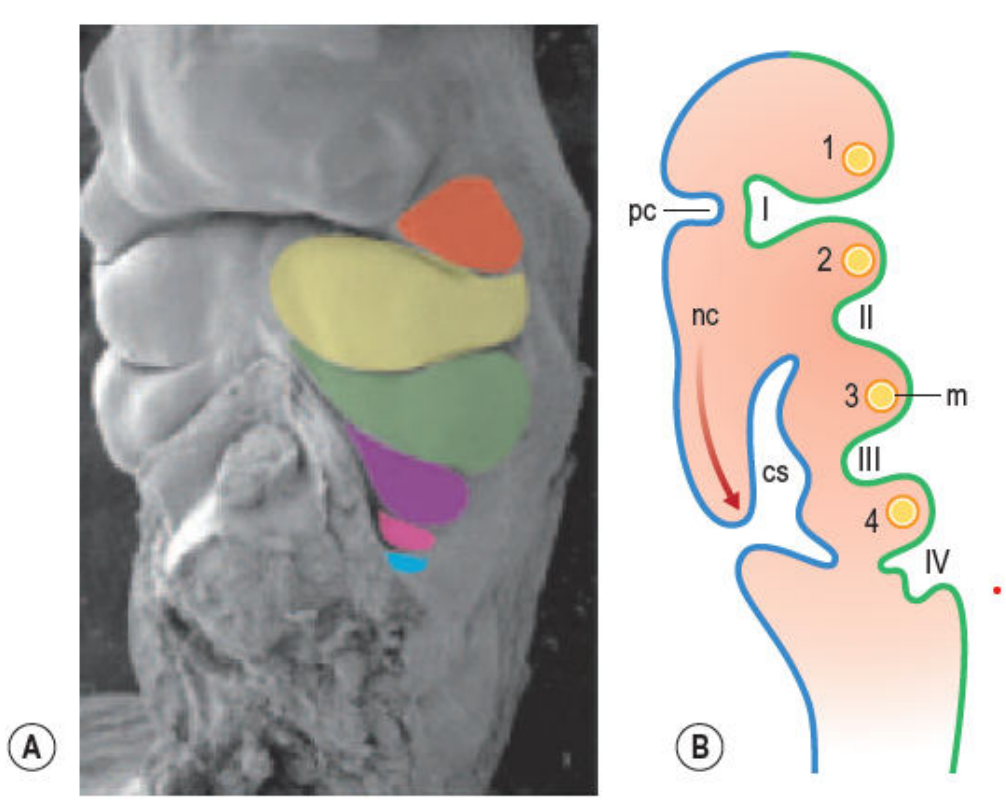


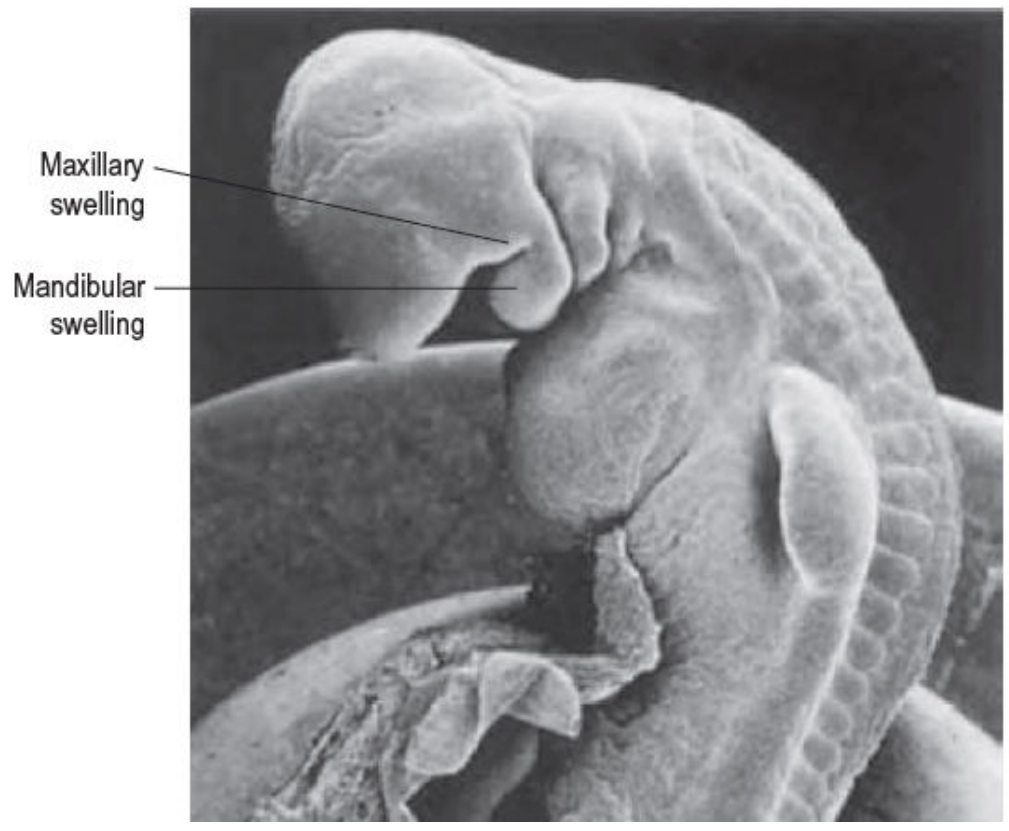


The mandibular prominences

- Lower jaw and lip
- Very rare
- Wide array of phenotypes

Pharyngeal arches





Arch	Skeletal element	Musculature	Nerve	Artery
1st (mandibular and maxillary)	Incus, malleus, zygomatic, squamous. Part of the temporal, mandible and maxilla	Muscles of mastication	Trigeminal	Maxillary artery
2nd (hyoid)	Stapes, styloid process of temporal bone, stylohyoid ligament. Lesser horn and body of hyoid bone	Muscles of facial expression	Facial	Stapedial artery
3rd	Greater horns and lower body of hyoid	Muscles of the stylopharyngeus (throat)	Glossopharyngeal	Common carotid/internal carotid
4th and 6th	Cartilages of the larynx	Muscles of pharynx constriction, muscles of phonation, palatoglossus (tongue), muscles of upper esophagus	Vagus	Arch of aorta, right subclavian artery, original sprouts of pulmonary artery, ductus arteriosus, roots of pulmonary arteries

ETIOPATHOGENESIS

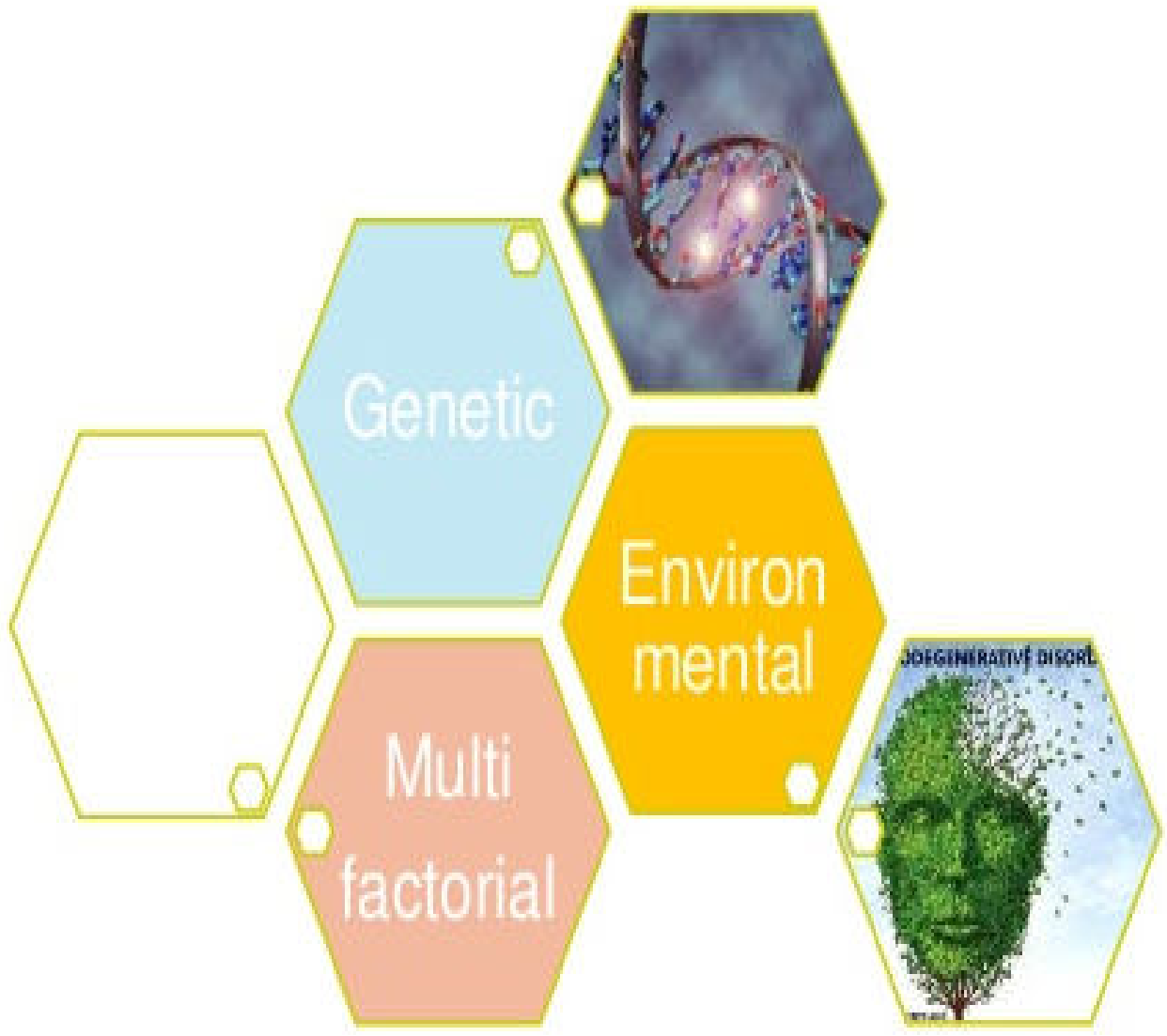
Early Chinese

- Eating rabbit "hare lip"
- Bad karma or wrong doings

Philippines

- Force to the fetal face

Familial or "In the blood"



ETIOPATHOGENESIS

1] Genetic

They can be classified in to 4 groups

1. Chromosomal
2. Single gene
3. Multifactorial
4. Mitochondrial

ETIOPATHOGENESIS

2] Multifactorial because:

1) Chances increases if more than one family member if affected
2) More the severity, greater the chances of recurrence in sibling
3) Higher risk if affected individual is of less affected sex
4) Risk decreases in remotely related individuals
5) Consanguinity increases the rate because of sharing of genes

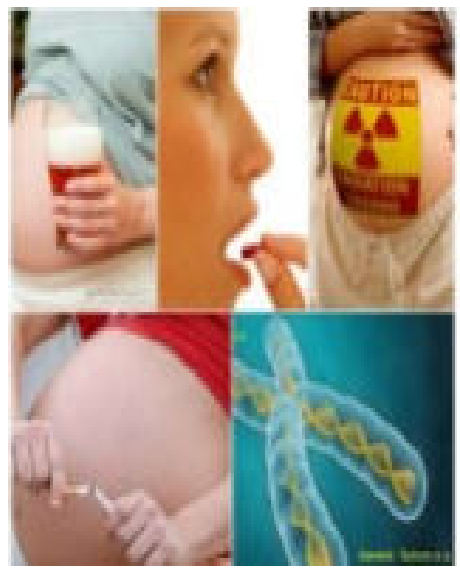
Etiopathogenesis

2 unaffected parents with 1 child affected
- Risk for future children:
- 4.4% for CL+/-palate
- 2.5% for CP only

1 parent affected
- Risk for future children
- 3.2% for CL+/-palate
- 6.8% for CP only

1 parent affected with 1 child affected
- Risk for future children
- 15.8% for CL+/-palate
- 14.9% for CP only

3. Environmental factors



Maternal Smoking or tobacco exposure
Viral infections
Poor nutrition
Drugs
Teratogens- Rubella, cortisone/steroids/ mercaptopurine
Methotrexate, Valium, Dilantin

PREDISPOSING FACTORS

- High maternal age
- Diabetes
- Toxemia
- Reduced blood supply
- Folic acid deficiency
- Racial – mongoloids
- Radiations

Davis and Ritchie's classification (1922)

Group I: Prealveolar process cleft (clefts affecting the lip)

1. Unilateral (right/left: complete/incomplete)
2. Bilateral (right: complete/incomplete; left: complete/incomplete)
3. Median (complete/incomplete)

Group II: Postalveolar process cleft (clefts affecting the palate)

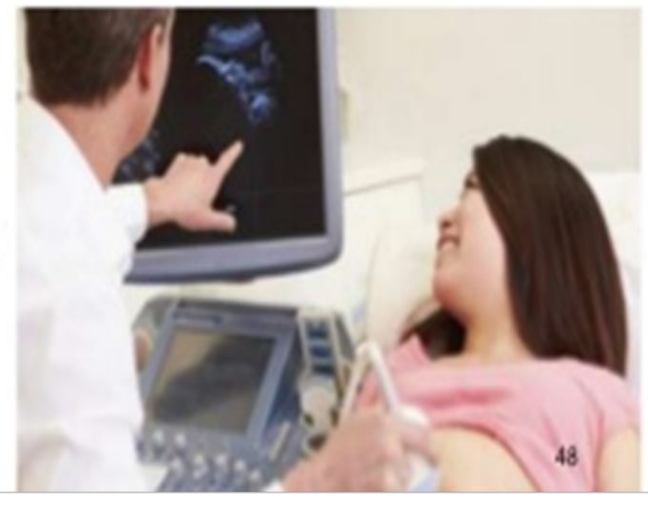
1. Soft palate
2. Hard palate

Group III: Alveolar process cleft (any cleft involving the alveolar process)

1. Unilateral (right/left: complete/incomplete)
2. Bilateral (right: complete/incomplete; left: complete/incomplete)
3. Median (complete/incomplete)

Diagnosis

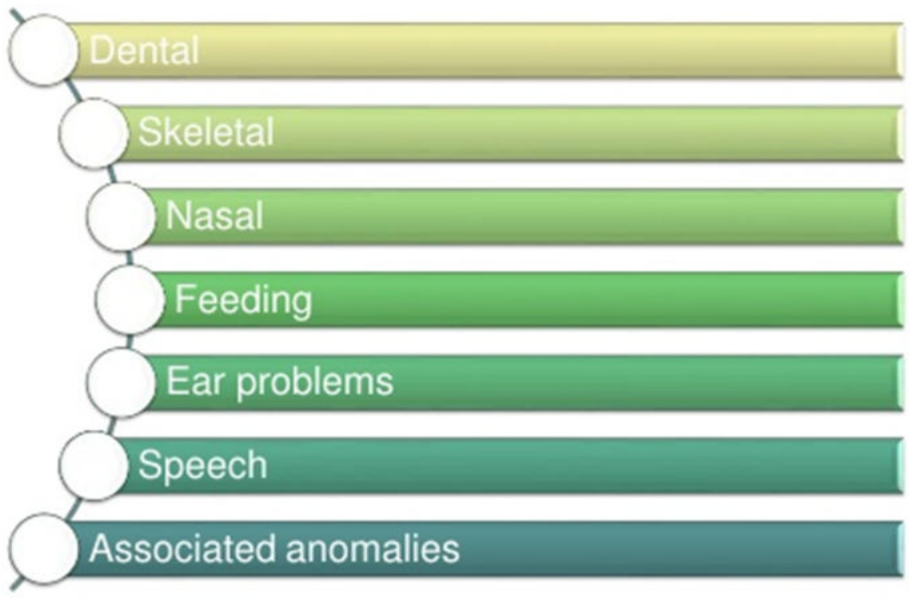
- Prenatal ultrasound – 2D or 3D
- Prenatal counselling
- 22% to 33% rates for detecting facial clefts
- 73% - fetal cleft lip
- 1.4% - isolated cleft palate
- Color Doppler ultrasonography can also be used



USG

- Non-invasive diagnostic tool
- Confirm fetal viability
- Determine gestational age
- Establish number of fetuses and their growth
- Check placental location
- Examine fetal anatomy for detecting malformations

Problems with clefts



DENTAL

- Tooth agenesis, hypodontia (most common)
- Supernumerary teeth (2nd most common)
- Enamel hypoplasia (CI)
- Crossbites
- Ectopic eruption, transposition
- Taurodontism, dilacerations

SKELETAL

- Maxillary deficiency
- Mandibular prognathism
- Class III malocclusion
- Concave profile

Feeding problems

Oronasal fistula
Nasal regurgitation
Bottle cup, spoon feeding
30-45 deg angle to aid swallowing

Syndromes

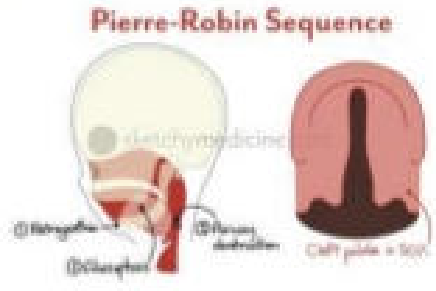
- Around 400 syndromes
- **Chromosomal anomalies**
  - Trisomy 13 (Patau)
  - Trisomy 18 (Edward)
  - Trisomy 21 (Down's)
  - Velocardiofacial syndrome (22q11 deletion)

➢ **Inherited syndrome**

- Sticklers (Autosomal dominant)
- Treacher Collins (AD)
- Van der Woude (AD)

➢ **Non-inherited syndrome**

- Pierre Robin Syndrome – triad of cleft palate, glossoptosis, retrognathia
- Goldenhar syndrome



Teratogenic syndrome

- Fetal alcohol syndrome
- Fetal phenytoin syndrome
- Fetal valproate syndrome

Management by Multidisciplinary approach

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Cheiloplasty

- Goal: improve facial aesthetics by restoring nasal and lip contour
- Timing: 3 to 6 months
- **Millards "RULE OF TEN"**

  [term coined by Wilhelmmesen and Musgrave in 1969]
  - 10 weeks (age)
  - 10 pounds (weight)
  - 10 gm/dl (Hb)

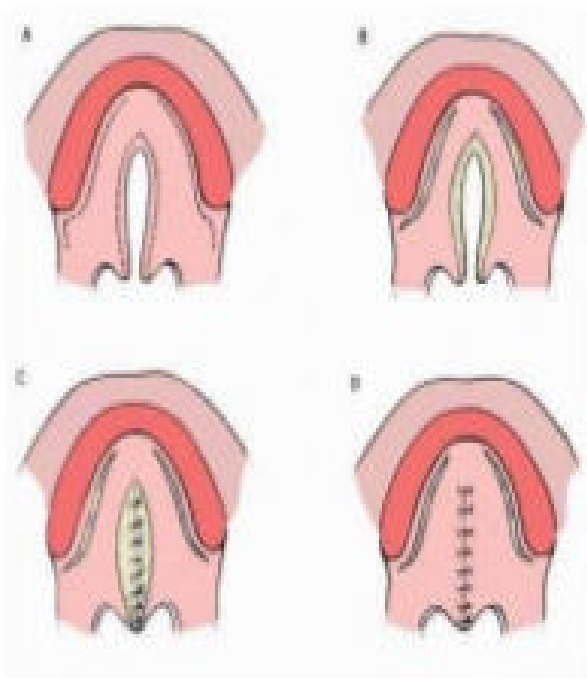
Cleft palate surgery

Von langenback

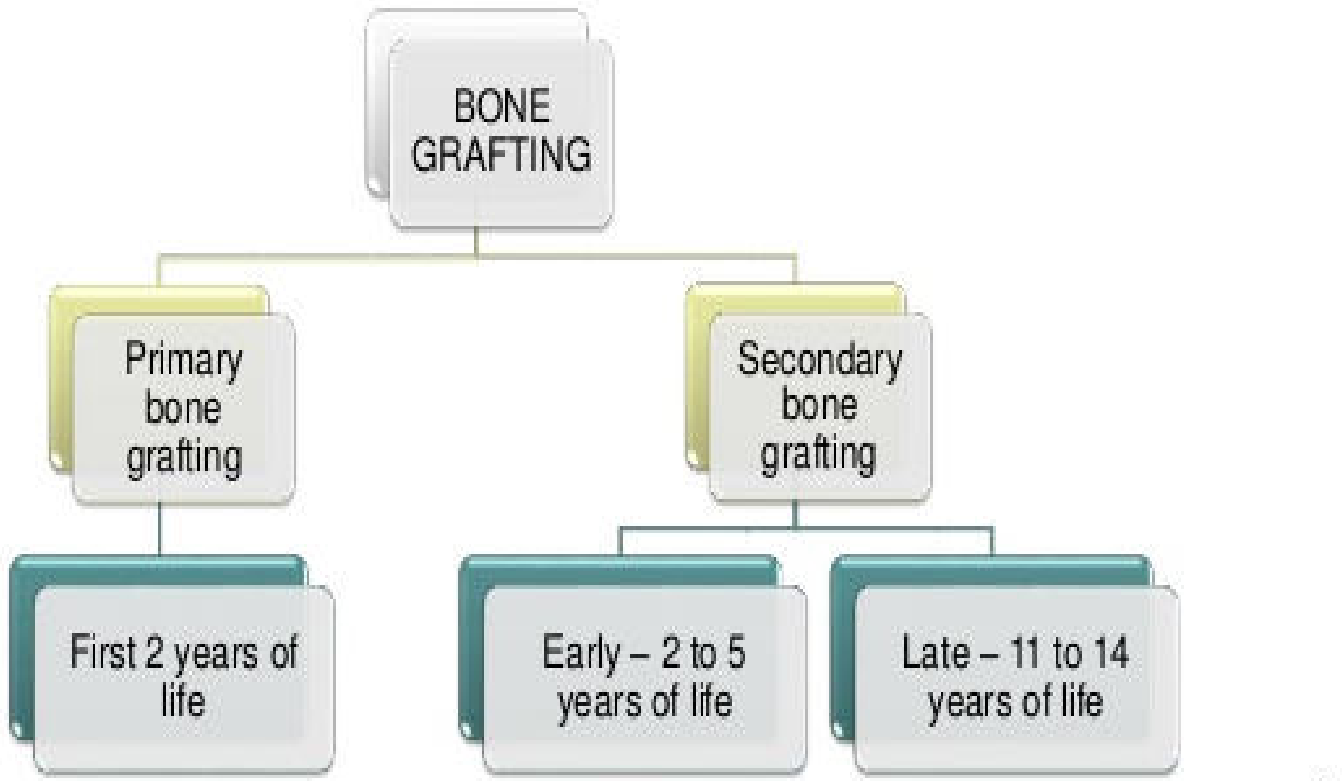
VY palatoplasty by Veau

Furlows palatoplasty

Wardil-kilner Pushback palatoplasty



Bone grafting



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Recent advances

Fetal endoscopic approach

Fetal surgery in intrauterine life (less than 20 weeks)

Open fetal surgery

Conclusion

- Second most common congenital anomaly
- Embryogenesis and etiology to be kept in mind
- Team approach
- Research to be aimed at Epigenetic modification

Thank You