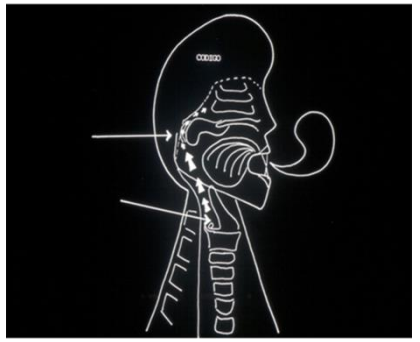
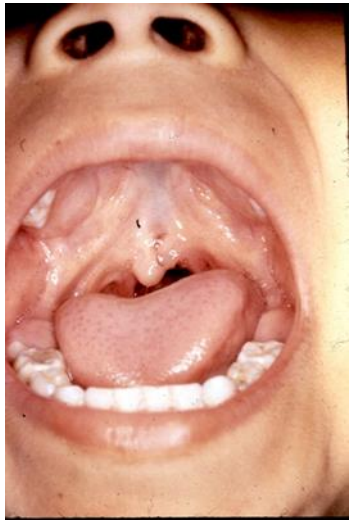
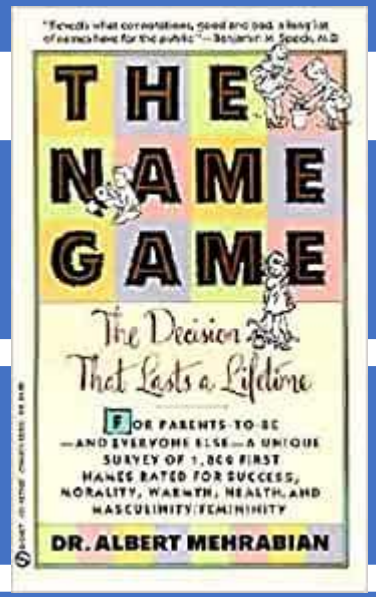


Common Craniofacial Syndromes



With Speech, Language, Swallowing, Voice disorders

22q11.2 Microdeletion Syndrome



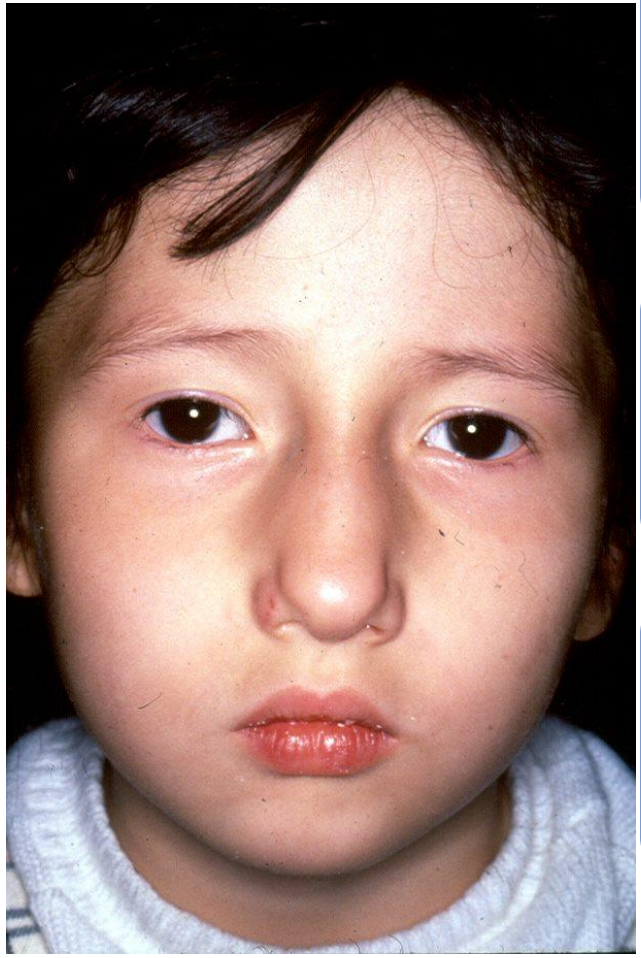
Shprintzen Syndrome

DiGeorge Syndrome

Conotruncal Anomaly Face Syndrome

Etc.

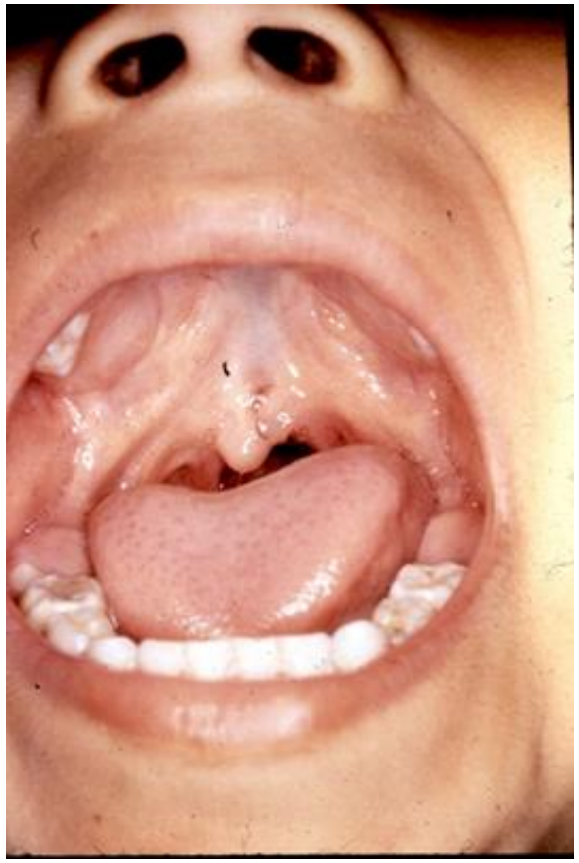
22q11.2 Microdeletion Syndrome



Most cases de novo Mutations

Autosomal Dominant

OSCSP-VPI-CA



DiGeorge

Sequence

Not Syndrome

Defective develolpment of Neural Crest Cells (NCC)

NCC colonize pharyngeal arches

lower jaw, neck and heart tissues.

DiGeorge

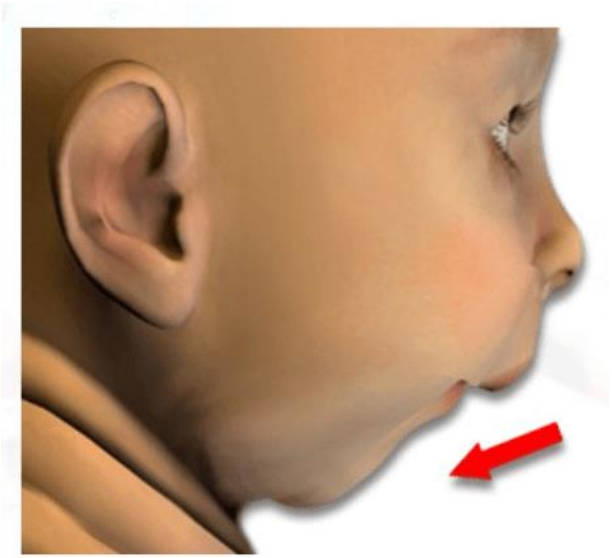
DiGeorge sequence with 22q11.2 microdeletion

DiGeorge sequence WO 22q11.2 microdeletion

22q11.2 microdeletion WO DiGeorge Sequence

Pierre Robin

- Sequence
- Micrognathia
- Glossoptosis
- Total or subtotal cleft of secondary palate



Pierre Robin

Isolated sequence (Non – Syndromic)

PRS in a syndrome (Syndromic)

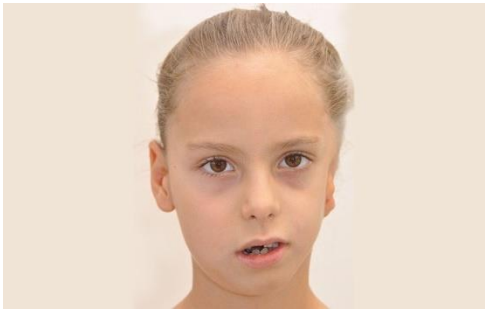
Hemifacial Microsomia

Autosomal Dominant

Unilateral

Disturbance of the blood supply to the first and second branchial arches in the first 6 to 8 weeks of pregnancy.

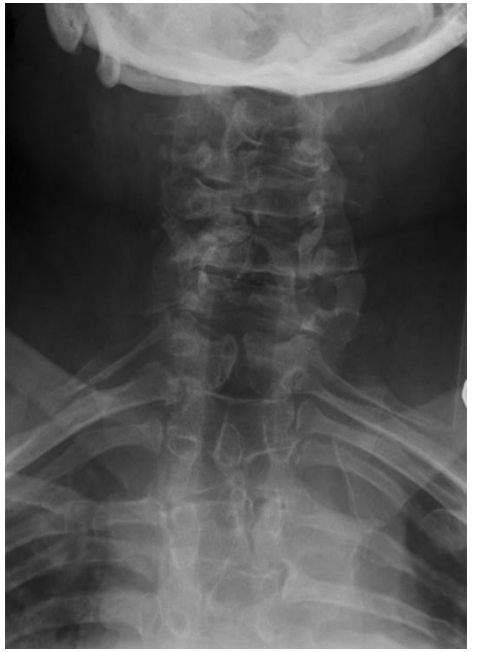
Unilat. Microtia; Unilat. Micrognathia (Possible PRS); Facial Palsy (Freq. buccal and mandibular branches)



Goldenhar Syndrome

Autosomal Dominant

- Similar to HM but bilateral
- Cervical vertebrae fusion
- Autosomal Dominant



Oculo – Auriculo – Vertebral Spectrum

Features of Goldenhar and Hemifacial Microsomia

Inheritance P. (?)

Craniosynostosis

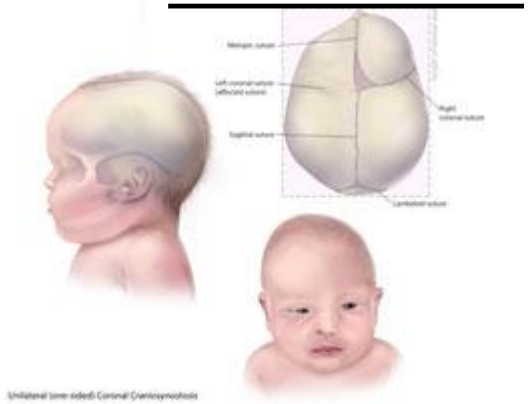
Premature fusion of cranial sutures

Metopic craniosynostosis (malformation)

Crouzon Syndrome (Aut. Dom.)

Apert Syndrome (Craniosynostosis + syndactilia) (Aut.Dom.)

Fontanelles

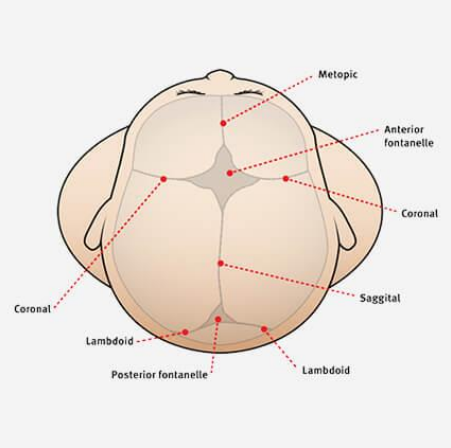


Craniosynostosis

Hyponasality

Cognitive – Language issues

Fontanelles



Treacher Collins Syndrome

- Most cases are de novo mutations
- Autosomal Dominant
- Underdevelopment of facial bones
- Microtia - Atresia (uni o bi)
- Conductive Hearing Loss
- PRS
- Coloboma

TCS

- Multiple Craniofacial Surgeries
- Palatoplasty
- VPI surgery complicated by OSA
- BAHA

Moebius

Congenital bilateral paralysis facial and abducens.

Feeding problems

Dysphagia

Sialorrhea

Strabismus

Lack of facial expression.

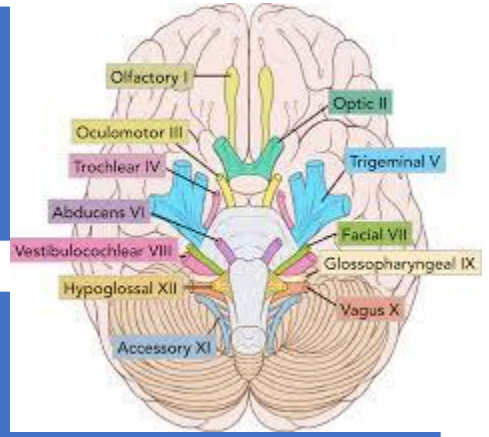
Moebius

- Severe flaccid dysarthria
- Hypernasality
- Facial reanimation by gracilis muscle free transfer.
- Palatoplasty if Palatal paresis (Additional X palsy)
- Speech Treatment by compensation
- Excision of salivary glands or Botox

video

video

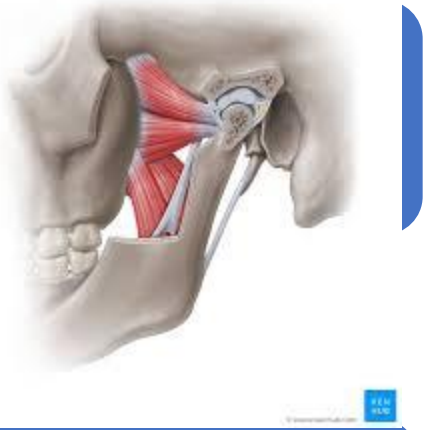
Moebius



V and XII not affected

X may be affected (not frequently)

Moebius



Lingual muscles w normal mobility and strength

Pterygoid muscles (Medial and lateral) with normal mobility and strength