# The ENT Management of Syndromic Children

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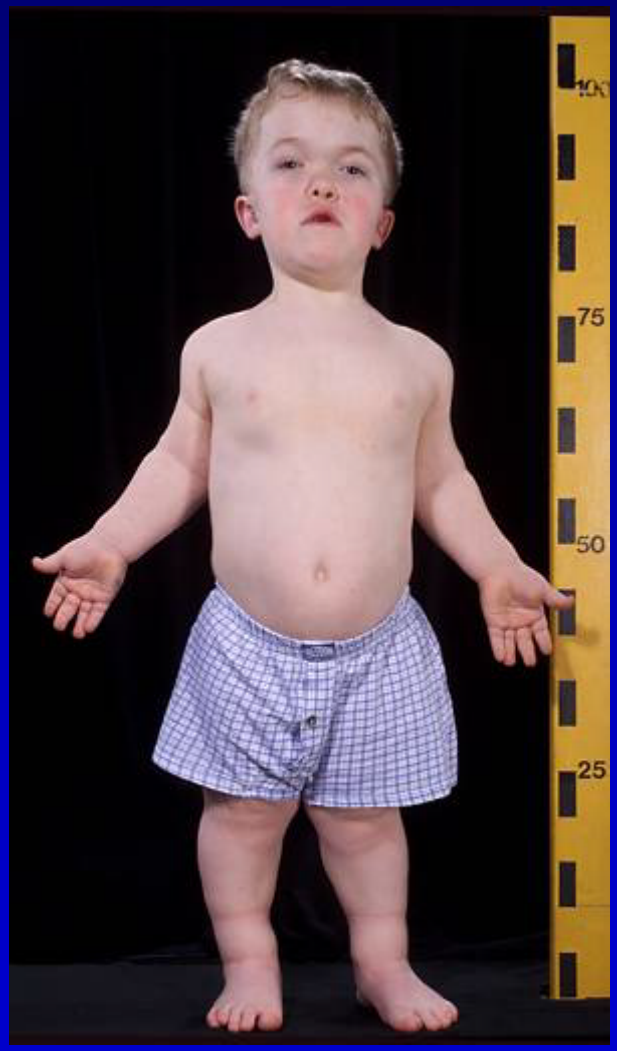
‘You don’t need to know all about the syndrome to treat the child’

# Definitions

- Syndrome = collection of traits / health problems / birth defects which usually have a single underlying cause e.g. Down
- Sequence = all features results from a single abnormality e.g. Pierre Robin
- Association = recognisable pattern without a single cause e.g. CHARGE

# Achondroplasia

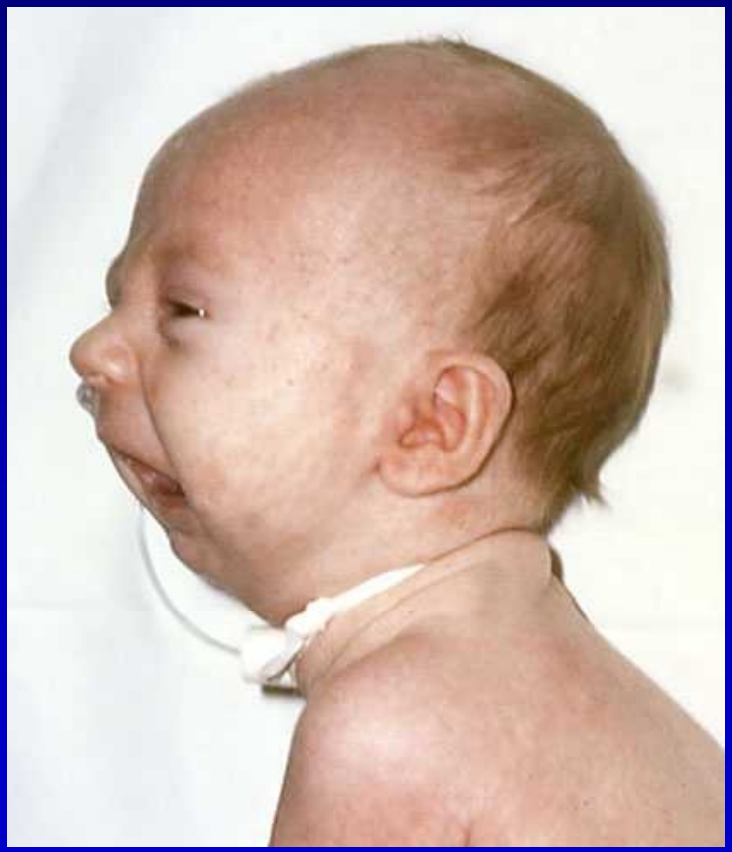



- Short stature
- Large head
- Short extremities
- Mid face hypoplasia
- Frontal / mandibular prominence
- Cervical instability

# Achondroplasia - ENT

- OSA
  - Beware cervical instability
- Mixed hearing loss
- Fused ossicles

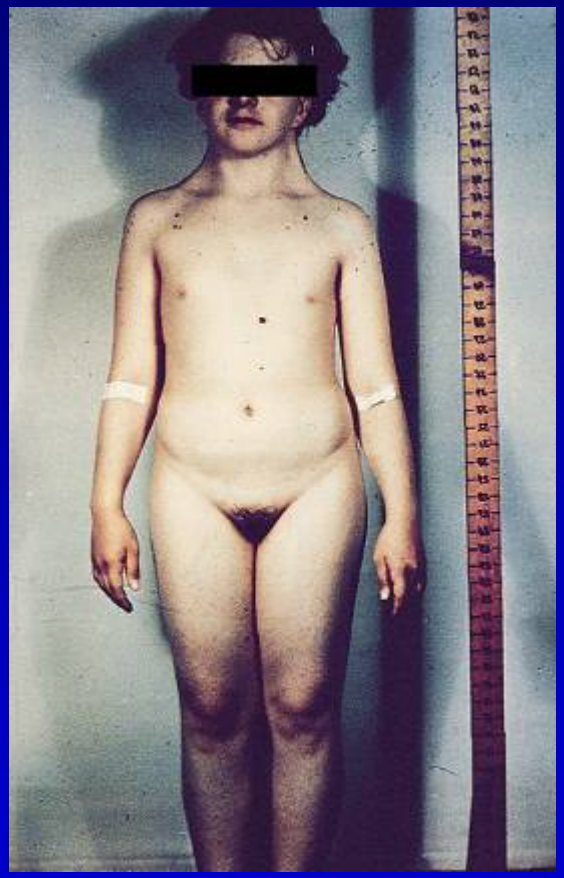
# Pierre Robin Sequence



- Small jaw
- Glossoptosis
- Cleft palate

Feeding and airway issues

# Turner's Syndrome - XO

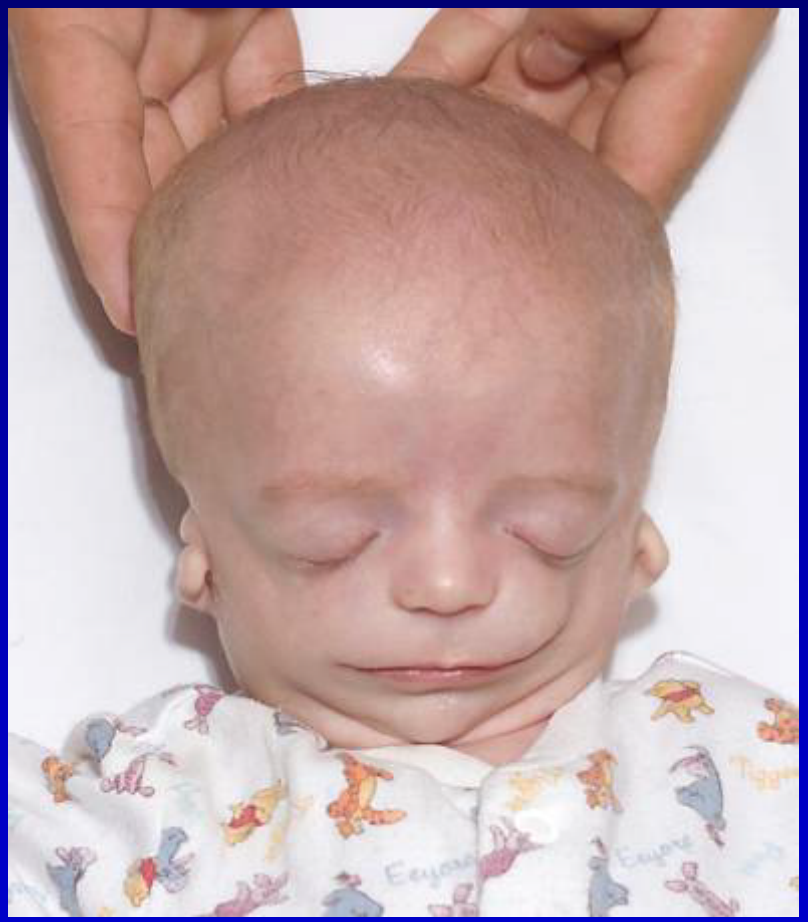


- Short stature
- Low hairline
- Down turned mouth
- Webbing neck/digits
- Widely spaced nipples
- Cubitus valgus
- Gonadal aplasia

# Turner's Syndrome - ENT

- Mixed hearing loss
- Ossicular deformities
- Micrognathia

# Treacher Collins Syndrome



- Mandibular / maxillary hypoplasia
- Microtia
- Down slanting palpebral fissures
- Cleft palate
- Choanal atresia

# Treacher Collins - ENT

- Airway obstruction / Difficult intubation
- Choanal atresia
- Conductive deafness
- Mastoid hypoplasia
- Absent parotid
- Absent / Small Sinuses

# Goldenhar's Syndrome and Associated Conditions

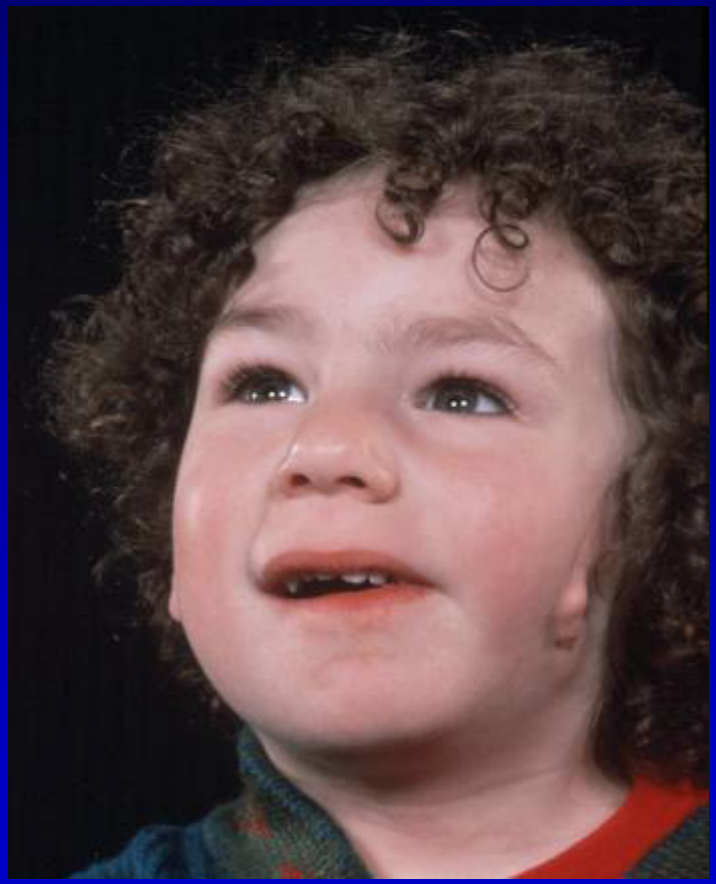
Unilateral Dysmorphogenesis in 1st and 2nd branchial arch development

- Hemifacial microsomia
- Oculoauricular vertebral dysplasia
- Branchio-oto-renal syndrome

# Goldenhar's Syndrome

- Facial asymmetry
  - Mandibular hypoplasia
  - Micrognathia
  - Macrostomia
- Microtia
- Epibulbar dermoid
- Associated with vertebral, renal and cardiac anomalies

# Hemifacial Microsomia



- Facial asymmetry
  - Mandibular hypoplasia
  - Micrognathia
  - Macrostomia
- Microtia

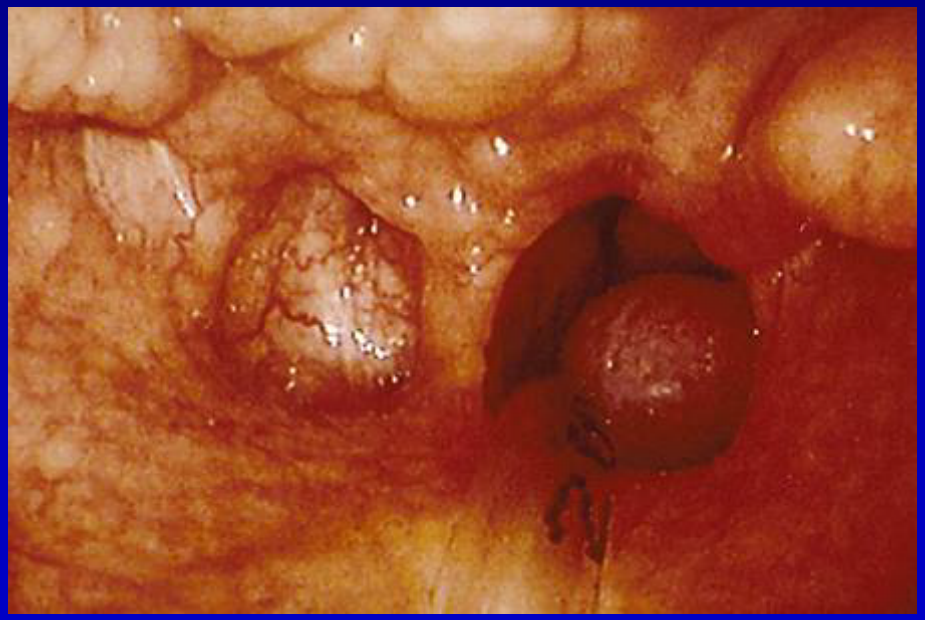
# Goldenhar's Syndrome / Hemifacial Microsomia - ENT

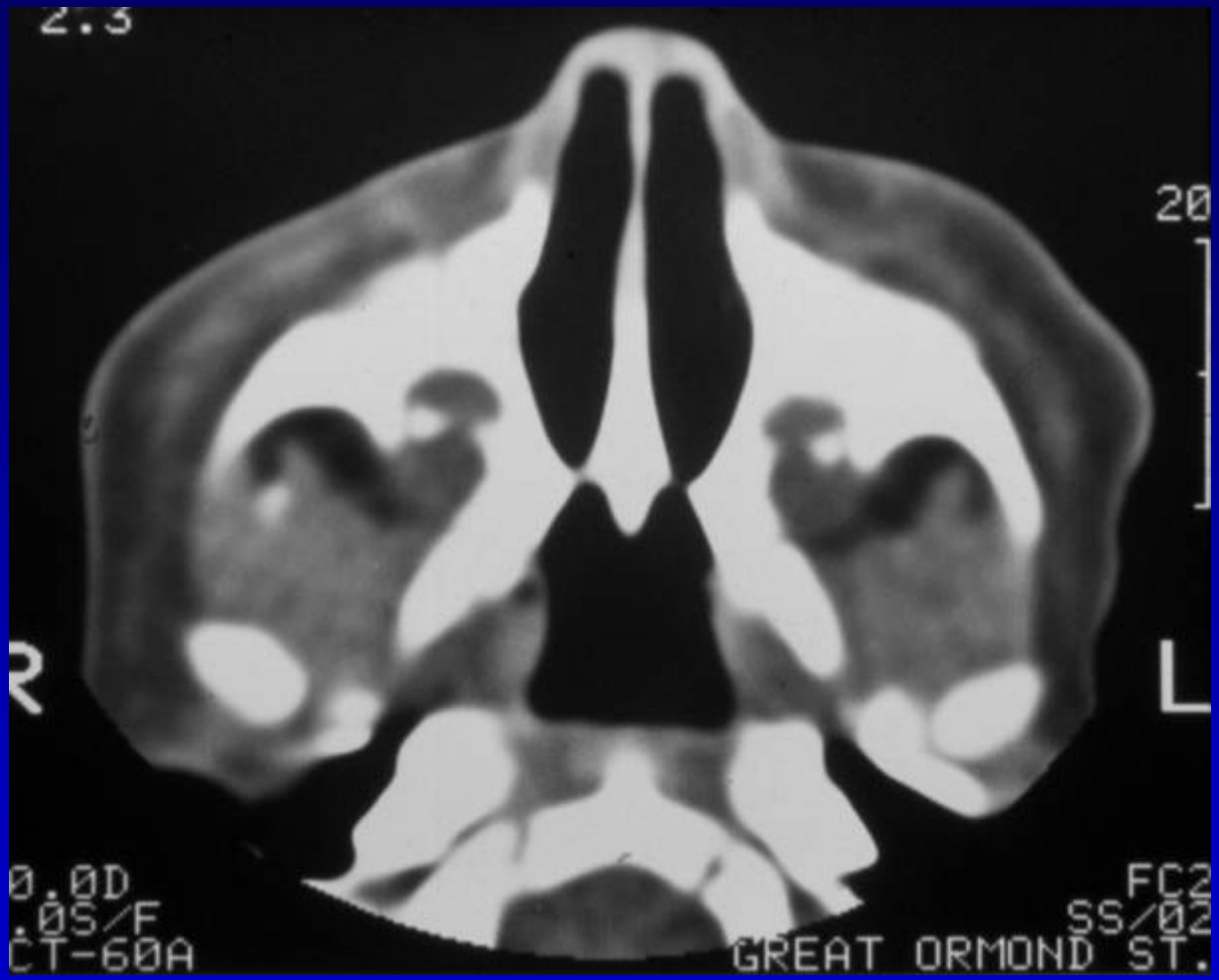
- Airway obstruction
- Facial nerve weakness
- Ossicular malformations
- Inner ear abnormalities
- Reduced / Absent parotid secretion
- Branchial cleft remnants in antero-lateral neck

# CHARGE Association

- Coloboma
- Heart defects
- Atresia choanae
- Retarded growth
- Genital hypoplasia (male)
- Ear anomalies (deafness, pinnae)

# Choanal Atresia

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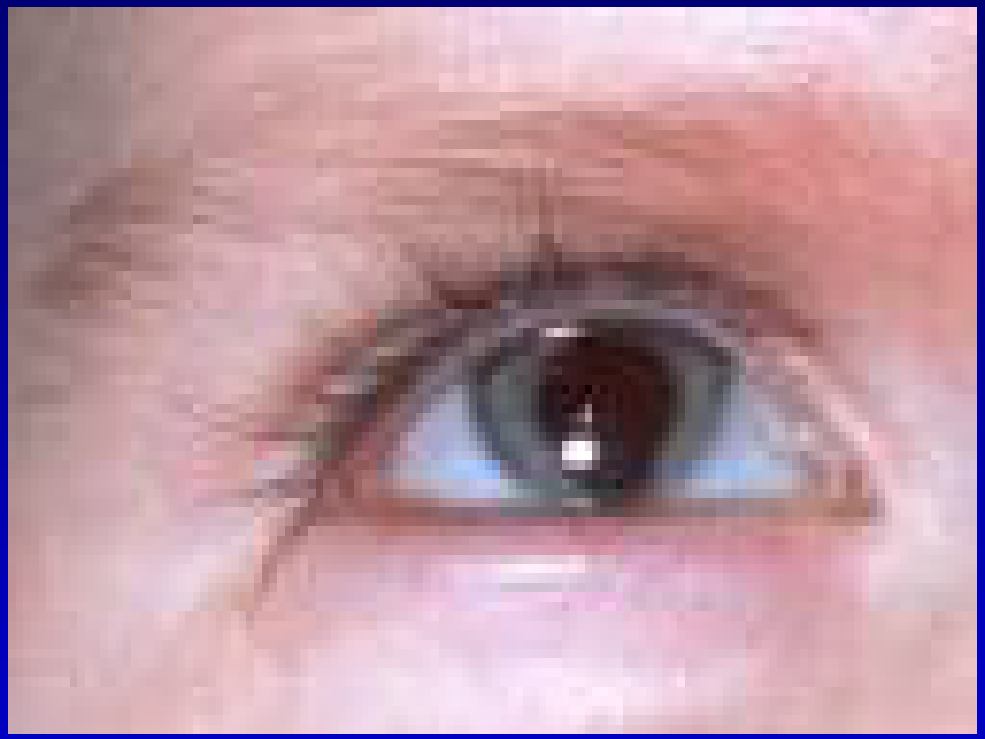
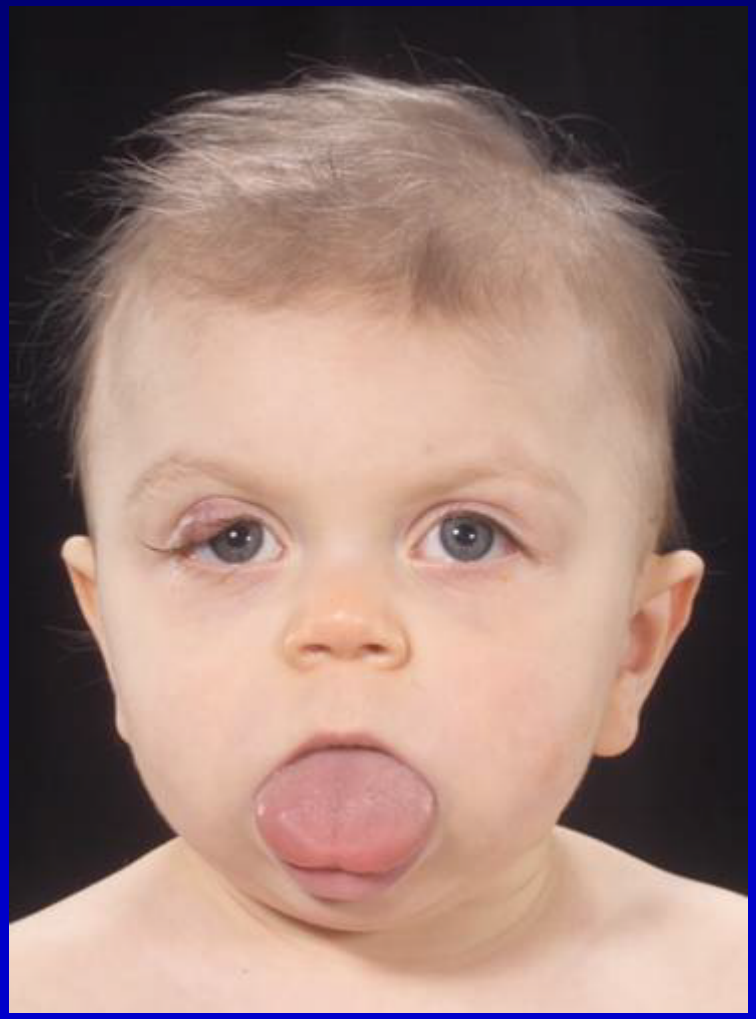


# Beckwith Weidemann Syndrome



- Generalised overgrowth
- Hemihypertrophy
- Macroglossia

# Beckwith - Wiedemann - ENT

- Airway obstruction
- Feeding difficulties

Consider anterior tongue reduction

# Mucopolysaccharidoses

- Genetic enzyme defect
- Glycosaminoglycans accumulate in lysosomes
- Coarse skin
- Skeletal dysplasia
- Hepatosplenomegaly
- Progressive condition
- Hunter's / Hurler's most common

# MPS - ENT

- OME - otorrhoea common post G's
  - MPS deposits in middle ear
- Ossicular abnormality in Hunter's
- SNHL
- Rhinosinusitis / Recurrent chest infections
  - poor mucociliary clearance
  - RS unresponsive to treatment

# MPS - ENT

- Airway Obstruction
  - large tongue
  - adenotonsillar hypertrophy (cervical instability)
  - high larynx - obligate nasal breathers
  - narrow trachea (weakened cartilage)
  - thick secretions
  - Progressive condition - 44% T&A pts need subsequent tracheostomy

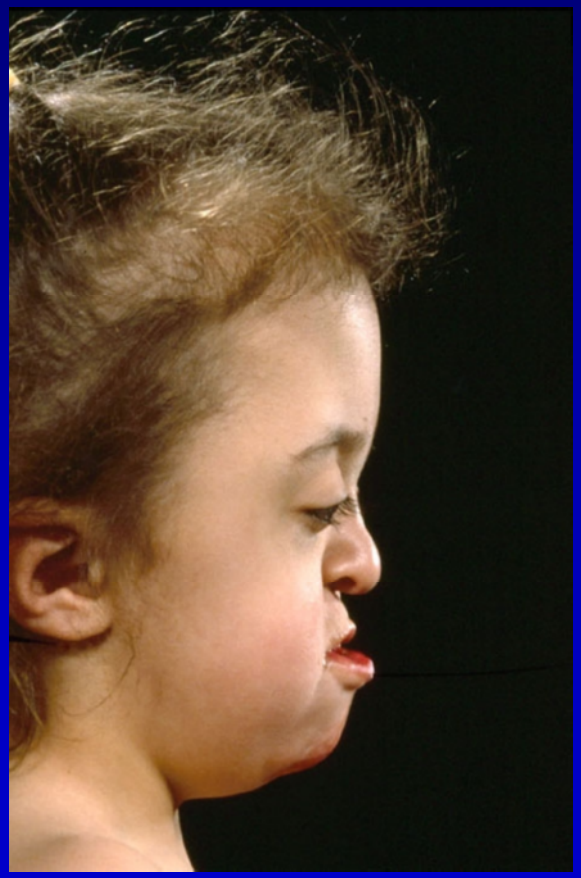
# Craniosynostoses

- Premature fusion of cranial vault/base sutures
- Midface hypoplasia
- High arched palate
- Exorbitism
- Raised ICP/papilloedema
- Skeletal abnormalities

# Craniosynostoses

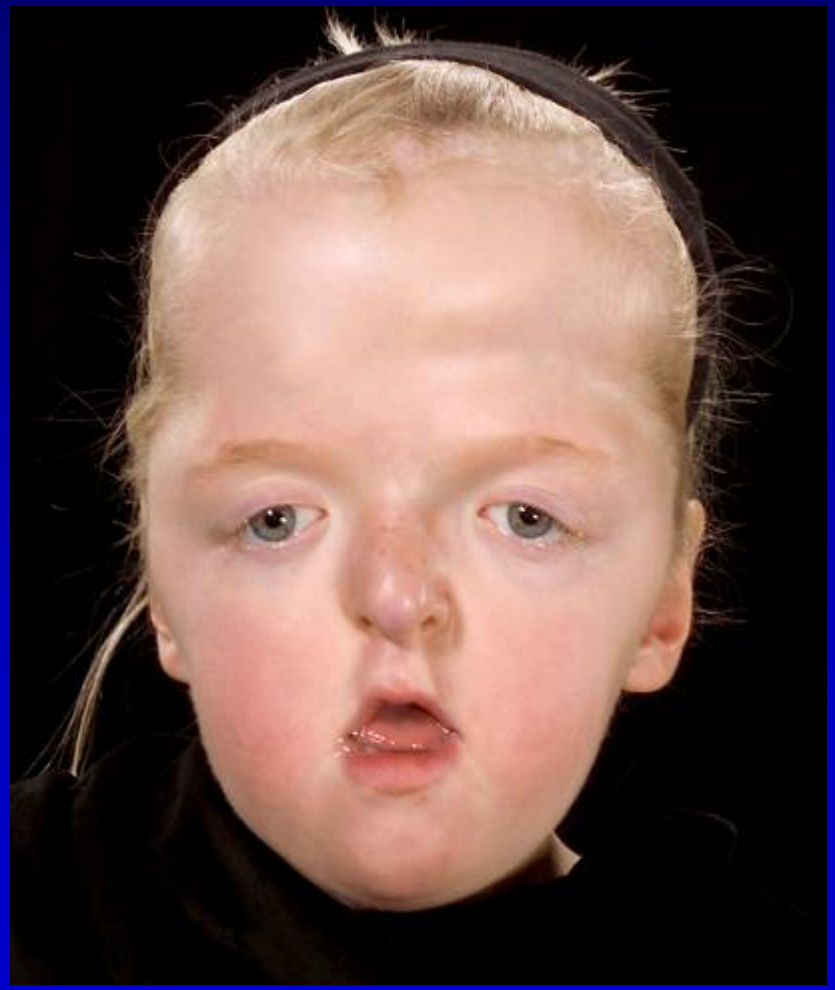
- Non-syndromic
  - 94% of total
  - isolated single suture (sagittal) synostosis most common
- Syndromic
  - fibroblast growth factor receptor mutations
  - Crouzon's, Apert's, Pfeiffer's, Saethre Chotzen

# Crouzon's Syndrome



- Occular proptosis
- Maxillary hypoplasia
- Class 3 malocclusion
- Hypertelorism
- Normal hands / feet
- Normal devt.

# Apert's Syndrome



- Tower shaped skull
- Mid facial retrusion
- Hypertelorism
- Complete syndactyly - hands and feet
- Acne
- Cleft palate

# Pfeiffer's Syndrome

- Mid face retrusion
- Broad short thumbs
- Large toes
- Down slanting eyes
- Low set ears
- Various phenotypes
  - Clover leaf most severe

# 22q11 deletions

- Various defects – ‘CATCH 22’
- Cardiac anomalies
- Abnormal facial features
- Thymic aplasia
- Cleft palate
- Hypocalcamia
- Groupings of defects called DiGeorge Syndrome, Velocardiofacial Syndrome

# Osteogenesis Imperfecta

- Autosomal dominant
- Blue sclera
- Long bone fractures
- Fusion ossicles
  - Role of stapedectomy ?

# Congenital Hearing Loss

- Waardenburg – heterochromia irides / white hair
- Pendred – thyroid abnormalities
- Alport – renal problems
- Usher – retinitis pigmentosa
- Jervel / Lange – Nielson – prolonged QT interval

# General Otological Management in Syndromic Children

- Mixed losses - early diagnosis
- Multidisciplinary approach
  - audiological physicians
  - speech/language therapist
- SNHL
  - hearing aids
  - cochlear implantation

# Otology - Continued

- Conductive hearing loss
  - BC aids (soft bands)
  - BAHA
  - Ossiculoplasty
- Microtia
  - BAAP
  - Auricular reconstruction

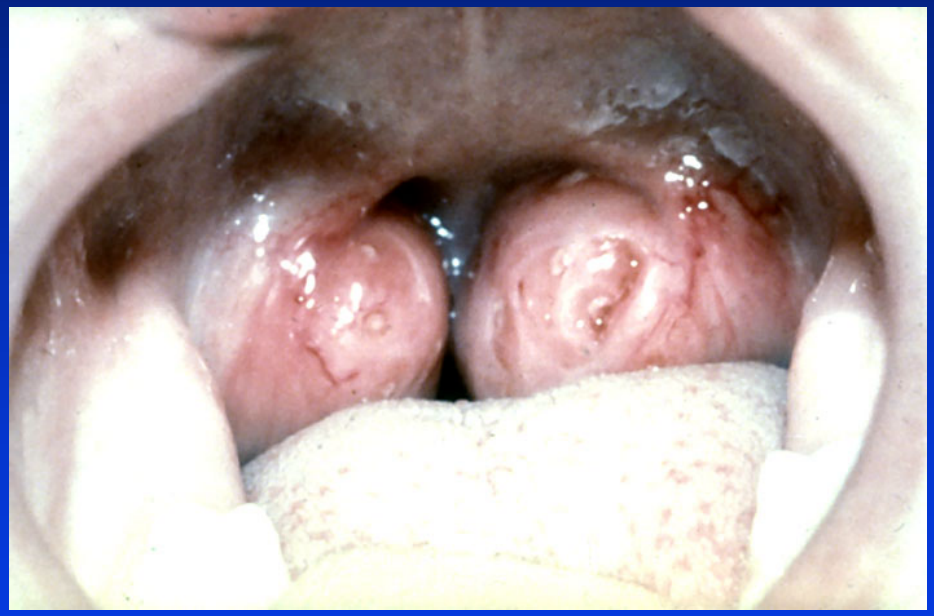
# General Rhinological Management

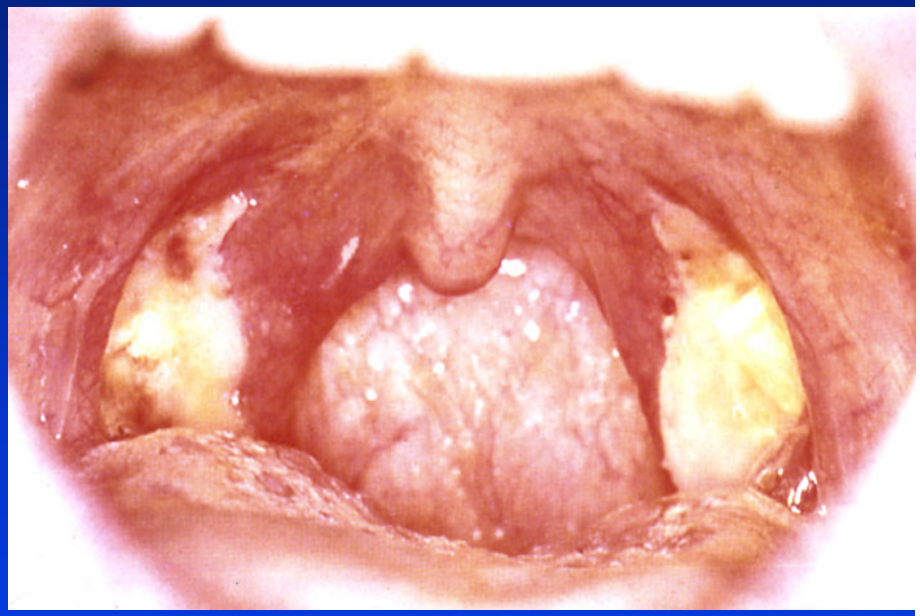
- History
  - ? allergy
- Maximal Medical Treatment
- CT scanning
  - ‘C’ shaped mucosal thickening in CF
- Role of FESS

# General Airway Management

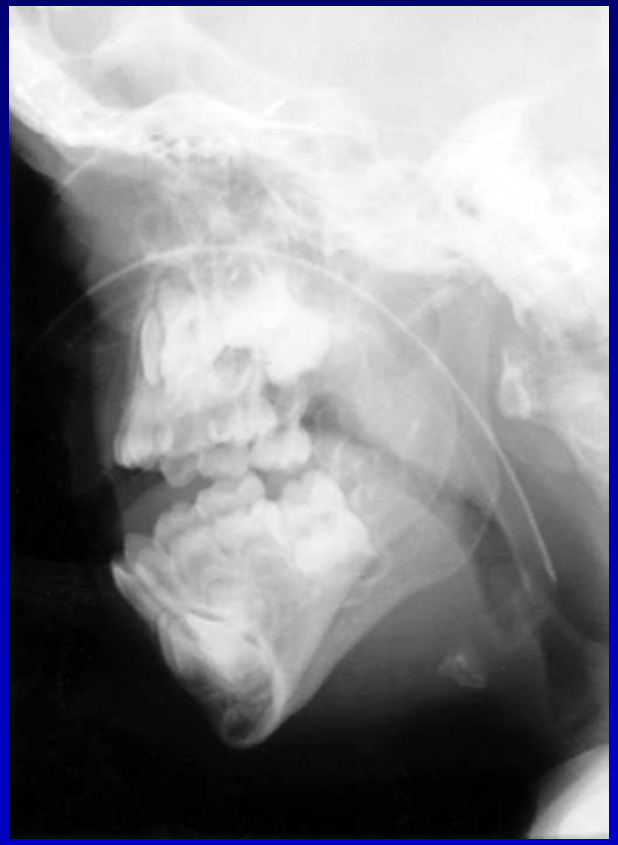
- Stridor vs. Stertor
- Exclude airway anomalies (MLB)
- Remove enlarged TsAs
- Nasopharyngeal tube
- CPAP
- Tracheostomy
- Osteotomies / midface advancement

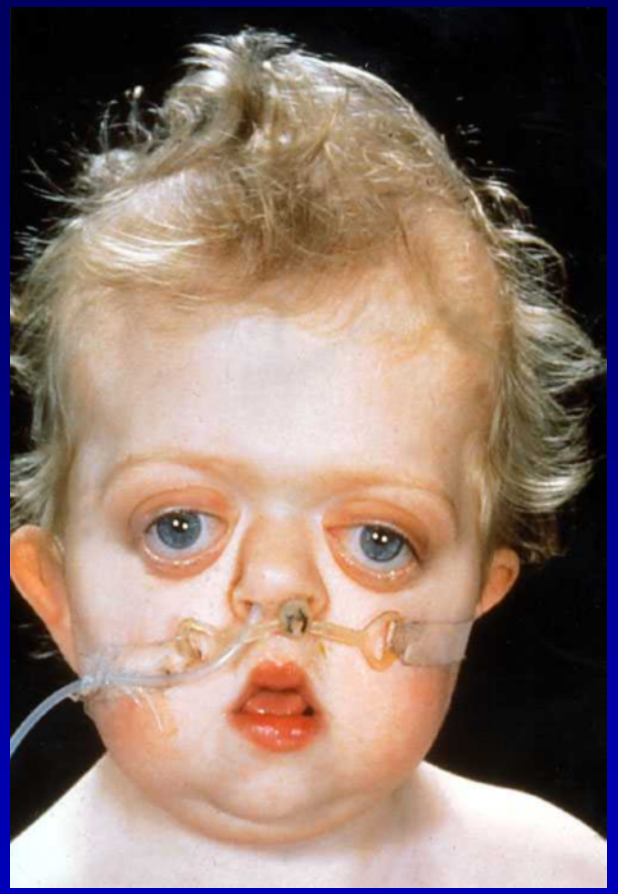
# Adenotonsillectomy

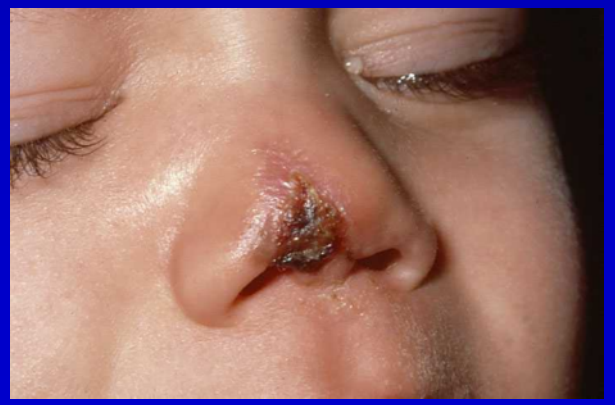




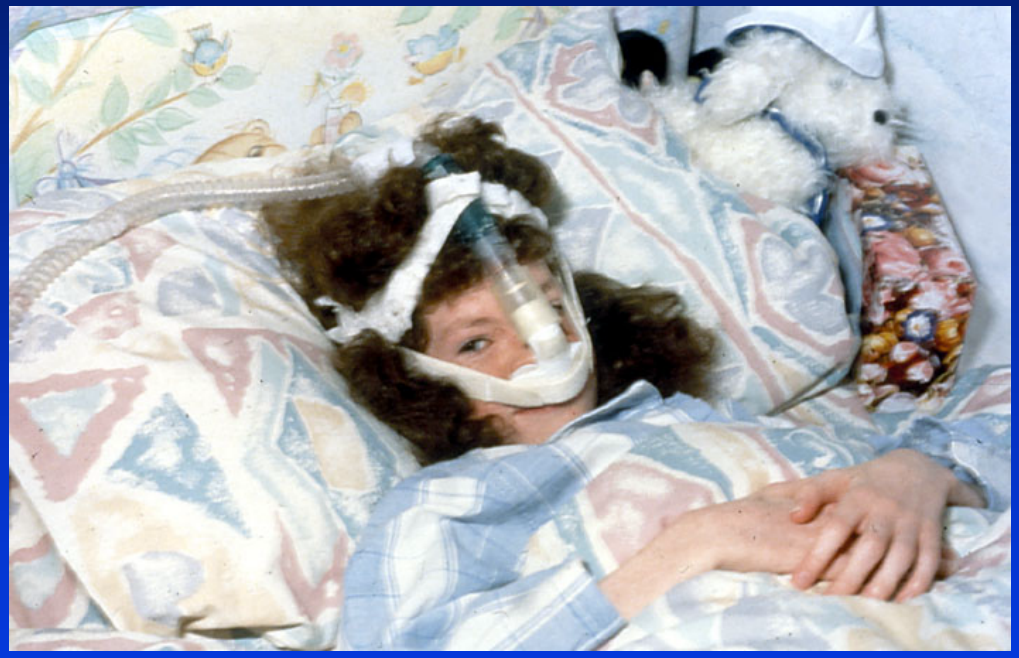
# Nasopharyngeal Airway



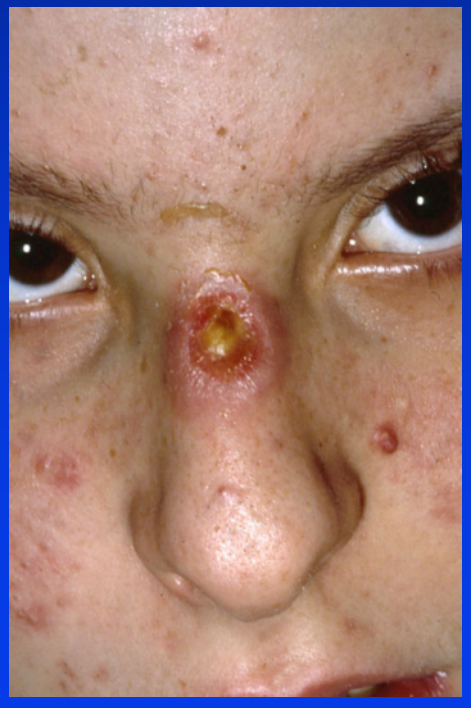




# Nasal CPAP



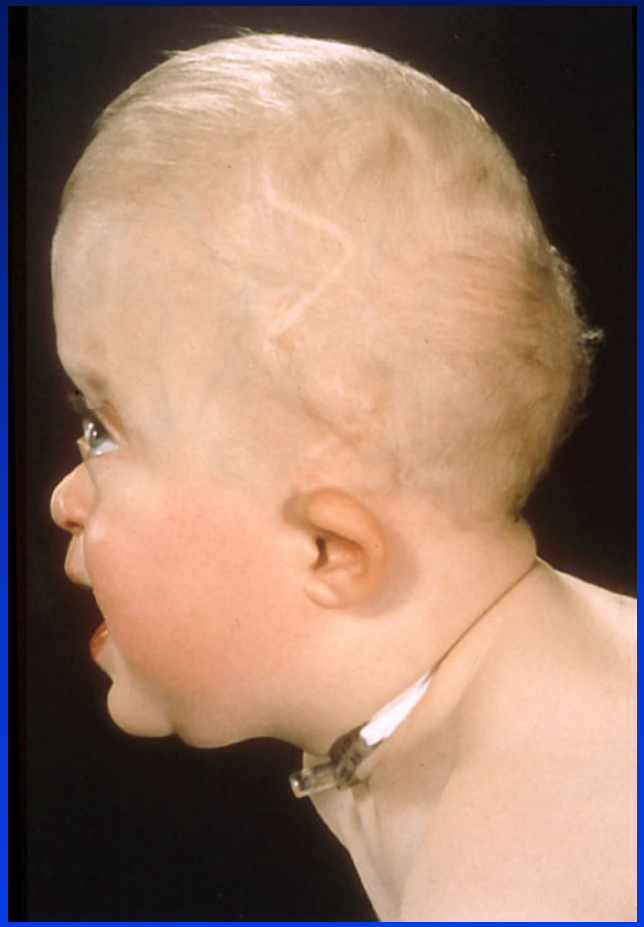
acclimatisation
complications
compliance



# Management - Surgery

- Adenotonsillectomy
- Tracheostomy
- Mid – facial advancement

# Tracheostomy



# QUESTIONS