Congenital Hearing Loss

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Etiology

- Congenital HL
  - 50% Genetic
  - 50% Acquired
- Childhood Onset HL
  - 50% Genetic
  - 25% Acquired
  - 25% Unknown

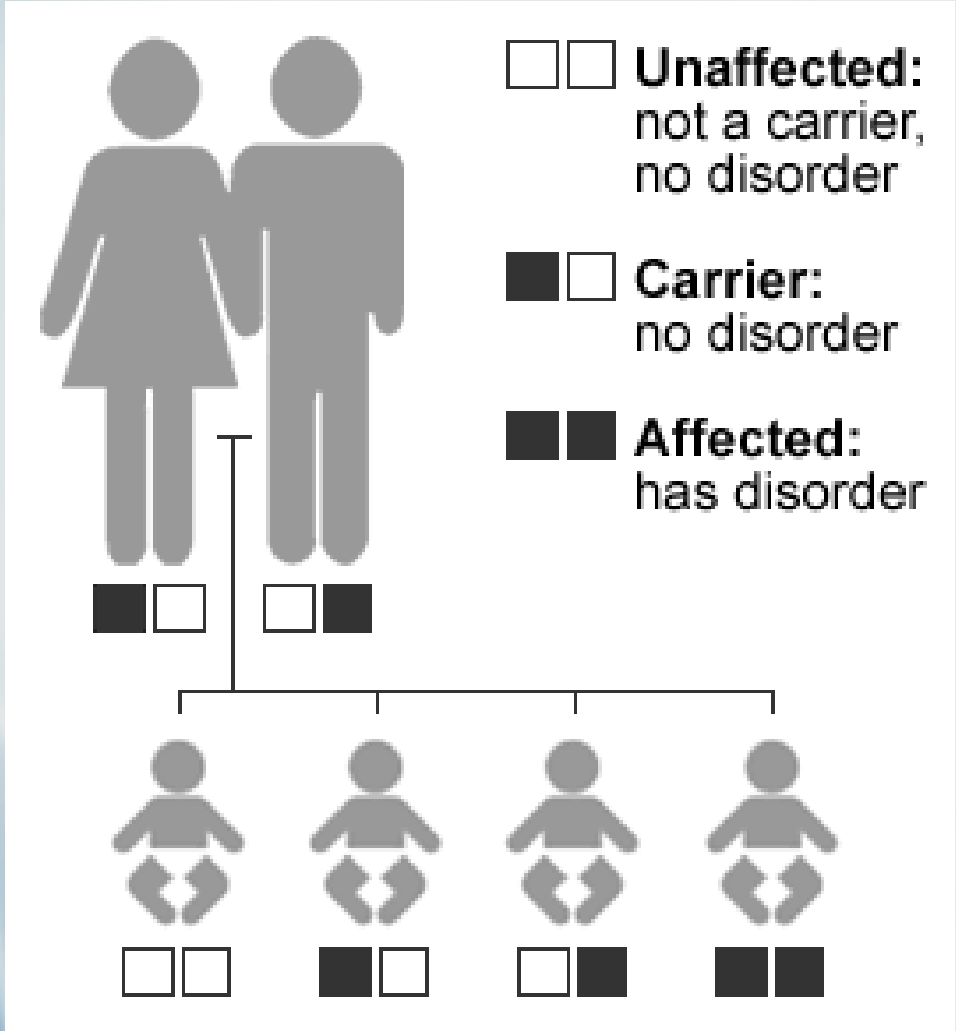
Genetic HL

- 75% non-syndromal
- 25% syndromal

- 75% autosomal recessive (AR)
- 25% autosomal dominant (AD)
- 1-2% X-linked
- Rare mitochondrial

Autosomal recessive HL

- Monogenic, 25% risk to offspring if both parents are carriers
- Severe to profound SNHL, prelingual onset



Autosomal recessive syndromal HL

- Usher syndrome
- Pendred
- Jervel and Lange Nielsen
- Goldenhar (Oculoauriculoverterbral spectrum)

Usher Syndrome

- Retinitis pimentosa and SNHL
- Night blindness > field cut > central blindness
- Most common cause of congenital deafness
- Dx: electroretinography

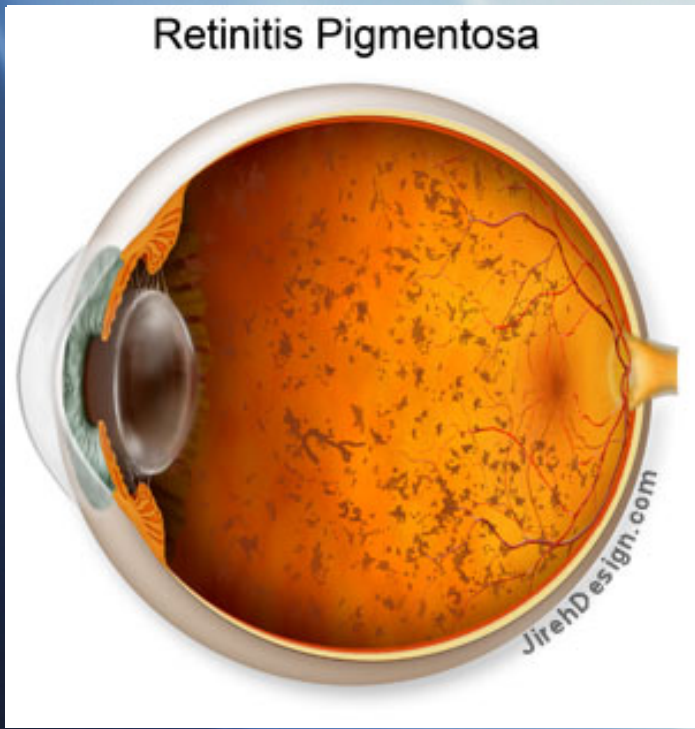
Usher Types

- Type I (most common):
  - Profound SNHL, no vestibular fxn
  - RP onset in early childhood
  - Atypical myosin (myosin 7A): interferes with mechanoelectrical transduction in labyrinthine hair cells
- Type II:
  - Congenital sloping SNHL
  - Normal vestibular fxn
  - RP onset in teens

Usher Types

- Type III:
  - Progressive SNHL and vestibular dysfunction
  - Vestibulocerebellar ataxia
- Type IV:
  - Mental retardation and hypotonia

Usher



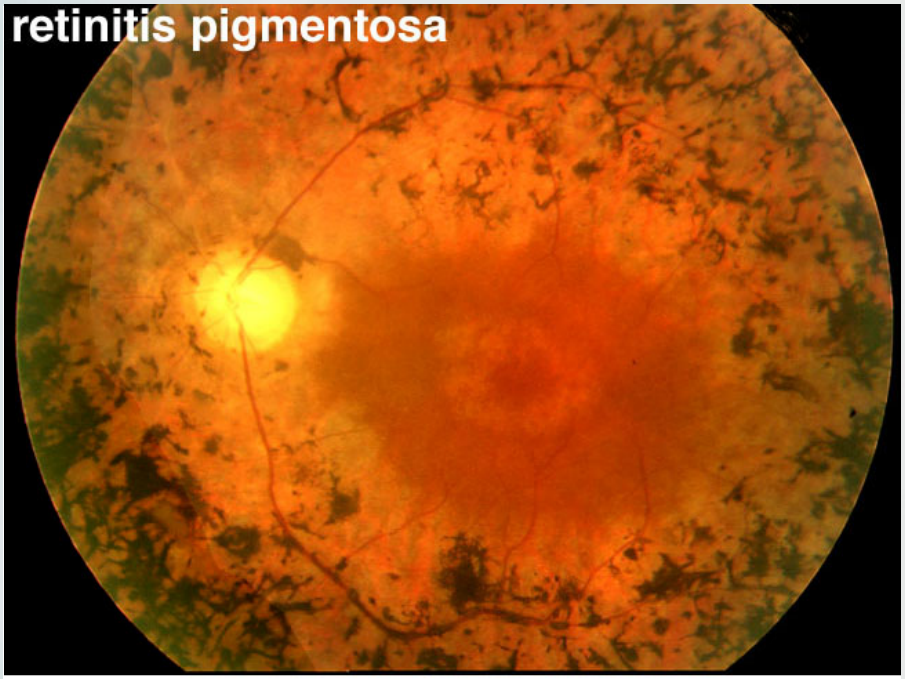
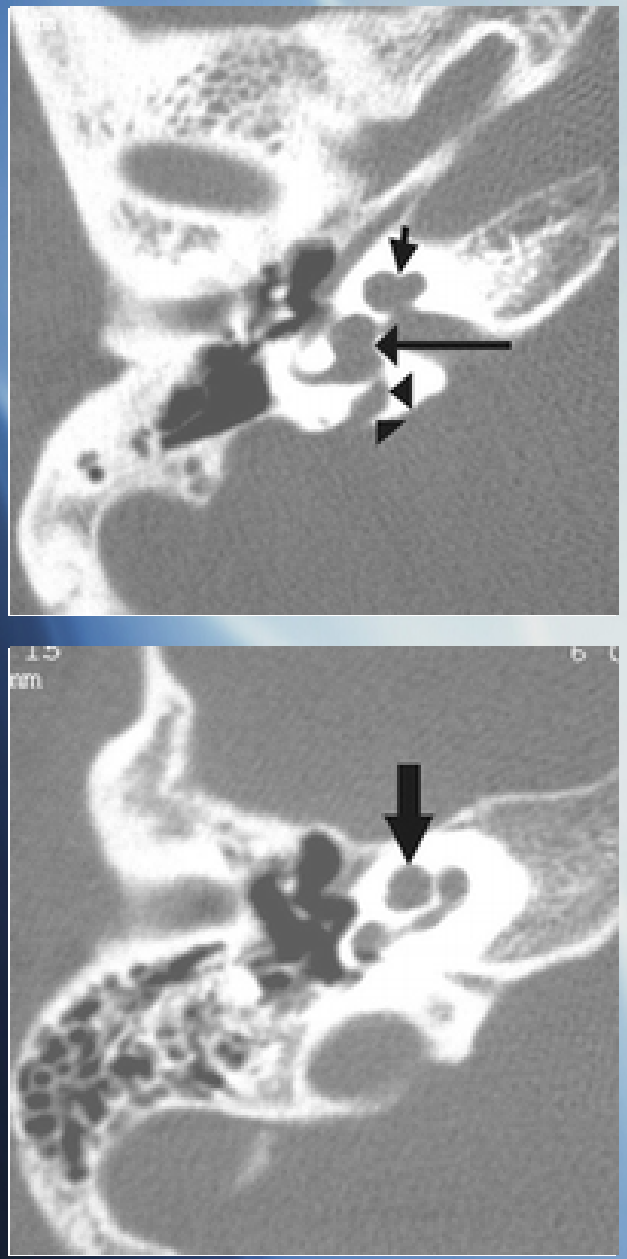


Fig. 12b. Fundus photo of a patient with retinitis pigmentosa.

Pendred Syndrome

- Defect in tyrosine iodination
- Gene mutation: affects pendrin, molecule involved in chloride-iodine transport
- Sx: severe to profound SNHL, multinodular goiter in childhood
- Assoc with Mondini malformation and enlarged vestibular aqueduct
- Dx: (+) perchlorate test
- Tx: thyroid hormone to suppress goiter



- Transverse CT scans of the middle ear in a 47-year-old patient with Pendred syndrome.
- (a) Modiolus is not discernible (short arrow). Vestibular aqueduct (arrowheads) and vestibule (long arrow) are enlarged.
- (b) Interscalar septum between upper and middle turn of the cochlea is absent (arrow).

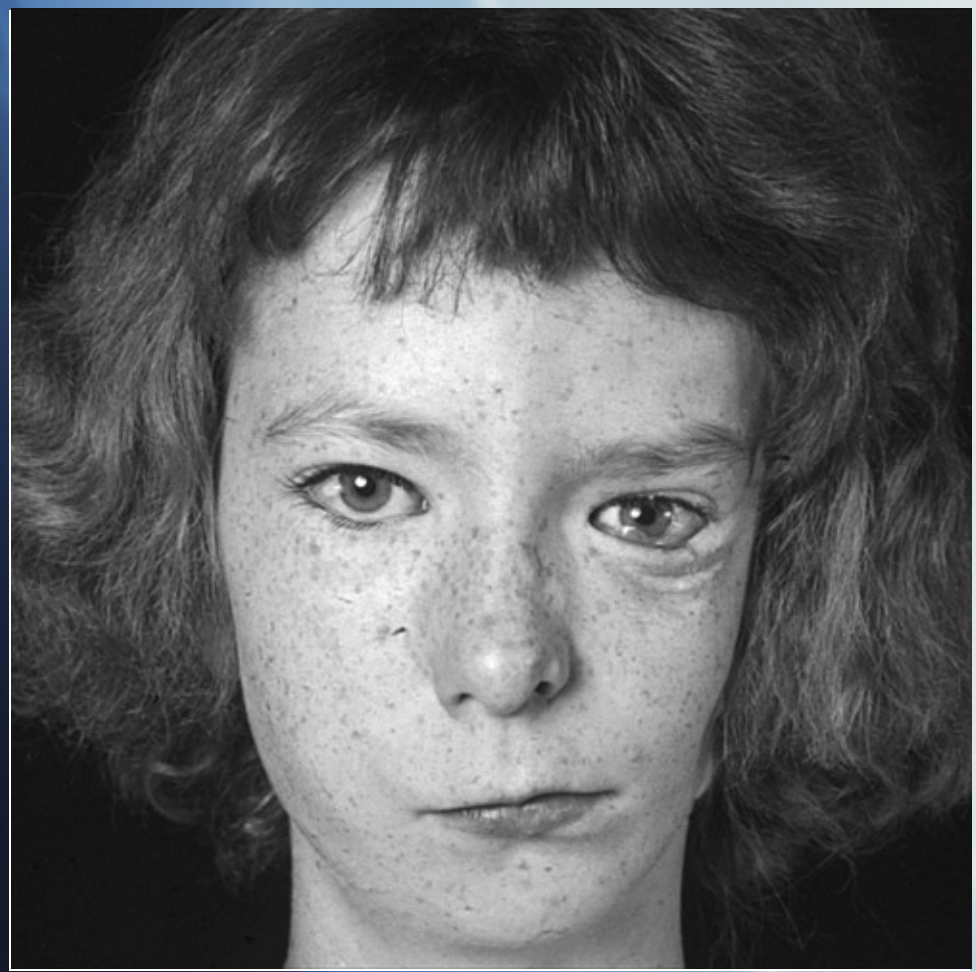
Jervell and Lange Nielsen

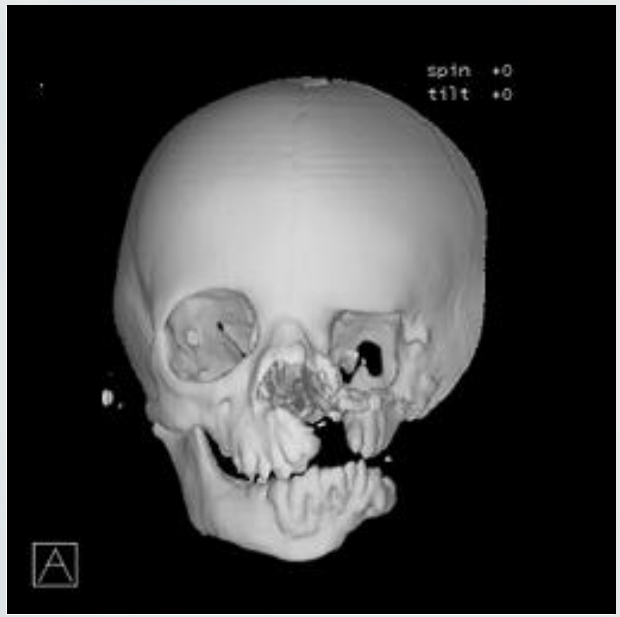
- Congenital profound SNHL
- Prolonged QT interval with syncope, sudden death
- Gene mutation: KVKQT1 = abnormal K+ channel
- Dx: EKG
- Tx: Beta blockers, hearing aids

Goldenhar Syndrome

- First and second arch derivatives, hemifacial
- CHL and SNHL (mixed)
- Ocular: epibulbar dermoids, colobomas
- Auricular: preauricular appendages, pinna abnormalities, EAC atresia, ossicular malformation/absence, abnormal facial nerve, stapedius, semicircular canals and oval window
- Vertebral: fusion/absence of cervical vertebrae

Goldenhar Syndrome





Autosomal Dominant

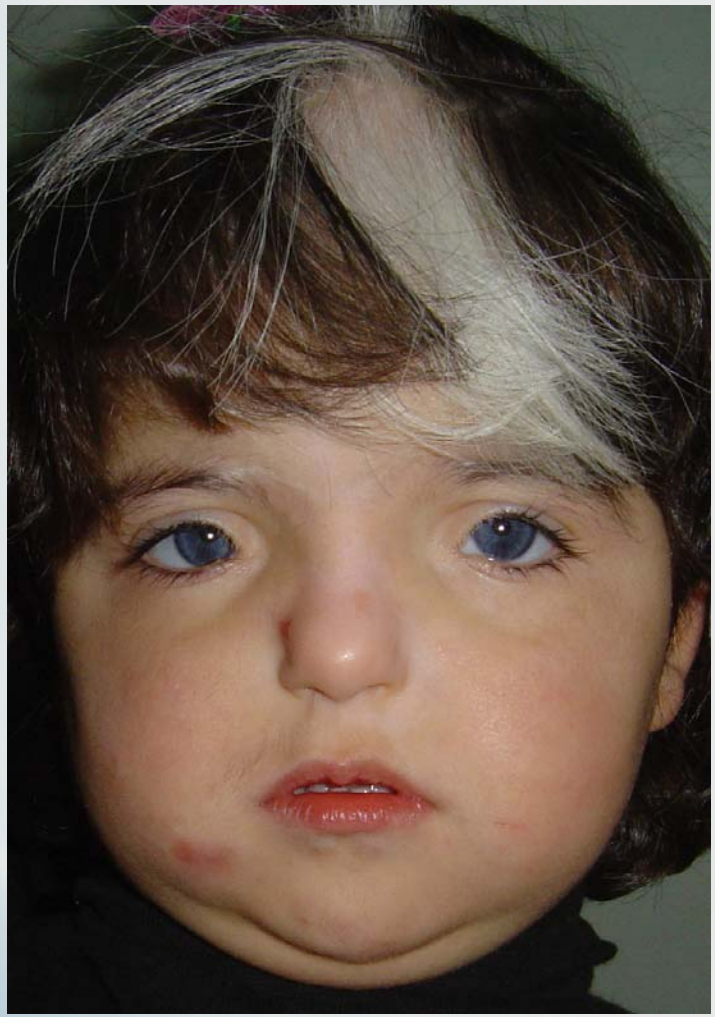
- Vertical pattern of inheritance
- Risk to offspring of 50% if 1 parent affected
- Variable penetrance and expressivity
- Often postlingual hearing loss, progressive

AD Syndromes

- Waardenburg
- Treacher Collins
- Apert
- Crouzon
- Stickler
- Neurofibromatosis
- Brancio-oto-renal

Waardenburg Syndrome

- Abnormal tyrosine metabolism
- Pigment abnormalities: heterochromic iriditis, white forelock, patchy skin depigmentation
- Craniofacial abnormalities: dystopia canthorum, synophrys, flat nasal root



Waardenburg Types

- Type I:
  - Dystopia canthorum, pigment and craniofacial abnormalities, 20% with SNHL
  - Mutation in PAX3 gene
- Type II:
  - No dystopia canthorum, 50% with SNHL but not as severe
  - MITF mutation

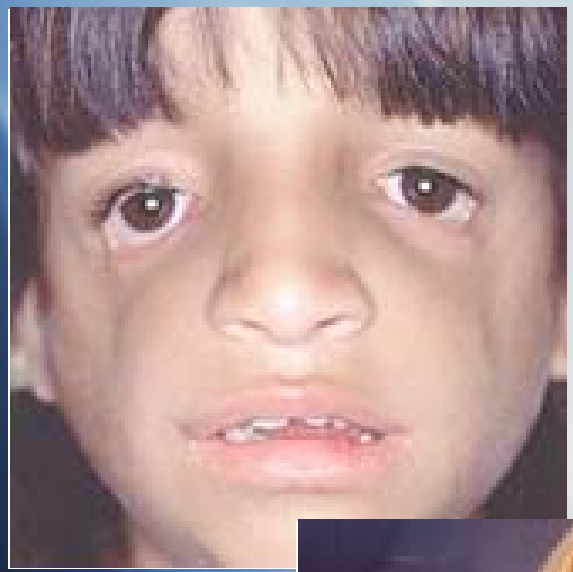
Waardenburg Types

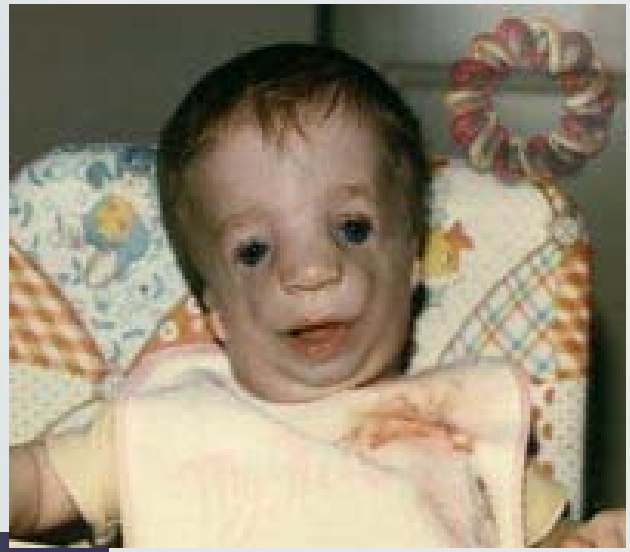
- Type III (most severe):
  - Unilateral ptosis and skeletal abnormalities
  - PAX3 mutation
- Type IV:
  - Type II plus Hirschsprung's disease (aganglionic megacolon)

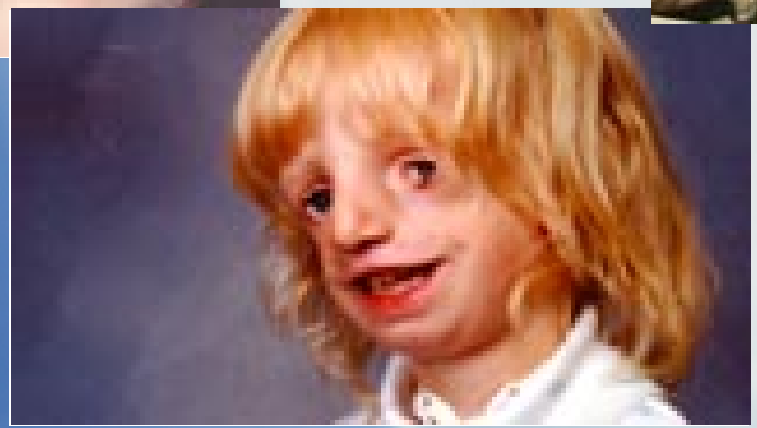
Treacher Collins (Mandibulofacial dysostosis)

- Hypoplasia of mandible and facial bones
- Downsloping palpebral fissures, colobomas
- Atretic external and middle ear
- Mixed HL
- Cleft palate (35%)
- Gene mutation on chr 5q: TCOF1 codes for a cell transport protein (treacle)
- Tx: BAHA, bone conduction HA, surgical correction of aural atresia

Treacher Collins



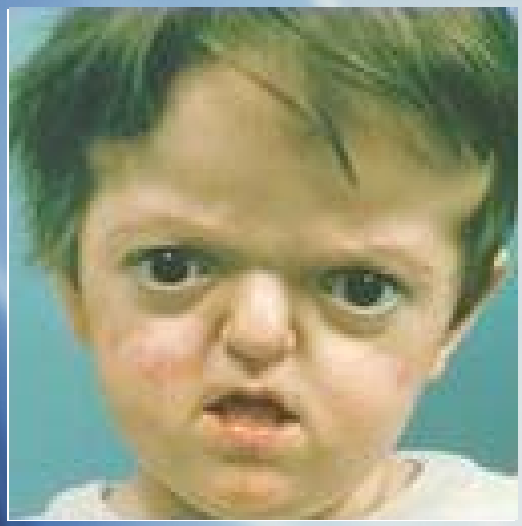


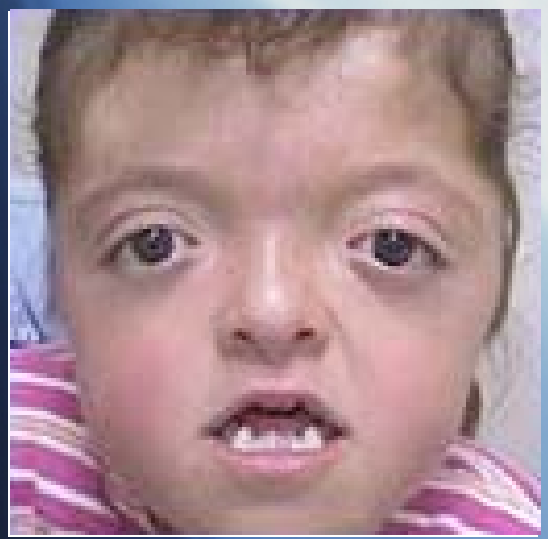


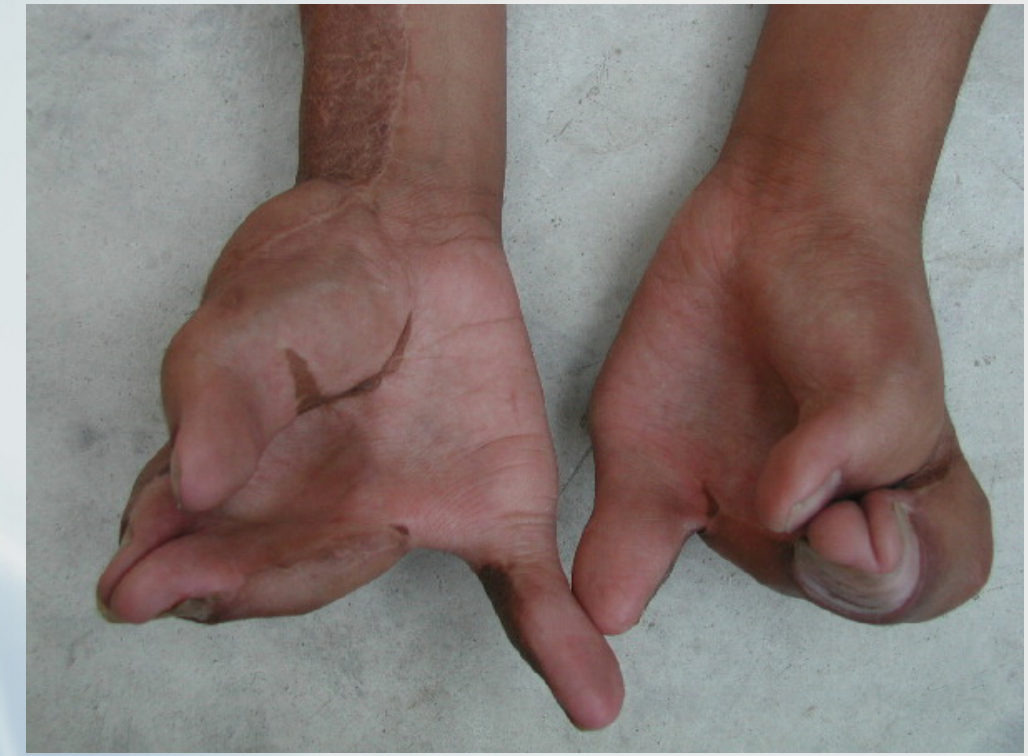
Apert Syndrome (Acrocephalosyndactyly)

- Middle and inner ear affected
- Stapes fixation (CHL), patent cochlear aqueduct, large subarcuate fossa
- Hand syndactyly, midface abnormalities, craniofacial dysostosis, trapezoid mouth

Apert



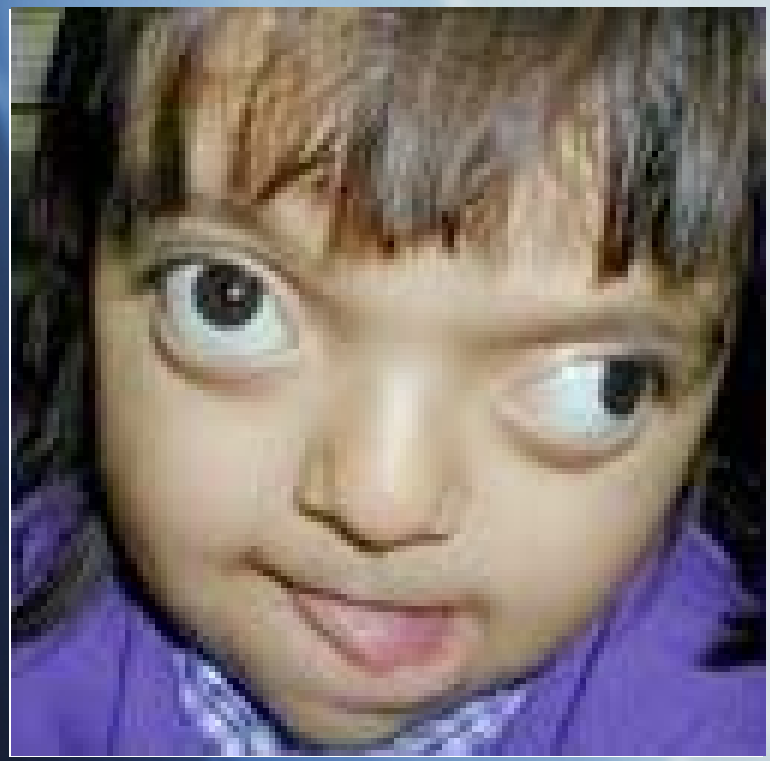




Crouzon Syndrome (craniofacial dysostosis)

- Atresia and stenosis of EAC, CHL, ossicular deformities
- Cranial synostosis, small maxilla, exophthalmos, parrot nose, short upper lip, mandibular prognathism, hypertelorism
- Abnormal FGF receptors

Crouzon



Stickler Syndrome

- Progressive Arthro-Ophthalmopathy
- Progressive SNHL (80%)
- Marfanoid body habitus
- Severe myopia, retinal detachment
- Flat midface
- Hypermobile joints
- Pierre Robin sequence: micrognathia, glossoptosis, cleft palate

Neurofibromatosis

- NF-1 (Von Recklinghausen Disease)
  - Café au lait spots, neurofibromas, Lisch nodules, 5% risk of unilateral acoustic neuroma
  - NF-1 gene on Chr 17
- NF-2 (central neurofibromatosis)
  - Bilateral acoustic neuromas or unilateral with 1st degree relative with NF-2 or multiple central schwannomas
  - NF-2 gene Chr 22q12 (tumor suppressor gene mutation)