Congenital Hearing Loss

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Introduction

- Deafness affects 0.2%
- SNHL attributed to
  - 50% genetic factors RC
  - 20-25% environmental factors
  - 25-30% sporadic
- Genetic deafness attributed to
  - 75% AR inheritance
  - 20% to AD
  - 5 % X-linked or chromosomal disorders
- 70% nonsyndromic
- Over 400 syndromes

Neonates at risk for SNHL

1. family hx of hereditary childhood SNHL
2. in utero infection TORCH
3. craniofacial anomalies
4. birthweight < 1500 grams
5. Apgars 0-4 at 1 minute or 0-6 at 5 minutes
6. hyperbilirubinemia
7. on ventilator > 5 days
8. ototoxic meds
9. bacterial meningits

50 %

Inner Ear Malformation

- 9 weeks gestation Cochlea fully formed
- The deformities are classified into 5 different groups

RC??

Michel Aplasia

- Complete agenesis of petrous portion of temporal bone
- External and middle ear may be unaffected
- Thought to result from an insult prior to the end of the third gestational week
- Affected ears are anacusic
- Autosomal dominant inheritance has been observed

Mondini Aplasia

- Only the basal coil can be identified
- Interscalar septum is absent
- enlarged endolymphatic duct
- Thought to result from insult around the sixth week of gestation
- Also described in?? X3
  - Waardenburg's
  - Treacher Collins
  - Pendred's,

3ATP

Scheibe Aplasia

- Also known as
  - Cochleo-saccular dysplasia
  - Pars inferior dysplasia (IAC)
- Normal** bony labyrinth and superior portion of membranous labyrinth
- Organ of Corti is poorly differentiated
- Deformed tectorial membrane
- Collapsed Reissner's membrane
- **Most common form of inner ear aplasia**

* RC CT scan ?

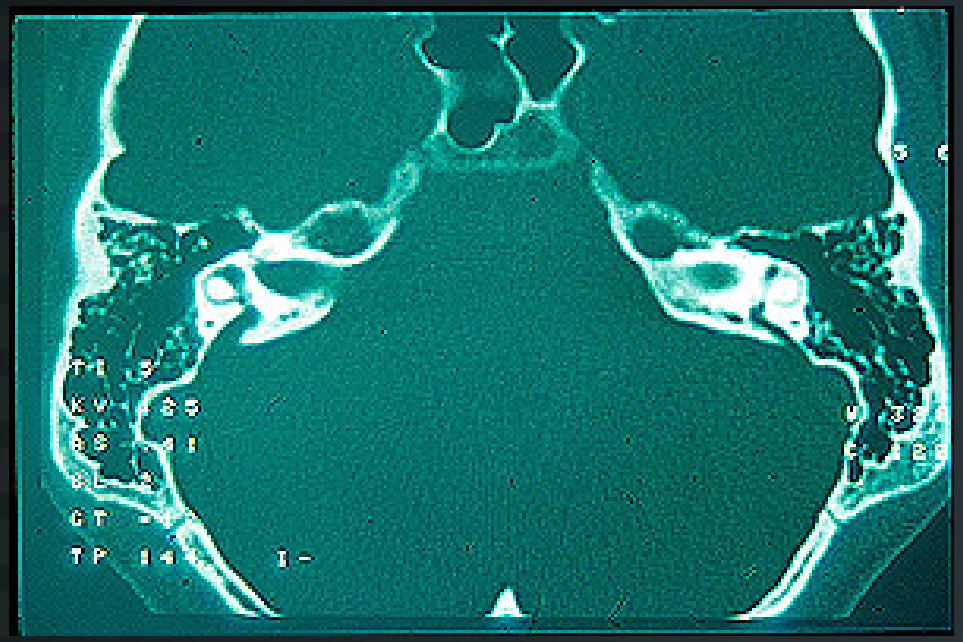
Alexander Aplasia

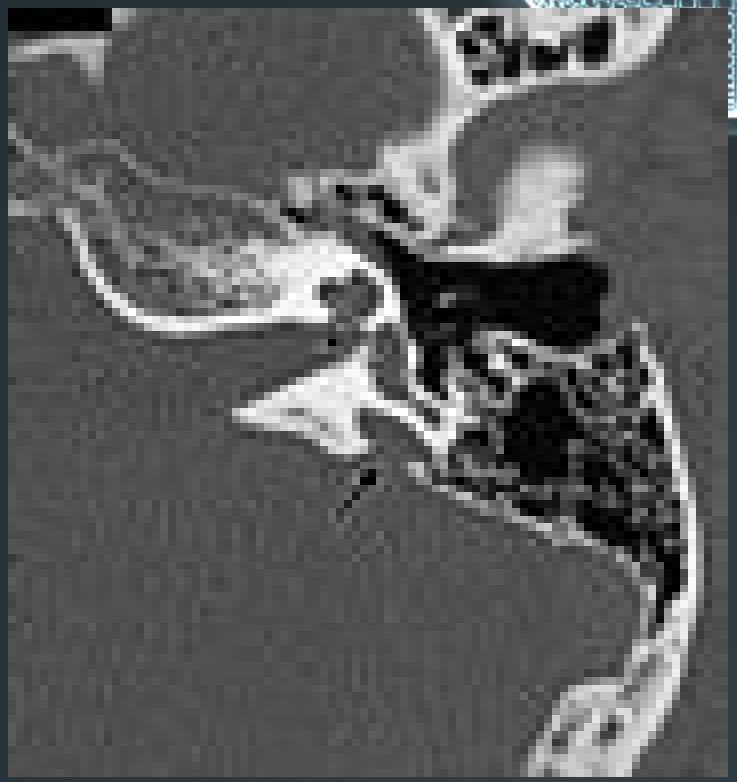
- Limited cochlear duct
- Effects on organ of Corti and ganglion cells
- High frequency hearing loss

Enlarged Vestibular Aqu

- Early onset SNHL,
- Usually bilateral progressive
- Hydrodynamic changes→labyrinthine membrane disruption
- May accompany cochlear and SCC deformities
- Familial cases suggest AD >AR
- AP diameter >2 mm
- Has been associated with ??? syndrome

Pendred's





RC

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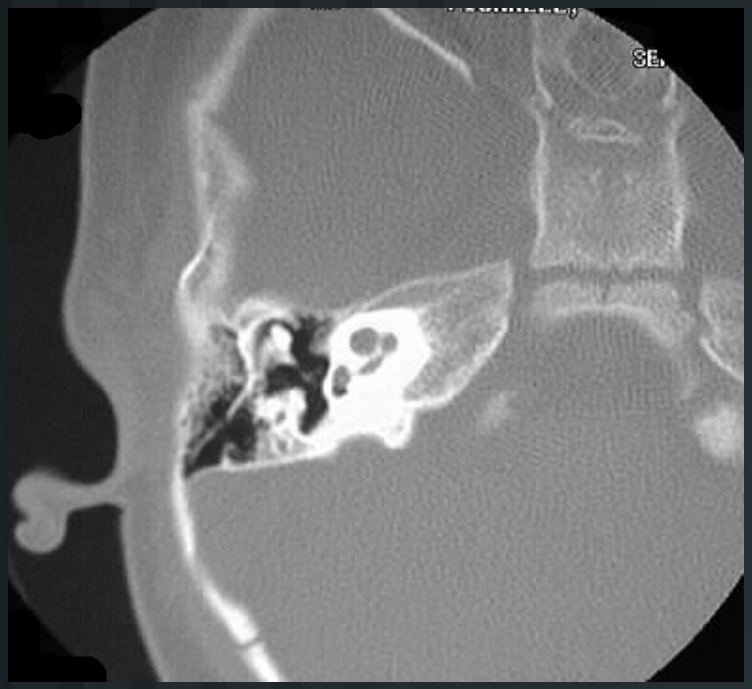
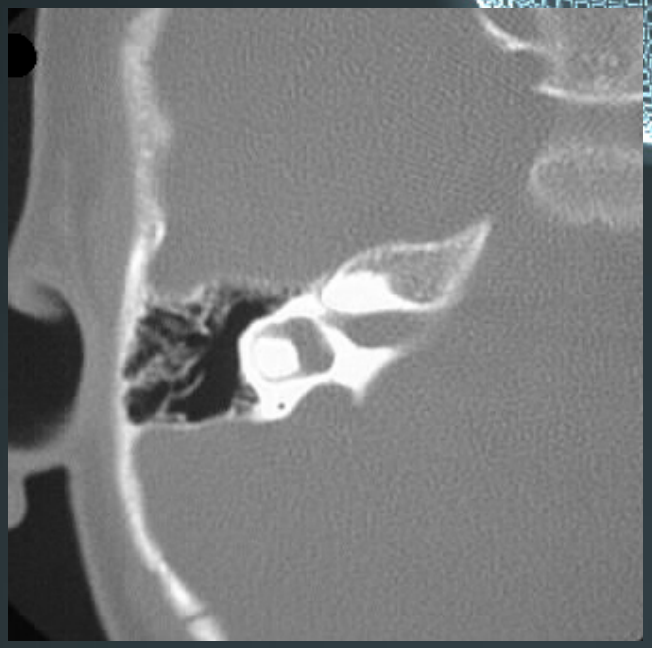




Mondini Malformation and Vestibular Aqueduct Syndrome

Semicircular Canal

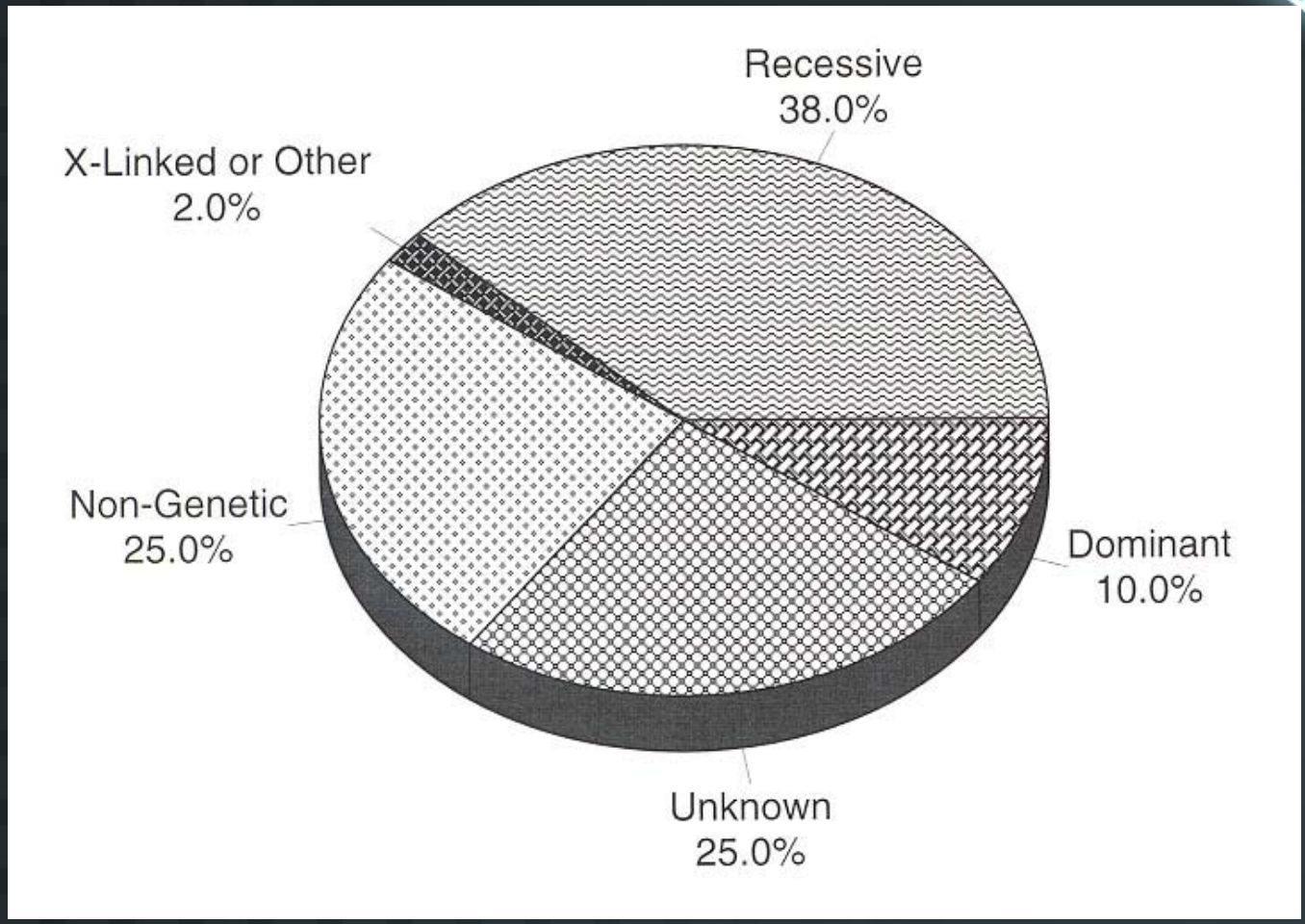
- Canal formation begins in the sixth gestational week
- Superior canal is formed first
- lateral canal is the last to be formed
- Isolated lateral canal deformities are the most commonly identified inner ear malformation on CT scans* (? VA)**

RC
C 425
C 426+419

Classification

- Syndromic and nonsyndromic forms
- Up to 30% due to syndromic forms
- Also divided into groups by mode of inheritance:
  - Autosomal Dominant
  - Autosomal Recessive
  - X-Linked Disorder
  - Mitochondrial Disorder
  - Multifactorial Disorder

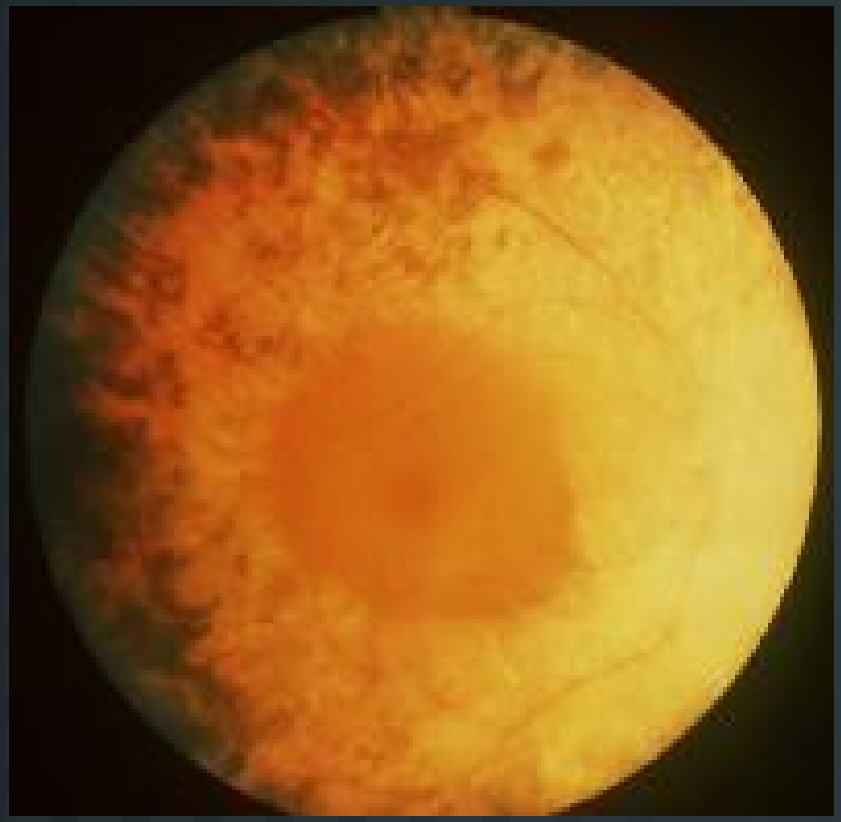
Etiology of Deafness



Recessive Disorders

- Most common pattern
- 80% of hereditary hearing loss
- 5 Syndromes
    1. Usher
    2. Pendred
    3. Goldenhar
    4. *Alport*
    5. Jervell and Lange-Neilsen

4 years old with congenital bil prof HL and no vestibular function



Usher Syndrome

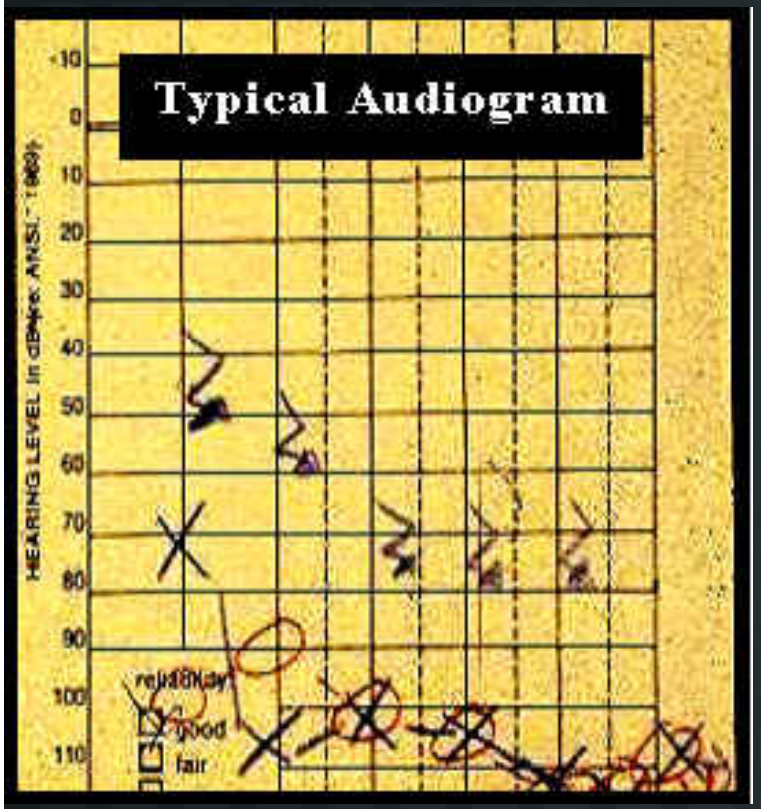
- Prevalence of 3.5 per 100,000
- SNHL and retinitis pigmentosa
- Three subtypes
  - Type 1

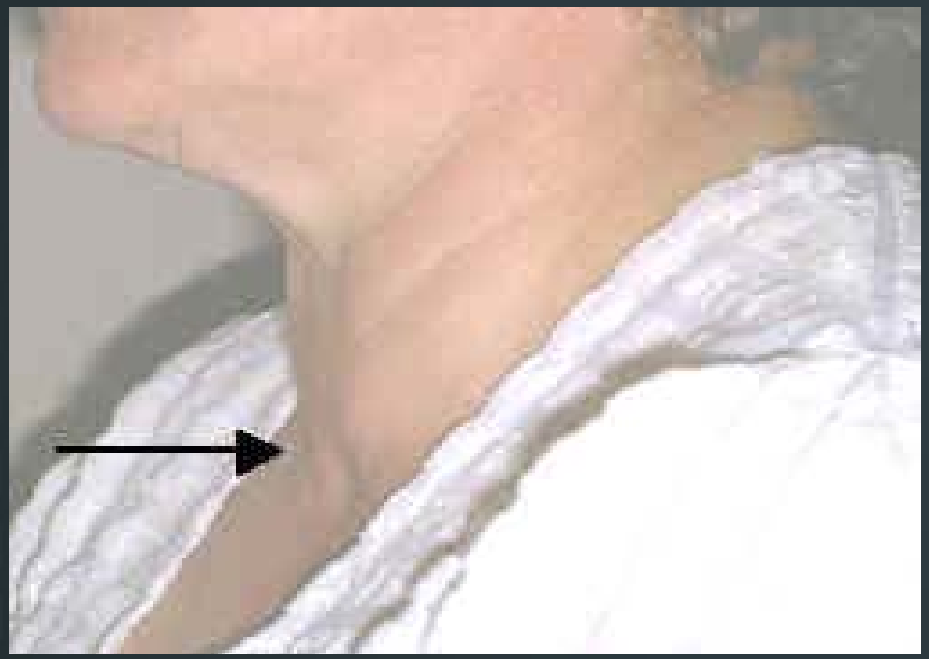
    congenital prof HL and no vestibular
  - Type 2

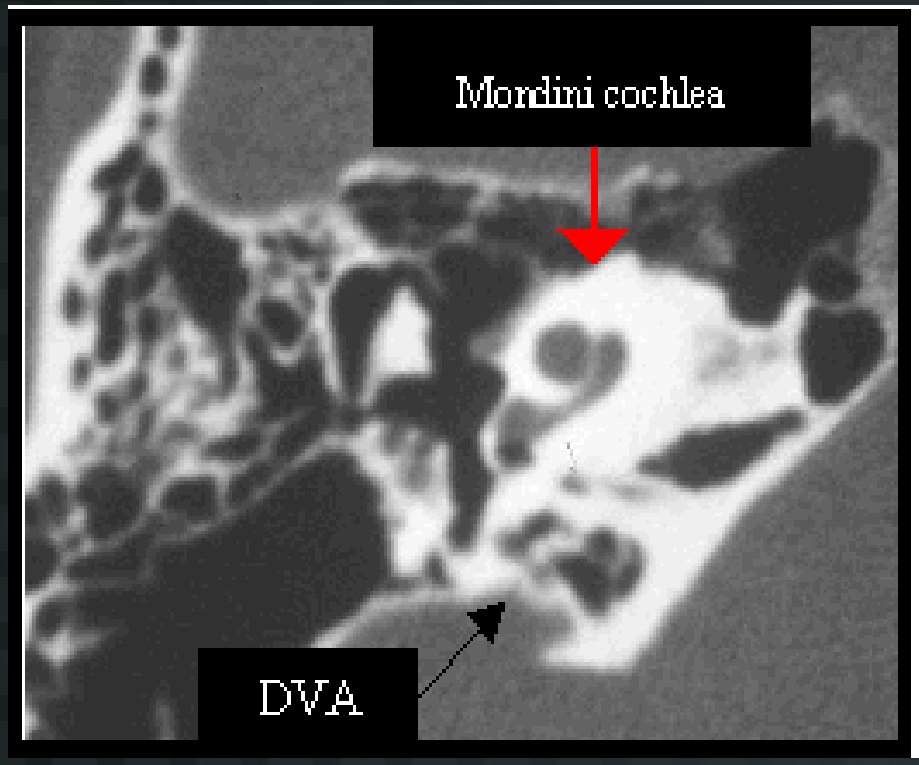
    moderate losses and normal vestibular
  - Type 3

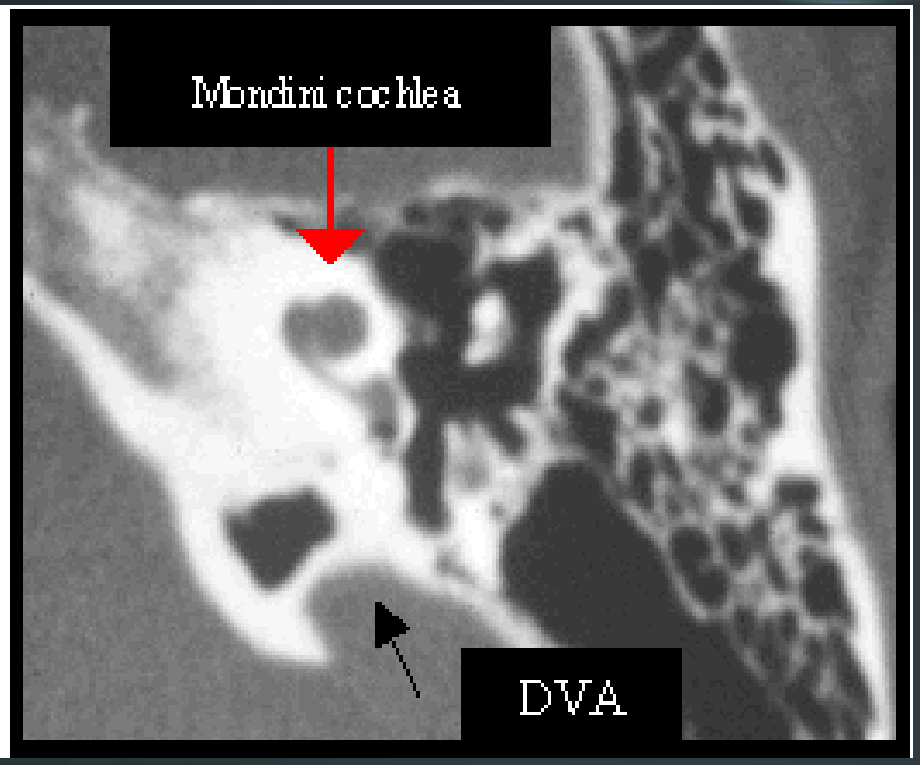
    progressive HL and variable vestibular

18 y lady with profound SNHL and thyroid goiter






Mondini cochlea
DVA


Mondini cochlea
DVA

Pendred Syndrome

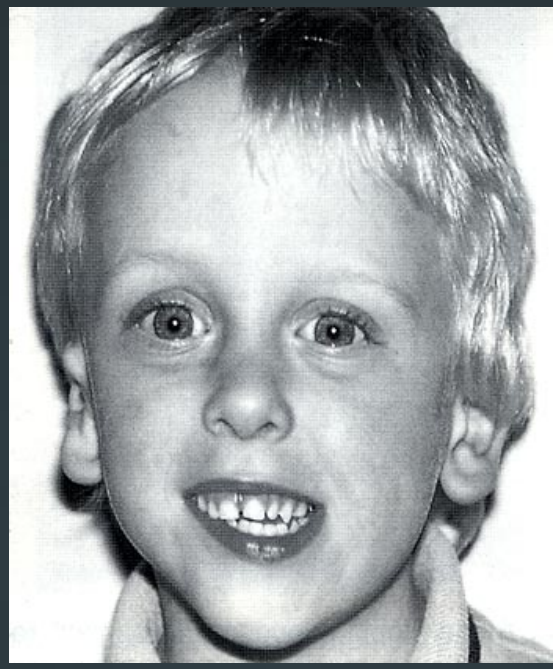
- CT scan
  - Mondini deformity or
  - enlarged vestibular aqueduct
- abnormal tyrosine* iodination
- euthyroid goiter
- treated with exogenous thyroid hormone
- definitive Dx Perchlorate** discharge test
- chromosome 7**

•Waardenburg

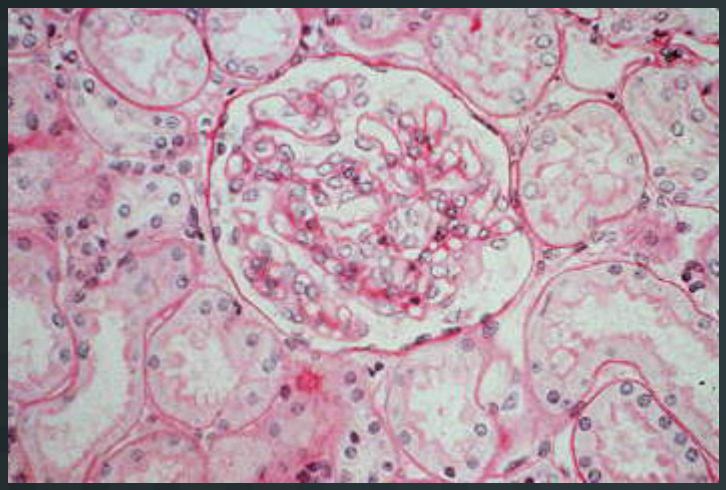
• Cystic fibrosis sweat chloride value of more than 60 mEq/L + Ch 7

Goldenhar syndrome

- Oculo-auriculo-vertebral
- Usually conductive
  - Auricular malformations
  - EAC stenosis
  - ossicular abnormalities
- Sensorineural occasionally
- **Unilateral** abnormalities of 1st and 2nd branchial arches
- Sporadic (most)
- vertebral malformations
- Upper eyelid colobomas



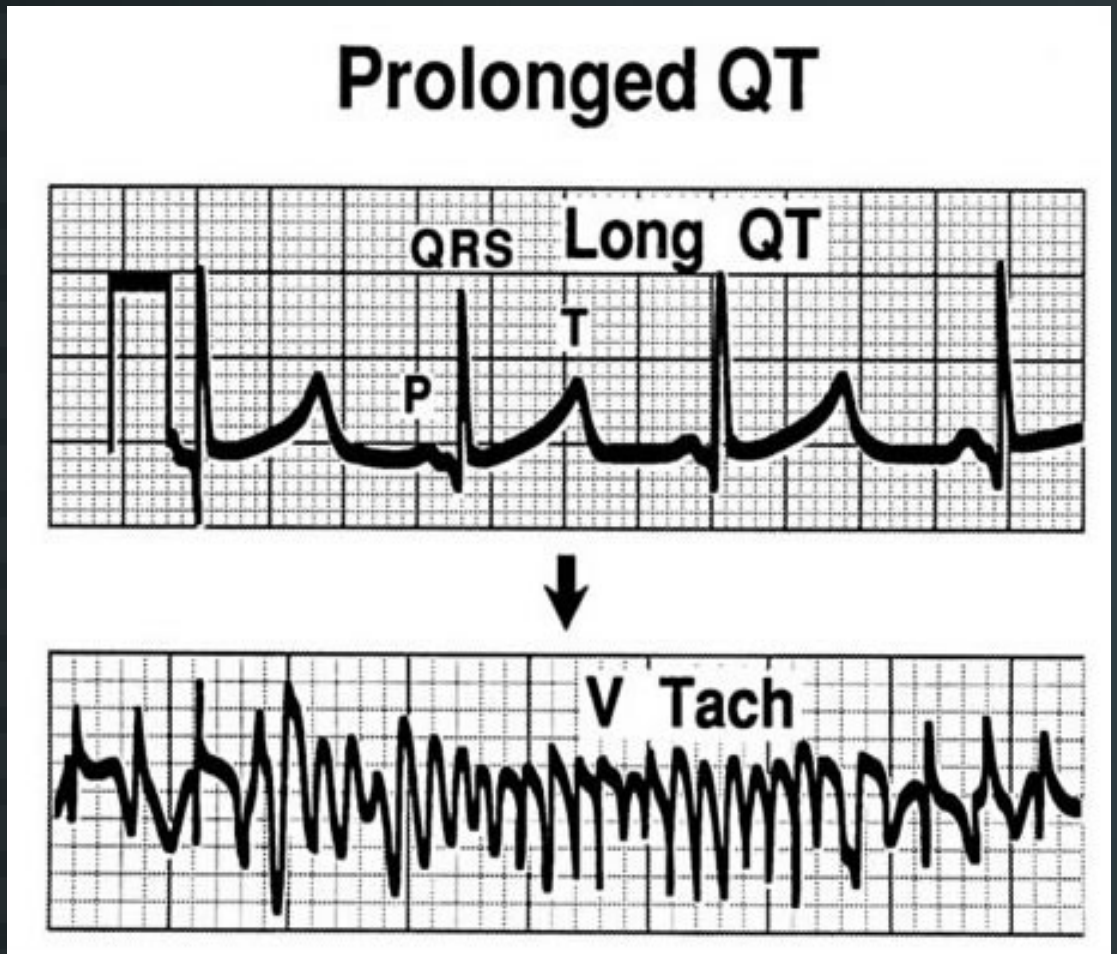
25 Y man with bil prof SNHL recently Dx with ESRF



Alport Syndrome

- hearing impairment
- may not become evident until the second decade
- Kidney
  - Asymptomatic → ESRF
  - hematuria
- Eye
  - posterior cataracts
  - corneal dystrophy
  - dislocation of the lens
- Defects in collagen type IV
- 20% are Autosomal recessive

2 months old failed screening test in NICU & has ECG



Jervell and Lange-Neilsen

- profound SNHL
- cardiac conduction defect → syncopal episodes
- ECG
  - large T waves
  - prolonged QT interval RC
- ECG should be done on all children with uncertain etiology of hearing loss
- Cardiac component treated with beta-adrenergic blockers
- chromosome 11

Autosomal Dominant Disorders

Waardenburg Syndrome

5 Y old with bil prof SNHL
+ve FHx

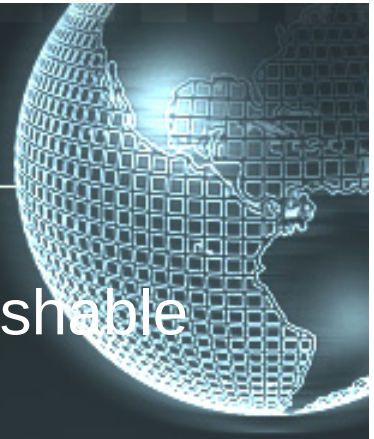
Waardenburg Syndrome

- Accounts for 3% of childhood hearing impairment
- Most common form of inheritable congenital deafness
- Incidence is 1 in 4000 live births
- May have unilateral or bilateral SNHL
- All features are variable in appearance

Waardenburg Syndrome

- Pigmentary features include (tyrosine* metabolism):
  - white forelock
  - Heterochromia irides
  - Premature graying
  - Vitiligo
- Craniofacial features include
  - dystopia canthorum
  - broad nasal root
  - synophrys

Waardenburg Syndrome



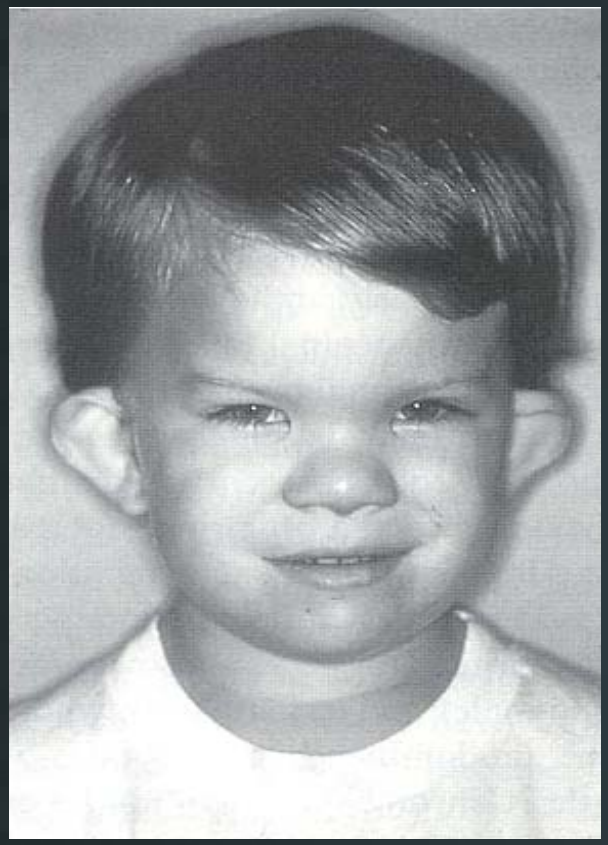
- Three different types that are clinically distinguishable
- Type1
  - Heterochromia irides
  - White forelock,
  - Patchy hypopigmentation,
  - Dystopia canthorum
  - SNHL seen in 20%
- Type 2
  - absence of dystopia canthorum
  - SNHL seen in 50%
- Type 3
  - microcephaly,
  - skeletal abnormalities
  - mental retardation,
  - + features in type 1

Stickler Syndrome

- Ear
  - Severe SNHL in 15%
  - Ossicular malformations
- cleft palate
- micrognathia
- Eye
  - Cataracts
  - severe myopia
  - retinal detachments

Branchio-oto-renal

3Y boy cup-ear, Branchial cleft fistula and one kidney



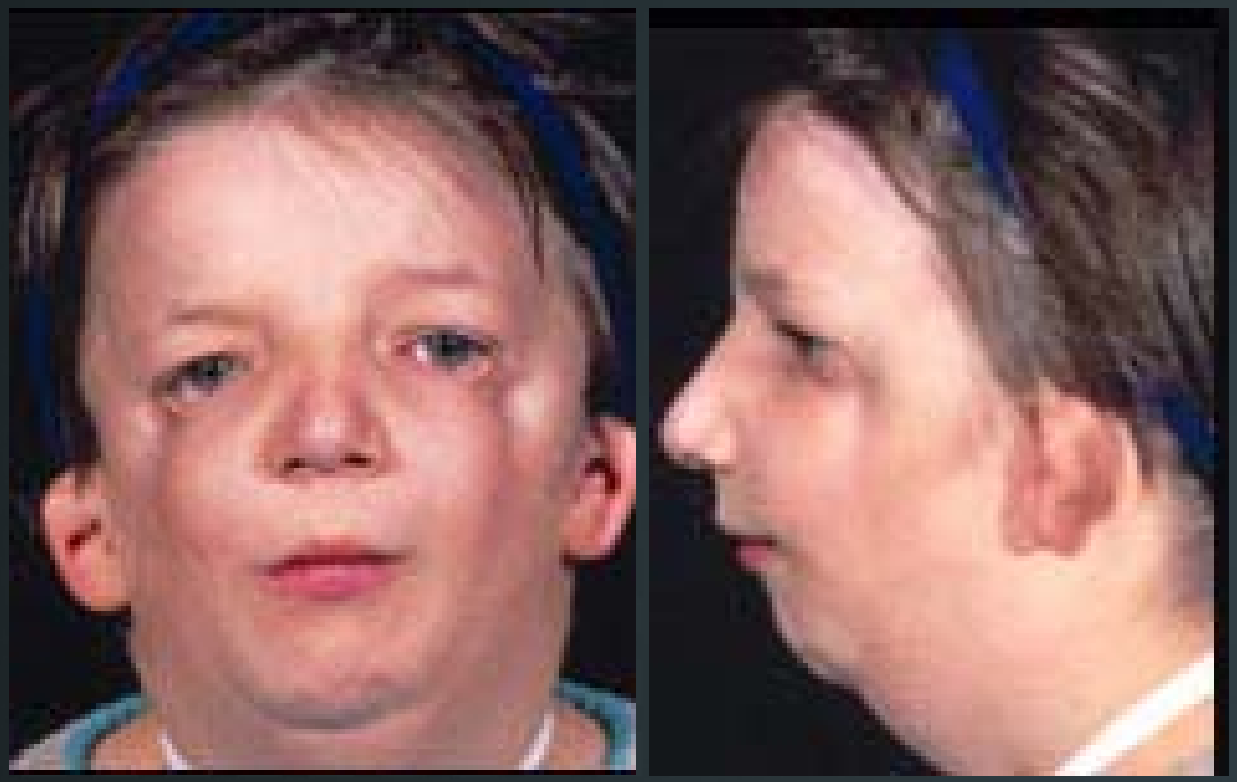
RC

Branchio-oto-renal

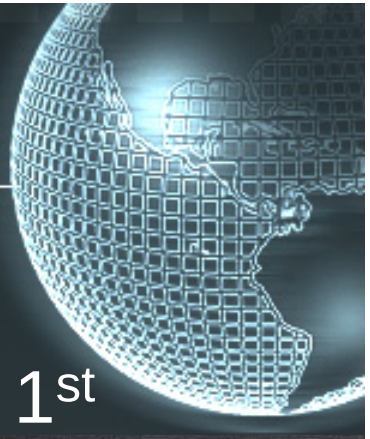
- Estimated to occur in 2% CHL
- 75% significant hearing loss
- Ear pits/tags
- Cervical fistula
- Renal failure
- Chromosome 8q

Treacher Collins

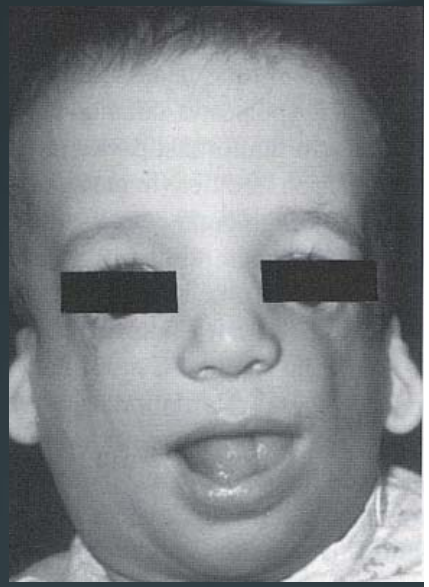
4 years old boy with mix-HL

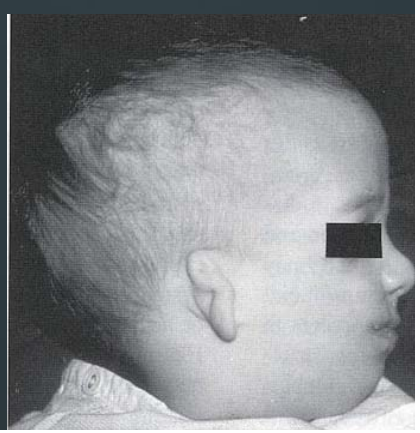


Treacher Collins



- **Bilateral** and symmetrical abnormalities of 1st and 2nd branchial arches
- EAC & Ossi- malformations are common
- Facial features are bilateral
  - malar hypoplasia,
  - downward slanting palpebral fissures,
  - coloboma of lower eyelids,
  - hypoplastic mandible,
  - cleft palate
- Transmitted AD with high penetrance





~Amie's Home~

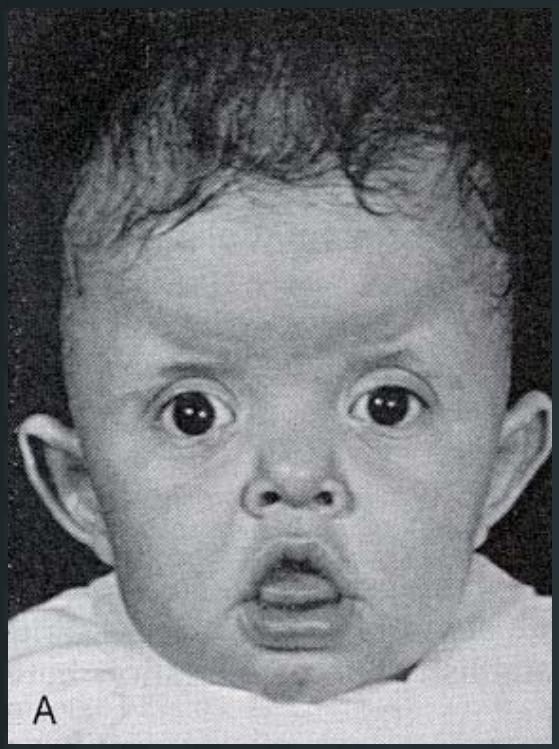
"What lies behind us and what lies before us are tiny matters compared to what lies within us."
-Ralph Waldo Emerson

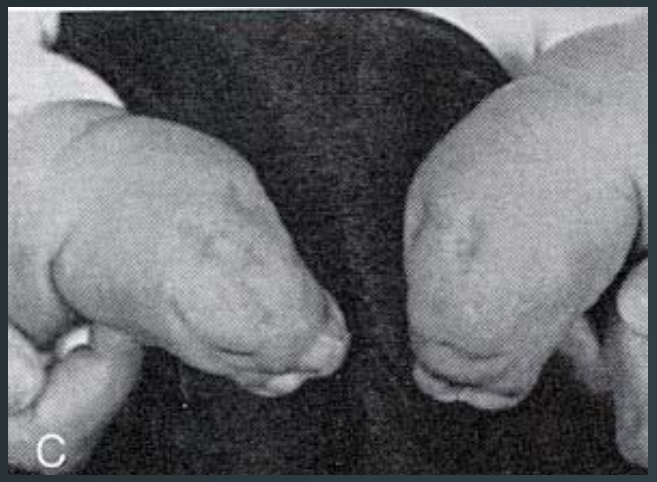
NEWS UPDATE: For the past 10 months, I have been participating in the making of a documentary about Treacher Collins syndrome that will air on the Discovery Channel. The title of the documentary is "Unmasked: Treacher Collins Syndrome" and it features the stories of six people with Treacher Collins syndrome, including my own story. For more info please see my webpage about the documentary, which includes air dates and times as well as info for how to order a copy of the show.

In most every way, I am your typical, garden-variety human being. I am a first year Med-Peds (i.e. Internal Medicine and Pediatrics) resident at Baylor College of Medicine in Houston, Texas. I graduated medical school from the University of Texas Health Sciences Center in Houston. However, many people assume right from the start that I must not be "normal" since I don't look "normal." I have Treacher Collins syndrome.

Apert

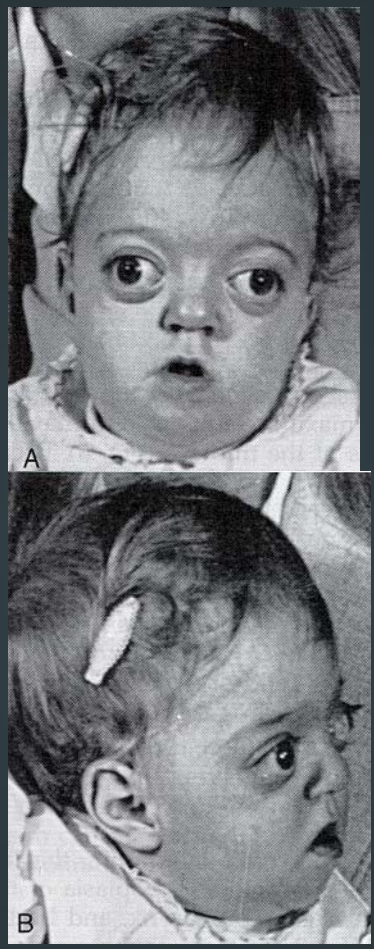
RC

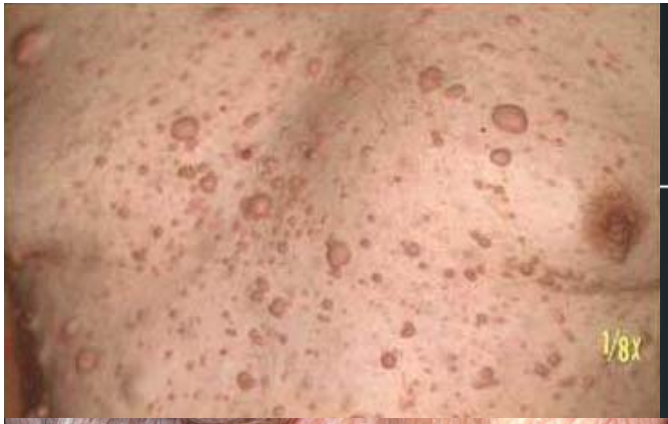




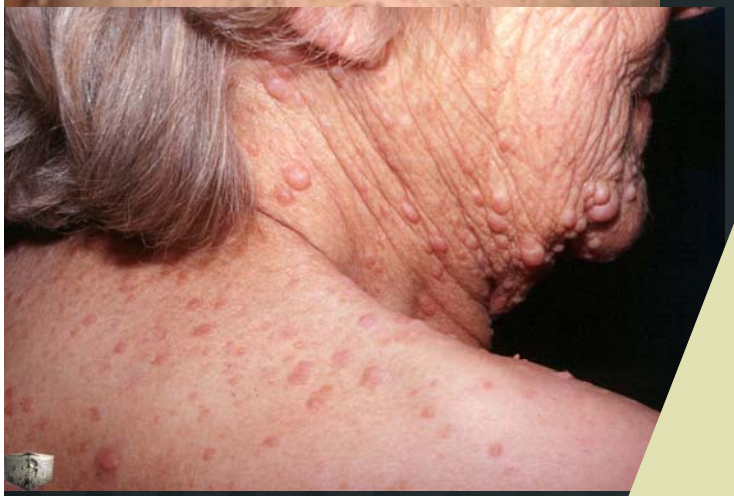
Apert and Crouzon

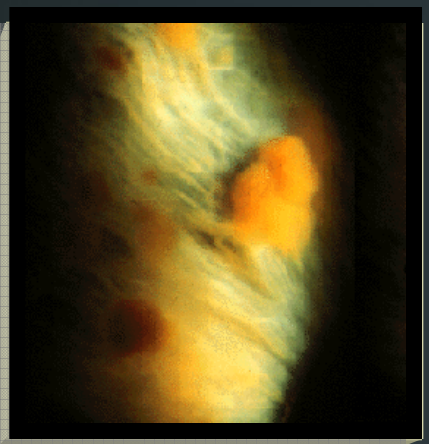
- Craniosynostosis
- Maxillary hypoplasia
  - prominent mandible
  - Prominent forehead
  - high arched palate
  - Hypertelorism
  - Exopthalmos
  - depressed nasal bridge
- congenital fixation of stapedial (A)
- Syndactyly (A)

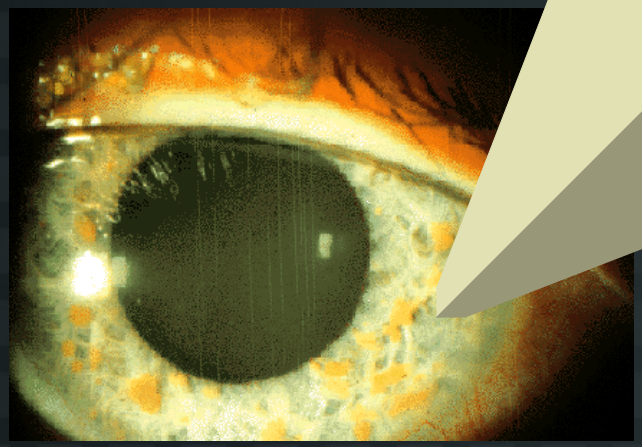




30 y Bil severe SNHL







Lisch nodules

Neurofibromatosis

- Classified into two types
- Type 1 is more common with an incidence of 1:3000 persons
- Type 1 generally includes
  - multiple café-au-lait spots
  - cutaneous neurofibromas,
  - plexiform neuromas
  - Lisch nodules of the iris
  - optic gliomas
- chromosome 17

Neurofibromatosis

Type 2 includes

- Chromosome 22
- Bilateral acoustic neuromas in 95%
- Unilateral with 1st degree relative or 2
  - subcapsular cataracts
  - Gliomas
  - Schwannomas
  - meningiomas

X-Linked

- X-linked inheritance is rare
- 1% to 2% of cases
- 6% of nonsyndromic profound losses in males
- Example ???

1. Norrie
2. Oto-palato-digital
3. Alport

Norrie Syndrome

- 30% progressive SNHL in second or third decade
- Eye
  - congenital or rapidly progressive blindness
  - Pseudoglioma
  - ocular degeneration→ microphthalmia

Oto-palato-digital Syndrome

- CHL seen due to ossicular malformations
- flat midface
- hypertelorism,
- small nose
- cleft palate
- short stature
- Broad, long fingers and toes

Mitochondrial Disorders

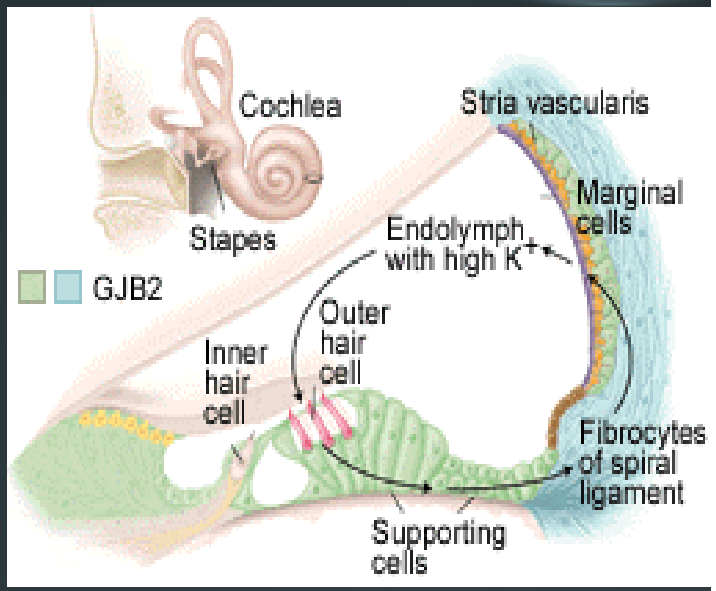
- progressive HL
- sensitivity to aminoglycosides
- affect energy production through ATP synthesis
- progressive neuromuscular degeneration
  - Ataxia
  - Ophthalmoplegia
- diabetes mellitus
- encephalopathy
- Nearly all mitochondria come from the mothers' egg and she transmits them to all offspring?
- sperm do not transmit mitochondria to the offspring

Nonsyndromic deafness

- Not restricted to the cochlea
- GJB2 (Connexin 26) RC
- GJB6 (Connexin 30)
- GJB3 (Connexin 31)
- GJA1 (Connexin 43)

Connexin 26

- Nonsyndromic
- AR or AD
- Chromosome 13* (26/2)
- 50 % nonsyndromic
- K+ Homeostasis



- RC connexin channels K+ ions

supporting cells → stria vascularis → endolymph

Connexin 26

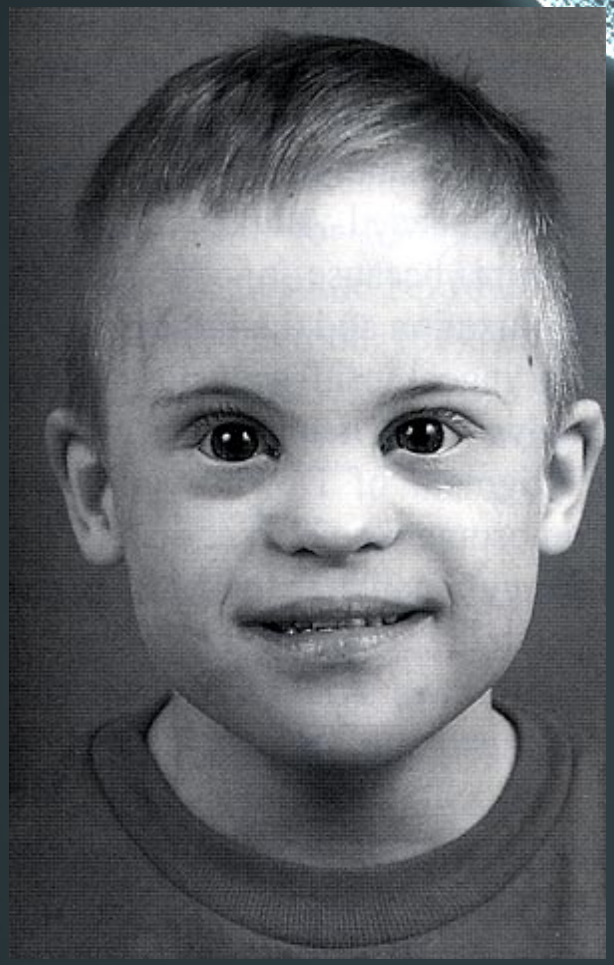
- Autosomal recessive
  - prelingual onset
  - severe to profound
  - all frequencies affected
- Autosomal dominant
  - postlingual
  - less severe.
  - Middle or high frequencies

Multifactorial disorders

- Genetic component is influenced by environment
- Often due to the interaction of several genes
- Genotype does not necessarily predict phenotypic outcome

Down Syndrome

- Trisomy 21
- 1 in 700 births
- Maternal age >35 carries increased risk



Hearing Concerns

- Conductive hearing loss
  - more common
  - small pinna
  - stenotic EAC
  - eustachian tube dysfunction
  - ossicular fixation
- Sensorineural hearing loss
  - less common
  - ossification of basal spiral tract

C.H.A.R.G.E. RC

C.H.A.R.G.E.

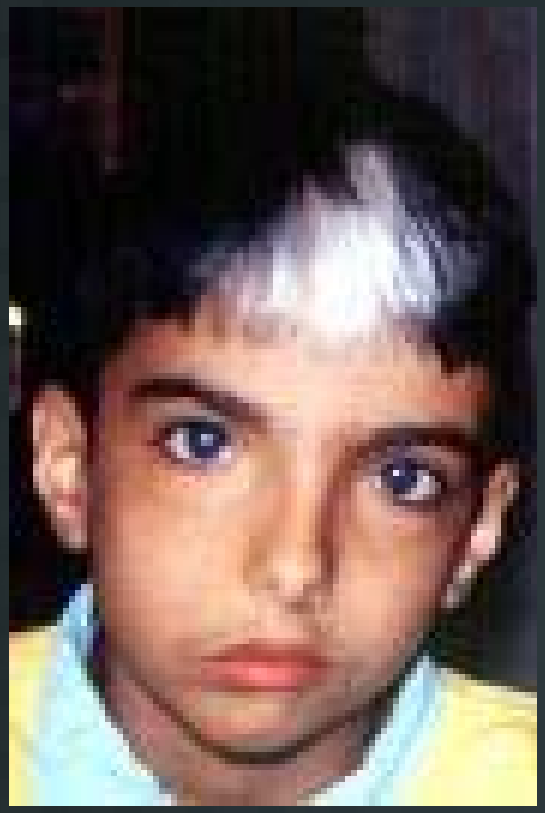
- Coloboma
- Heart defect (tetralogy of Fallot,ASD,VSD)
- Atretic choanae
- Retarded growth
- Genitourinary anomalies
- Ear malformations

Hearing Concerns

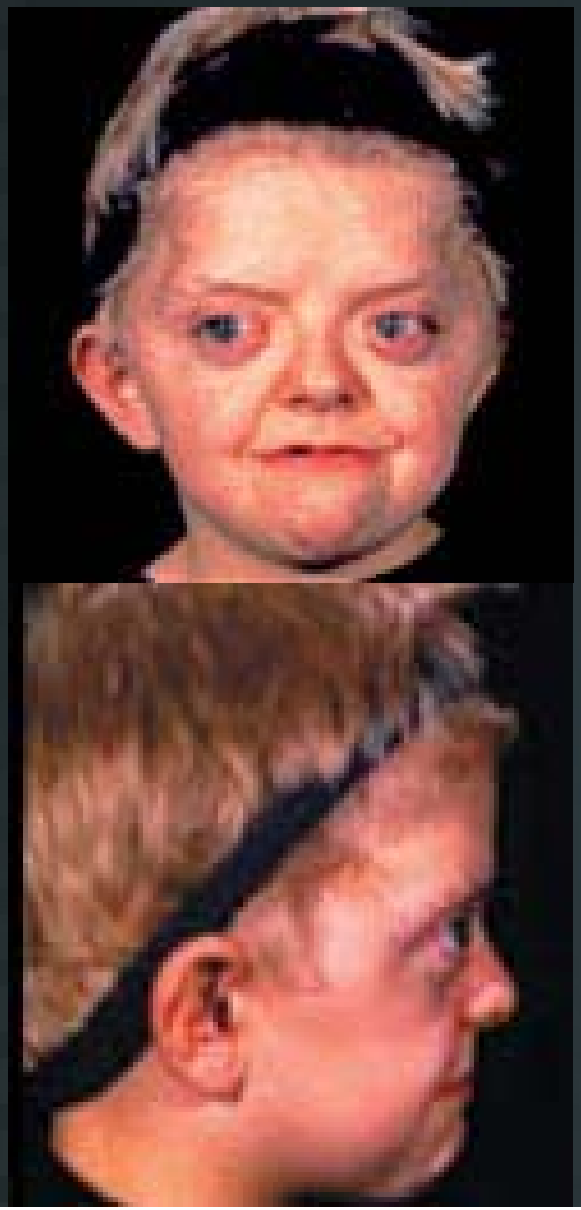
- External ear anomalies (vary widely)
- Deafness is of mixed type
- Middle ear anomalies
  - absence of stapes
  - abnormal incus
  - absence of oval window
- Inner ear anomalies
  - Mondini
  - dysplasia of pars inferior
  - absence of pars superior
  - short cochlea


What
do you
think?

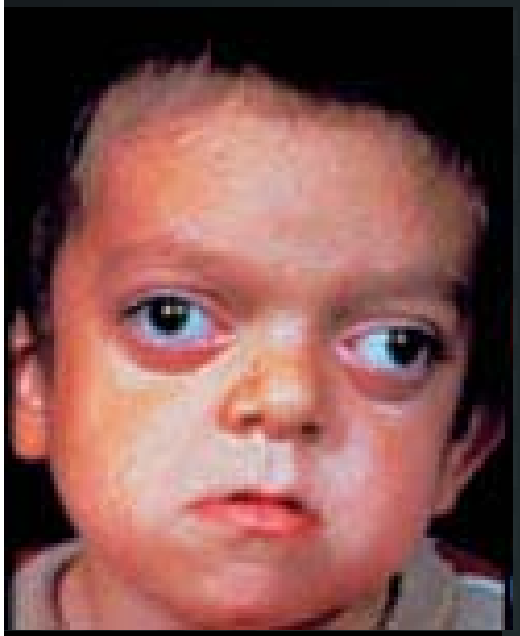
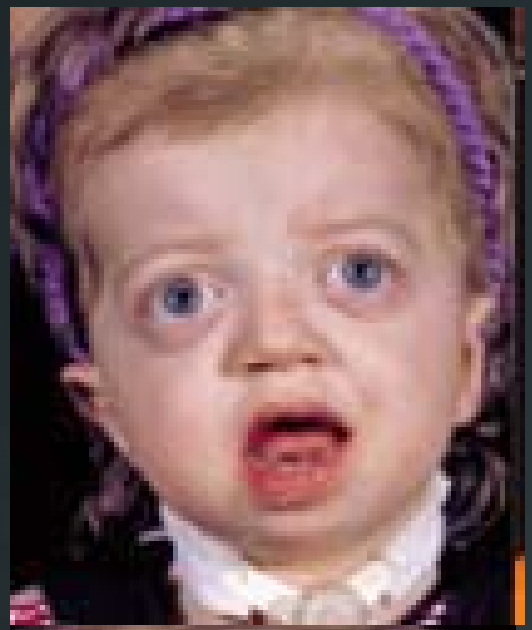
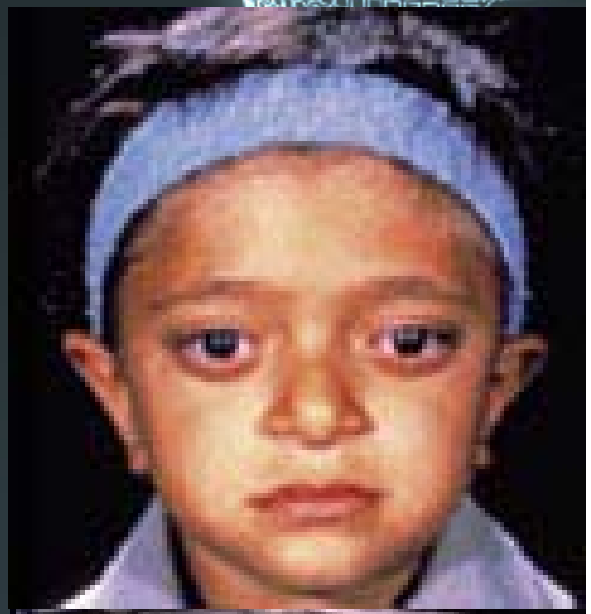
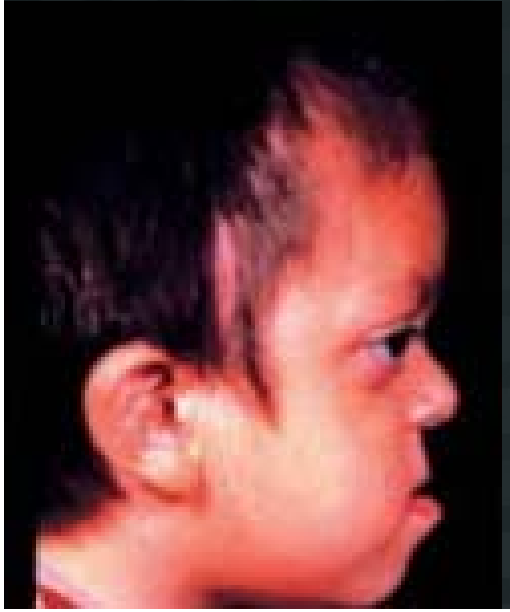
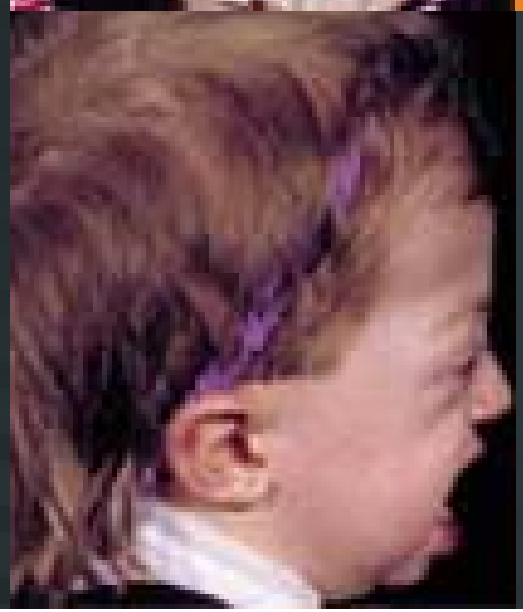
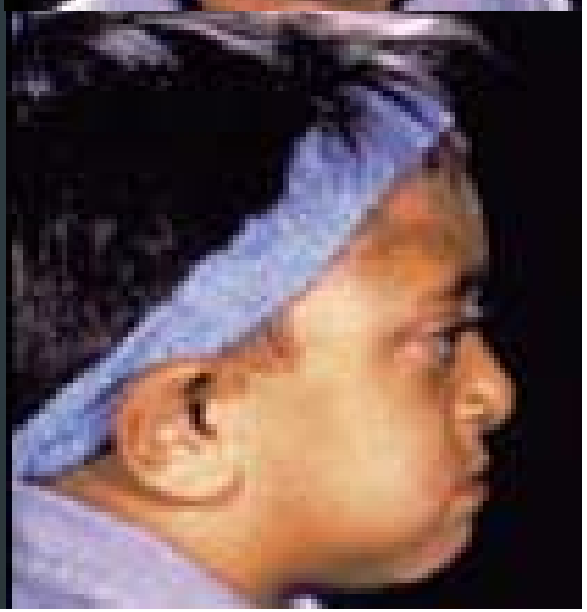
Waardenburg Syndrome



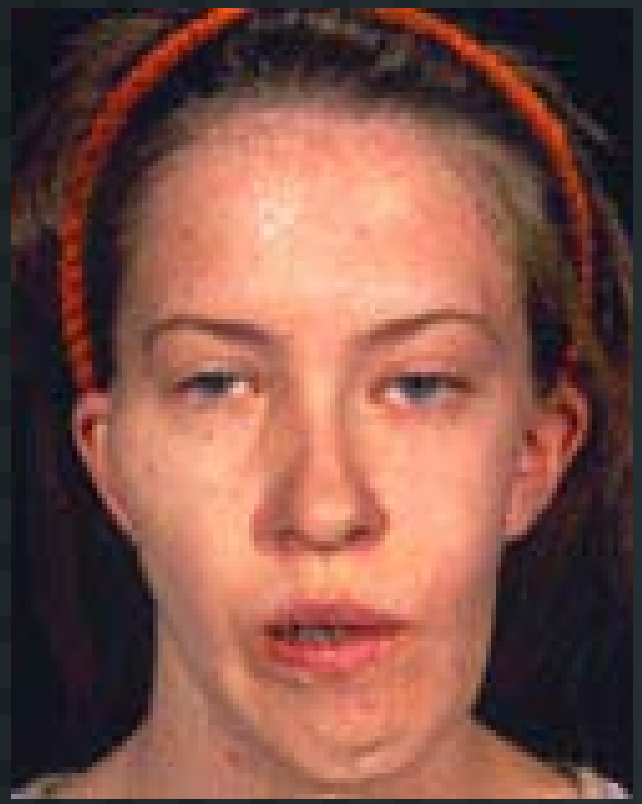
Aperts Syndrome

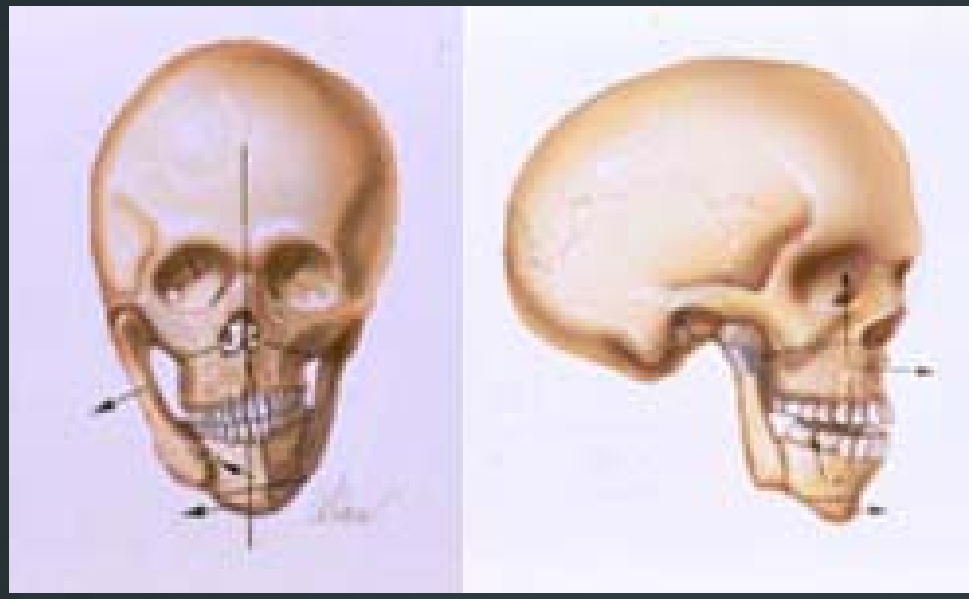


Crouzons Syndrome

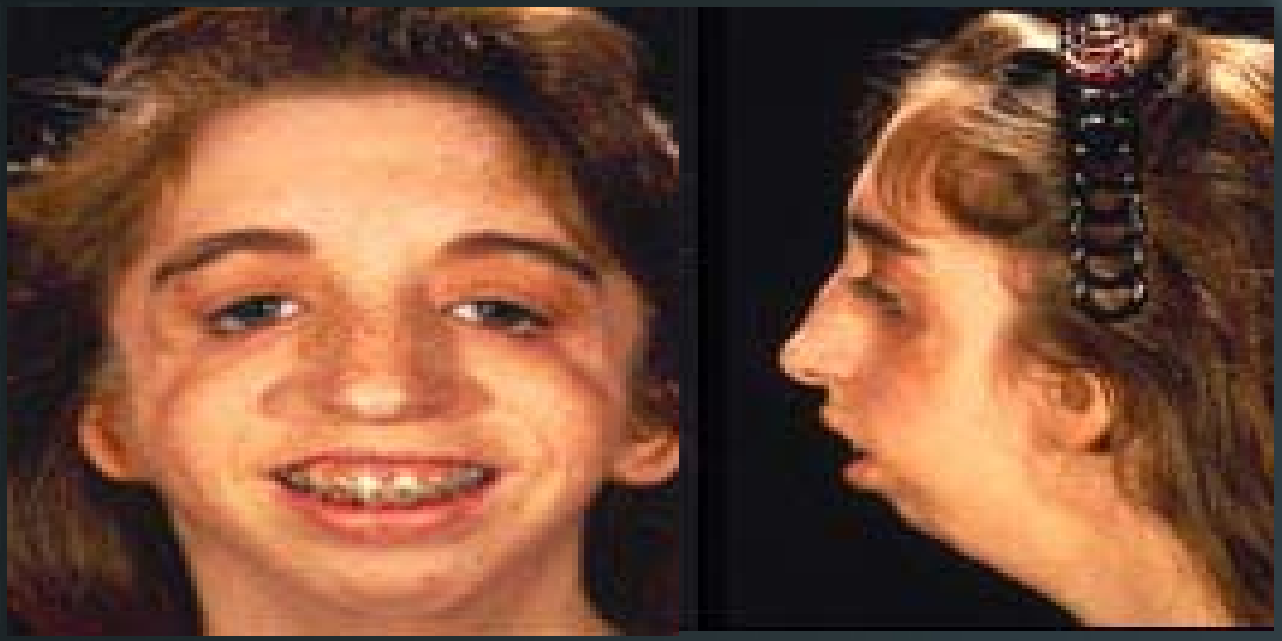







Goldenhar Syndrome





Treacher Collins



Autosomal Dominant

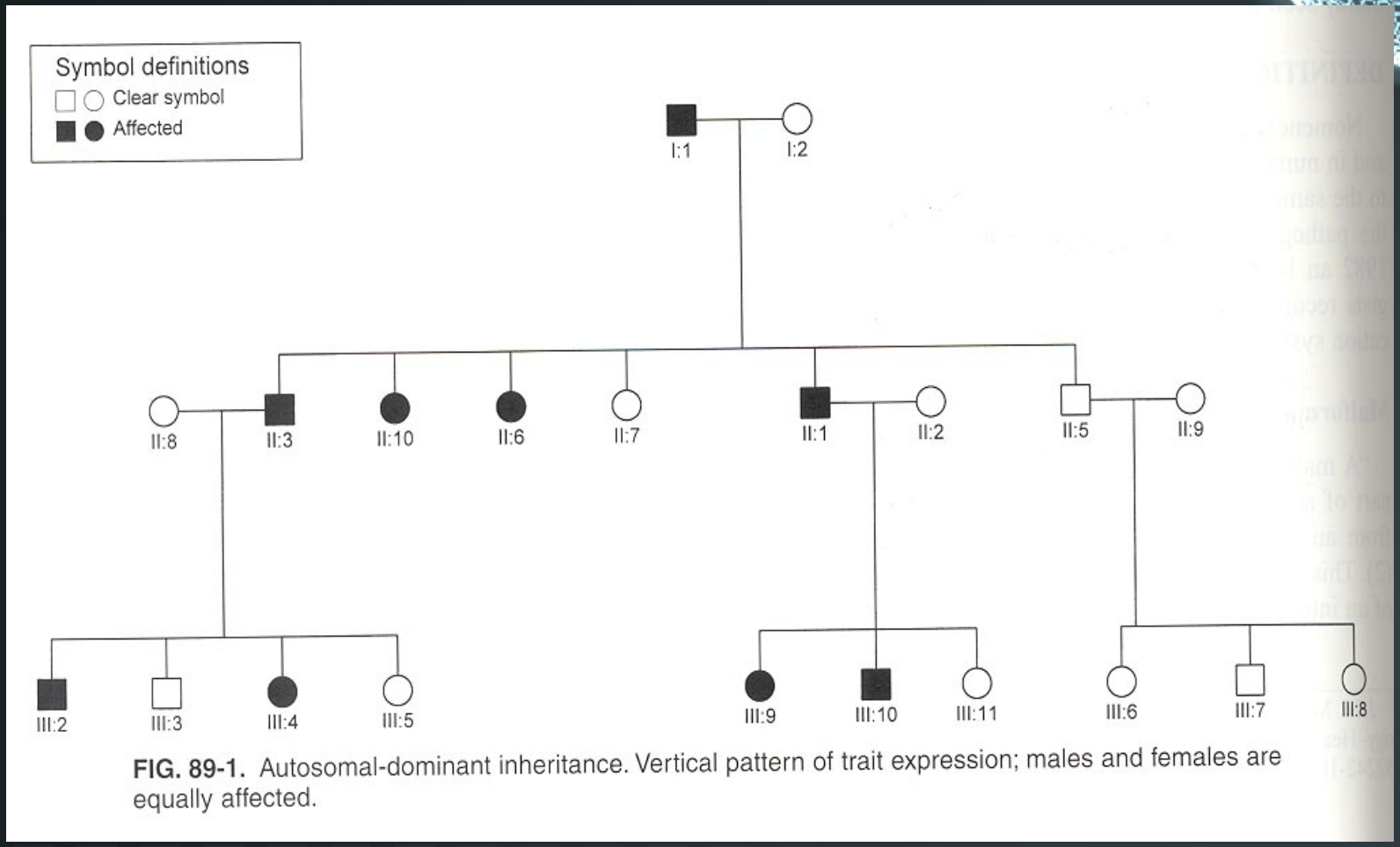
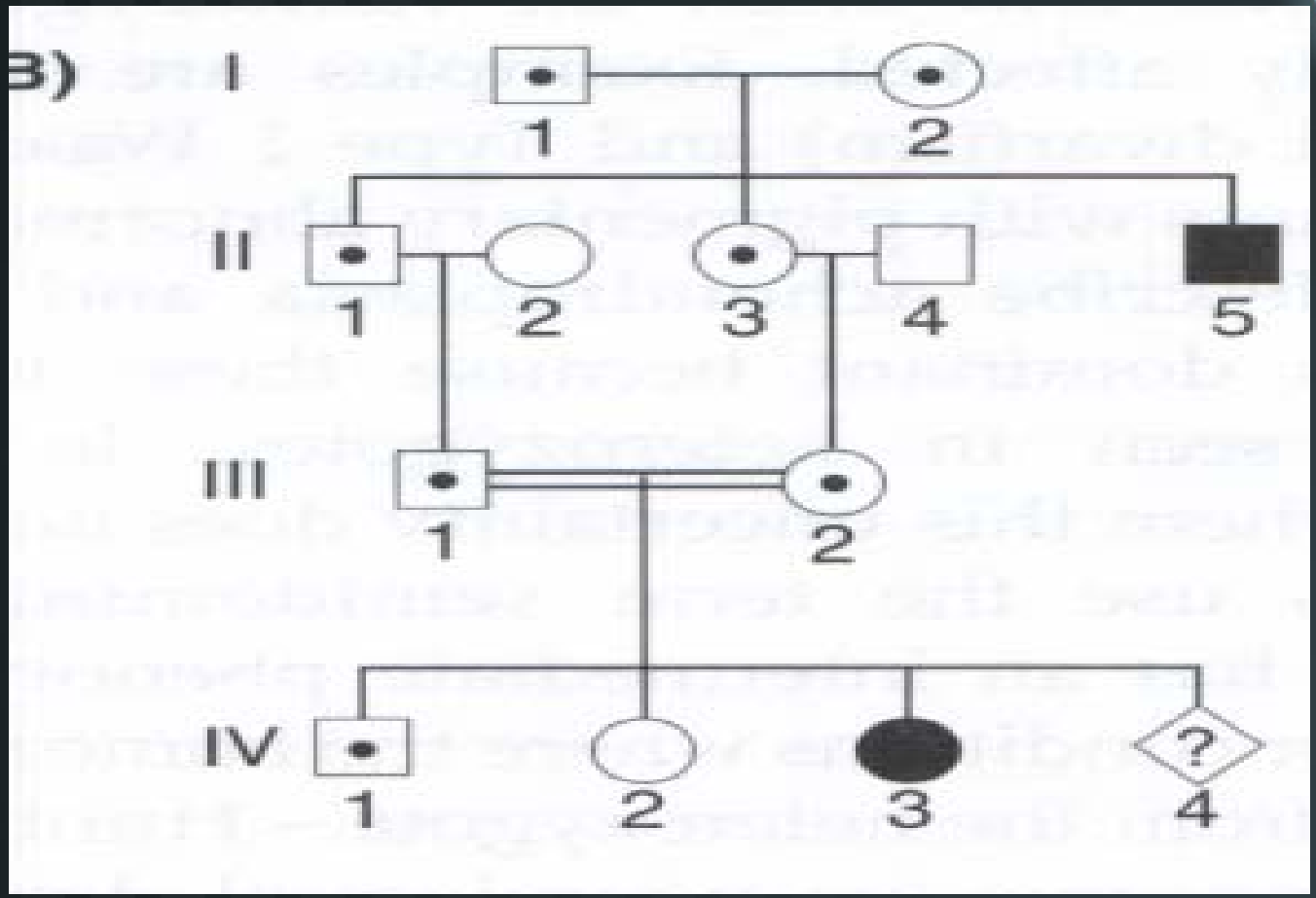


FIG. 89-1. Autosomal-dominant inheritance. Vertical pattern of trait expression; males and females are equally affected.

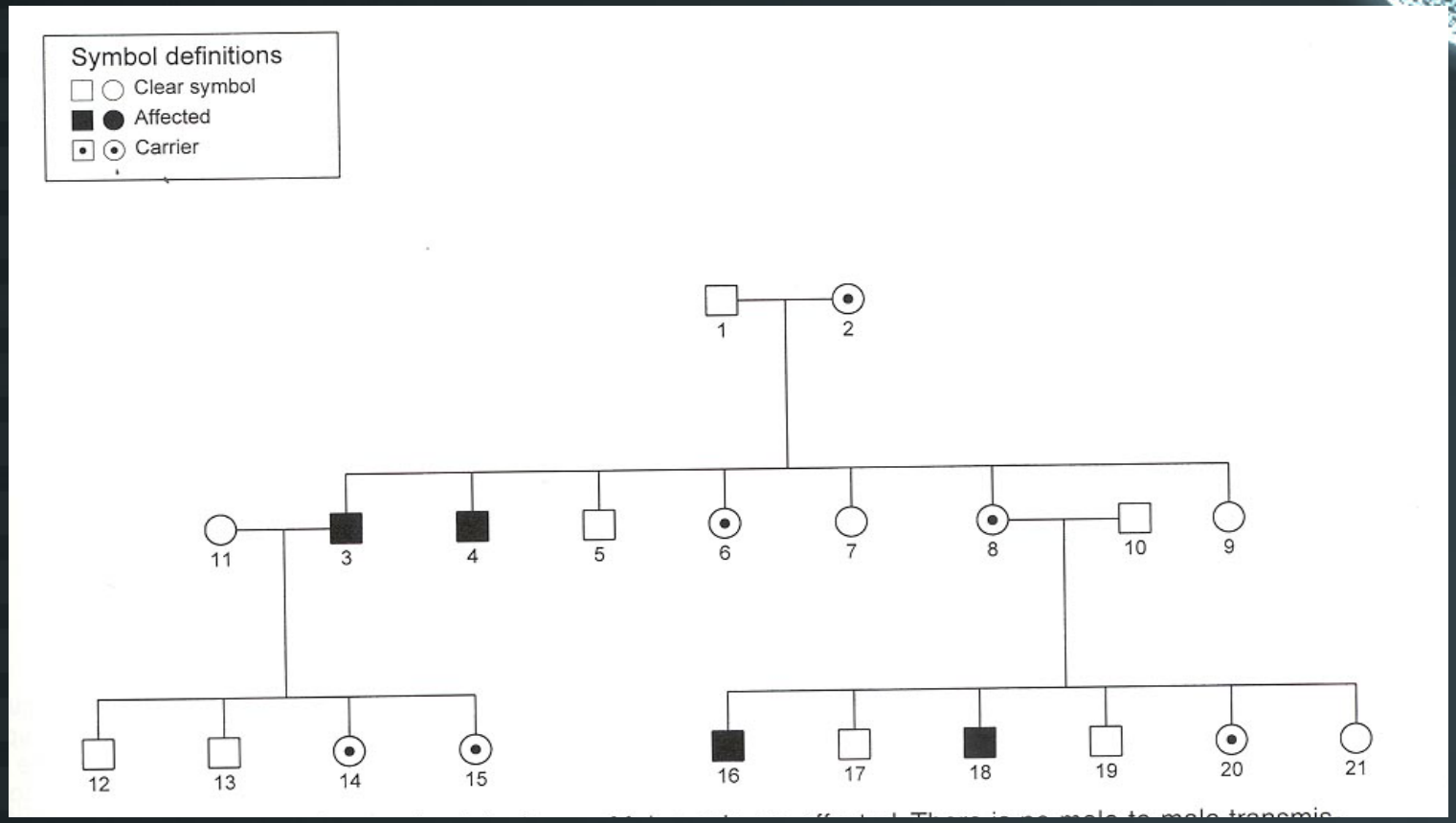
RC

Autosomal Recessive



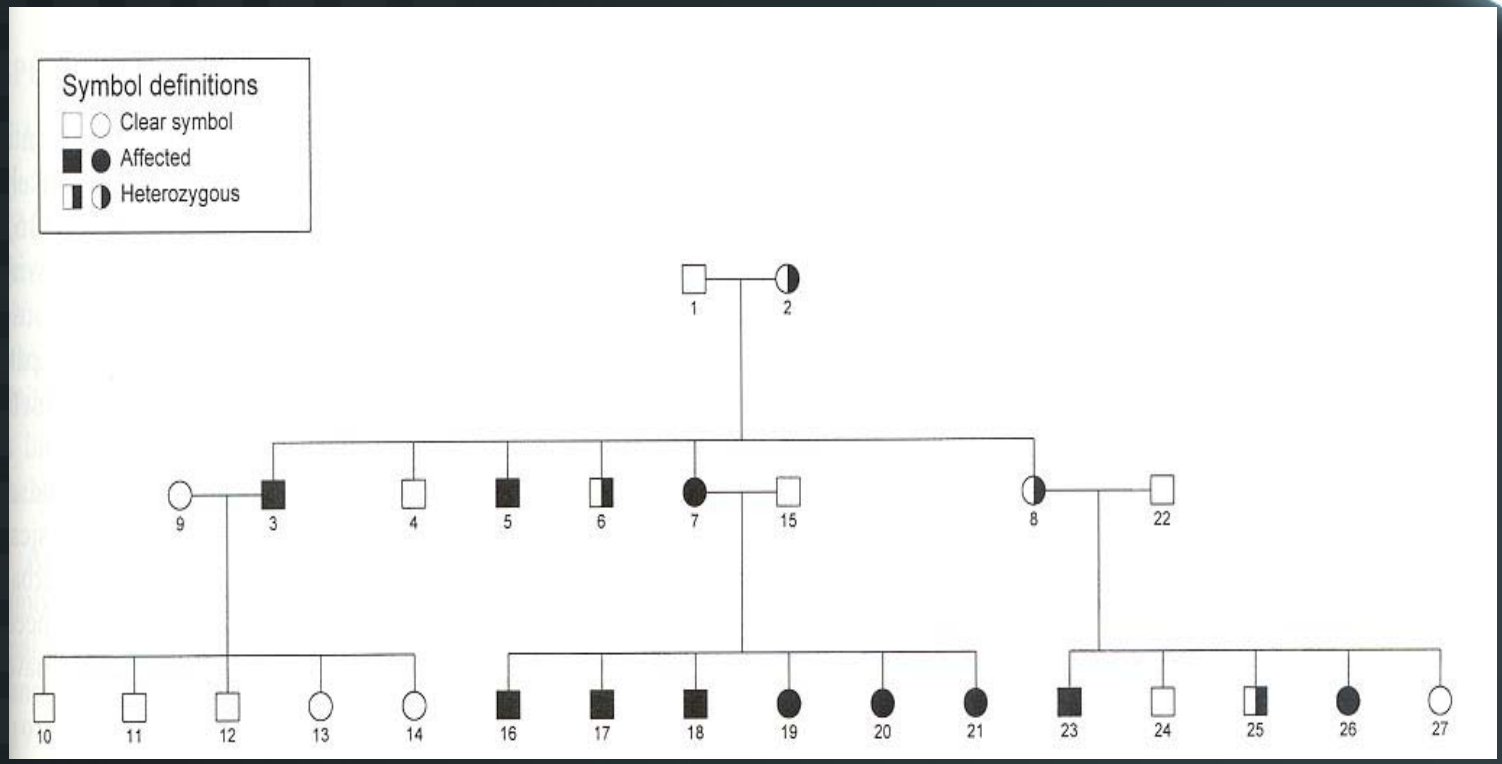
RC

X-Linked Inheritance



RC

Mitochondrial RC



RC


Thank
You