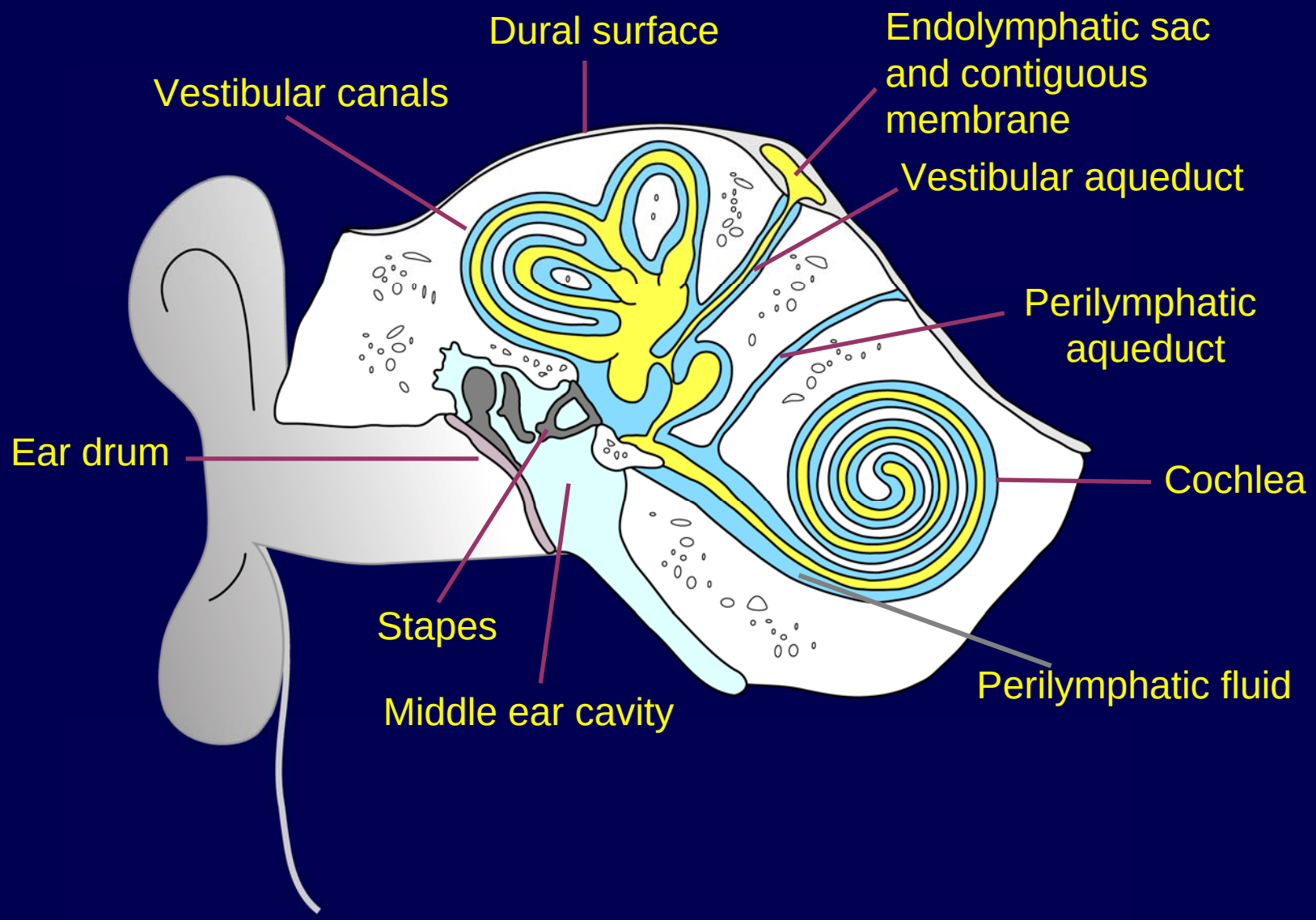
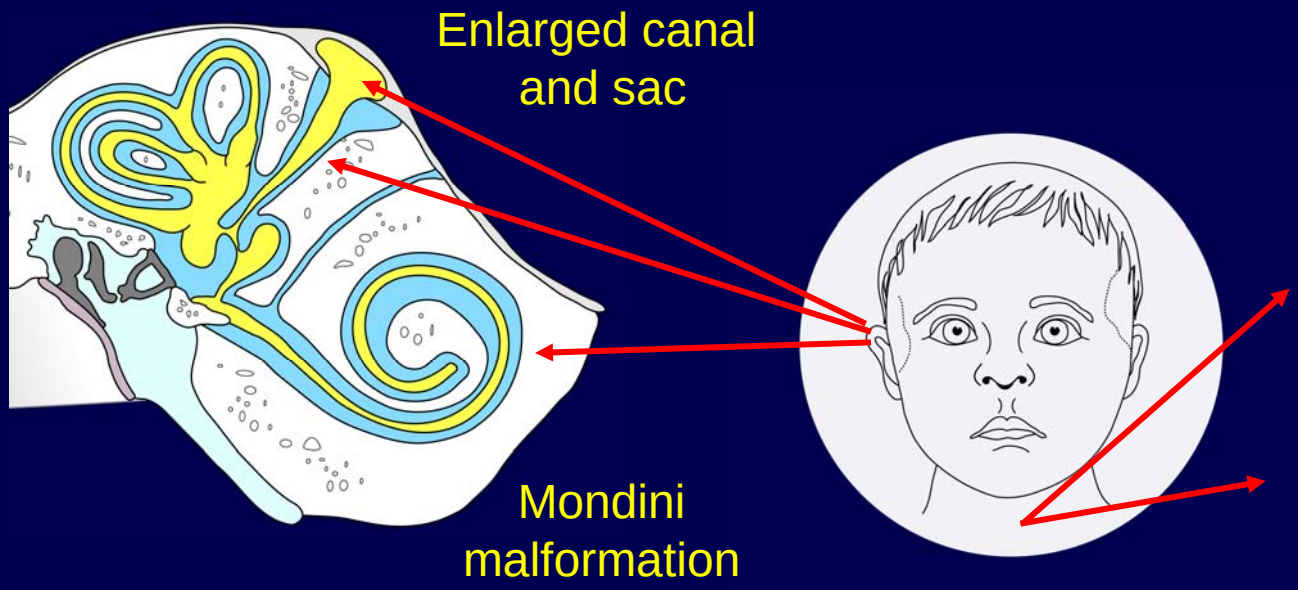
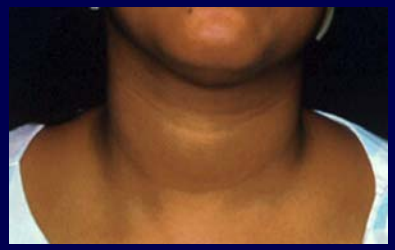
Hearing Loss Syndromes

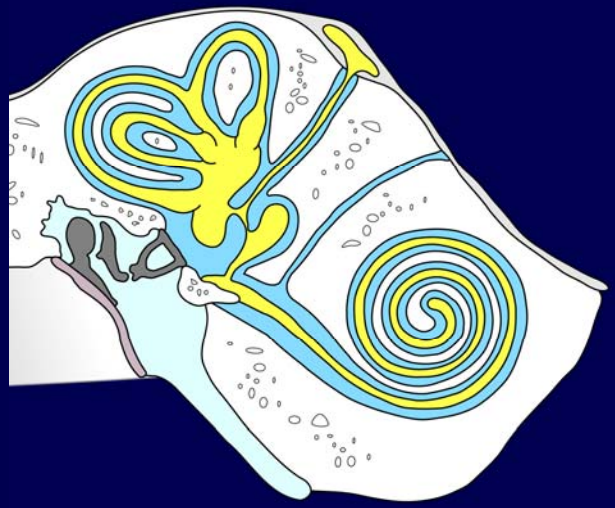
- Account for 20-30% of genetic HL
- >400 syndromes
- Each syndrome is relatively rare
- Syndromes identified by:
  - Physical examination findings
  - Internal ear malformations
  - Physiological traits (e.g., ECG changes)

Anatomy Review

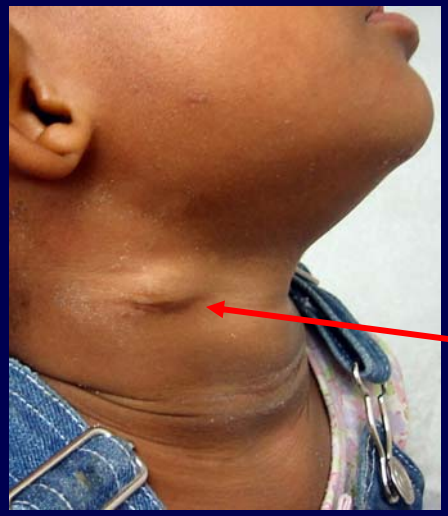


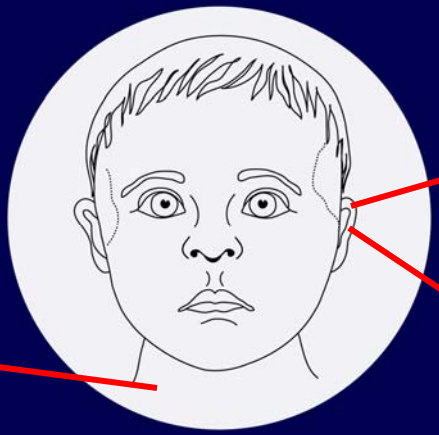


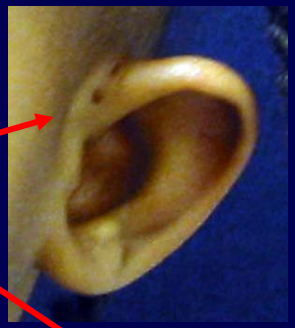


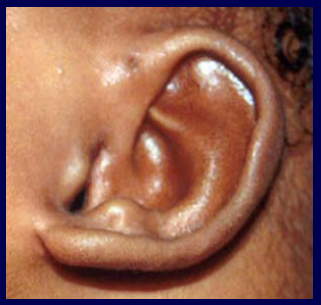


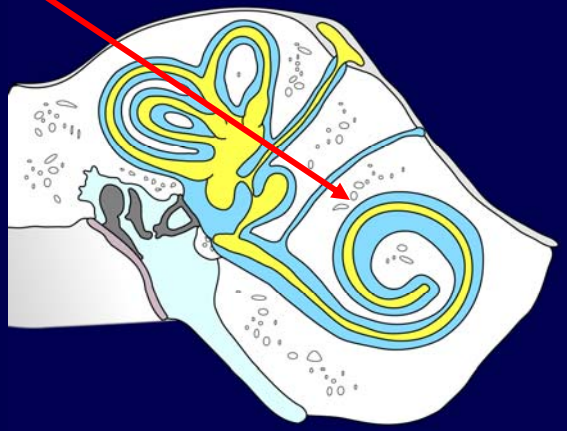
- Pendred Syndrome
  - Hearing loss, usually prelingual
  - Goiter in 2nd decade, most are euthyroid
  - Large vestibular aqueduct and endolymphatic sac
  - Upper 2/3 of cochlea is poorly formed









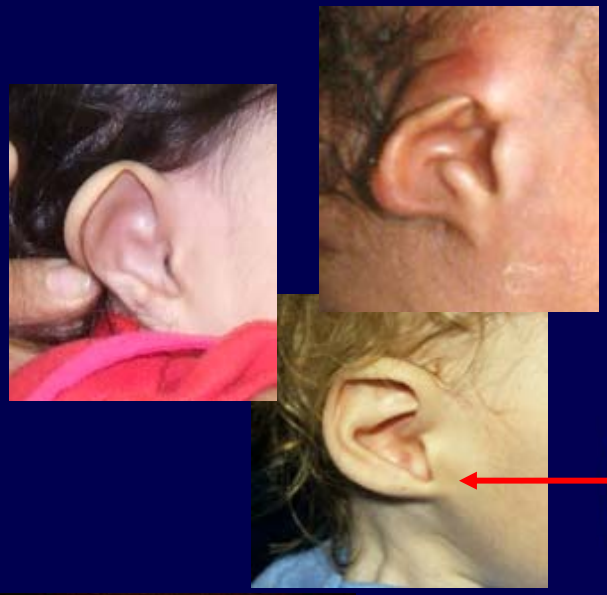


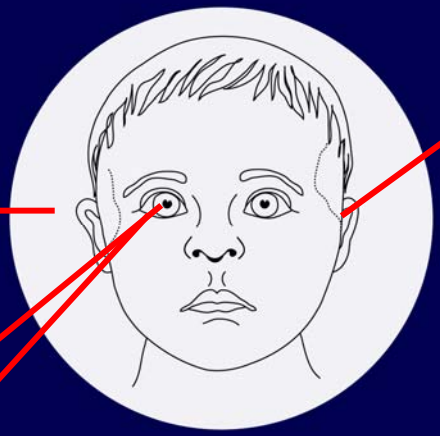
BOR syndrome

Branchio
- fistulas, sinuses, cysts

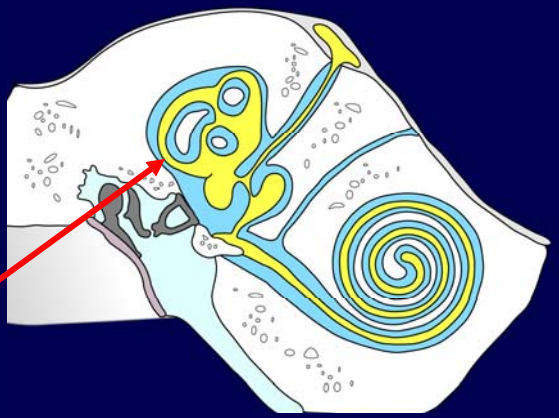
Oto
- cupped ears, pits, tags
- Mondini anomaly
- malformed middle ear ossicles
- semicircular canal hypoplasia

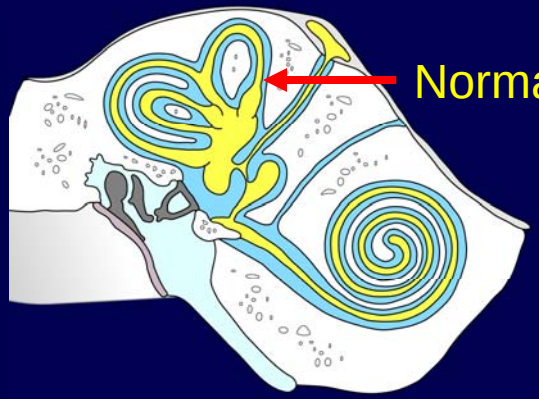
Renal
- hypoplasia to bilateral renal agenesis



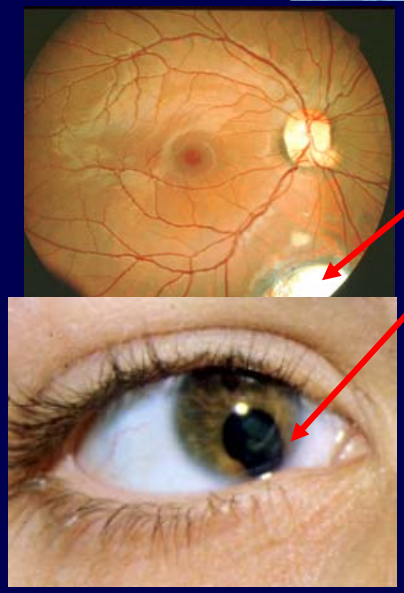


Semicircular canal hypoplasia





Normal



CHARGE Syndrome

Coloboma of the iris or retina

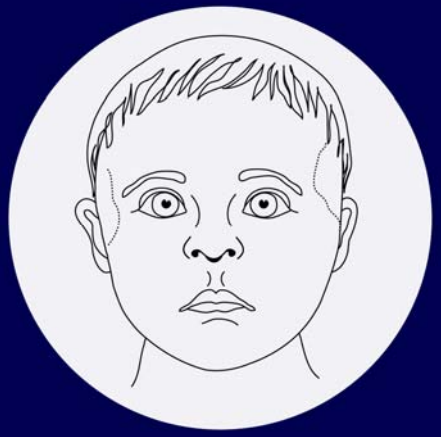
Heart defects

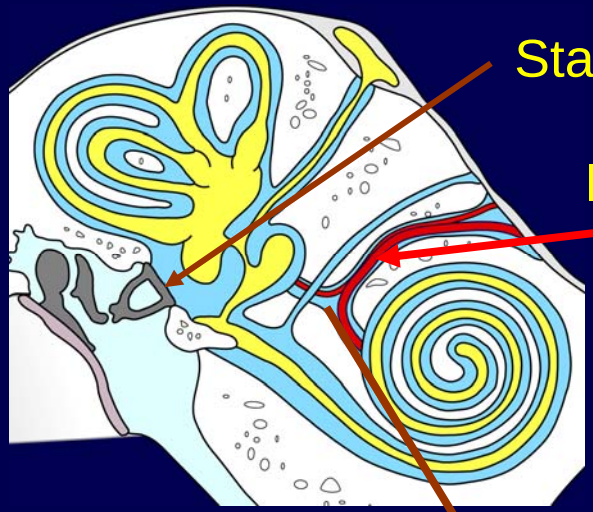
Atresia or stenosis of the choanae

Retarded growth and development

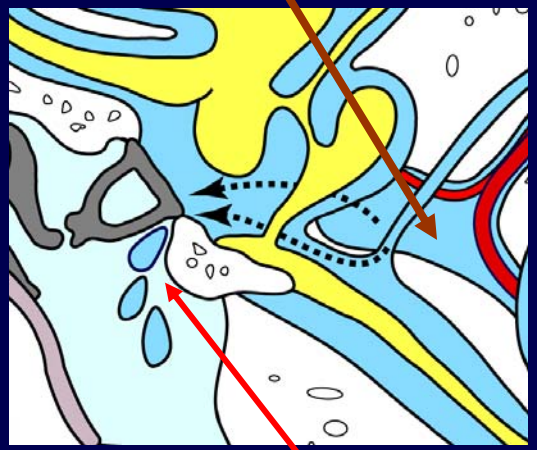
Genital hypoplasia in males

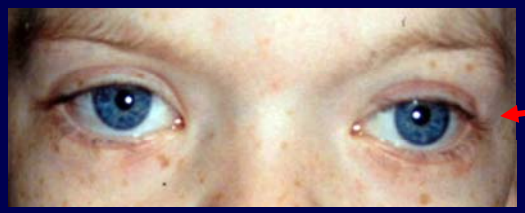
Ear anomalies

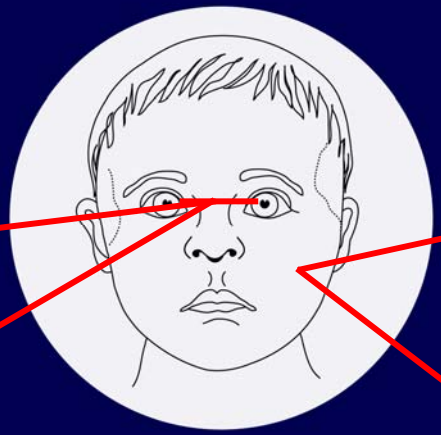


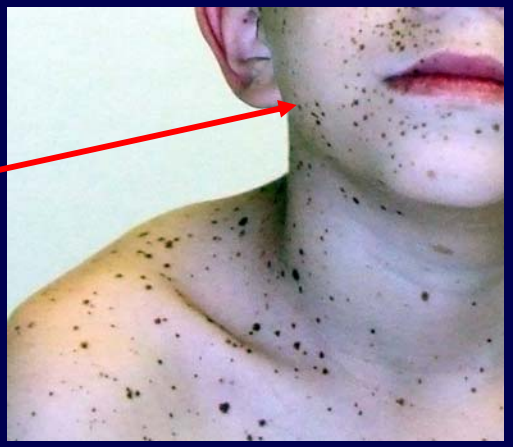


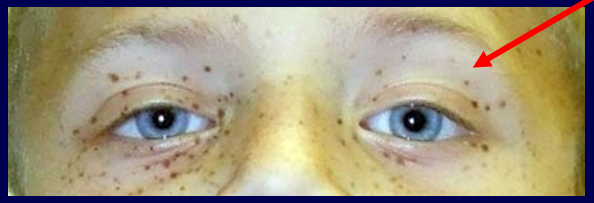
- Perilymphatic Gusher-Deafness syndrome
  - Conductive hearing loss
  - Stapes fixation
  - Perilymphatic gusher during stapes surgery

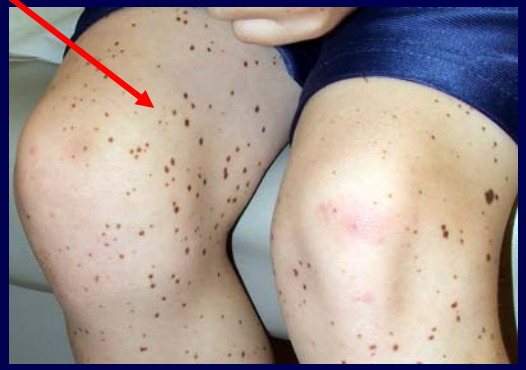






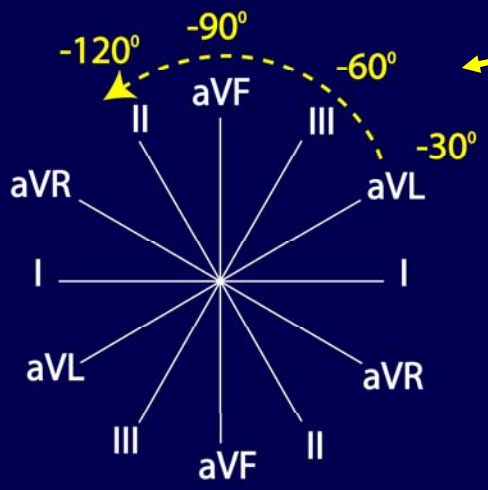




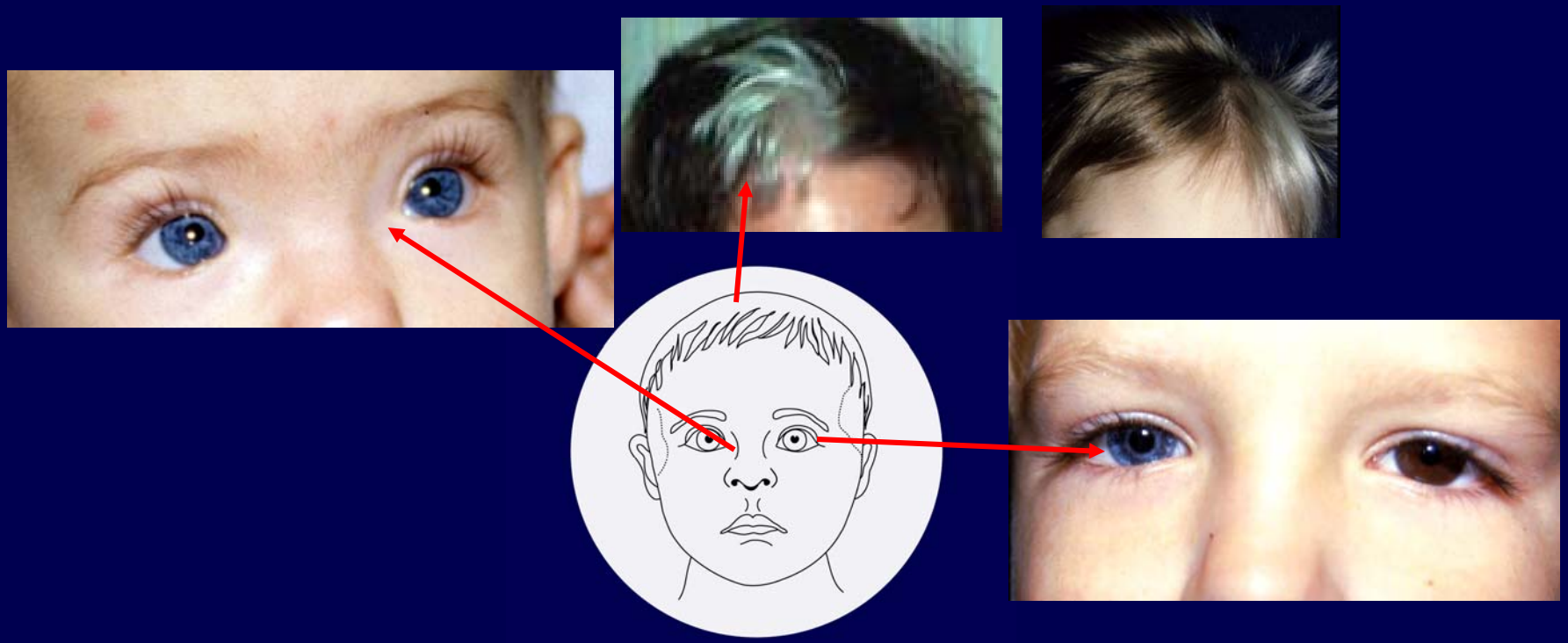


LEOPARD Syndrome

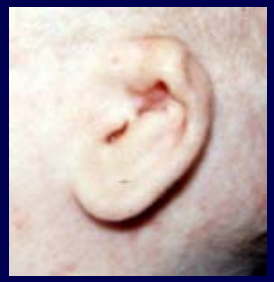
- Lentigines
- EKG abnormalities
- Ocular hypertelorism
- Pulmonic stenosis
- Abnormalities of genitalia
- Retardation of growth
- Deafness

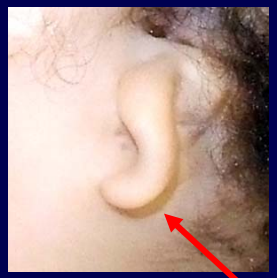


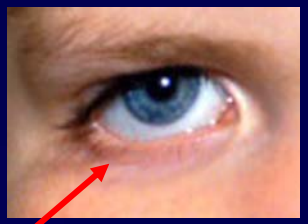
Left axis deviation

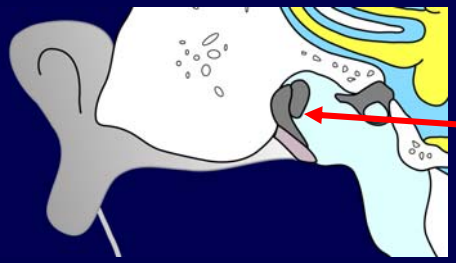


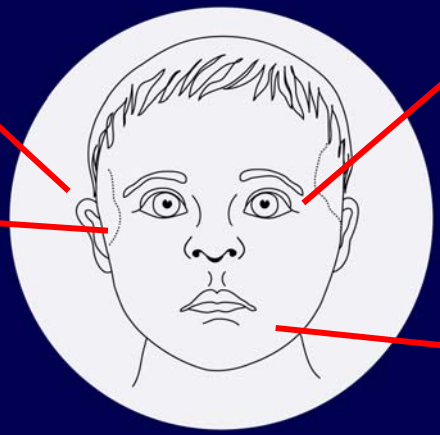
- Waardenburg Syndrome
  - Dystopia canthorum
  - White forelock
  - Hypochromic irides, heterochromia
  - Sensorineural deafness

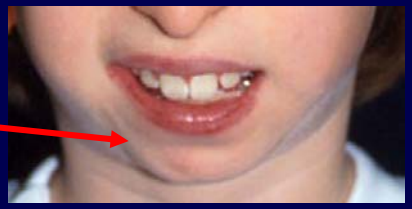


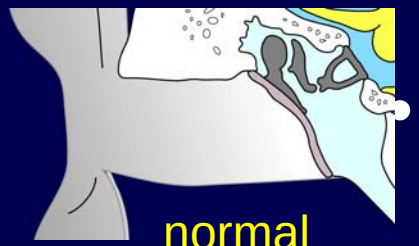












Treacher Collins Syndrome

- Down-slant of palpebral fissures, lower lid coloboma
- Hypoplasia of maxilla, micrognathia
- Dysmorphic ears
- Atresia of middle-ear ossicles, conductive hearing loss

Genetic Causes

Syndrome	Gene/s	Inheritance
Pendred	SLC26A4	AR
BOR	EYA1 SIX5	AR
CHARGE	CHD7	AD
Perilymph Gusher	POU3F4	X-linked
LEOPARD	PTPN11 RAF1	AD
Waardenburg	PAX3 MITF1 SNAI2 SOX10	AD
Treacher Collins	TCOF1	AD

Other Syndromic Conditions

- Alport
  - hematuria
- Stickler
  - retinal detachment
- Usher
  - retinitis pigmentosa
- Jervell and Lange-Nielsen
  - long PR interval on ECG
- KID (keratitis, ichthyosis, deafness)
  - Skin hyperkeratosis