

# HL due to Connexin-26 gene defects

- Account for 10% of all childhood hearing loss!
- Clinical features:
  - Normal physical exam
  - Normal vestibular function
  - Normal inner ear structure
- Most genetic HL conditions have a normal physical exam

# Common areas of involvement

- Craniofacial area
  - Ears
  - Eyes
  - Neck
- Skin and hair
- Other regions

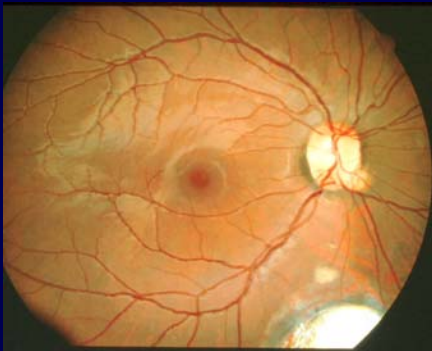
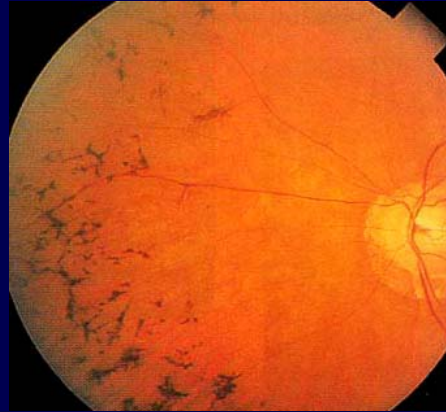
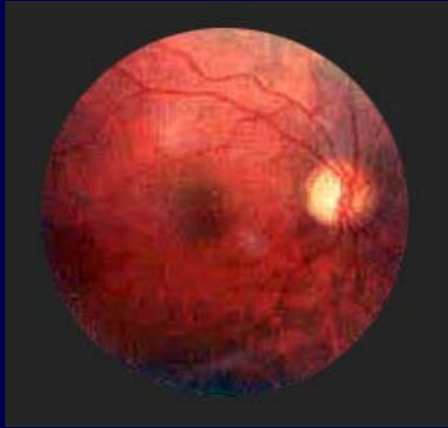
# Ear Abnormalities



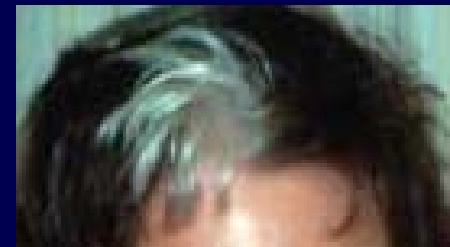
Normal



# Eye Abnormalities



# Skin and Hair



# Neck



# Evaluation of HL

- External physical exam
- Other factors
  - Middle and inner ear structure
  - Age of onset
  - Audiogram profile
  - Family history
  - Type of hearing loss
  - Environmental exposures