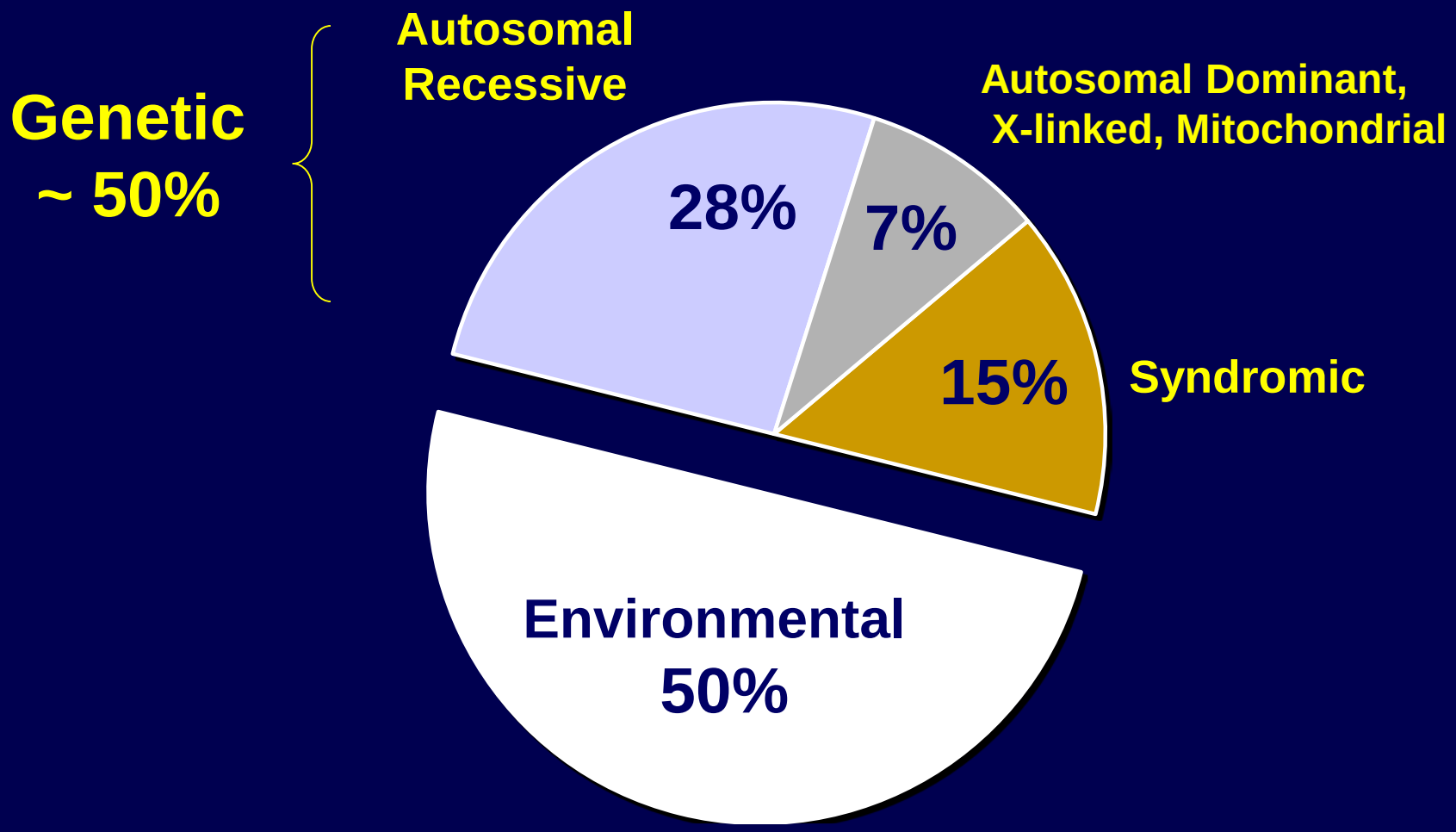
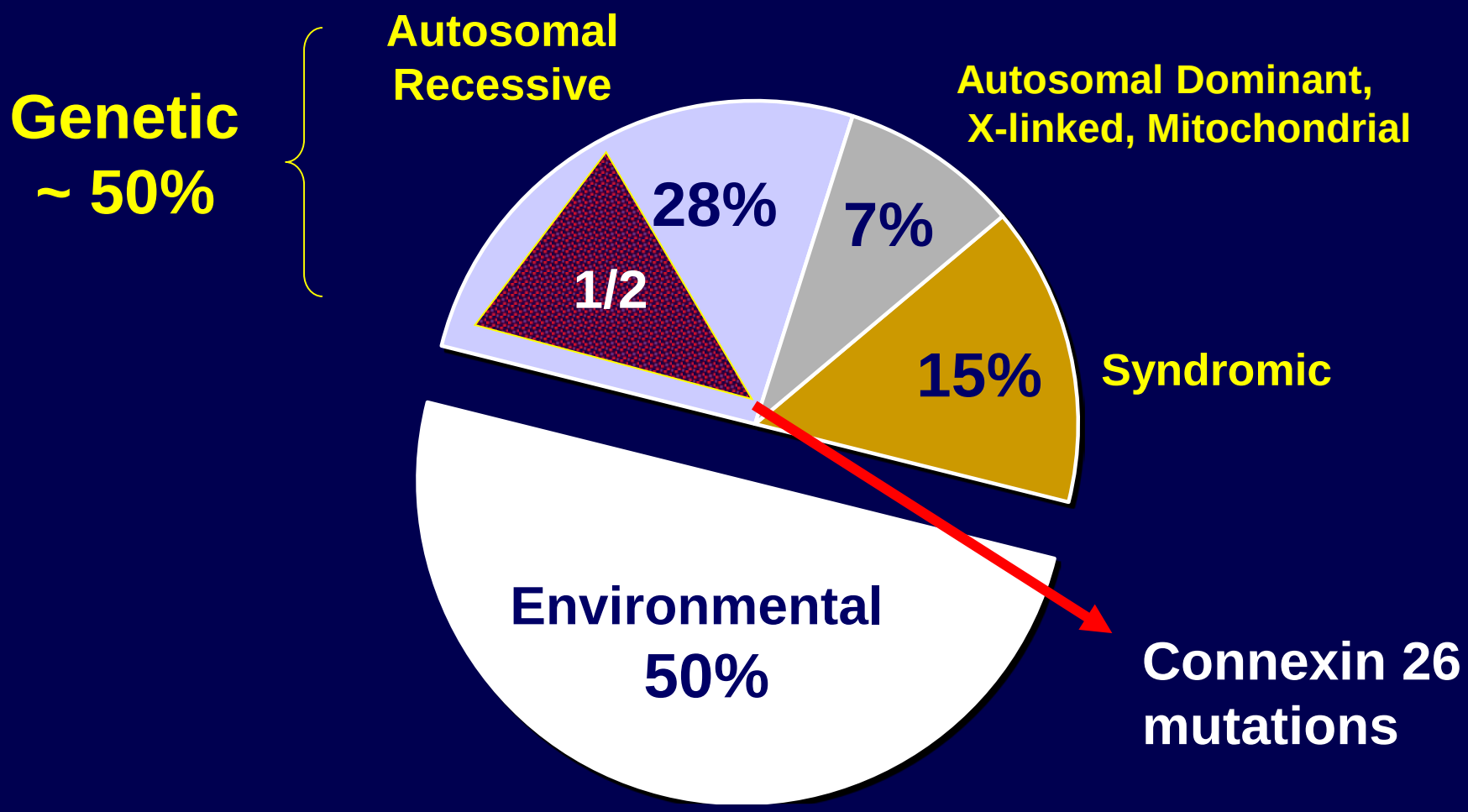
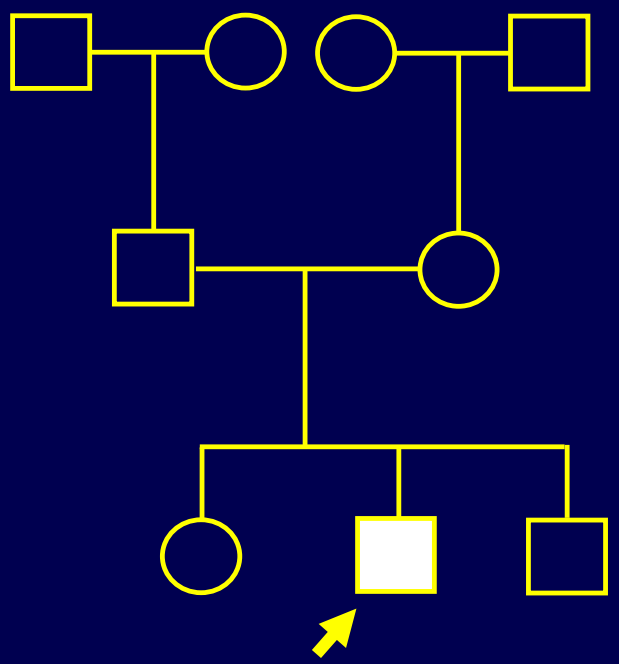
Hearing Loss and GJB2 (Connexin 26)

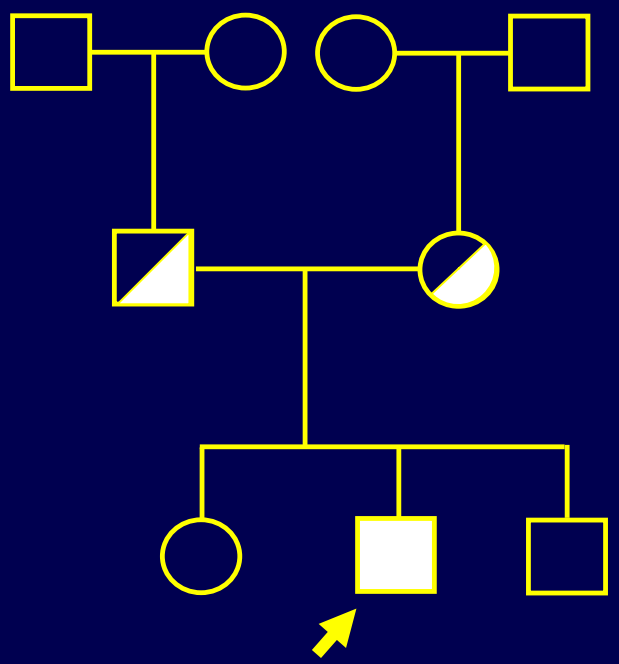
Causes of Hearing Loss

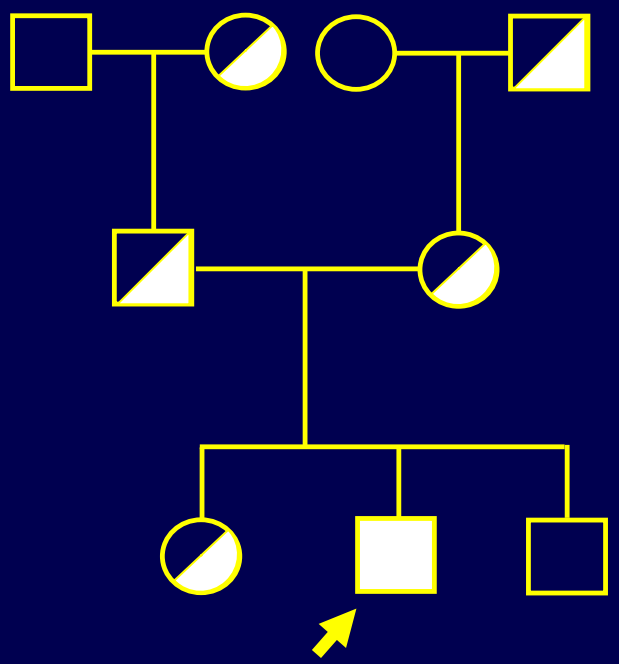


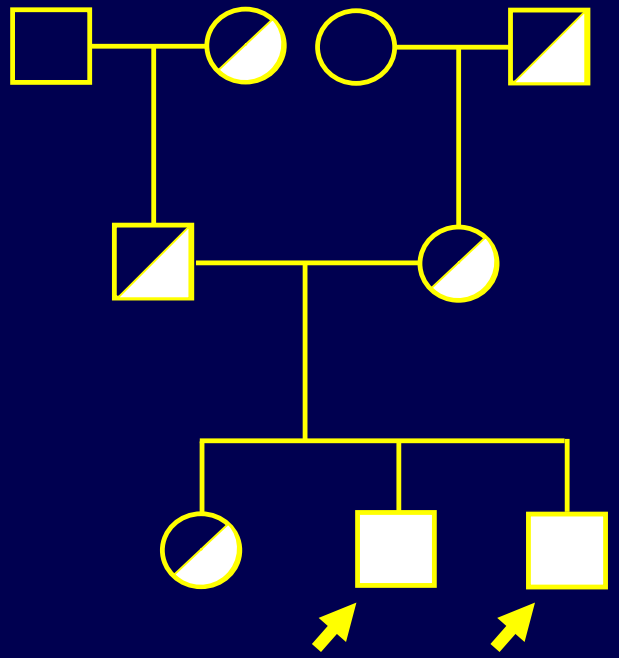
Causes of Hearing Loss





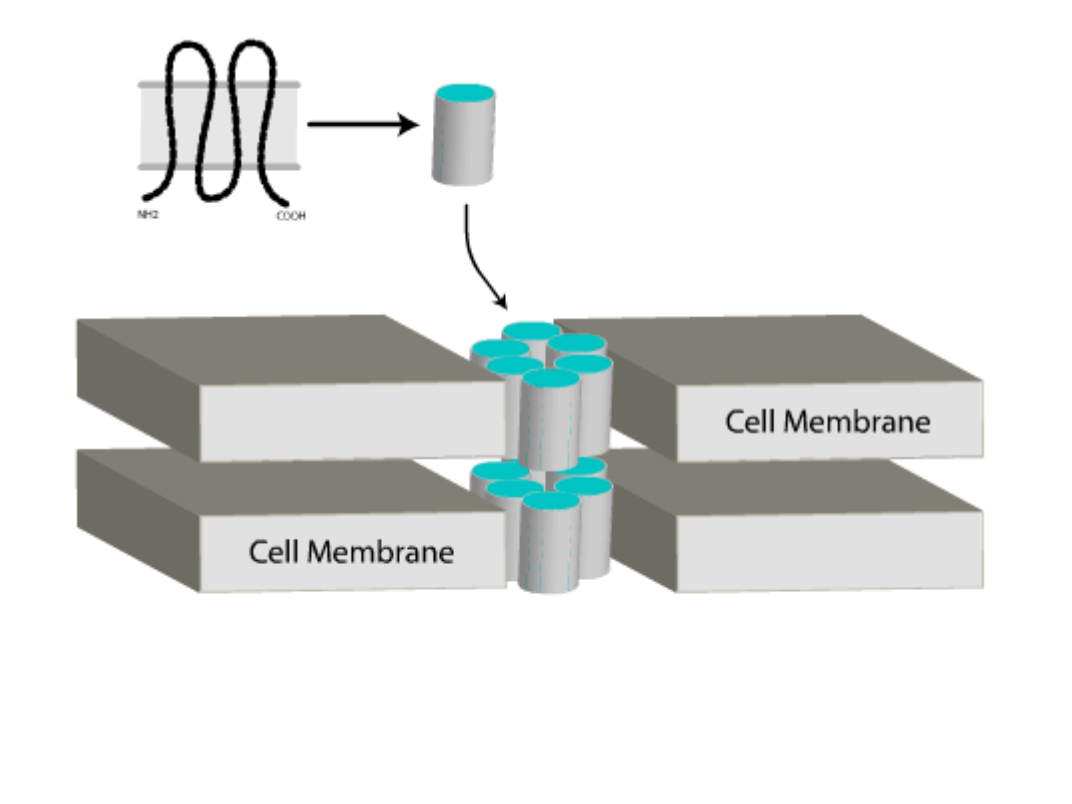


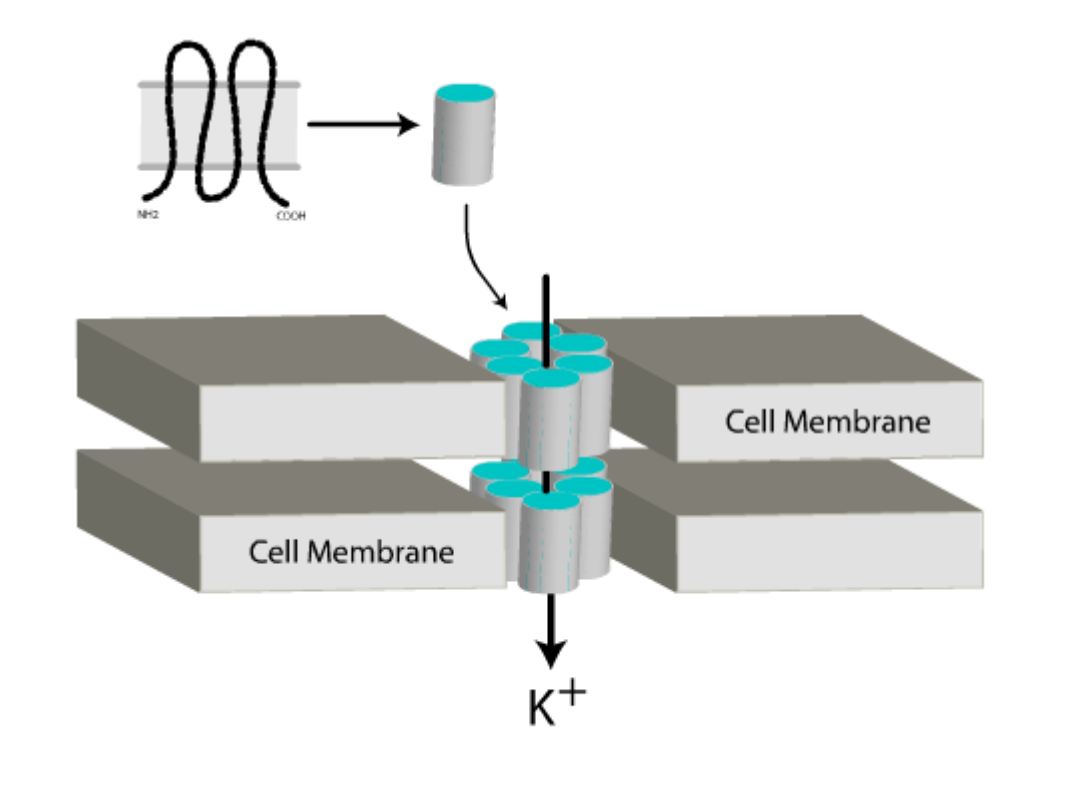


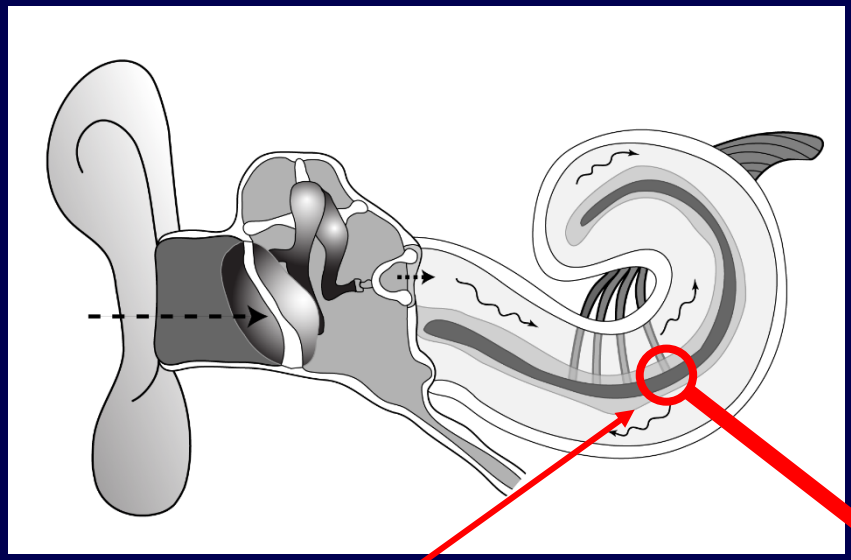


The Connexin-26 Gene and its Protein

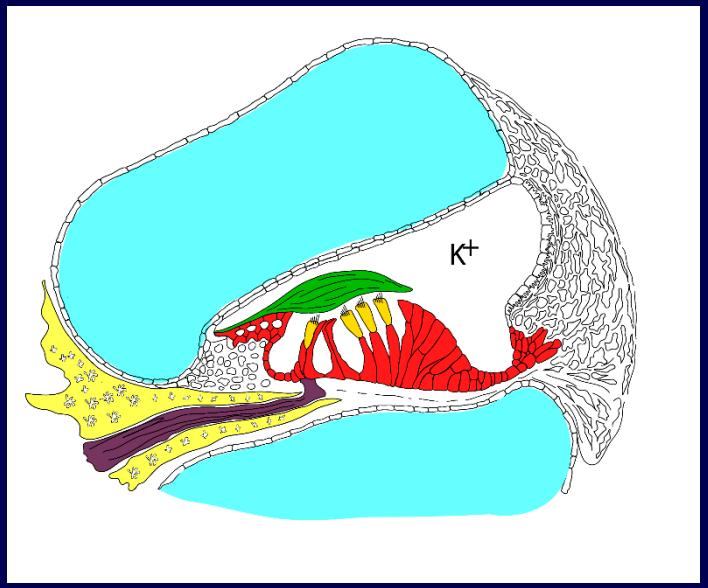
Connexin-26 is a gap junction membrane protein



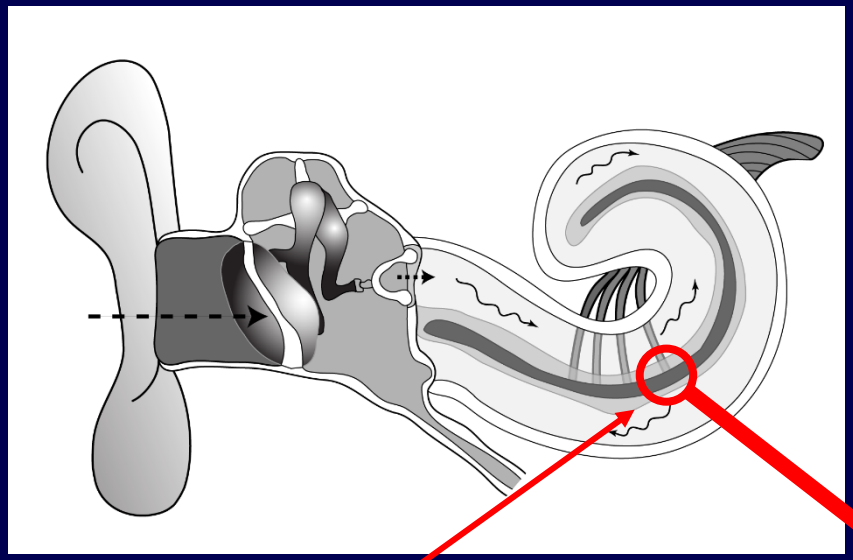

NH2
COOH
Cell Membrane
Cell Membrane
K+



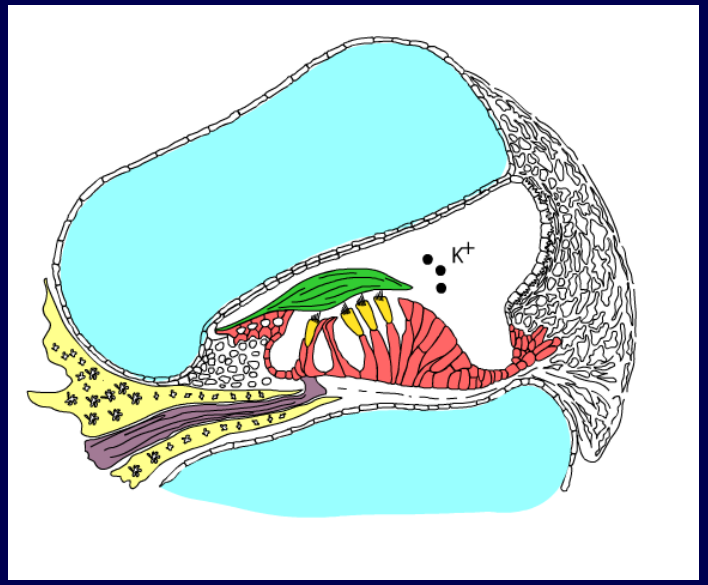
Organ of Corti in the cochlea



Recycling of K^+ in the inner ear

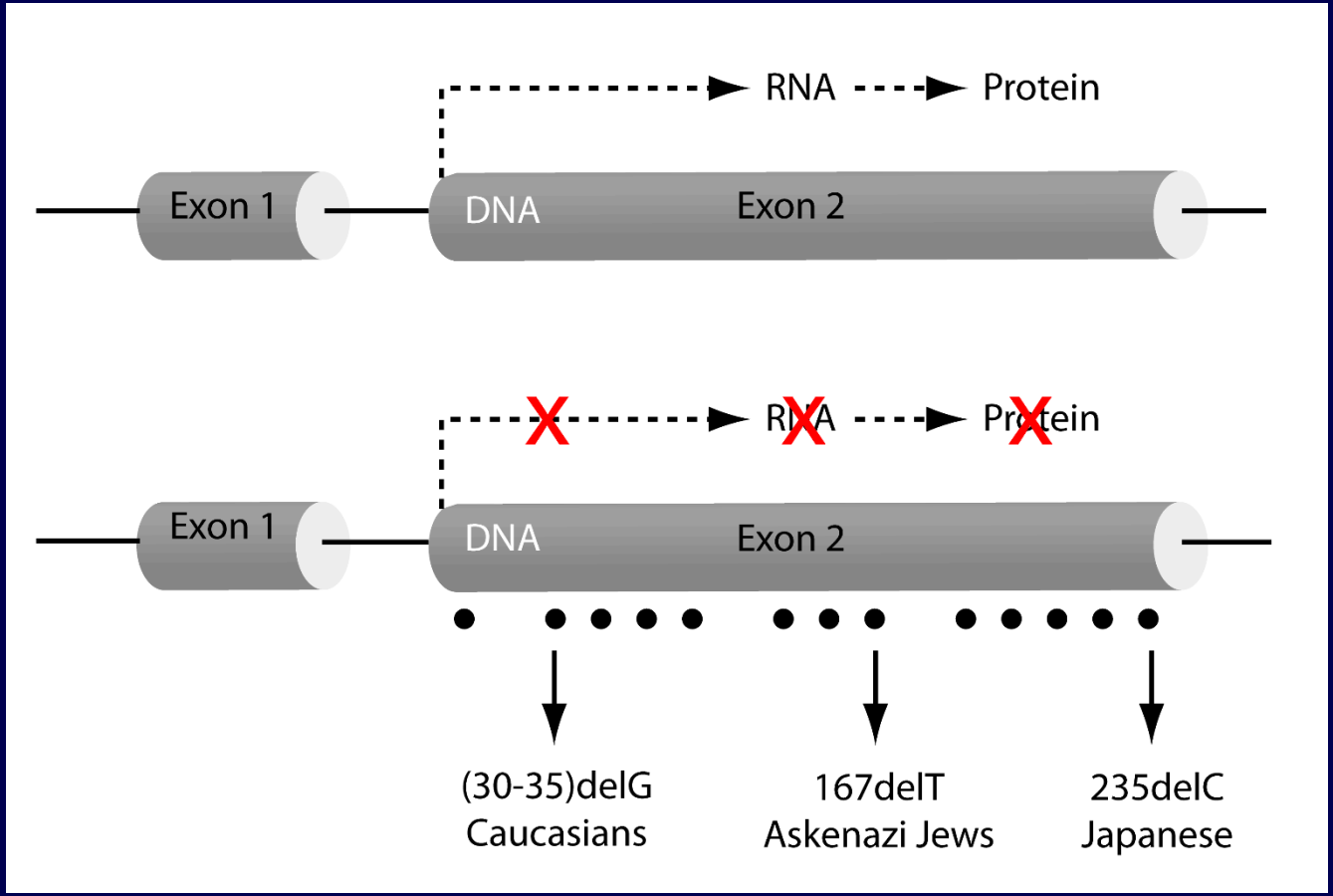


Organ of Corti in the cochlea



Recycling of K^+ in the inner ear

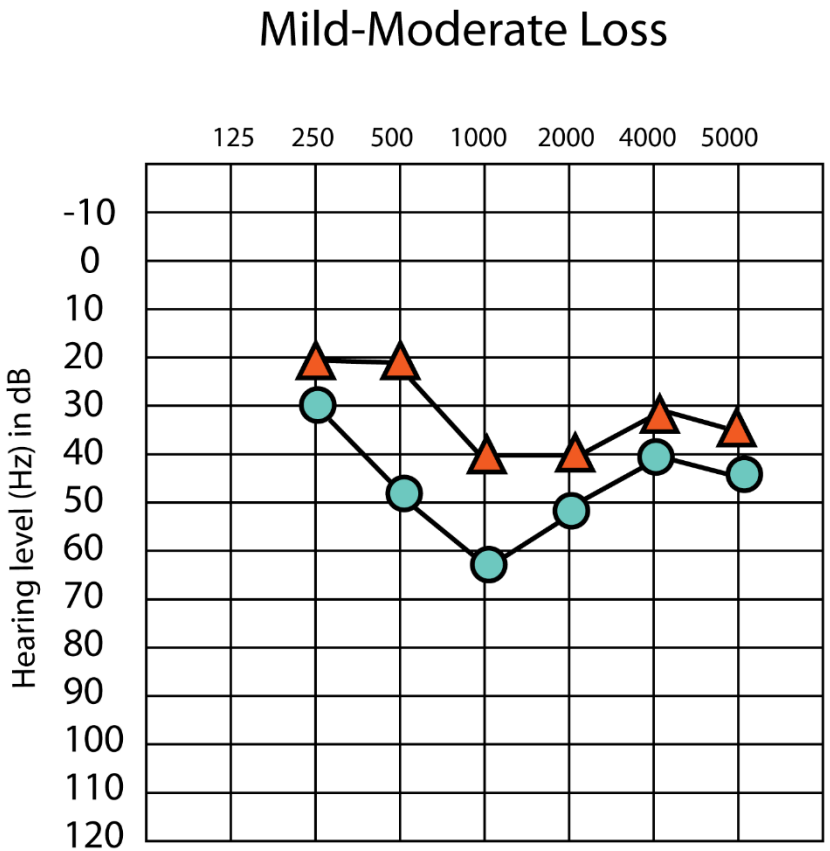
Connexin 26 mutations

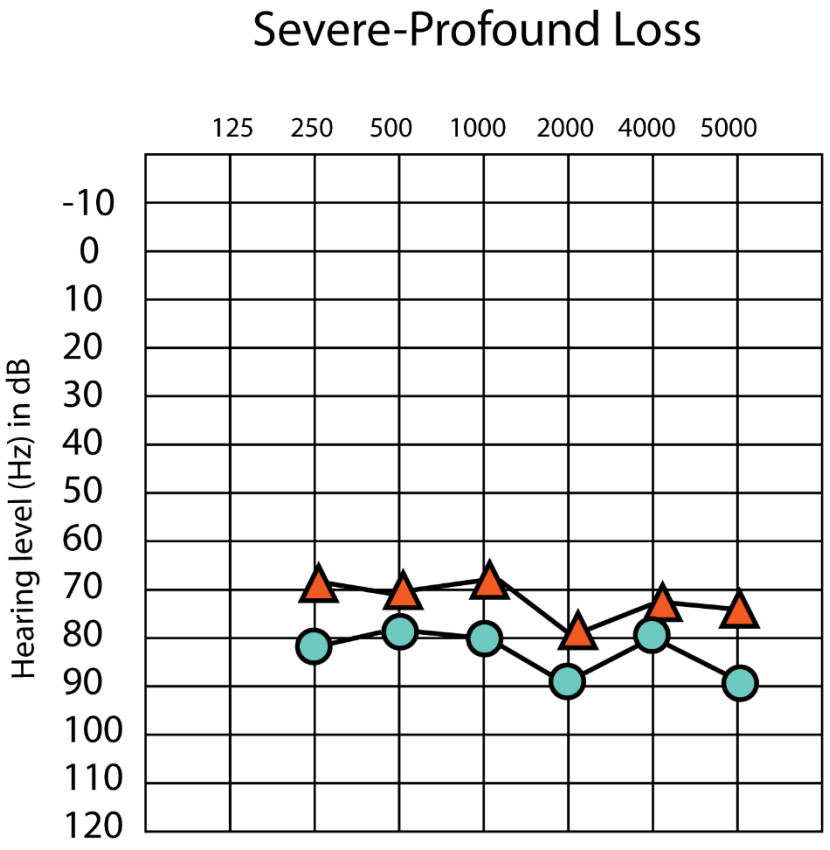


Clinical features C-26 hearing loss

- Always pre-lingual onset
- Non progressive
- Variable severity within families
- Normal vestibular function
- Normal inner ear structure
- Normal physical exam

Varied Degrees of Hearing Loss for C-26





High Prevalence of C-26 Mutations

- 10% of all hearing loss
- World wide distribution of carriers:
  - 2-3% of Caucasians (35delG)
  - 4% of Ashkenazi Jews (167delT)
  - .4% of African Americans (35delG)