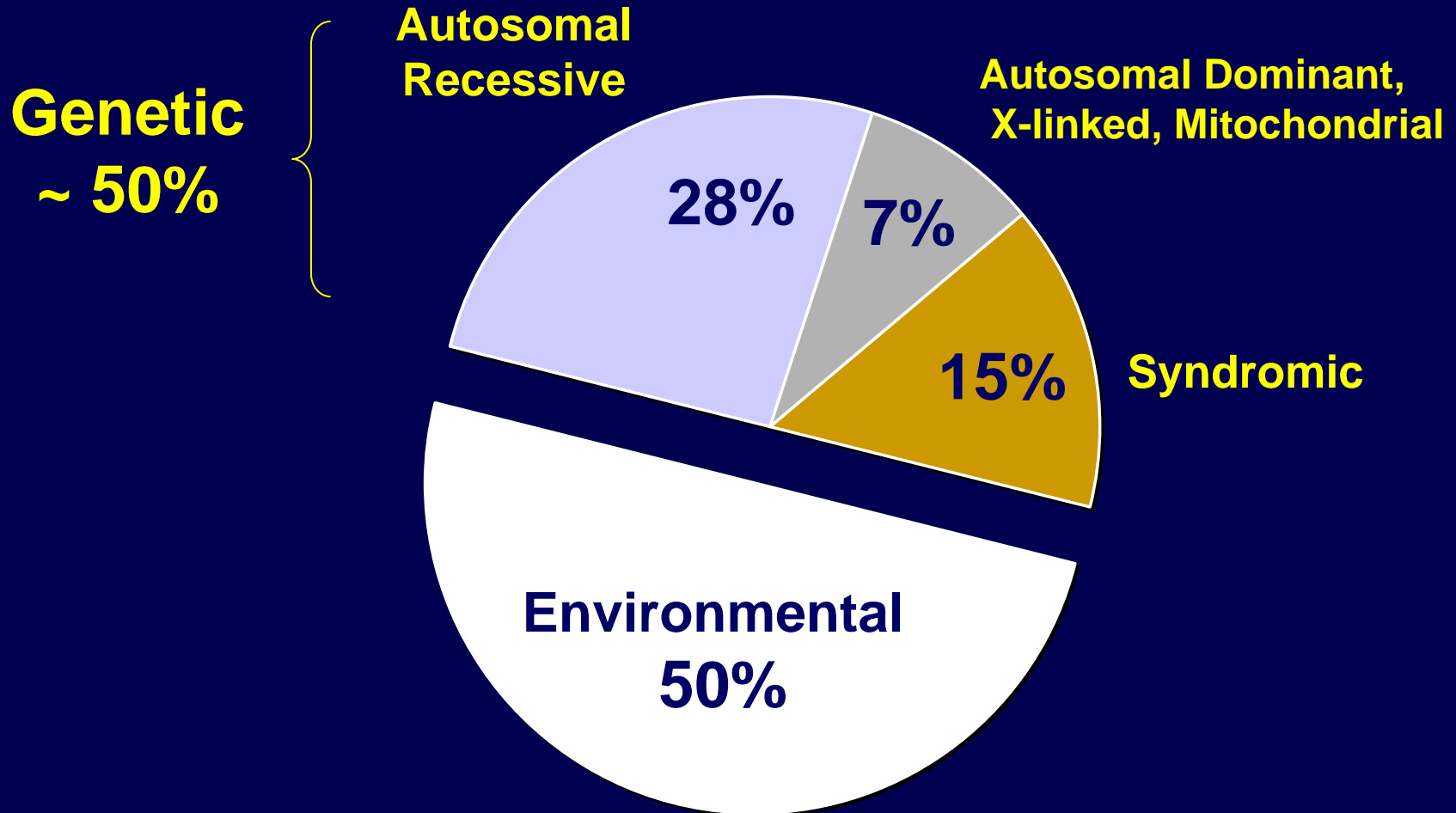


Hearing Loss and Connexin 26

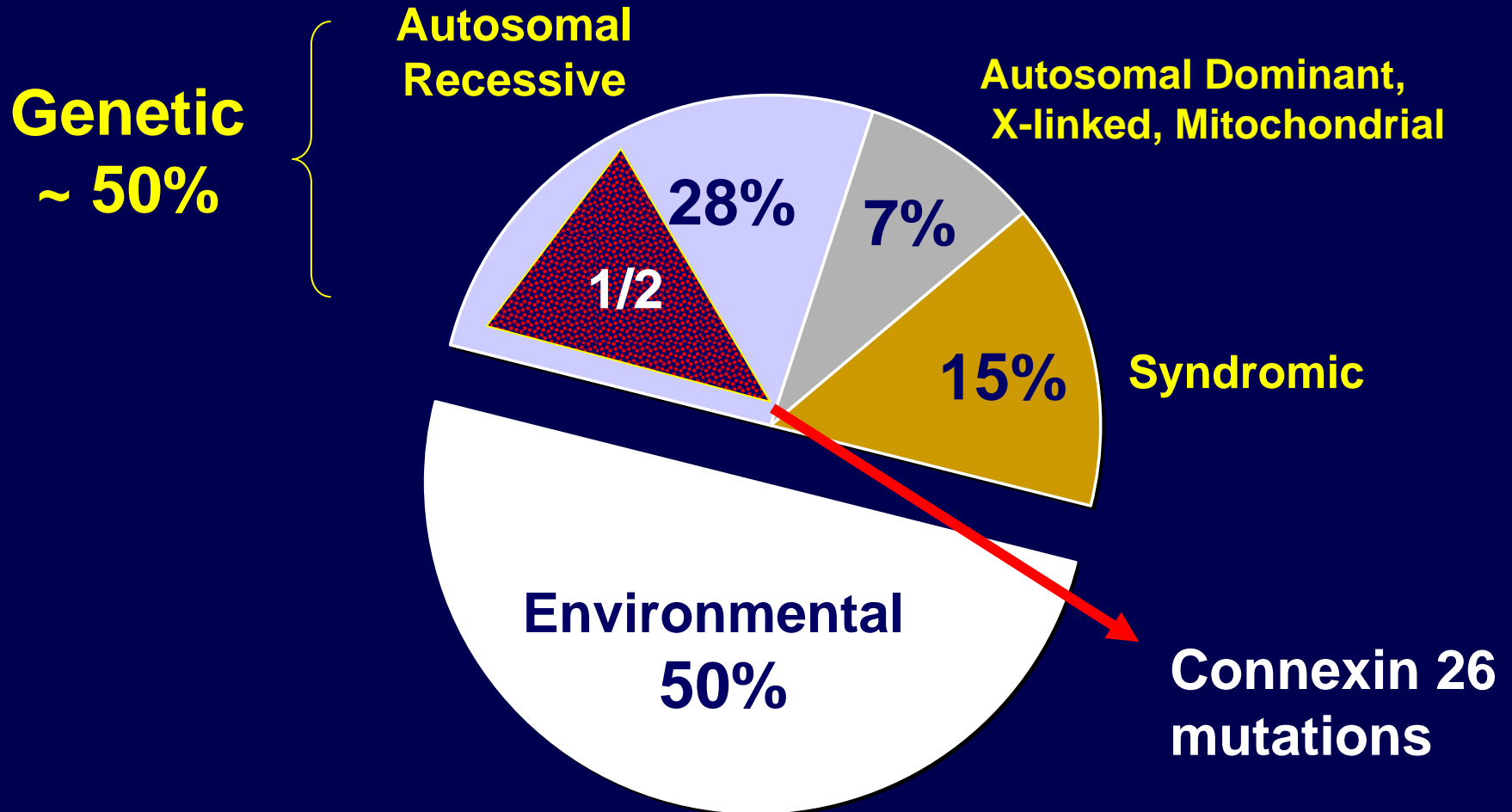


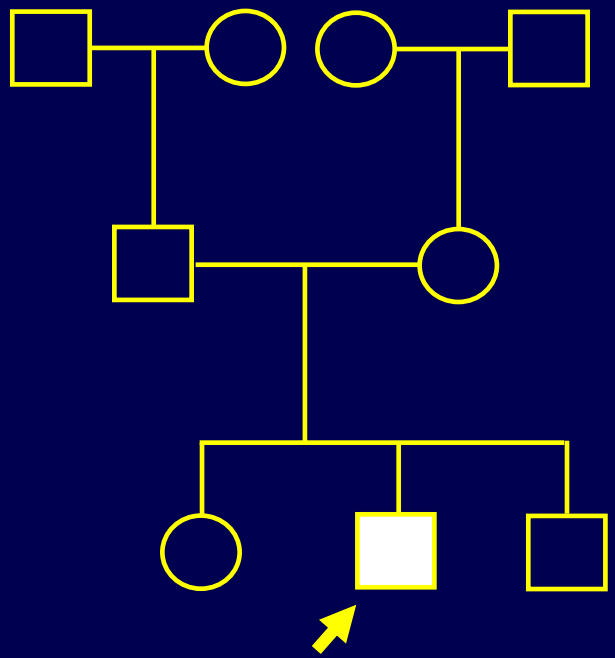
Charles Williams, MD
Division of Genetics and Metabolism
University of Florida

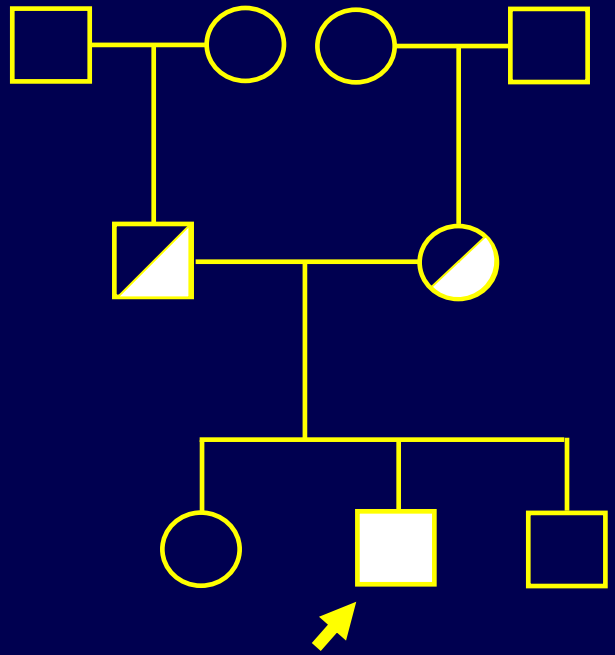
Causes of Hearing Loss

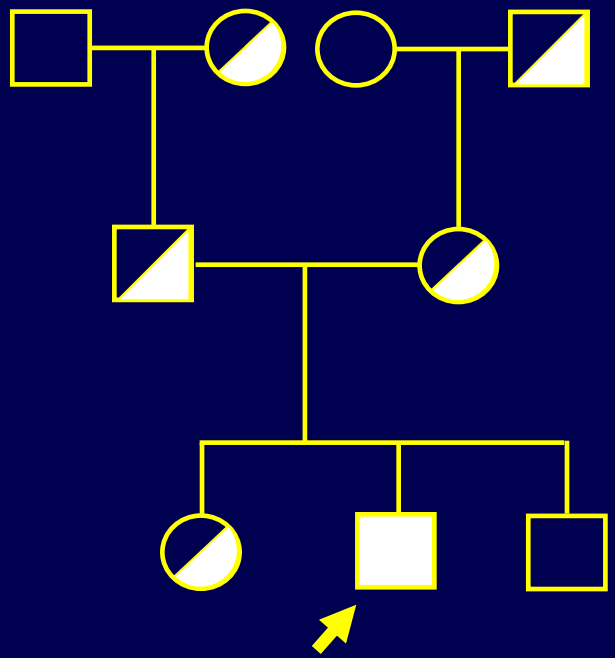


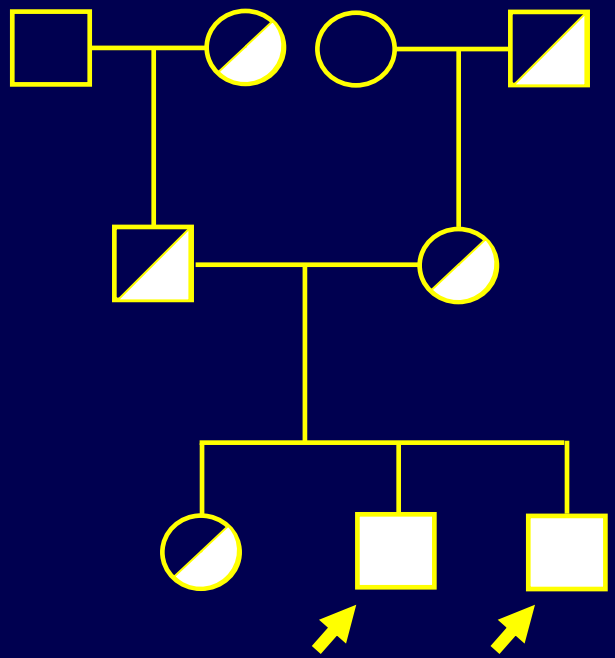
Causes of Hearing Loss





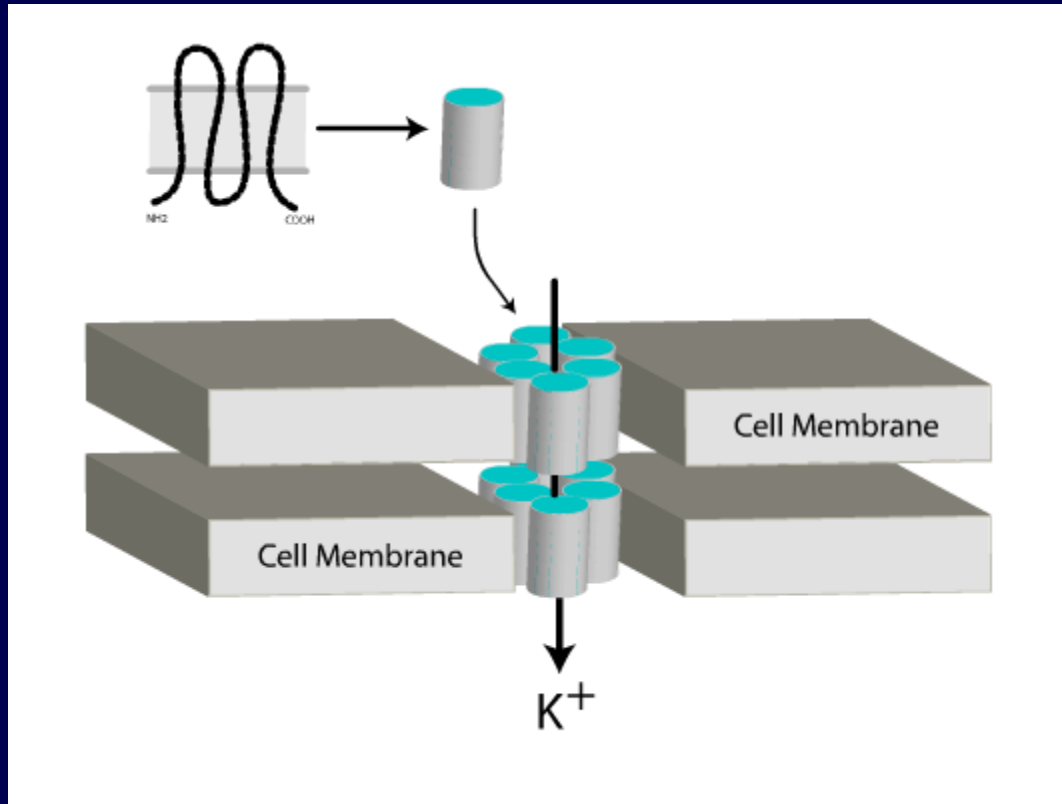


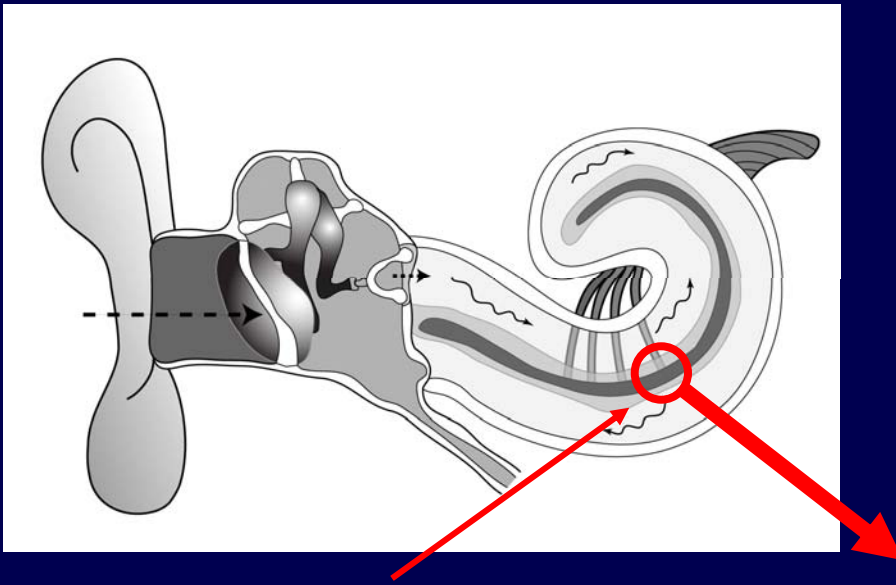




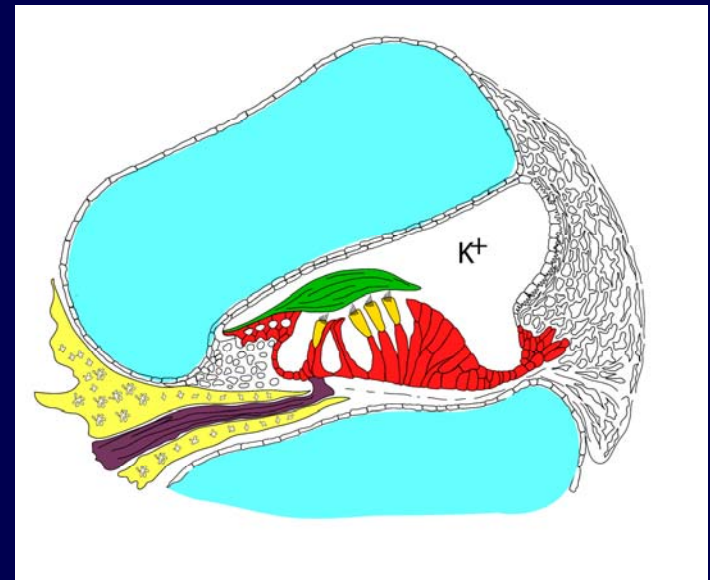
The Connexin-26 Gene and its Protein

Connexin-26 is a gap junction membrane protein



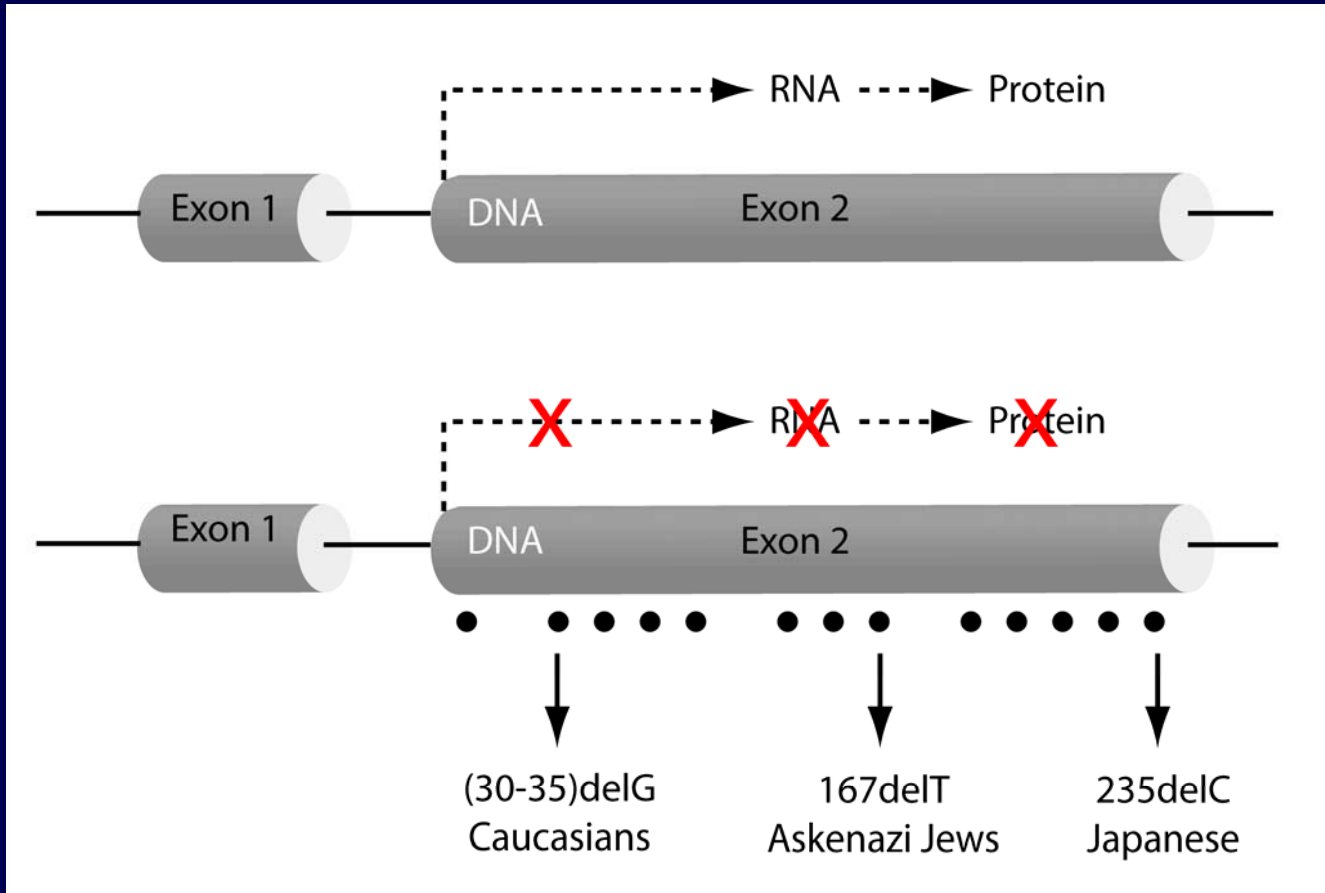


Organ of Corti in the cochlea



Recycling of K⁺ in the inner ear

Connexin 26 mutations

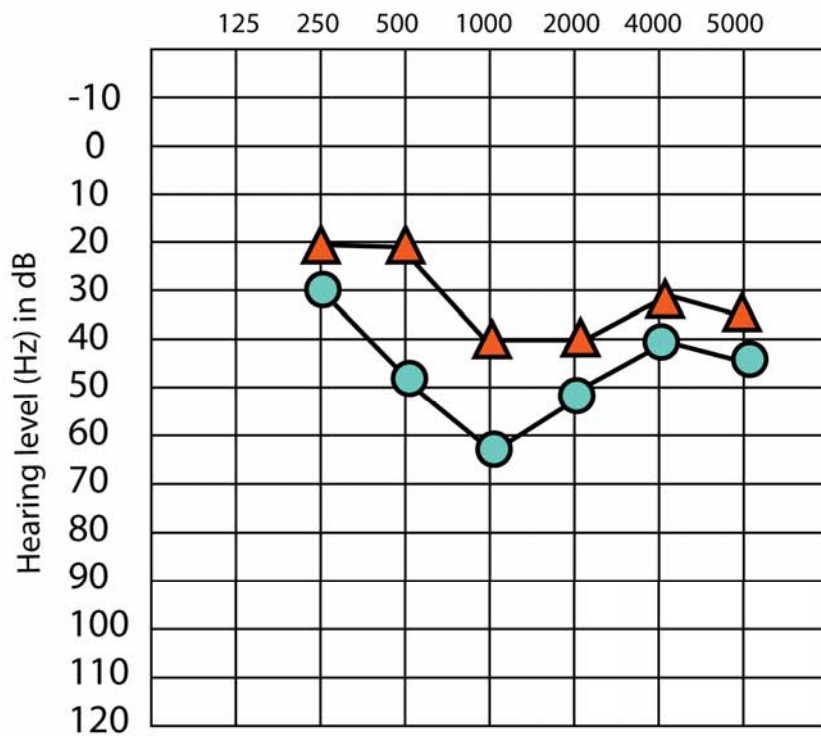


Clinical features C-26 hearing loss

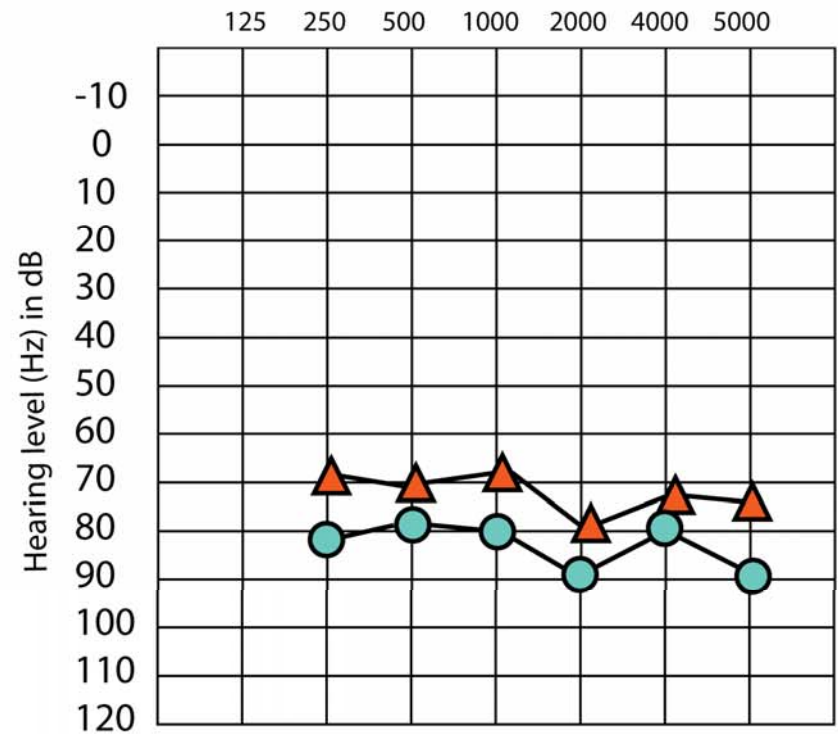
- Always pre-lingual onset
- Non progressive
- Variable severity within families
- Normal vestibular function
- Normal inner ear structure
- Normal physical exam

Varied Degrees of Hearing Loss for C-26

Mild-Moderate Loss



Severe-Profound Loss



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)