Hearing Loss and Connexin 26

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Causes of Hearing Loss

Genetic ~ 50%

- Autosomal Recessive: 28%
- Autosomal Dominant, X-linked, Mitochondrial: 15%
- Syndromic: 7%

Environmental: 50%
Causes of Hearing Loss

Genetic ~ 50%

Autosomal Recessive

Autosomal Dominant, X-linked, Mitochondrial

Syndromic

Environmental 50%

Connexin 26 mutations

15%
The Connexin-26 Gene and its Protein
Connexin-26 is a gap junction membrane protein
Recycling of K⁺ in the inner ear

Organ of Corti in the cochlea

Recycling of K⁺ in the inner ear
Connexin 26 mutations

DNA → RNA → Protein

Exon 1

DNA

Exon 2

(30-35)delG
Caucasians

167delT
Askenazi Jews

235delC
Japanese
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)