# Hearing Loss and Usher Syndrome

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## Usher Family Conference
## Portland, Oregon
## July 13, 2013

Seattle Children's
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# Overview

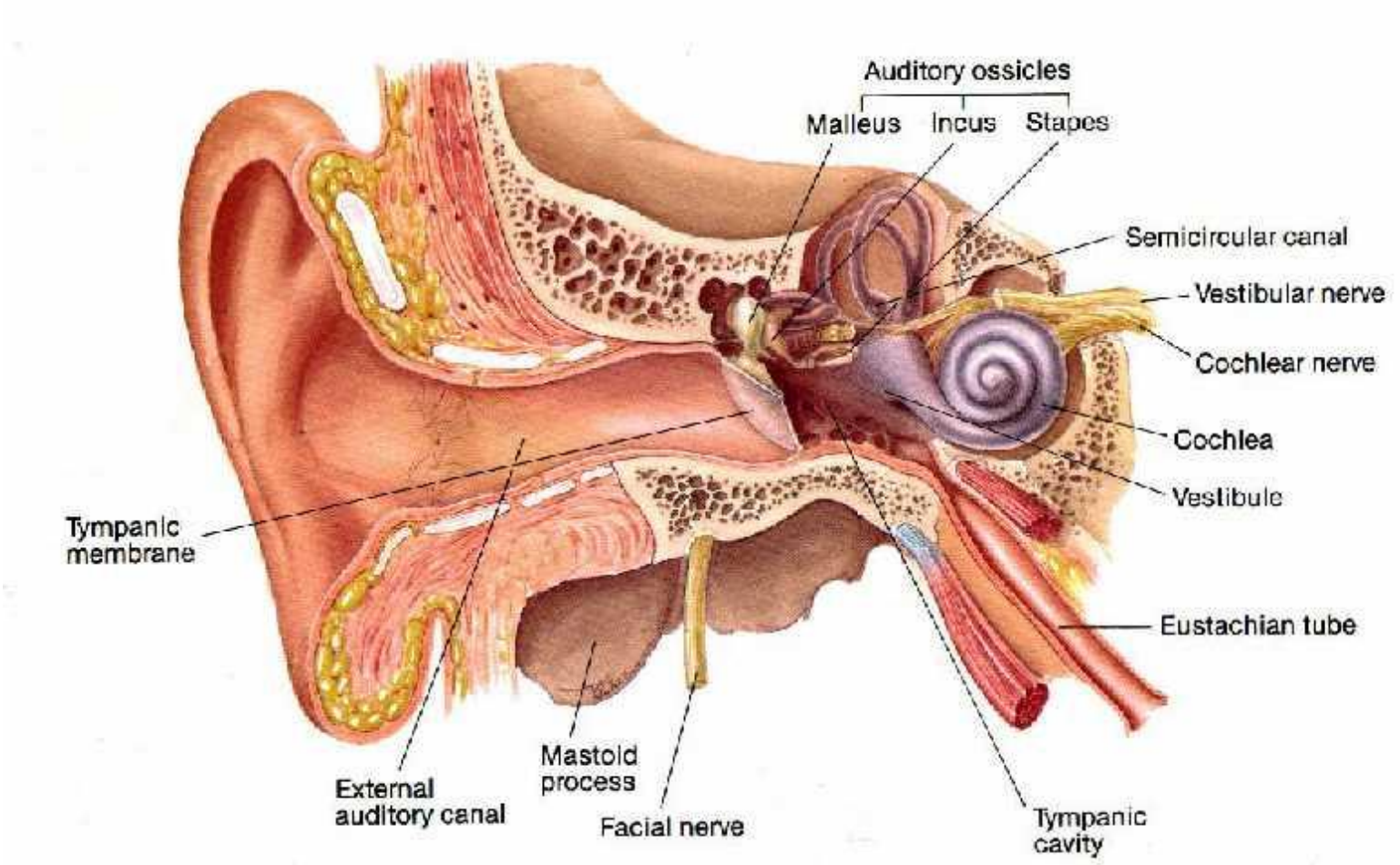
- **Childhood hearing loss**
  - How we measure hearing
  - Review of audiograms
  - Medical evaluation
  - Management of children with HL

- **Usher Syndrome and hearing loss**

# Milestones in diagnosis of childhood hearing loss

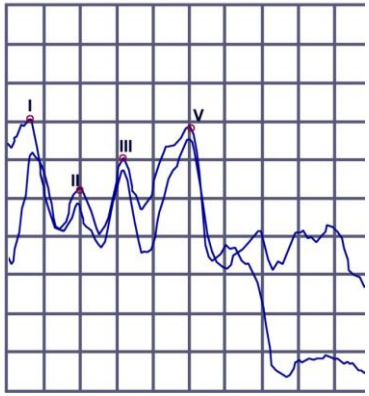
- 1960's Auditory brainstem response testing
- 1980s Automated auditory testing
  - ABR and EOAE
- 1999 Walsh Bill
- 2000's Early Hearing loss Diagnosis Detection and Intervention (EHDDI)
  - Screening by 1 month
  - Diagnosis by 3 months
  - Intervention by 6 months

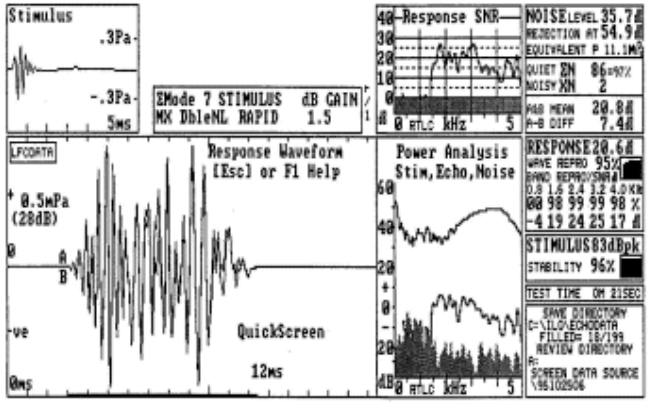
# Ears and Hearing 101



# How we measure hearing

| Type of test | Requirements | Advantages | Disadvantages |
|---|---|---|---|
| **Physiologic tests**<br><br>**ABR, BSER, BAER**<br><br>**EOAE** | Sleep or quiet | -Ear specific responses<br>-Does not require patient cooperation<br>-Correlates well with behavioral responses | -Requires sedation over 6 months of age<br>-physiologic response |



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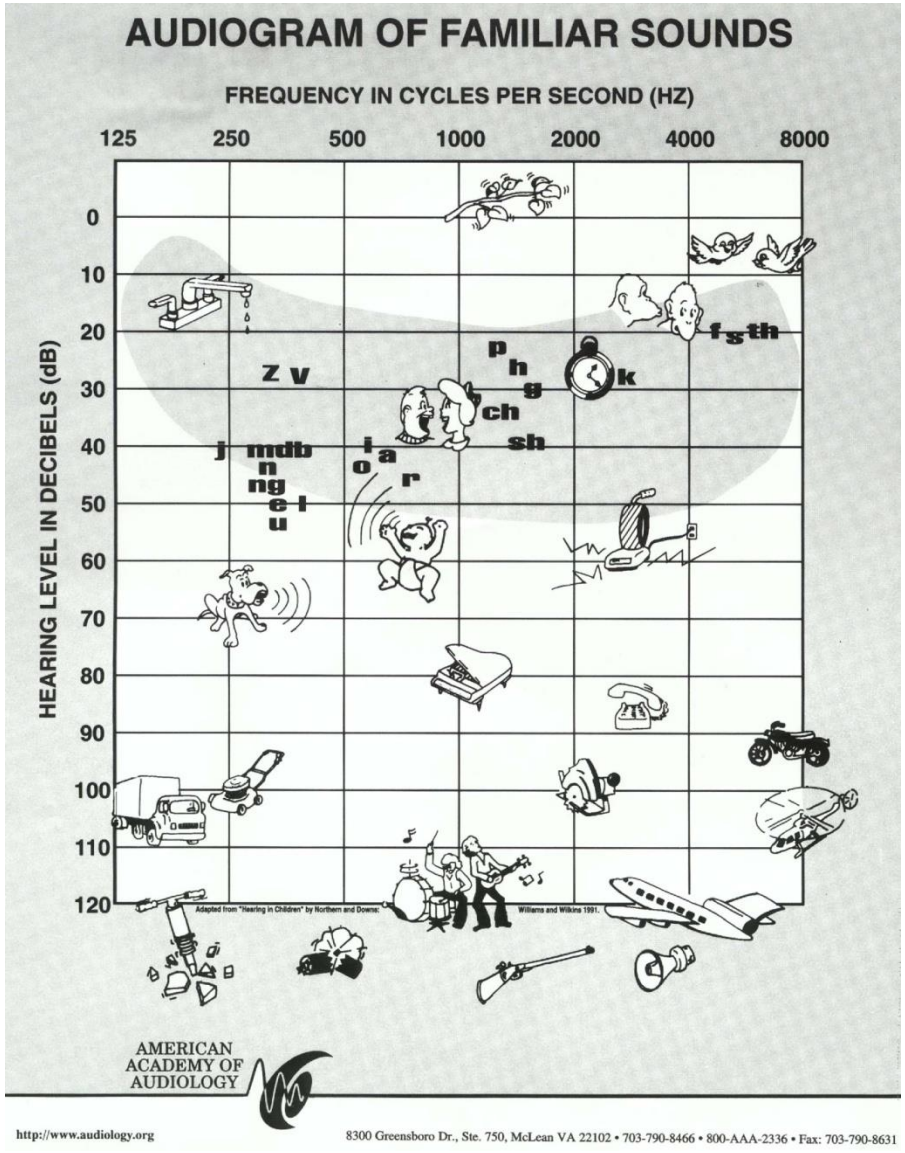
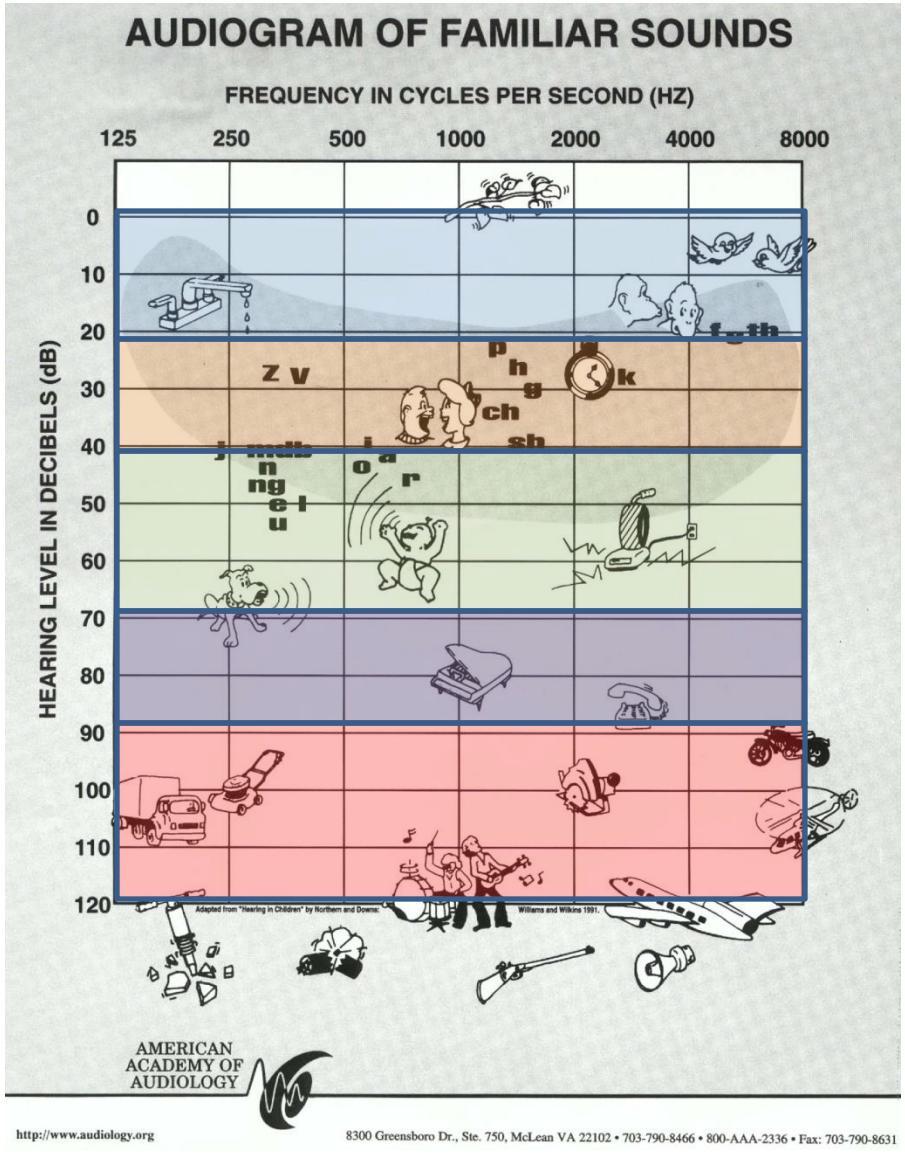
| **Behavioral**<br><br>**VRA**-visual reinforced audiometry<br>**CPA**-conditioned play audiometry<br>**CA**-conventional audiometry | >6 months old<br>Cooperative | Gold standard for assessment of hearing | Patient must be developmentally ready to understand the test |

# Ears and Hearing 101



# Ears and Hearing 101




NORMAL

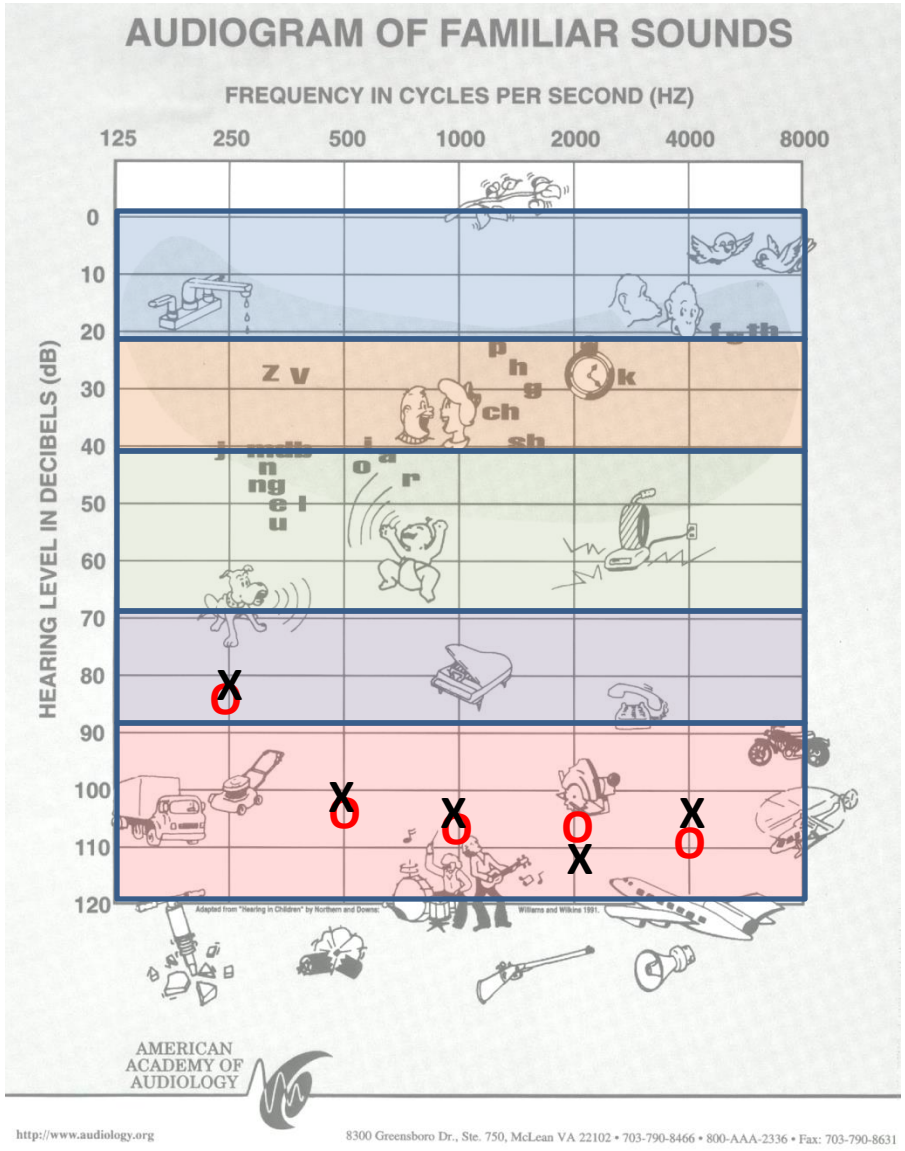
MILD

MODERATE

SEVERE

PROFOUND

# Ears and Hearing 101

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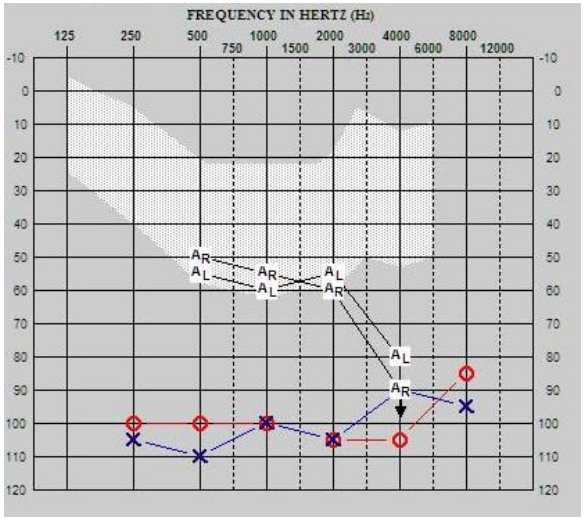
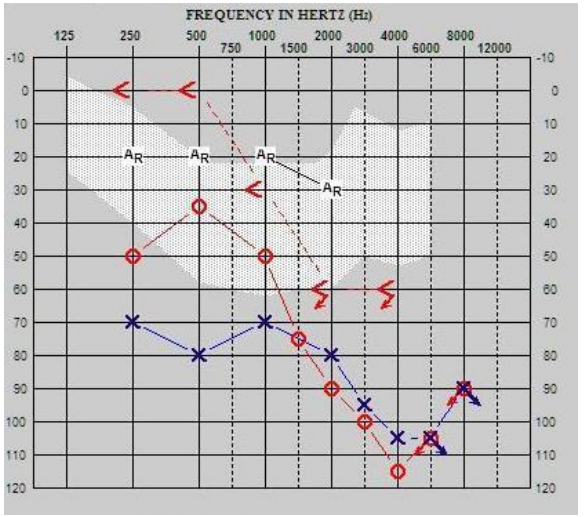
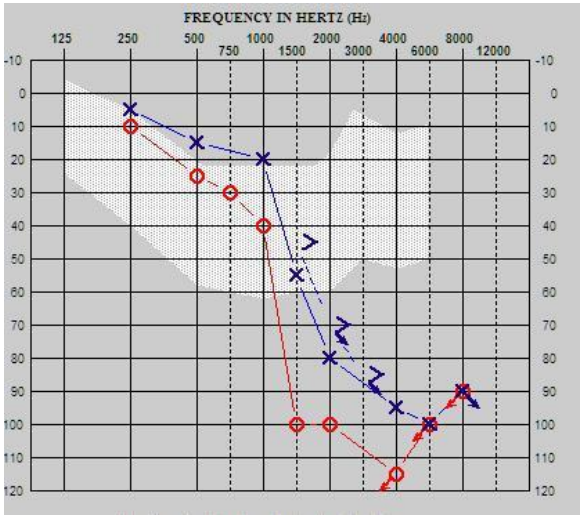
# Example Audiograms



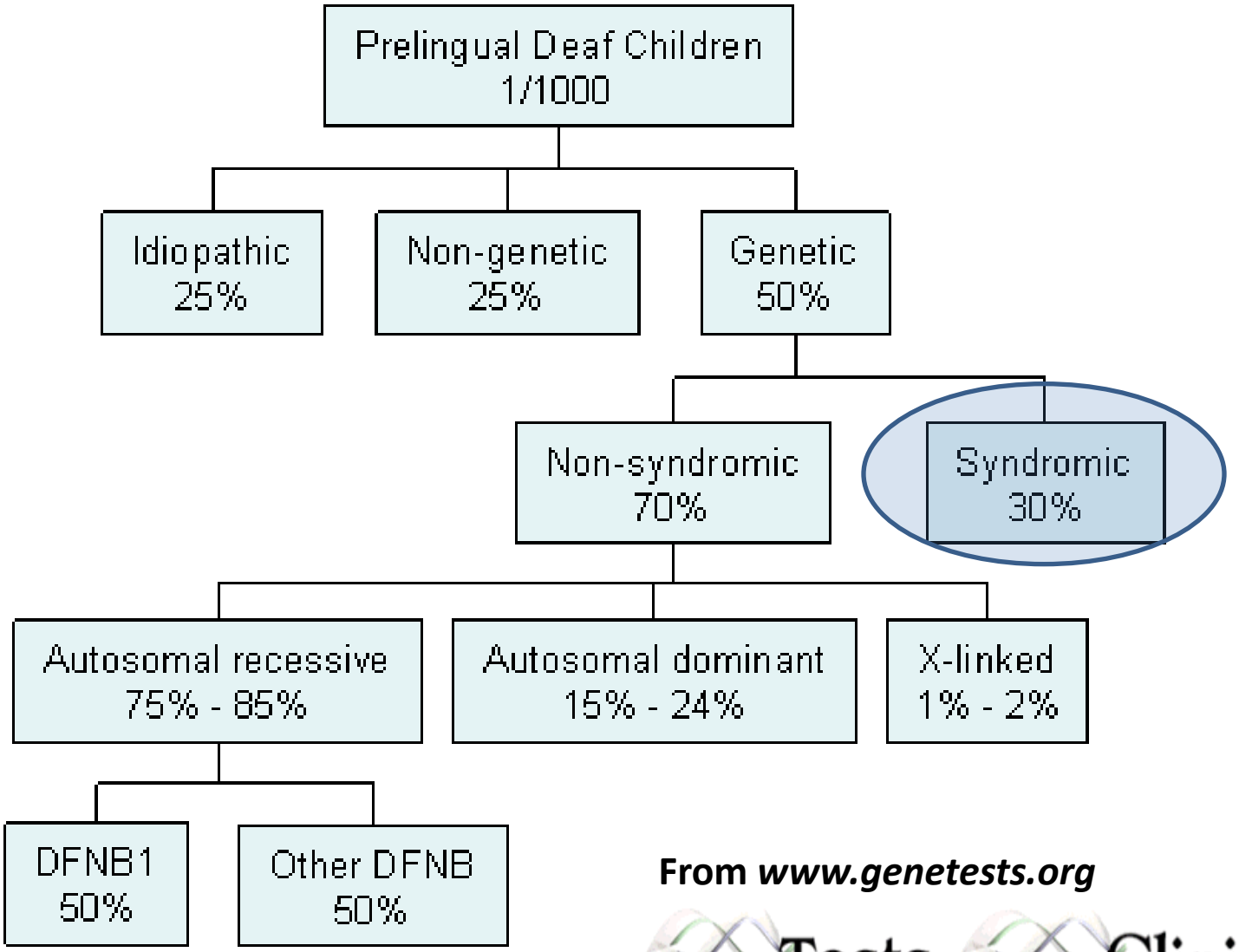



# Medical evaluation of childhood hearing loss

- **History**
- **Physical examination**
- **Characterization of hearing loss**
- **Imaging studies**
  - CT and/or MRI scans
- **Tests for specific causes of hearing loss**
  - CMV testing
  - Genetic tests
- **Tests to look for associated problems**
  - Balance testing
  - Ophthalmologic evaluation
  - Electrocardiogram
  - Renal ultrasound
  - Thyroid function studies
  - Others

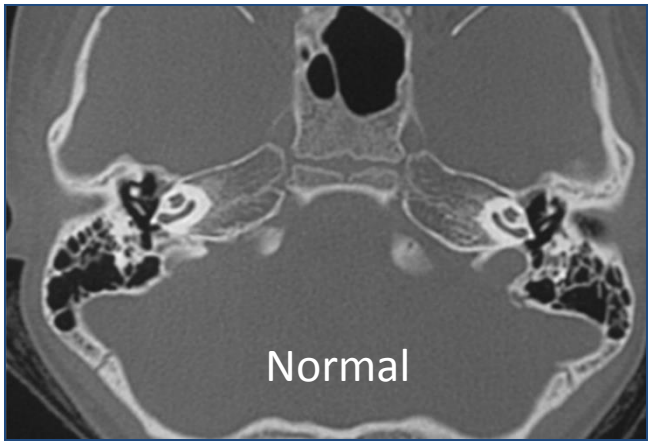
# Childhood Hearing Loss




From *www.genetests.org*

GENETests · GENEClinics

# CT scans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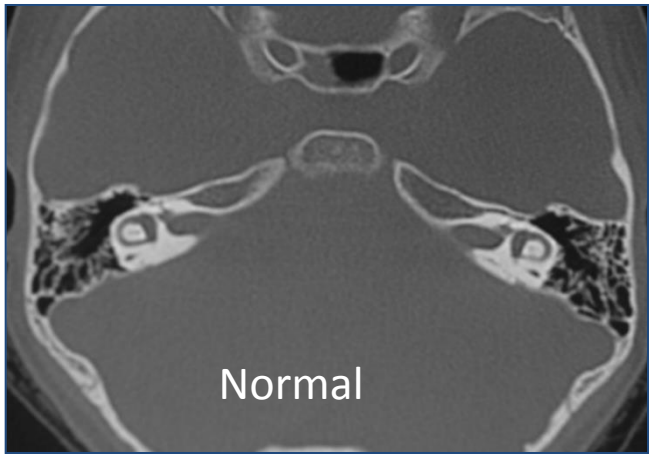
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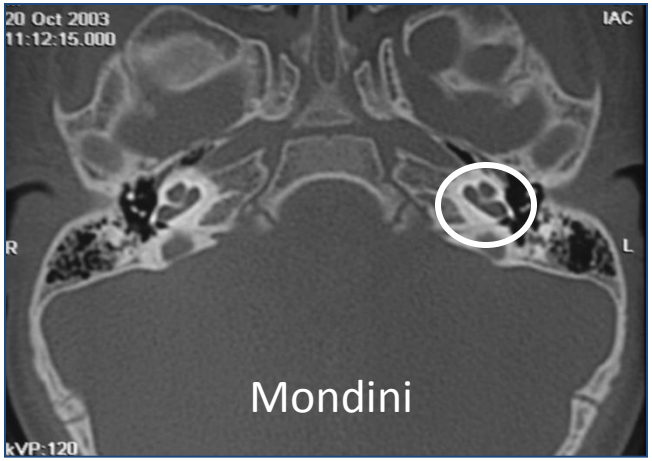
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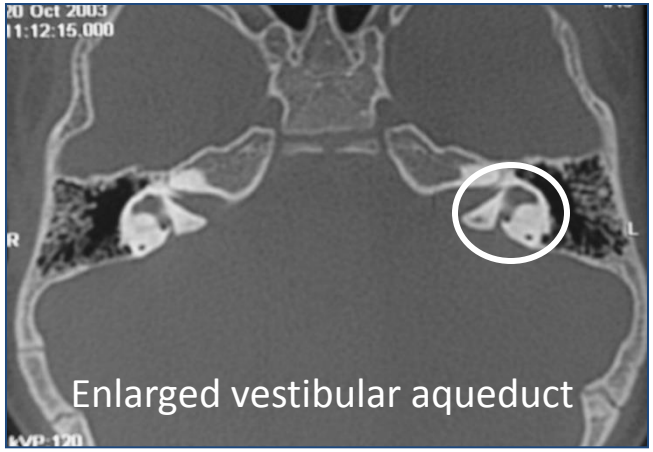
kVP:120


20 Oct 2003
11:12:15.000
R
L
Enlarged vestibular aqueduct

# Management of children with hearing loss

Early intervention/exposure to language

Amplification

- Hearing aids and cochlear implants
- FM systems

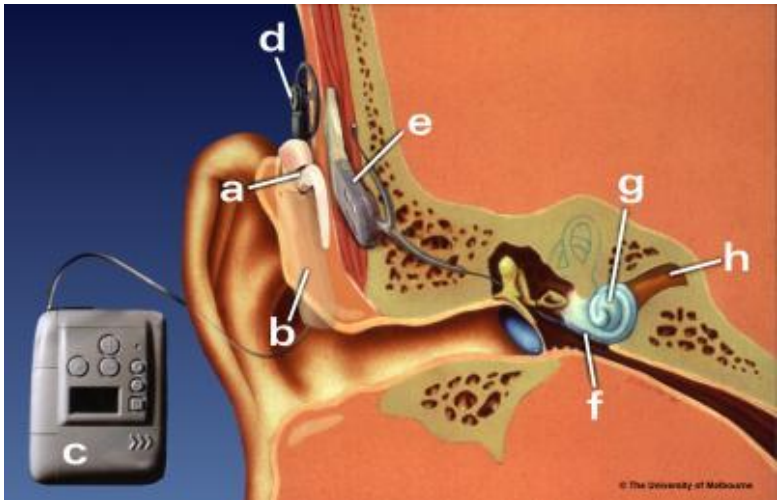
Accommodations in the school setting

# Diagnosis of Usher Syndrome

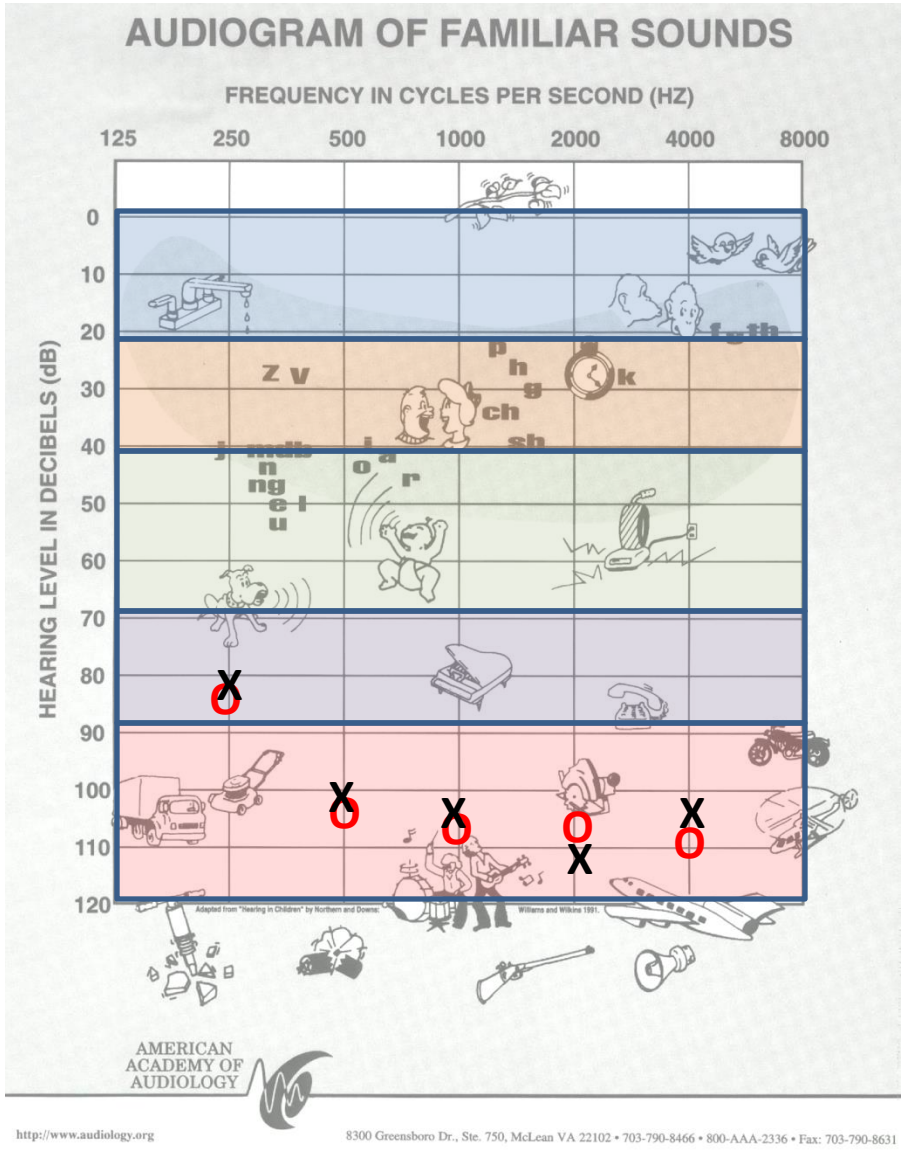
- Family history
- Congenital bilateral profound hearing loss and bilateral vestibular arreflexia
- ERG
- Clinical presentation
- Genetic testing (looking for one of 11 loci on 9 different genes)
  - Otochip
  - Otoscope

# Childhood Hearing Loss: Cochlear implantation

- Indications/guidelines
  - No significant speech benefit from appropriately fit hearing aids
  - 12 months of age
  - Absence of medical contraindications
- Emerging trends in CI
  - Bilateral CI
  - Earlier age
  - Lesser degrees of HL




# Hearing loss and US1

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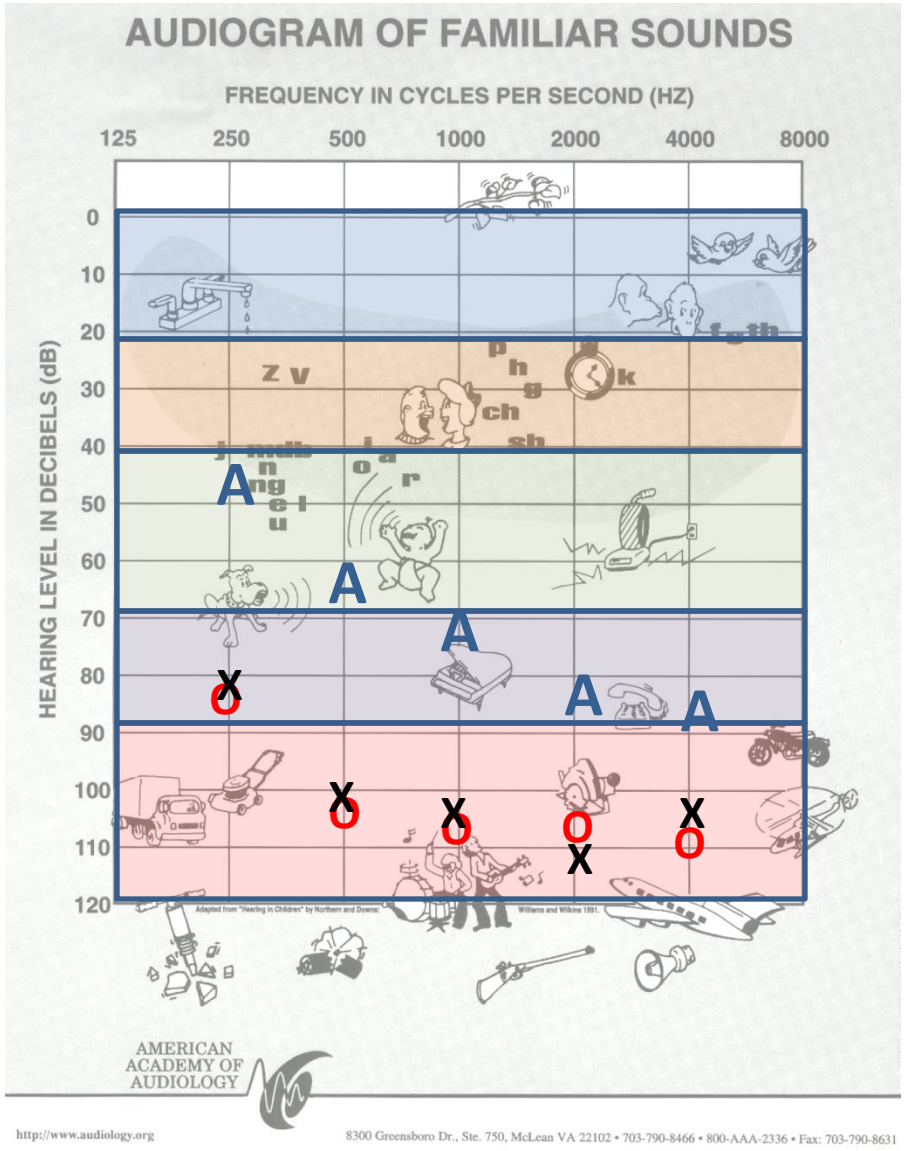
# Aided hearing and US1

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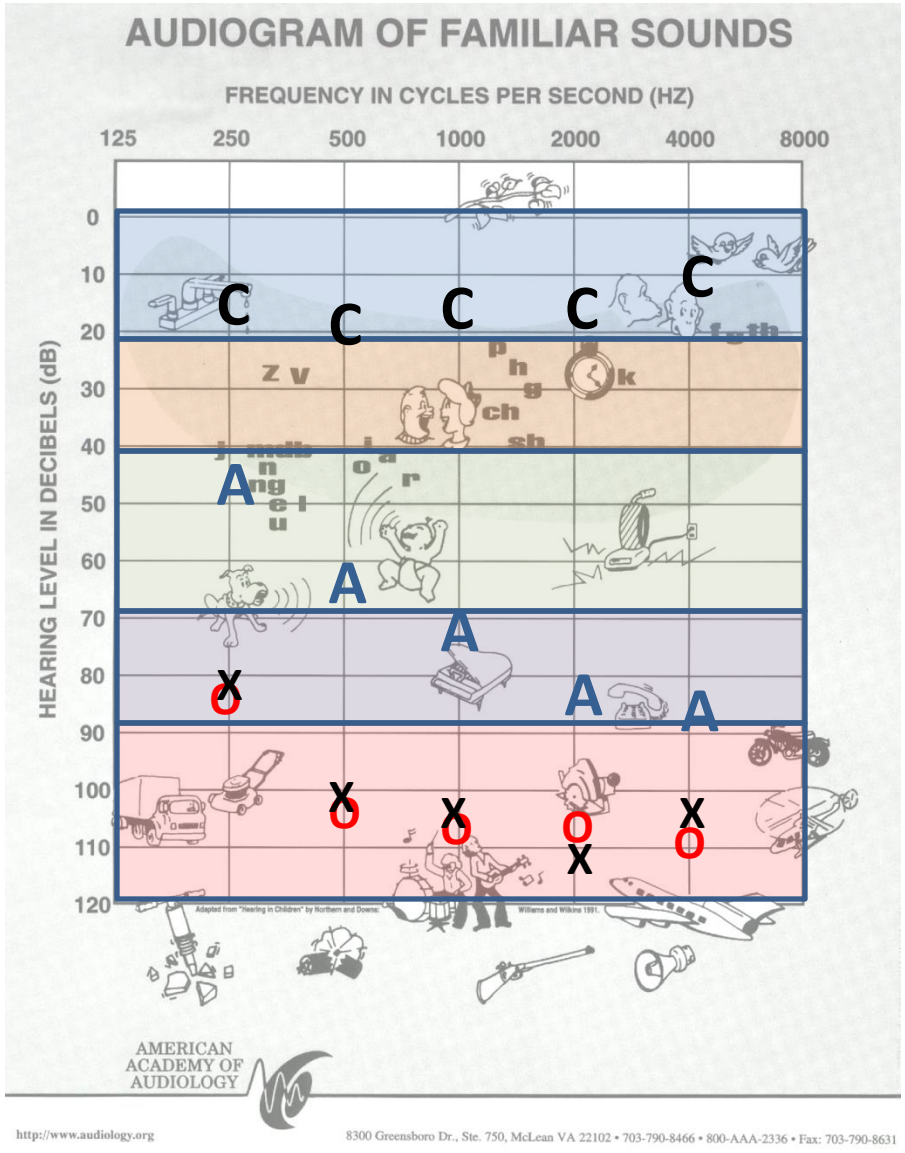
# CI and US 1

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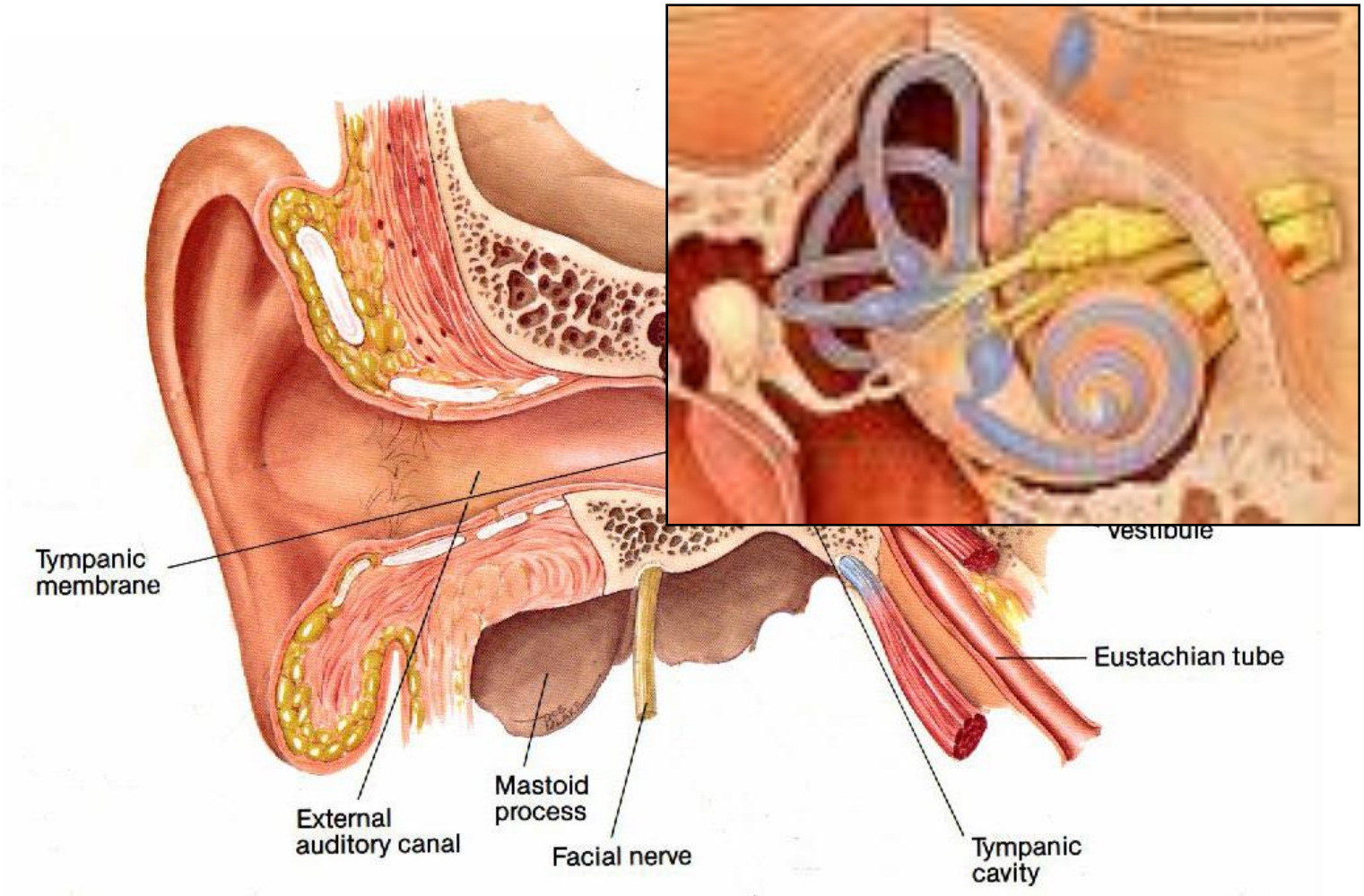
# Usher Syndrome and hearing loss

- CHILDHOOD HEARING LOSS IN USA
  - 1-3/1000 newborns have severe to profound HL
  - 2-5/1000 newborns have milder degrees of HL
  - over 90% of children with hearing loss have parents with normal hearing.
- USHER SYNDROME ACCOUNTS FOR:
  - about 1:23,000 in USA
  - 3-6% of children with hearing loss in USA
  - 50% of people with deaf-blindness in USA
  - the most common recessively inherited form of syndromic hearing loss.

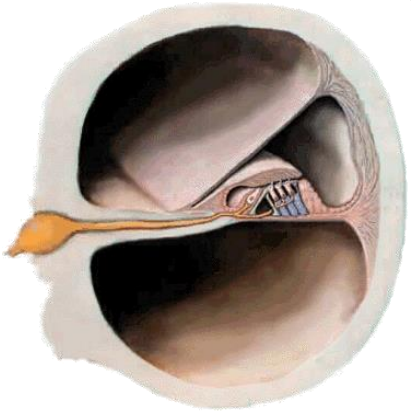
# Hearing loss and Usher Syndrome

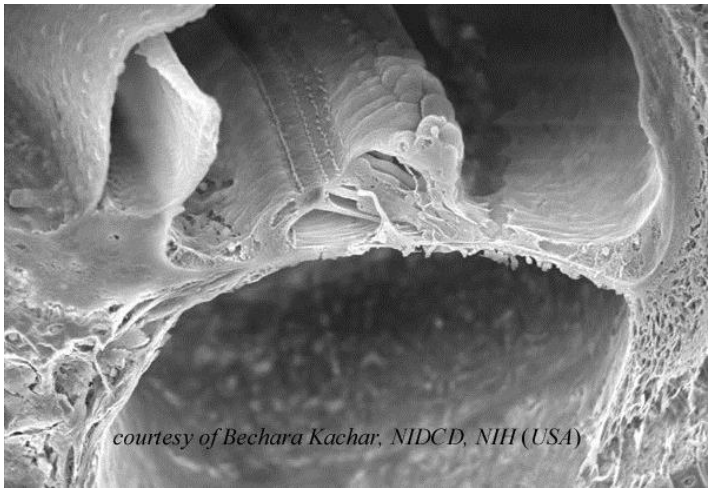
| US Type | Hearing | Vision | Balance | Genes |
|---|---|---|---|---|
| Type I<br>B,C,D,E,F,G,H,J,<br>K | Congenital<br>Bilateral<br>Profound | RP<br>Progessive loss | Congenital<br>Bilateral<br>Arreflexia | *MYO7A, CDH23, PCDH15, USH1C, USH1G* |
| Type II | Congenital<br>Bilateral<br>High frequency | RP<br>Adolescent to adult onset | Normal | *USH2A, GPR98, DFNB31* |
| Type III | Postlingual<br>Bilateral<br>Progressive | RP<br>Late onset | Variable<br>Progressive | *CLRN1* |

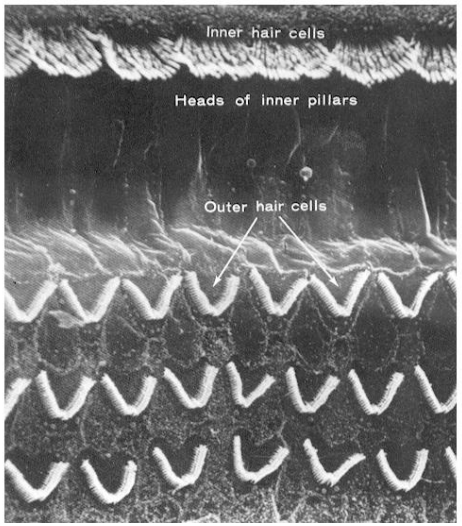
# How the ear functions



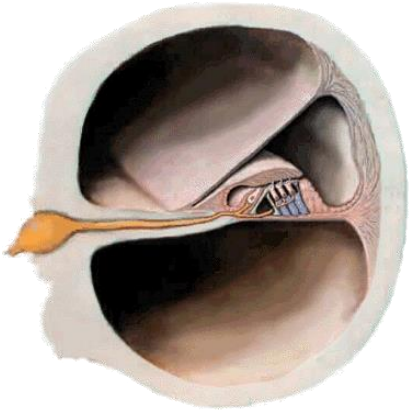
# How the ear functions – microscopically

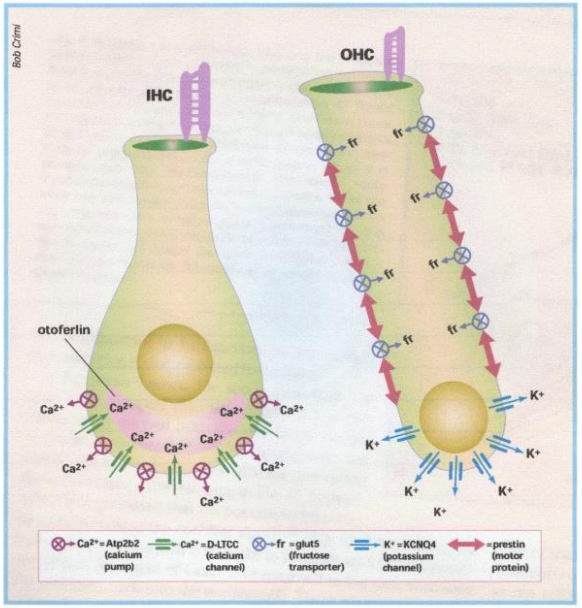


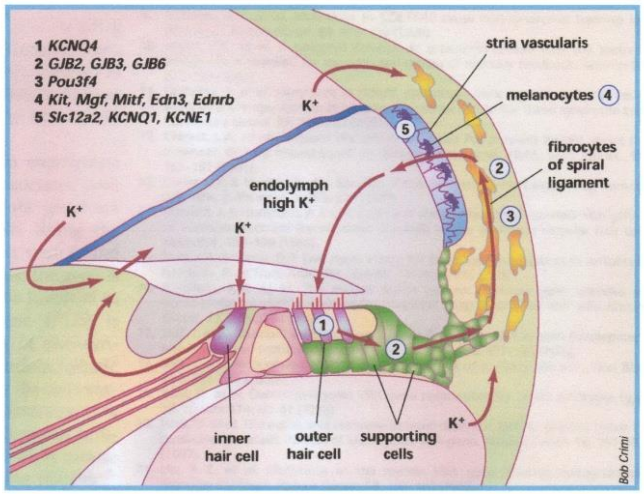


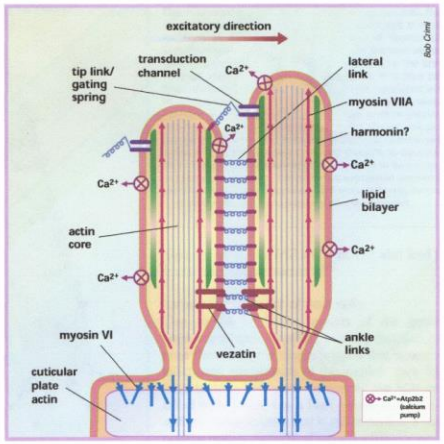



# How the ear functions – molecularly



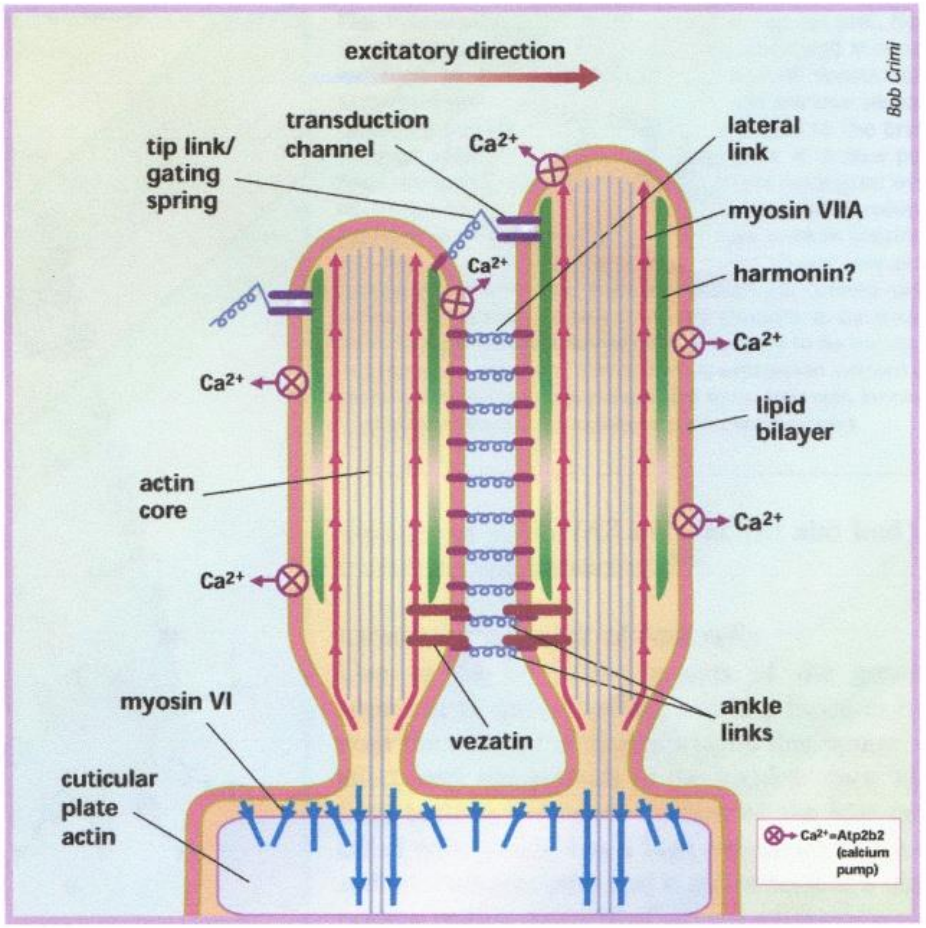


# Usher Syndrome Type 1

- USH 1B *MYO7A*
- USH1C *USH1C*
- USH1D *CDH23*
- USH1E *unknown*
- USH1F *PCDH15*
- USH1G *USH1G*
- USH1H *unknown*
- USH1J *CIB2*
- USH1K *unknown*



# Usher Syndrome and hearing loss

- Genetic therapies for US hearing loss are not yet available.

- Understanding the molecular mechanisms of hearing loss will pave the way for biologic interventions.

# Childhood hearing loss: Seattle Children's Hospital

- Hearing Loss Clinic
  - Audiologists
  - Counselor
  - Developmental pediatrician
  - Education specialist
  - Genetic counselor
  - Nursing
  - Otolaryngologist