Hearing Loss Syndromes

- Account for 20-30% of genetic HL
- >400 syndromes
- Each syndrome is relatively rare
- Syndromes identified by:
  - Physical examination findings
  - Internal ear malformations
  - Physiological traits (e.g., ECG changes)
Pendred Syndrome
- Hearing loss, usually prelingual
- Goiter in 2nd decade, most are euthyroid
- Large vestibular aqueduct and endolymphatic sac
- Upper 2/3 of cochlea is poorly formed
BOR syndrome

Branchio
  fistulas, sinuses, cysts

Oto
  cupped ears, pits, tags
  Mondini anomaly
  malformed middle ear ossicles
  semicircular canal hypoplasia

Renal
  hypoplasia to bilateral renal agenesis
CHARGE Syndrome
- Coloboma of the iris or retina
- Heart defects
- Atresia or stenosis of the choanae
- Retarded growth and development
- Genital hypoplasia in males
- Ear anomalies
- Perilymphatic Gusher-Deafness syndrome
  - Conductive hearing loss
  - Stapes fixation
  - Perilymphatic gusher during stapes surgery

- Internal Auditory Canal
- Auditory Canal Dilation
- Leakage of perilymph
LEOPARD Syndrome

- Lentigines
- EKG abnormalities
- Ocular hypertelorism
- Pulmonic stenosis
- Abnormalities of genitalia
- Retardation of growth
- Deafness

Left axis deviation
• Waardenburg Syndrome
  - Dystopia canthorum
  - White forelock
  - Hypochromic irides, heterochromia
  - Sensorineural deafness
Treacher Collins Syndrome

- Down-slant of palpebral fissures, lower lid coloboma
- Hypoplasia of maxilla, micrognathia
- Dysmorphic ears
- Atresia of middle-ear ossicles, conductive hearing loss
<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Gene/s</th>
<th>Inheritance</th>
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<tbody>
<tr>
<td>Pendred</td>
<td>SLC26A4</td>
<td>AR</td>
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<tr>
<td>BOR</td>
<td>EYA1, SIX5</td>
<td>AR</td>
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<tr>
<td>CHARGE</td>
<td>CHD7</td>
<td>AD</td>
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<tr>
<td>Perilymph Gusher</td>
<td>POU3F4</td>
<td>X-linked</td>
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<tr>
<td>LEOPARD</td>
<td>PTPN11, RAF1</td>
<td>AD</td>
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<tr>
<td>Waardenburg</td>
<td>PAX3, MITF1, SNAI2, SOX10</td>
<td>AD</td>
</tr>
<tr>
<td>Treacher Collins</td>
<td>TCOF1</td>
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</table>
Other Syndromic Conditions

- **Alport**
  - hematuria
- **Stickler**
  - retinal detachment
- **Usher**
  - retinitis pigmentosa
- **Jervell and Lange-Nielsen**
  - long PR interval on ECG
- **KID (keratitis, ichthyosis, deafness)**
  - Skin hyperkeratosis