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Hearing Loss In Children With 22q11.2 Deletion Syndrome

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HEARING LOSS IN CHILDREN WITH 22Q11.2 DELETION SYNDROME

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Introduction

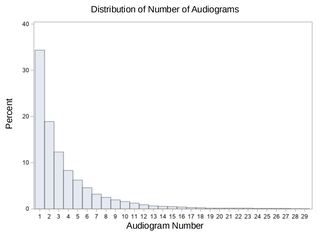
22q11.2 deletion syndrome (22q11.2DS) is the most common microdeletion syndrome, with most recent published prevalence of 1:2,140 live births. Hearing loss is common in children with 22q11.2DS and has been reported to occur in 32-60% of people. Hearing loss is typically conductive in nature secondary to chronic eustachian tube dysfunction and chronic otitis media with effusion (COME). Close monitoring of hearing and the middle ears is recommended to detect COME and the need for ear tubes. Additionally, there is a high prevalence of speech delay in children with 22q11.2DS, stressing the importance of close audiometric evaluation and medical intervention as appropriate. While prior studies confirm a relationship between hearing loss and 22q11.2DS in children, there is a lack of data regarding frequency, type, and specific features of hearing loss for children with this syndrome.

Methods

- Study Design and Participants
  - Retrospective chart review of data combined from two large 22q Centers based in children's hospitals
  - Total of 1,640 consecutive charts reviewed
  - Inclusion criteria of children with 22q11.2DS and documented audiological testing
- Statistical Analysis
  - Presence of speech delay, 22qDS breakpoints, cardiac defects, cleft palate, and need for amplification was assessed
  - All audiograms recorded for each patient were analyzed
  - Trajectory of the hearing loss (or normal hearing) was summarized
  - Number of tubes placed and timing between placement along with hearing sensitivity was investigated
- Audiologic Parameters
  - Hearing loss was defined as thresholds greater than 20 dB HL
  - Degree of hearing loss was classified as mild: 25-40 dB HL, moderate: 41-55 dB HL, moderately-severe: 56-70 dB HL, severe: 71-90 dB HL, profound: 91+ dB HL
  - Type of hearing loss was defined as unspecified, conductive (CHL), sensorineural (SNHL), and mixed (MHL)
  - Tympanometry was classified as Type A, Type B large volume, Type B small/normal volume, and Type C

Study Participants

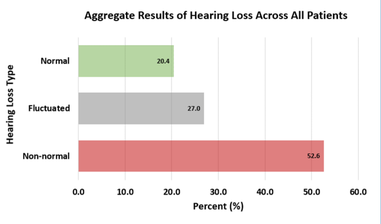
- Of the 1,640 charts reviewed, 775 children met inclusion criteria
  - 53% males; 47% females
- Total of 2,539 audiograms were reviewed
  - Median number of audiograms was 3 per person
  - Some individuals had up to 29 audiograms
- Timing of Audiograms
  - Average age at the first audiogram was 5.9 years
  - Largest interval between 1st and 2nd audiograms was 2.72 years for normal vs. 1.5 years for non-normal
- 86% of the children had documented history of speech delay
- 27% of the children presented with cleft palate



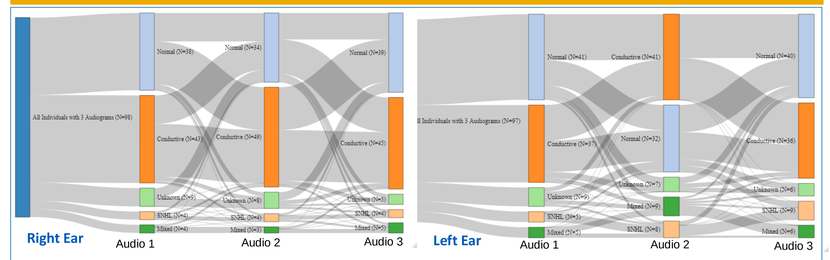
Audiometric Results

- **Collectively, 79.6% of all audiograms indicated some degree of hearing loss**
  - 20.4% Normal hearing
  - 52.6% Hearing loss
  - 27.0% Fluctuated between normal hearing and hearing loss
- Description of the Hearing Losses
  - Mild hearing loss was the most prevalent degree documented
  - 36% bilateral hearing loss; 26.3% unilateral hearing loss; 37.7% in soundfield
  - Greatest documented type of hearing loss was CHL
  - 3-10% audiograms showed SNHL with 90% remaining stable

		Right (n=673)	Left (n=672)	Sound Field (n=110)
Type	Conductive	511 (76%)	464 (69%)	14 (13%)
	SNHL	70 (10%)	104 (15%)	3 (3%)
	Mixed	51 (7%)	68 (10%)	1 (1%)
	Unknown	41 (6%)	36 (5%)	92 (83%)
		Right (n=900)	Left (n=856)	Sound Field (n=249)
Severity	Severe	74 (8%)	95 (11%)	17 (7%)
	Moderate	211 (23%)	188 (22%)	68 (27%)
	Mild	615 (68%)	573 (67%)	164 (66%)



Trajectory of Hearing Loss



This demonstrates the importance of consistent audiological monitoring.

Other Findings and Conclusions

- Anatomic abnormalities of the middle and inner ear were found in 37 children when imaged
  - Dysmorphia of the vestibule (35%), semicircular canal abnormalities (32%), ossicular abnormalities (27%), and cochlea anomalies (16%) were all found
- Tube placement was common in this population due to COME
  - 39% of the cohort required at least one set of tubes
  - Of those children, 55% needed multiple sets of tubes
- 80% of children with SNHL also had a cardiac defect
- Hearing loss for those with cleft palate was statistically the same as the group without palatal clefting

What's Next?

- Larger prospective studies are needed to determine if hearing loss continues to be a chronic issue or if there is an age at which hearing loss becomes less common
- Frequent hearing evaluations are critical due to speech and language delays which can be exacerbated by untreated hearing loss
- **New clinical practice recommendation**
  - **Annual otolaryngologic evaluation**
  - **Formal hearing evaluations every 1-2 years**

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