Investigating the Relationship Between Hearing loss in Girls with Turner syndrome and Developmental Concerns

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INTRODUCTION

- Turner syndrome (TS) affects ~1/2000 females and increases the risk of hearing loss (Geerardyn et al, 2021).
- Hearing loss may be linked to the loss of the X chromosome p arm. (Lin, et al, 2023).
- Karyotype may be used as a predictor for future hearing impairment even at young ages (Verner et al., 2011).
- Otitis media can cause hearing loss and is also common in TS. (Leach et al., 2020).
- There are several treatment options for hearing loss including medications, surgery, and hearing aids. (Atkinson, et al, 2015) (Rovers et al., 2005) (Gates et al., 1987).

AIMS

The goals of the current project:

1. Characterize hearing loss in a young cohort of patients with TS
2. Assess a relationship between hearing loss and speech delay

METHODS

- **Chart reviews of Patients with TS ≤6 years old and at least one audiology evaluation**
- N= 50 patients at Children's Hospital Colorado in the INSIGHTS Registry
- Excluding those with diagnoses of autism spectrum disorder (ASD).
- Data collected on the degree and type of hearing loss, ear affected (bilateral vs. unilateral), and the age at the audiology
- Speech delay was defined by the Ages and Stages Questionnaire (ASQ) or documentation by a developmental or speech specialist of a speech delay in the chart.

DEMOGRAPHICS

	Conductive Hearing Loss (CHL)	Sensorineural Hearing Loss (SNHL)	Mixed Hearing Loss	Normal Hearing
Karyotype	N=20	N=2	N=2	N=26
45,X	9 (45%)	1 (50%)	2 (100%)	11 (42.3%)
45,X/46,XX	3 (15%)	1 (50%)	0 (0%)	4 (15.4%)
45,X47,XXX	0 (0%)	0 (0%)	0 (0%)	4 (15.4%)
Isodicentric	5 (25%)	0 (0%)	0 (0%)	4 (15.4%)
Other	3 (15%)	0 (0%)	0 (0%)	3 (11.5%)
Race/Ethnicity				
White	16 (80%)	2 (100%)	0 (0%)	20 (76.9%)
Black or African American	3 (15%)	0 (0%)	0 (0%)	2 (7.69%)
Other	1 (5%)	0 (0%)	2 (100%)	4 (15.4%)
Current age (yrs) (Median & IQR)	7.3 (5.1)	12.1 (14.6)	15.1 (0.04)	7.2 (5.5)
# of audiology evaluations (Median & IQR)	4.5 (4)	2.5 (1)	5.5 (1)	3 (2)

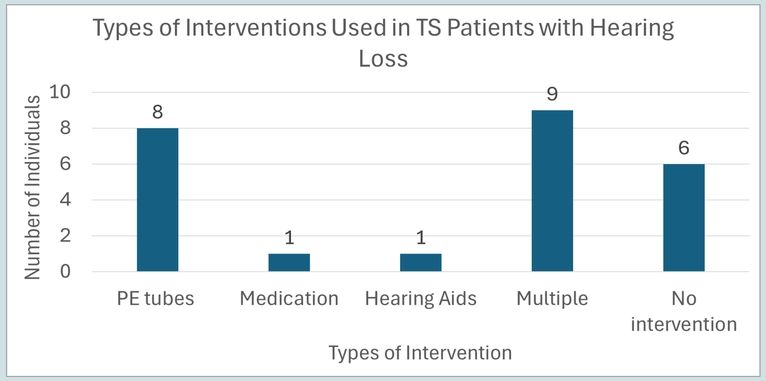
SPEECH DELAY RESULTS

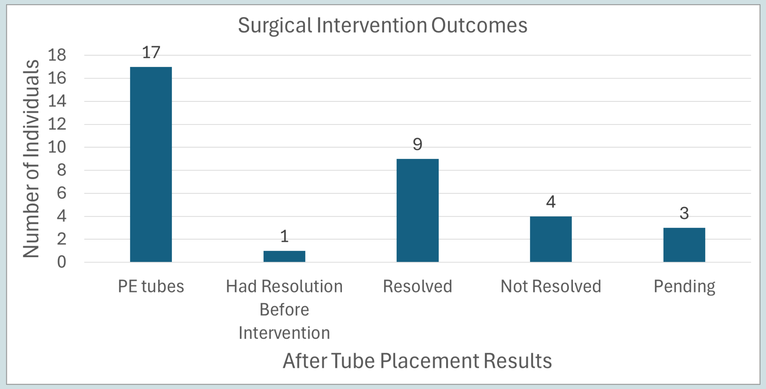
Speech History & Status	Conductive Hearing Loss	Sensorineural Hearing Loss	Mixed Hearing Loss	Normal Hearing
	N=20	N=2	N=2	N=26
Had Speech Delay Prior to Intervention	15 (75%)	2 (100%)	1 (50%)	16 (61.5%)
Speech Delay Status After Intervention				
Yes	9 (60%)	1 (50%)	1 (50%)	NA
No	4 (26.7%)	1 (50%)	1 (50%)	NA
Not evaluated yet	2 (13.3%)	0 (0)%	0 (0%)	NA

Hearing Loss Results Compared to Speech Delay

Audiology Result	No Speech Delay (N = 16)	Speech Delay (N = 34)
Normal Hearing	10 (62.5%)	15 (44.1%)
Inconclusive	1 (6.25%)	1 (2.94%)
Abnormal	5 (31.3%)	19 (55.9%)

HEARING LOSS RESULTS





HEARING LOSS RESULTS (CONTINUED)

Hearing Loss & Interventions	Conductive Hearing Loss (CHL)	Sensorineural Hearing Loss (SNHL)	Mixed Hearing Loss	Normal Hearing
	N=20	N=2	N=2	N=26
Age at HL diagnosis (mean ± SD)	2.9 ± 1.8	3.1 ±1.3	9.8 ±1.8	NA
Intervention				
Surgery (ie: tubal insertion)	15 (75%)	1 (50%)	1 (50%)	2 (7.7%)
Medication	6 (30%)	0 (0%)	0 (0%)	0 (0%)
Hearing aid	2 (10%)	1 (50%)	2 (100%)	0 (0%)
No intervention	5 (25%)	0 (0%)	0 (0%)	24 (92.3%)
Current HL Status				
HL Persist	4 (20%)	2 (100%)	2 (100%)	NA
Resolved	9 (60%)	0 (0%)	0 (0%)	NA
Recurred	4 (26.7%)	0 (0%)	0 (0%)	NA
Pending Follow-up Exam	3 (15%)	NA	NA	NA

Reasons for hearing loss not resolved after tubes: n=4

- Mild to moderate mixed/CHL (bilateral)
- Normal to mild SNHL (unilateral)
- CHL (left-sided) and Inconclusive (right-sided), but pt also has trisomy10q.
- Mild to moderate mixed (unilateral)

DISCUSSION

- Our data shows there is an increase in both hearing loss and speech delay in the TS population
- Further research is still needed if hearing loss correlates with speech delay
- This data suggests that individuals with TS benefit from additional audiology exams

LIMITATIONS AND FUTURE DIRECTIONS

- This is a small pilot analysis further analyses with larger sample size is needed
- This is a cross-sectional study and thus causal link between hearing loss and speech delay cannot be made.
- Missing data may exist in patient charts, such as evaluations, interventions, or follow-up done outside of our hospital.

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