Cholesteatoma In Children With Sotos Syndrome
A Case Series and Literature Review

Leonard CG, Ranguis S, Cushing SC, Blaser S, James AL

Introduction

Sotos syndrome, is a rare genetic disorder characterised by neurodevelopmental delay and excessive childhood growth including macrocephaly. We present our experience of children with Sotos syndrome and cholesteatoma and compare the prevalence in our institution with other known risk factors for cholesteatoma.

Methods

a) Children with Sotos syndrome who were treated for cholesteatoma between May 2002 and January 2020 were identified from a prospectively collated database.

b) Comparison was made with those children in the database known to have risk factors for cholesteatoma.

Results

Case	Cholesteatoma side	Age (yrs)	Sex	EAONO/JOS Stage	Ossicular Status
1	Left	8.5	M	2	Incus and Stapes Crura eroded
2	Left	9.6	M	1	Incus eroded
3	Left	8.2	M	2	Incus and Stapes Crura eroded
4	Right	10.9	F	Precholesteatoma	Incus erdded
5	Bilateral (Right)	4.1	M	N/A	N/A
6	Bilateral (Left)	4.5	M	N/A	N/A

Table 1 showing demographics at time of surgery and extent of cholesteatoma.

	Number of children with acquired cholesteatoma	% of total (n=400 children)	Number of bilateral cases (%)	Published prevalence estimate of condition in population	Published prevalence estimate of cholesteatoma in condition
Total	400	100%	60 (13%)	-	1:10,000
Sotos syndrome	5	1%	1 (20%)	1:10,000-14,000	NA
Cleft palate	42	11%	6 (14%)	9:10,000	2%
Turner syndrome	3	1%	1 (25%)	1:2000-2500	4.4 - 6%
Trisomy-21	5	2%	2 (40%)	13.5:10,000	NA

- Cleft palate is more common in both the overall population and within our database by the same factor as Sotos Syndrome
- Cholesteatoma may be as common in children with Sotos syndrome as in children with cleft palate, and more common than in other syndromes associated with cholesteatoma.
- Only 33/86(38%) children with Sotos syndrome who underwent CT or MRI had normal middle ears and mastoids bilaterally.

Conclusion

Children with Sotos syndrome appear to be at increased risk of developing acquired cholesteatoma. Impaired temporal bone pneumatization is a common incidental finding in Sotos syndrome in keeping with this risk.