

Late-Onset Permanent Childhood Hearing Impairment

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Background & Methods

- NHSP universally offered to all babies since 2006 in the UK
- NHSP aims to identify moderate to profound PCHI
 "Moderate or worse" PCHI – defined from average hearing thresholds of 40dB in the better ear in 0.5, 1, 2 and 4kHz (16)
 -May miss mild HL + isolated HL (e.g. mid-frequency loss) + high frequency HL (20)
 -Late-onset or progressive HL can also be missed
- There is scarcity of literature on late-onset PCHI diagnosis (9)

- Late-onset PCHI cases identified within a 3-year period (January 2022-December 2024).
- Registration with Hospital Clinical Governance Department.
- Retrospective review of eligible patient notes including demography, hearing loss profile, age at diagnosis, hearing loss progression, auditory amplification, medical deafness aetiological investigation results and vestibular profile.
- R version 4.5.0 (11/04/2025) used for statistical analysis. Statistical significance was set at a =0.05 threshold.

Demographics

- In total, 104 patients were included in the analysis.
- 41 female (39.4%) and 63 male (60.6%).
- Median age: 6 years.
- 20 patients (19.2%) were ≤ 5 years old.
- Mean age at diagnosis: 7.41 years.

Subgroup analysis included 2 age groups (≤ 5 years old and > 5 years old), 2 severity groups (max moderate including mild-moderate HL and max profound including moderate/severe-profound HL) and left and right ears separately.

Presence of Additional Comorbidities

- | | | |
|---|---|---|
| <ul style="list-style-type: none"> • ASD • ADHD • Learning disability • Developmental delay • Ex-prematurity | <ul style="list-style-type: none"> • Dyslexia • Cerebral palsy • Atrial septal defect with previous surgical closure • Hirschsprung disease • Large bowel hypoganglionosis | <ul style="list-style-type: none"> • Asthma • Dyspraxia • Growth hormone insufficiency • Phelan-McDermid syndrome |
|---|---|---|

Results

Results - CMV

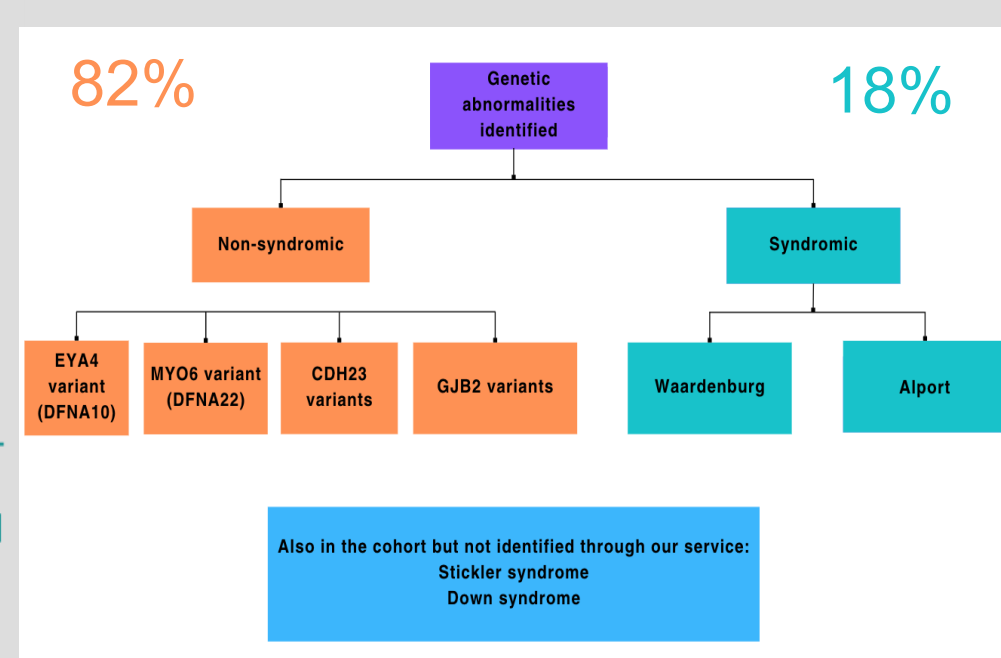
- Performed in 41 cases (39.4%)
- Positive in 10 cases (24.4%):
 - acquired in 8 cases (80%)
 - congenital in 2 cases (20%)

- B/L profound SNHL.
 - Vestibular testing not yet performed.
 - Evidence of CMV infection on MRI.
 - Normal R28, R65 and R67 gene panels.
 - Non-verbal and awaiting ASD pathway assessment.
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- B/L mild-moderate SNHL.
 - L utricular hyperfunction with large amplitudes and low thresholds on o-VEMP.
 - Normal MRI head and IAM.
 - Normal R28, R65 and R67 gene panels.
 - No comorbidities.

Results – Genetic Testing

HL genetics performed in 36 cases (35.3%)

HL causative genetic abnormalities identified
HL genetics performed
= 30.6%



Genetics diagnostic yield comparison did not have a statistically significant difference among patients with different hearing loss severities.

HL gene panels have proven cost-effectiveness and high diagnostic yield (up to 40%) across all ages, HL grades and ethnicities (Sloan-Heggen et al., 2016).

However, diagnostic rates in late-onset HL patients are lower than those in early-onset patients (28% and 16% respectively) (Sloan-Heggen et al., 2016; Mori et al., 2016).

Results - Imaging

Imaging performed in 77 cases (76.2%)

Imaging abnormalities identified
imaging performed = 36.4%

Imaging abnormalities diagnostic for HL
imaging abnormalities identified = 75%

Imaging abnormalities diagnostic for HL
imaging performed = 27.3%

Wide range of diagnostic yields for MRI (2–60%).
Greater yield of CT than MRI for EVA (yield difference 16.7% [95% CI, 9.1%–24.4%]) and borderline advantage for cochlear anomalies (4.7% [95% CI, 0.1%–9.5%]).
Studies more likely to report brain findings with MRI. (12)

% of abnormalities identified of the total imaging performed as well as % of abnormalities diagnostic for HL of the total imaging performed were statistically significantly larger in the patients with more significant hearing losses (moderate/severe-profound) (p-value=0.002 & 0.001 respectively).

- Commonest abnormality identified was cochlear nerve hypoplasia/aplasia.
- This was followed by EVA, SCCD, middle ear pathology (ossicular abnormalities and erosion 2° cholesteatoma) and lastly infective sequelae (white matter changes and labyrinthine ossification).

Results - Vestibular

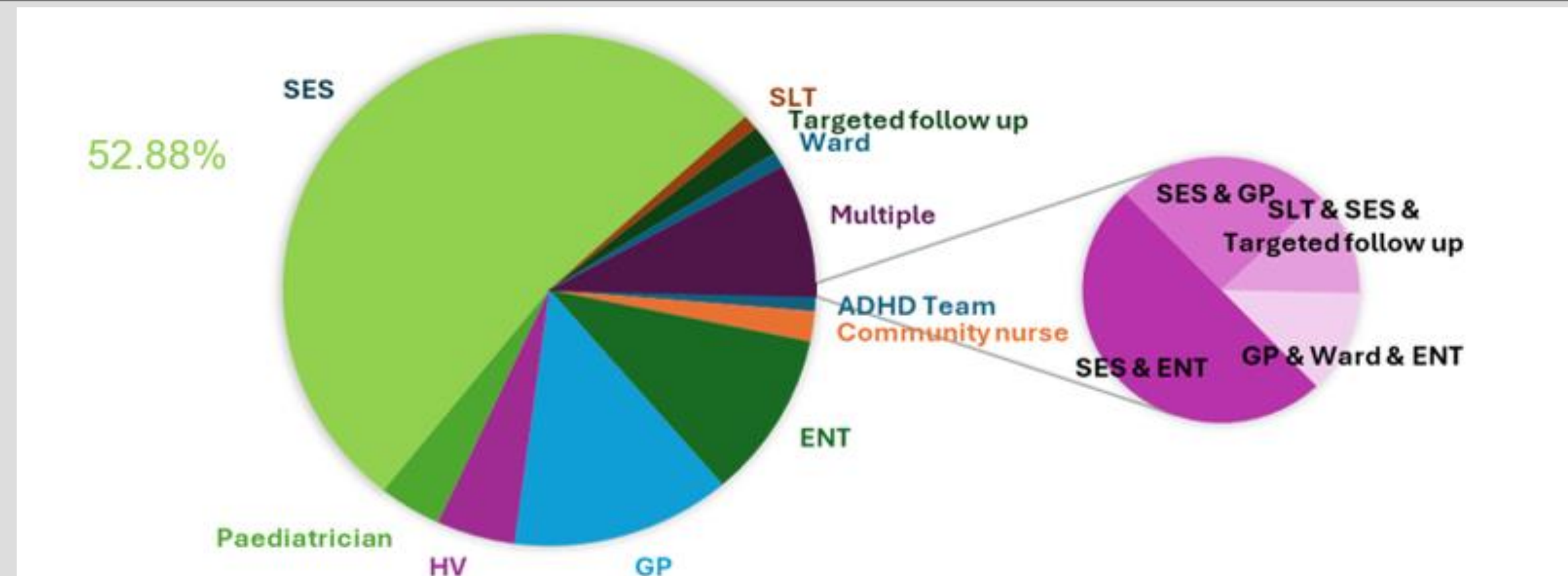
- Vestibular testing was performed in 74 cases (71.2%) **31.1%** of cases had abnormalities.
- Commonest to least common vestibular test abnormality identified:
o-VEMP > c-VEMP > v-HIT > SHIMP > VNG
- For right ears, groups with more severe hearing losses had statistically significant more abnormalities identified on vestibular testing in total (p-value=0.041).
- For both right and left ears, v-HIT was the only vestibular test to have statistically significant differences in terms of severity between hearing loss and test abnormalities (p-value=0.001 and 0.010 respectively)
 - High risk of vestibular dysfunction in children with SNHL, with reported prevalence ranging between 20-85% (10; 1; 11; 17)
 - Even mild unilateral hearing loss may be accompanied by vestibular dysfunction (13)

Results – Aetiological Profile

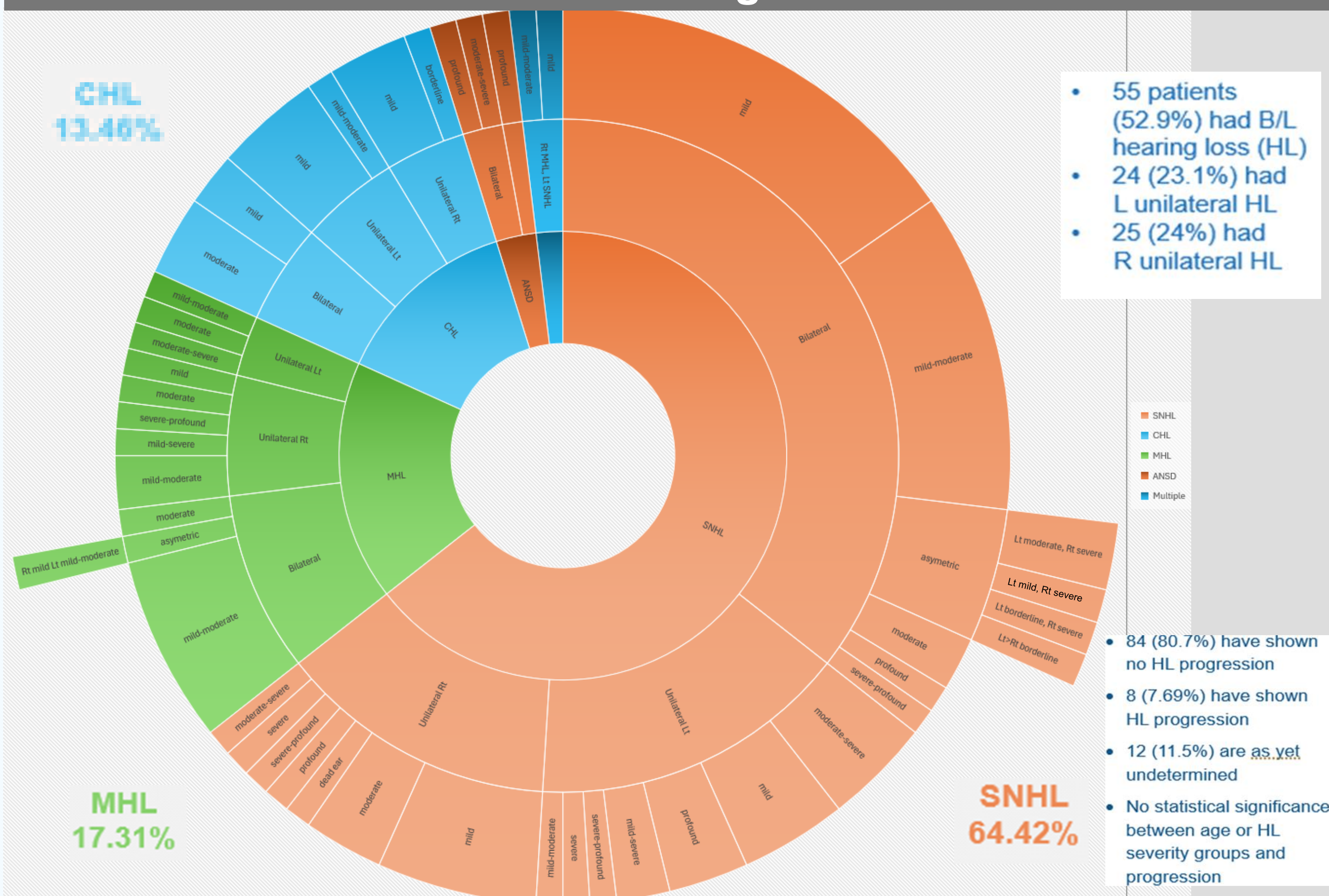
- 20 (19.2%) children had normal aetiological investigations.
- 35 (33.6%) had confirmed aetiology.
- 49 (47.1%) had aetiology not yet determined due to:

- 33 incomplete/pending investigations (67.3%)
- 8 awaiting aetiological appointments (16.3%)
- 4 declining further input (8.1%)
- 1 non-attending (2%)
- 2 discharged due to transition to adulthood (4%)
- 1 moving out of catchment area (2%)

Results - Referral Pathways



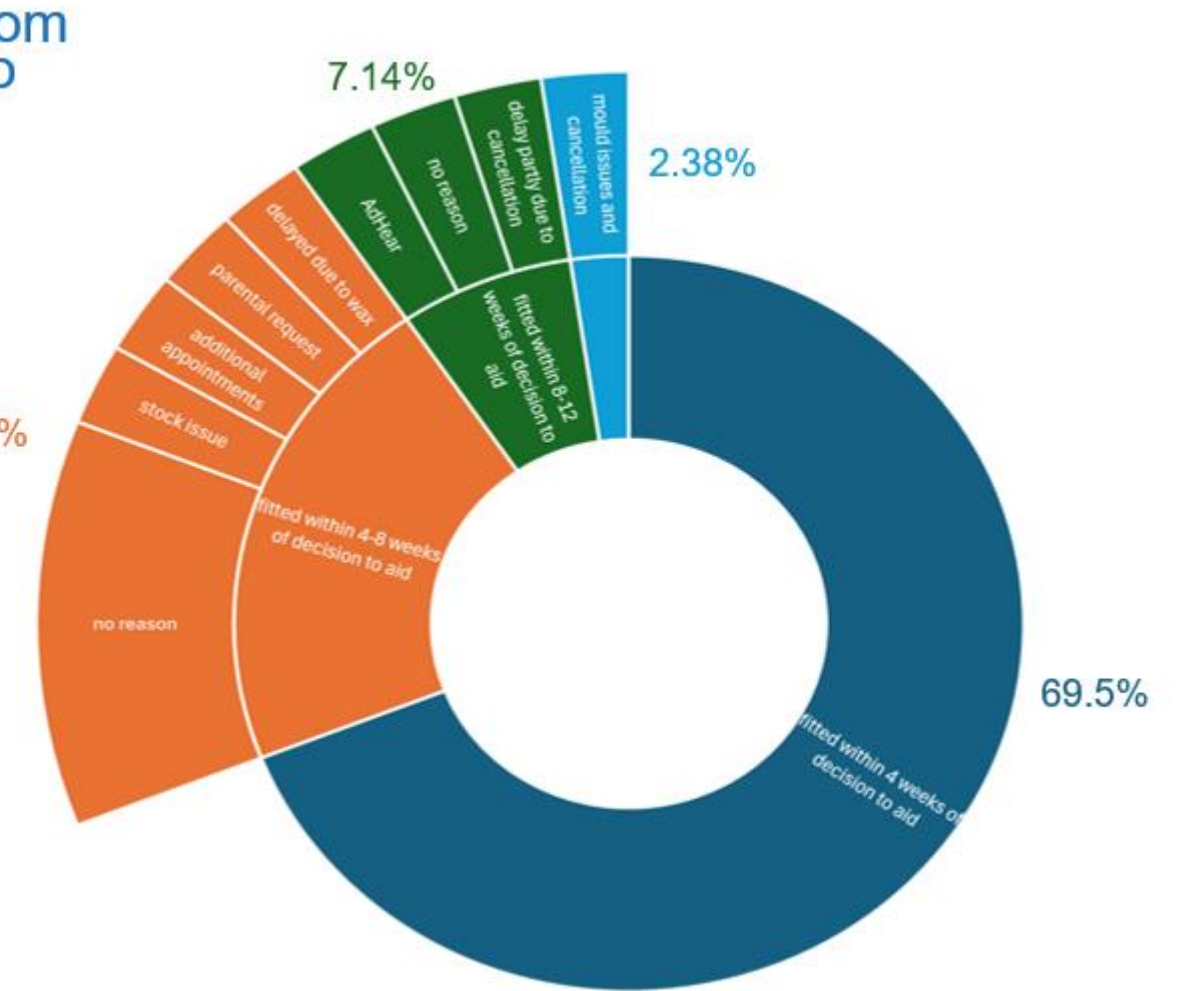
Results - Audiological Profile



Results - Hearing

- **Comparison between ≤ 5 year old and > 5 year old groups:**
 - Hearing loss in the right ear was found to be more severe in the ≤ 5 year old group than in the older patients (30.0% vs 10.7%, p-value=0.019).
 - ANSD and CHL hearing loss types were more frequent in the younger patients in both ears (10% vs 1.2% & 25% vs 4.8%, p-value=0.001 for the right ear and 10% vs 0% & 15% vs 8.3%, p-value=0.012 for the left ear).
- **Intervention:**
 - Of the 104 children reviewed 42 (40.38%) were eligible and provided with intervention of some kind.
 - Intervention included contact mini (CM), hearing aids (HA), CROS aids, bone conduction hearing aids (BCHA) and cochlear implant (CI).
 - Of these 42 children, 23 (54.76%) were provided with intervention within 4 weeks of confirmation of hearing loss, including decision to aid time.

Results - Time from Decision to Aid to Fitting in Weeks



Recommendations

- Number of children were identified as overdue for review and aetiological investigations. Each case will be escalated for appointment as soon as possible.
- Recording of PCHI cases (including late-onset), although improving in recent years, has been somewhat patchy, with multiple documents and spreadsheets used by different clinicians. Going forward, the department should consider alternative methods of monitoring/recording PCHI cases to make future audit easier. Auditable parameters/alerts could be one such method.
- Best efforts to ensure amplification fittings are offered within 4 weeks of diagnosis/decision to aid in line with BSA/BAA recommendations should continue. If a patient cannot be provided with an appointment for intervention within 4 weeks, then the reasons why should be clearly documented.
- Importance of continuing to ensure consistency in aetiological investigation requests and clear documentation of any clinical deviation from BAAP guidelines.

Conclusions

- Late-onset PCHI is under-recognised, under-studied and often missed.
- School entry screening programmes are invaluable in identification of late-onset PCHI cases.
- Multidisciplinary efforts are paramount in ensuring optimal and timely diagnosis, decision making and bespoke management.
- Aetiological investigations, including vestibular assessments work synergistically to improve yield and inform diagnosis of late-onset PCHI.
- Consistency in documentation is crucial to minimise missed cases and delays in diagnosis and management.
- Late-onset cases present with a diverse audiovestibular profile.
- In late-onset PCHI, natural history of aetiology is crucial in explaining clinical timelines, prognosis and need for ongoing monitoring.

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