Diagnostic yield of identifying a genetic cause of hearing loss in children after the introduction of the Next Generation Sequencing Panel
R67 gene panel for monogenic hearing loss

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BACKGROUND

- 50% of permanent hearing loss in children can be due to genetic factors. In our study, NGSP achieved 30.91% diagnostic yield.

AIMS AND OBJECTIVES

- To determine the diagnostic yield of NGSP R67 in identifying a genetic cause of SNHL in children with bilateral SNHL.
- Obtain a practical and a realistic idea about the diagnostic yield when compared with existing published evidence (25-33% yield).
- Establish new audit standards incorporating the yield, the logistics of genetic referrals and outcomes.
- Adopt the test as standard practice to investigate all cases of paediatric bilateral SNHL.

RESULTS

- The R67 panel is recommended for those with confirmed, bilateral sensorineural hearing loss (SNHL).
- Since the introduction of R67 in 2021 in UK, this is the first audit of its kind in an Audiovestibular Department to explore the diagnostic yield for bilateral PCHI in children.

FUTURE STEPS

- Re-audit in another year to see if the diagnostic rates of the region increase for the year, after implementing NGSP as first line aetiological investigation alongside existing audiological and medical tests.
- Share this audit with national genetic services
- Educate and improve awareness and knowledge to improve HL clinical practice.
- Publish this audit.

REFERENCES:


DISCUSSION AND CONCLUSION

- In our department, NGSP achieved 30.91% diagnostic yield.
- This is a high diagnostic yield, indicating the beneficial application of NGSP as a standard.
- Our reference papers for diagnostic yield came from two studies, one using single-gene and custom gene panels, and the other using whole-exome sequencing, which had diagnostic yields of 25 and 33% respectively.
- However, the most important observation was that it picked up rare and uncommon genes that would have been impossible to diagnose before NGSP.
- We suggest that NGSP is a beneficial tool to add to hospital guidelines, in a tiered approach to diagnosing HL aetiology. This should be used in combination with a comprehensive clinical history, examination, first-line investigations (audiology, neuro-otology and scans) and MTX advice from different specialties including Clinical Geneticists. Effective utilisation of R67 is recommended.