Estimating the prevalence of genetic testing among children who are deaf or hard of hearing using healthcare claims data

Authors: Sana N. Charania, MPH[1]; Scott D. Grosse, PhD[1]; Kelly Dundon, AuD, MPH[2]; Stuart K. Shapira MD, PhD[1]

[1] National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA

[2] GenTech Associates, Inc., Indianapolis, IN

Background

The Joint Committee on Infant Hearing (JCIH) recommends that infants who are confirmed as deaf or hard of hearing (D/HH) are referred for a genetic evaluation.

Objectives:

1. To estimate the prevalence of genetic testing among insured young children who are D/HH compared to children not identified as D/HH.
2. To compare the prevalence of genetic testing among subgroups of children who are D/HH.

Methods

Data Source: IBM MarketScan Commercial claims database for two 3-year time periods, 2014-2016 and 2017-2019.

- Children identified as D/HH were aged 0-3 years and continuously enrolled for at least the first year in an employer-sponsored health plan.
- D/HH defined based on the presence of three or more claims on separate dates with an ICD-9 or ICD-10 diagnosis code for hearing loss.
- Identified claims associated with any type of cytogenetic or molecular genetic testing (e.g., karyotyping, chromosomal microarray, and tests for DNA variants in genes known to be specific to hearing loss).
- Calculated prevalence estimates of genetic testing among children with and without hearing loss.

Results

Figure 1. Study Sample

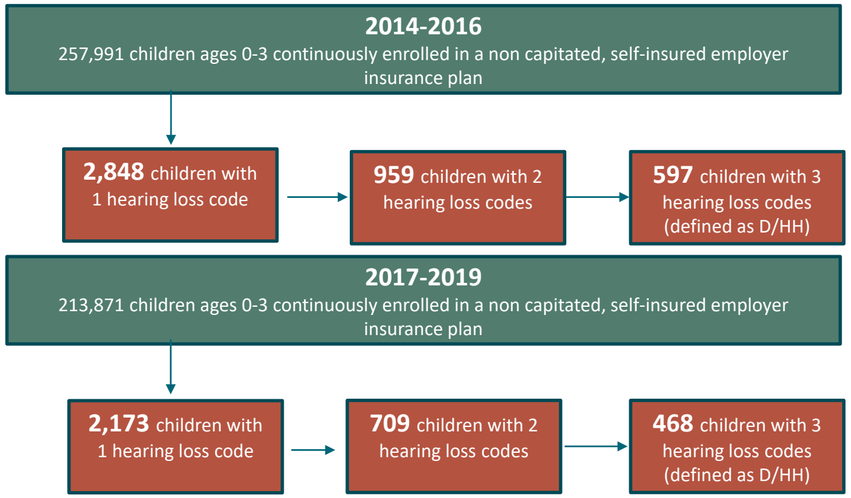
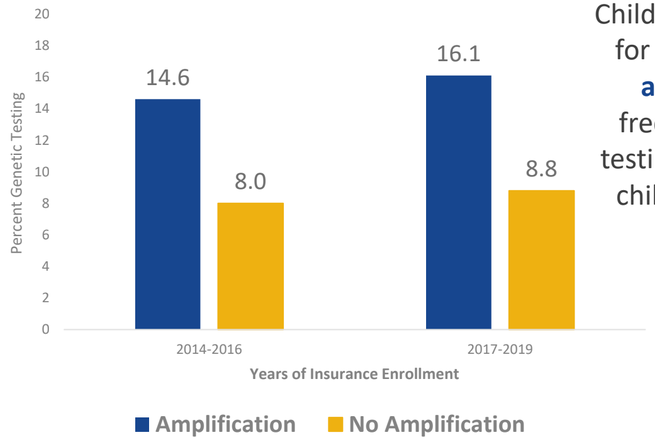


Figure 2. Prevalence of genetic testing among insured children who are D/HH vs children who are not D/HH (2017-2019)

13.0% of children who are **D/HH** received genetic testing vs **0.8%** who are **not D/HH**

Figure 3. Prevalence of genetic testing among children identified as D/HH who had claims for amplification



Children who had claims for **amplification** had **almost twice** the frequency of genetic testing relative to other children identified as D/HH

Conclusions

This analysis of-children identified as D/HH indicates a frequency of genetic testing among children identified as D/HH that is elevated relative to other children but low relative to JCIH guidelines. These findings suggest a need to increase genetic testing among children who are D/HH.

The data presented here are provisional and have not been published.

Contact Info

Sana Charania, NCBDDD
YJE9@cdc.gov