A Sponsored Genetic Testing and Counseling Program for Sensorineural Hearing Loss Reduces Barriers to Genetic Testing

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Sponsored by Eli Lilly and Company

PROGRAM OBJECTIVE

- Determine if removing some of the key barriers to genetic testing for individuals who are deaf or hard of hearing will increase genetic testing utilization and facilitate earlier and more accurate diagnosis

BACKGROUND

- Over 60% of congenital sensorineural hearing loss (SNHL) cases are due to genetic causes, yet few eligible individuals undergo genetic testing[1-2]
- A genetic diagnosis may inform care[3-7]:
  - Prognosis for hearing (stable vs. progressive)
  - Evaluation for syndromic causes
  - Personalized medicine (communication and treatment options, gene therapy clinical trials)
  - Empowerment/acceptance
- International Pediatric Otolaryngology Group (IPOG) and American College of Medical Genetics and Genomics (ACMG) recommend a comprehensive gene panel for bilateral SNHL and auditory neuropathy[8-9]
- Key barriers to genetic testing include insurance reimbursement, patient costs, provider education gaps, availability of genetic services, and logistical challenges[10-13]
- Hearing healthcare provider's role related to genetic testing may involve referral of individuals for genetic testing, counseling of individuals on potential benefits and limitations of genetic testing, guiding individuals through decision-making on intervention choices and potential need for change depending on the genetic form of SNHL

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METHODS

- A no-charge genetic testing and counseling program for SNHL was initiated in June 2024 and data reported through December 2025
- Testing was performed by targeted panel sequencing of 274 hearing loss associated genes by a CLIA-certified laboratory
- Individuals <40 years of age in the U.S. with at least one of the following are eligible:
  - Bilateral or unilateral SNHL (by auditory brainstem response or behavioral testing) of mild, moderate, moderately severe, severe, or profound degree
  - Auditory neuropathy
- Individual/parent informed consent was obtained, and demographic and clinical history were recorded on an intake form by the provider at the time of ordering genetic testing
- Deidentified data and genetic findings were reported to the program sponsor. No protected health information was given to the sponsor

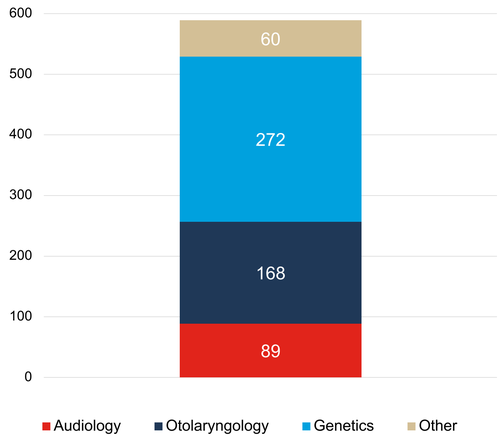
DEMOGRAPHICS

- Age ranged from 0 to 39 years with 84% under 18 years of age
- Hearing loss onset was predominantly congenital, accounting for 55% of cases
- Hearing level classification was 51% mild/moderate/moderately severe and 37% severe/profound
- Ethnicity/ancestry was consistent with the U.S. population

PROGRAM UTILIZATION

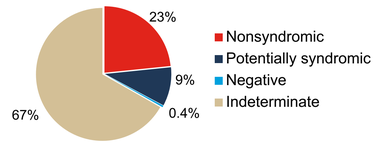
44% of referrals for genetic testing/counseling came from otolaryngologists and audiologists

Number of Ordering Providers by Specialty (n=589)

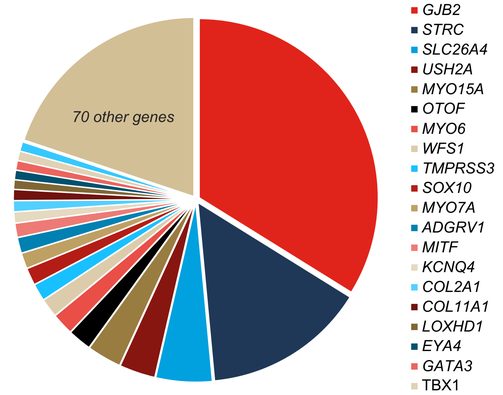


GENETIC DIAGNOSIS

Diagnosis (n=2,190)



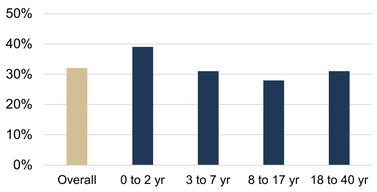
Positive Diagnosis (n=709)



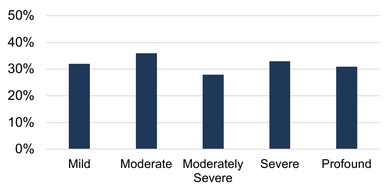
Other includes genes with ≤5 individuals diagnosed.

DIAGNOSTIC YIELD

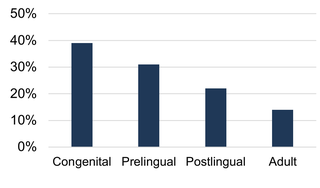
Age Group (n=2,190)



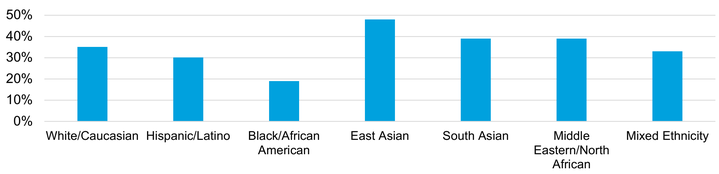
Hearing Level (n=2,190)



Hearing Loss Onset (n=2,190)

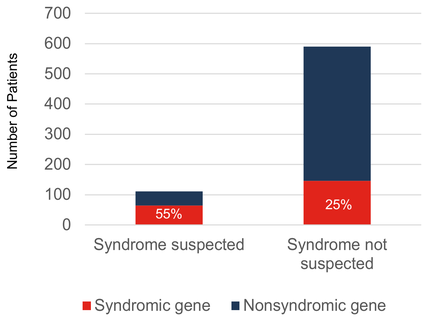


Ethnicity/Ancestry (n=2,190)

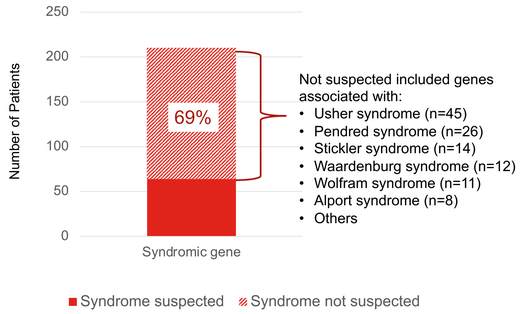


SYNDROMIC CONDITIONS

Positive Diagnosis (n=709)



Diagnosis Associated with a Syndromic Gene (n=212)



DISCUSSION

- Implementation of a sponsored genetic testing and counseling program for SNHL resulted in rapid uptake and yielded a meaningful diagnostic rate
- Accurate genetic diagnosis informs care through:
  - Identification of potentially syndromic conditions prior to symptom onset
  - Prognostication for changes in hearing levels over time (*STRC* typically stable vs. *SLC26A4*, *MYO15A*, *TMPRSS3* usually progressive)
  - Identification of potential gene therapy trial participants

References

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ACCESS GENETIC TEST ORDERING