Lend me your ears, the deafness continuum – navigating access to genetics education and services

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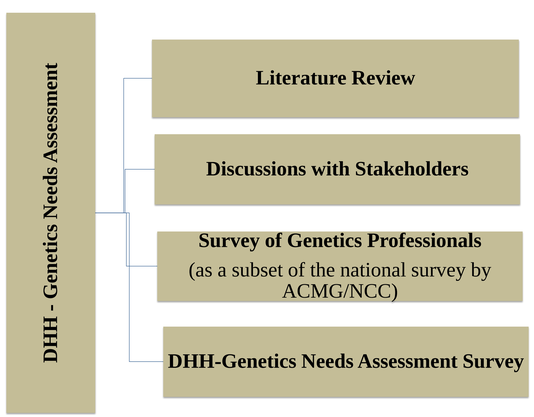
Background:

- Deafness affects 2-3/1,000 newborns. Early identification of Deaf or Hard of Hearing (**DHH**) infants and early intervention (**EI**) are critical to speech-language development.
- >100 genes are associated with hearing impairment. The spectrum of genetic testing for deafness ranges single gene testing (e.g., GJB2 and GJB6) to multi-gene panel testing.
- ~50% of infants with nonsyndromic deafness have identifiable gene changes in connexin 26 and/or 30 (GJB2/GJB6). Families with DHH children can benefit from timely genetic services.
- 12 LEND Audiology Programs (2 in NYMAC region) continue to augment pediatric audiology training efforts.
- HRSA continues to engage and promote family leadership in the EHDI system through the Universal Newborn Hearing Screening and Intervention program.
- Early Hearing Detection and Intervention (**EHDI**) programs support families with DHH children and their providers in appropriate screening (by the age of 1 month), diagnostic (3 mos) & intervention services (6 mos).
- Adding “9” (mos) to the 1-3-6-”9" goals could be a reminder to apply timeliness to the sharing of genetic educational resources & referring for genetic evaluation for deafness.
- NYMAC and the National Center for Hearing Assessment and Management (**NCHAM**) initiated a DHH-genetics needs assessment.

Methods:



Objectives of Deaf or Hard of Hearing (DHH) - Genetics Survey:

- Determine the unmet needs of physicians & health care providers related to genetics services for DHH children in the screening, diagnosis, and referral continuum.
- Identify areas where appropriate assistance can be provided to support physicians to increase family education about and genetic referrals for deafness.

The **long-term goal** is to use the findings to improve services for infants and children that are DHH as well as their families by integrating genetic services into the management of patients and families with deafness.

DHH-Genetics Needs Assessment-Survey Findings

- 219 providers in DC, DE, MD, NJ, NY, PA, VA, and WV completed the DHH-Genetics Survey.

Demographics of Respondents:

- Pediatrician - 43.1%; Family Medicine - 18.3%; Nurse Practitioner - 18.3%; Physician Assistant - 13.9%; Neonatologist - 3%; Other - 3.5%.
- Large metropolitan – 37.2%; Small metropolitan – 33.2%; Small Town – 18.4%; Rural Area – 11.2%.

Gaps or challenges:

- 80% did not know about EHDI (1-3-6) before this survey.
- ~41% respondents did not refer families to genetics.
- ~47 % were uninformed about DHH genetics.
- 55% were not confident about the genetics causes of DHH.
- ~44% never consulted genetics professionals.
- ~60% never ordered connexin testing for nonsyndromic DHH.
- ~37% were not confident about genetic referral importance.

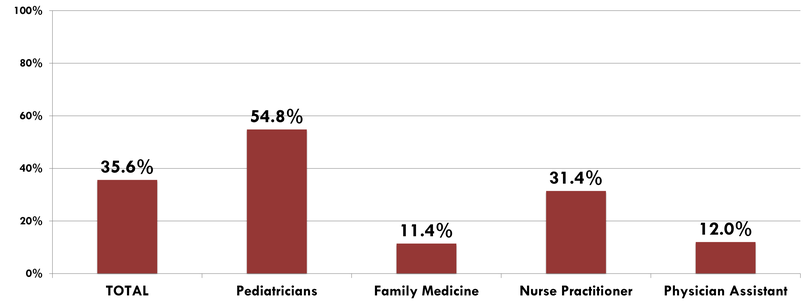
Referrals:

- ~49% & 24% referred parents of a DHH child identified via NBS to genetics often & sometimes respectively.
- ~40% cited FH as 1^{0}reason for genetics referral.
- ~31% and 21% thought lack of genetics professionals was always and sometimes a challenge when referring.
- ~70% referred a family with confirmed deafness to genetics.

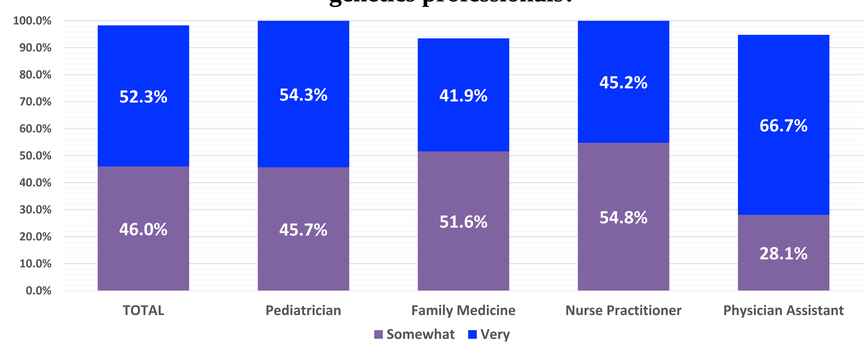
Strategies to facilitate tracking of genetics referral:

- System to follow-up with families (~93%).
- Increase genetics education of providers (~92%).
- System to follow-up with providers (~87%).
- Engage case managers in EHDI programs (~79%).

Have you referred parents of a child with hearing loss identified through NBHS to genetic professionals?



How important do you think it is to refer children with hearing loss to genetics professionals?



Next Steps:

- Continue interdisciplinary collaborations to provide technical assistance and supports to families and providers to manage genetic referrals for deafness.
- Determine the areas of appropriate intervention regarding genetics education.

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