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Where Do We Go From Here? The Need for Genetic Referrals in Patients who are Deaf or Hard of Hearing: Findings from a Regional Survey

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Abstract: Purpose. The purpose of this study was to assess primary health care providers' knowledge and use of genetic services for children whose hearing screening indicates they may be deaf or hard of hearing (DHH) and identify areas in which health care providers can be supported to increase family education and referral of families for genetic consultation.
Methodology. A survey was developed on current practices, knowledge, and perceived beliefs regarding genetic education and referrals for deafness. The surveys were distributed to pediatricians, family medicine physicians, nurse practitioners, and physician assistants in Washington DC, Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia.
Results. Among 266 respondents, 80% were uninformed about Early Hearing Detection Intervention (EHDI) 1-3-6 guidelines prior to taking the survey. Approximately 55% were not confident about the genetic causes of deafness, 44% rarely consulted genetics professionals, 41% had not referred families to genetics, and 37% were not confident about the importance of genetic referrals.
Conclusions. Integrated, targeted, and user-friendly genetics education strategies in the existing EHDI framework are needed to ensure adequate awareness and delivery of genetics services for children who are DHH.

Key Words: genetics, hearing loss, deafness, EHDI, provider survey, needs assessment

Acronyms: AAP = American Academy of Pediatrics; ACMG = American College of Medical Genetics and Genomics; CDC = Centers for Disease Control & Prevention; DHH = deaf or hard of hearing, EHDI = Early Hearing Detection and Intervention; EI = early intervention; HL = hearing loss; HRSA = Health Resources and Services Administration; LEND = Leadership Education in Neurodevelopmental and Related Disabilities; MCHB = Maternal & Child Health Bureau; MMS = Medical Marketing Services, Inc.; NBHS = Newborn Hearing Screening; NCHAM = National Center for Hearing Assessment and Management; NYMAC = New York–Mid-Atlantic Consortium for Genetic & Newborn Screening Services

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Approximately 3 to 4 per 1000 infants are born each year in the United States whose hearing thresholds indicate they are moderately, severely, or profoundly deaf or hard of hearing (DHH; Mercer, 2015). Early Hearing Detection and Intervention (EHDI) systems work to ensure timely identification and intervention for infants and toddlers who are DHH and include public health surveillance of newborn hearing screening (NBHS). The EHDI system has promoted 1-3-6 guidelines, recommending newborn screening no later than 1 month of age, evaluation of those infants who do not pass their hearing screening by 3 months of age, and enrollment in early intervention by 6 months of age. 1-3-6 has been shown to positively impact speech and language development for those identified as DHH (Moeller, White, & Shisler, 2006). More than 95% of all newborns[1] in the United States have their hearing screened at birth through NBHS with federal and state support (Muñoz, Shisler, Moeller, & White, 2009).

Progress in genetics has led to the identification of multiple genes causing non-syndromic and syndromic impacts on hearing levels, with over 400 genes now identified (Toriello, Reardon, & Gorlin, 2004). The majority of genetic causes (about 70%) are non-syndromic with more than half of identifiable variants or gene changes in two genes, *GJB2* and *GJB6*. These genes are associated with moderate to profound bilateral, sensorineural, and non-progressive impacts on hearing levels (Shearer, Hildebrand, & Smith, 2017). About 30% of children who are DHH with a genetic component have associated physical and clinical features such as retinitis pigmentosa (Usher syndrome), inner ear deformities and thyroid goiter (Pendred syndrome), cardiac arrhythmias (Jervell and Lange-Nelson syndrome), and renal malformations (Branchiootorenal syndrome; Shearer et al., 2017). The co-morbidities associated with these syndromes warrant additional medical assessments in newborns who are DHH. Previous studies have described how genetics evaluations can be incorporated into the EHDI process and benefit the parents of children with hearing impairment (Mercer, 2015; Muñoz et al., 2009; Schimmenti et al. 2004; White, 2004).

The benefits of an early genetic evaluation of newborns who are identified as DHH are numerous. First, a genetic evaluation can help families understand why their child is DHH, potentially reducing unnecessary concerns. Second, genetic evaluation can provide families with additional information about the services that might be helpful for their child and family. Given this information, families may consider reaching out to other families who have children with similar genetic backgrounds, building a stronger support system. Third, genetic evaluation can support a more thorough formation of a personalized medical care plan, thereby empowering families to obtain better care. This formation of a medical care plan can also provide additional connections to necessary medical and psychosocial support services. Further, an early genetic evaluation may help families better understand the link between hearing loss and genetics, and can provide an opportunity to discuss recurrence risk with genetic professionals.

Recent literature indicates that genetic services are under-utilized. A 2005 survey conducted by the National Center for Hearing Assessment and Management (NCHAM) indicated only 8.8% of physicians (n = 1,968) routinely refer a child who is DHH to a geneticist (Moeller et al., 2006). The referral rate for audiologists and otolaryngologists was higher, with 56% of audiologists reporting that they would *often* or *always* refer a hearing couple with one child who is DHH for genetic counseling (Connelly, 2010). When asked what initial set of tests they use in an infant with confirmed nonsyndromic sensorineural hearing loss, 49% of otolaryngologists reported they order a genetics evaluation (Duncan, Prucka, Wiatrak, Smith, & Robin, 2007). A simulation-based survey found that 37% of otolaryngologists and geneticists ordered a genetic consultation on the first encounter of sensorineural hearing loss and 30% did so on the second encounter (Jayawardena, Shearer, & Smith, 2015). Although physicians recognize the importance of genetics evaluation for children that are DHH, various challenges continue to persist in making physician referrals to genetics services. The 2005 NCHAM survey found that 90% of physicians perceived there being *somewhat of a need* or *a great need* for training and/or resources on genetics and DHH (Moeller et al., 2006). Furthermore, physicians have cited lack of appropriate education or training to make referrals and uncertainty about the usefulness of genetics (Connelly, 2010).

Because of the known under-utilization of genetic services by primary care providers caring for individuals who are DHH, we sought to assess primary health care providers' knowledge and use of genetic services for children who are DHH. We further sought to identify areas in which health care providers can be supported in increasing family education and referral of families for genetic consultation. The long-term goal of this study is to improve services for infants and children who are DHH as well as their families by integrating genetic services into the management of patients who are DHH.

Method

Instrument

The hearing loss needs assessment was developed through a collaboration between the New York–Mid-Atlantic Consortium for Genetic & Newborn Screening Services (NYMAC) and the National Center for Hearing Assessment and Management (NCHAM). NYMAC is one of seven regional genetics collaboratives funded by the Health Resources and Services Administration (HRSA). The region encompasses the District of Columbia, Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia. Two previous NCHAM surveys (distributed in 2005 and 2012),

[1]An updated percentage of newborns receiving hearing screening can be found at https://www.cdc.gov/ncbddd/hearingloss/ehdi-data2015.html

literature reviews, and ongoing discussions with the NYMAC region EHDI coordinators and the Leadership Education in Neurodevelopmental and Related Disabilities (LEND) faculty guided the design of the survey. The survey was designed to identify the unmet needs of children, families, and professionals regarding use of genetics in hearing loss screening, diagnosis, and referral.

The 10–20 minute survey was available in paper and electronic format and consisted of 32 questions divided into four sections: (a) demographic information, (b) knowledge and beliefs about genetic referrals for children with hearing loss, (c) current practice regarding referrals for children with hearing loss, and (d) resources and strategies needed (see Appendix for a copy of the survey). The survey also included links to available resources, including the American College of Medical Genetics and Genomics (ACMG) Action Sheet on patients who are DHH and genetics. Notably, DHH was defined as permanent, bilateral or unilateral, sensorineural or conductive hearing loss of an average loss of 30 decibels or more in the frequency range important for speech recognition.

The Johns Hopkins University and the Utah State University Institutional Review Boards approved the study.

Participants and Procedures

EHDI coordinators in the NYMAC region were contacted by NCHAM with study information, the paper survey, a pre-addressed and stamped envelope, instructions for completion, and a URL to the electronic version of the survey. EHDI coordinators then contacted pediatricians, family medicine physicians, nurse practitioners, and physician assistants within their states to invite them to complete the needs assessment. Respondents were encouraged to share the survey link with fellow providers to enhance uptake. Respondents were asked to complete only one survey (paper or electronic). Contact information for NYMAC and NCHAM was provided.

A follow-up email reminder to the target population was sent via Medical Marketing Services, Inc. (MMS), a professional service provider. MMS estimated that 17,974 physicians, 4,837 advance practice nurse or nurse practitioners, and 1,373 physician assistants received the email blast. Two written reminders and one e-mailed reminder were sent to EHDI coordinators to encourage providers in their states to complete the needs assessment. The paper survey was re-sent to providers in the state of Delaware only.

Analytic Strategy

Responses to demographic questions, questions related to knowledge and beliefs about genetic hearing loss referrals, current practices for hearing loss referrals, and resources and strategies are reported. All comparisons across groups (i.e., disciplines) herein were carried out using chi-square tests of independence. All analyses were conducted using the R statistical environment version 3.4.2 (R Core Team, 2018).[2]

Differences in the number of genetic referrals, timing of genetic referrals, and reasons for genetic referrals were examined by professional discipline. Differences in the perceived frequencies of challenges (*not a challenge, sometimes a challenge, always a challenge, not applicable*) relating to typical challenges experienced by clinical professionals were examined by reasons for the genetic referrals as well.

Results

Sample Characteristics

A total of 266 participants across 8 states and the District of Columbia completed the survey. Participants were allowed to skip questions; therefore, the response rate varied per question. Table 1 presents demographic information regarding the participants, including their specialty, experience, and practice information. Of the respondents, 47% were pediatricians, 53% worked in private practice, most (68%) worked in either a large or small metropolitan area, and 42% had more than 20 years of experience.

Table 1
Demographic Information about the Participants

	Percent Total *N* = 266
Specialty	*n* = 247
Family Medicine Physician	18%
Neonatologist	3%
Nurse Practitioner	17%
Pediatrician	47%
Physician Assistant	11%
Other	4%
Practice Setting	*n* = 244
Community clinic	13%
Federally Qualified Health Centers	7%
Hospital setting	15%
Medical school or parent university	3%
Private practice	53%
Other	9%
Practice Location	*n* = 240
Large metropolitan area	34%
Small metropolitan area	34%
Rural area	11%
Small town	20%
Years of Experience	*n* = 239
Less than 1 year	5%
1-2 years	7%
3-5 years	9%
6-9 years	13%
10-19 years	25%
More than 20 years	42%

[2]All code and data used in the present study are provided at: https://osf.io/8thwf

Provider Knowledge and Beliefs

A total of 266 participants across 8 states and the District of Columbia completed the survey. Participants were allowed to skip questions; therefore, the response rate varied per question. Table 1 presents demographic information regarding the participants, including their specialty, experience, and practice information. Of the respondents, 47% were pediatricians, 53% worked in private practice, most (68%) worked in either a large or small metropolitan area, and 42% had more than 20 years of experience.

Table 2 shows the responses regarding the participants' levels of confidence in speaking with parents of a child with permanent hearing loss about the genetic causes (52% not confident), the importance of genetic referrals (33% not confident), the logistics of genetic referrals (3% not confident), and the significance of genetics in hearing loss due to ototoxic medication exposure (43% not confident).

Current Practice

Approximately 41% of the participants have referred a family to a genetics specialist because there was a family history of hearing loss, 4.3% because the parents

Table 2
Responses about the Confidence Level in Speaking with Parents of a Child with Permanent Hearing Loss about Genetic Causes of Hearing Loss and Genetic Referrals

	Percent Total *N* = 266
Genetic causes of hearing loss	*n* = 225
Not confident	52%
Somewhat confident	40%
Very confident	8%
The importance of genetic referrals	*n* = 222
Not confident	33%
Somewhat confident	44%
Very confident	23%
The logistics of genetic referral for hearing loss	*n* = 221
Not confident	39%
Somewhat confident	43%
Very confident	18%
The significance of genetics in hearing loss due to ototoxic medication exposure	*n* = 221
Not confident	42%
Somewhat confident	39%
Very confident	17%

Table 3
Referrals by Discipline

	Pediatrician *n* = 116	Nurse Practitioner *n* = 41	Family Medicine Physician *n* = 45	Physician Assistant *n* = 27	Neo-natologist *n* = 8	Other *n* = 10	*P*-value
How many genetic referrals have been made							0.323
0–10	97%	100%	100%	100%	75%	100%	0-10
11–20	1%	0%	0%	0%	25%	0%	11-20
50+	1%	0%	0%	0%	0%	0%	50+
When genetic referral is made							0.178
At diagnosis	6%	22%	43%	0%	0%	0%	
Enroll in EI	71%	67%	29%	50%	100%	100%	
Screening	14%	0%	29%	50%	0%	0%	
Other	9%	11%	0%	0%	0%	0%	
Reason for Genetic Referral							<.001
I have not referred families to a genetics professional for hearing loss	16%	49%	54%	88%	14%	50%	
Parents are discussing another pregnancy	5%	5%	5%	0%	0%	0%	
Parents ask for a referral	3%	5%	7%	4%	0%	0%	
There is a family history of hearing loss (parents, siblings, cousins, etc.)	58%	33%	22%	4%	43%	38%	
Other	18%	8%	12%	4%	43%	13%	

Note. EI = Early Intervention. Due to missing values in responding discipline, this table includes 247 responses.

were discussing another pregnancy, and 3.9% because the parents asked for a referral. However over a third (37.1%) of the participants stated that they had not made a genetic referral. Nearly 40% of all participants indicated they have not consulted with a genetics specialist without a formal referral, with another 39% rarely consulting with a genetics specialist. The majority of participants (59%) had never ordered eConnexin 26 testing in infants with nonsyndromic hearing loss.

Table 3 shows the number of referrals, when referrals are made, and for what reason by discipline. Although there was a statistically significant difference of the reason for referral by discipline ($p < .001$), further analyses showed that physician assistants (12%), family medicine physicians (46%), and nurse practitioners (51%) were less likely to have made a referral as compared to pediatricians (84%; $p < .001$). Beyond this, there were no differences between disciplines regarding reasons for a referral ($p = .435$).

In addition, several participants indicated that they did not have tracking in place for failed newborn hearing screening (30%), failed newborn blood spot screening (26%), delayed developmental milestones (31%), and follow-up after referrals (32%). Of those that received reports of the newborn hearing screening, nearly 80% say they sometimes or often refer children to a genetics referral. Notably, however, this differed by discipline with family medicine physicians rarely making a genetic referral in these situations compared to other professionals ($p = .002$).

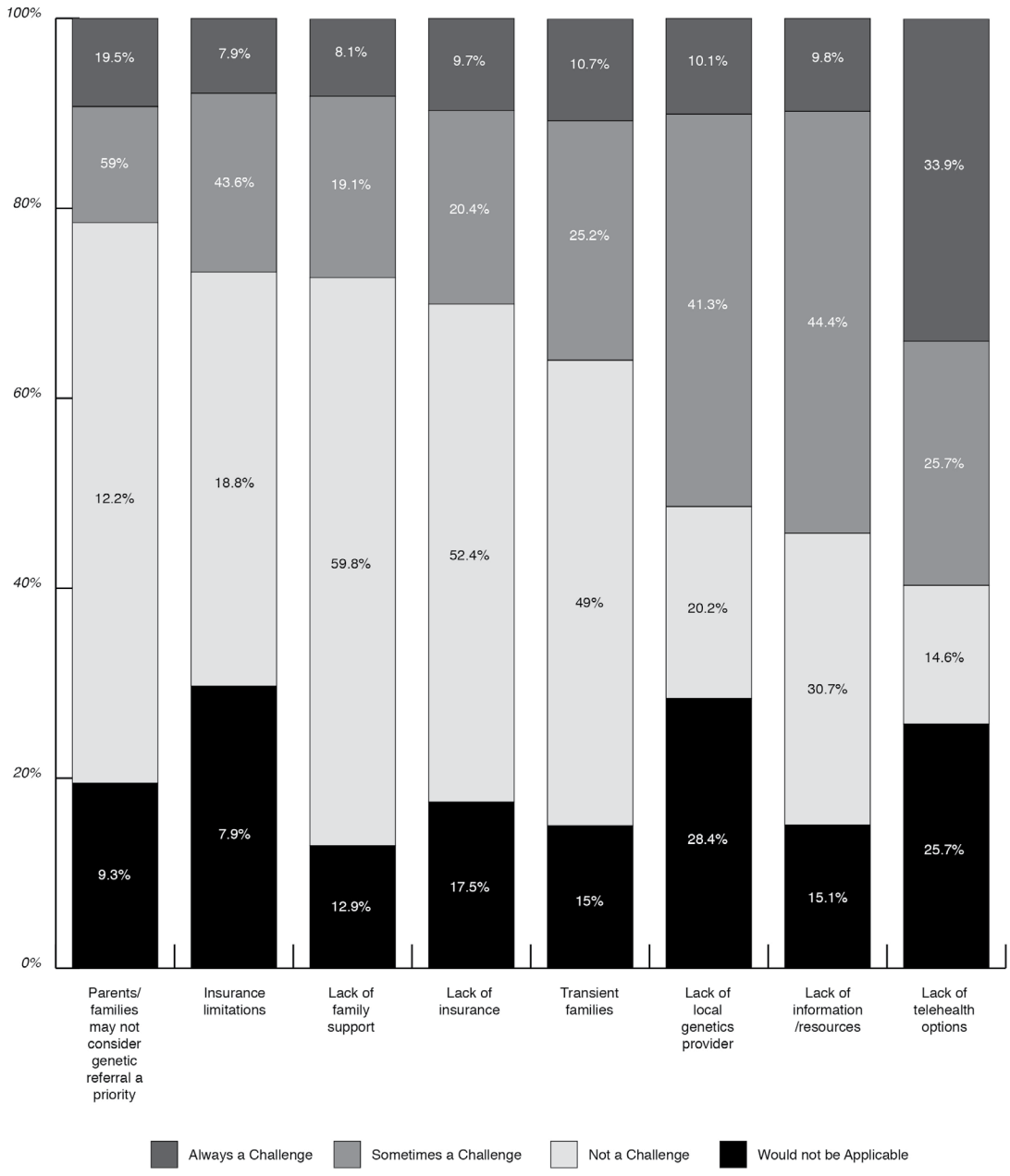

Figure 1. **Responses regarding challenges faced about the genetics of hearing loss.**

Challenges and Opportunities

A final question was asked regarding challenges professionals face in respect to making genetic referrals for hearing loss (see Figure 1). The greatest challenge perceived by the survey participants when making genetic referrals were the parent/family priorities about genetic referrals (78.5% indicating *always or sometimes a challenge*) followed by the family's inability to attend an appointment due to logistics, such as transportation or child care coverage (72.7% saying *always or sometimes a challenge*). More than half of participants indicated that lack of information to make the referral was a challenge as well. Neonatologists (83%) and pediatricians (65%) said lack of information/resources was not a challenge compared to family medicine physicians (11%), nurse practitioners (21%), and physician assistants (24%; $p < .001$).

A majority (84%) reported that a handout with resources on genetics of hearing loss for providers and families would be very helpful (Figure 2). Similar responses were given for both a quick reference guide about genetic referrals (82%) and contact information about genetic professionals in their area (74%). Educational webinars were far less popular with only 36.3% of participants indicating that webinars would be very helpful.

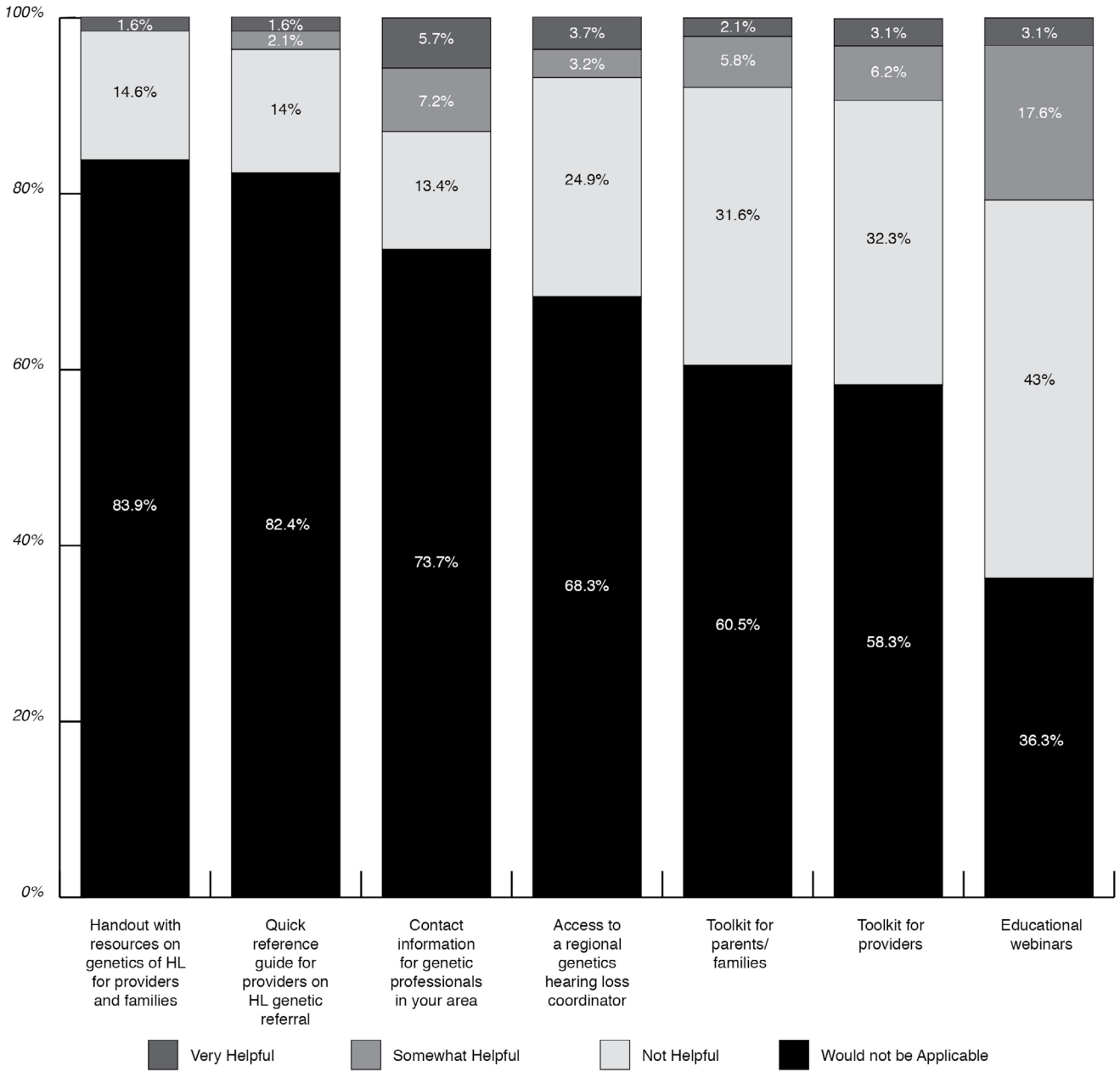

Figure 2. **Perceived needs of the participants regarding training, information, and other resources about genetics and hearing loss (HL).**

Discussion

These results strongly support the need for education on genetics and patients who are DHH for primary care providers, as 43% of respondents indicated they lack information on this topic. Data from the current study supports a prior physician survey on congenital hearing loss, in which approximately 40% of respondents perceived a great need for training and/or resources on genetics and DHH (Moeller et al., 2006).

In addition to education on genetics and patients who are DHH, there is a need for public health surveillance to ensure newborns identified as DHH through hearing screening have a genetic evaluation. As noted in our results, many participants do not have tracking mechanisms in place for NBHS and other infant health processes. However, several states in the NYMAC region do have existing mechanisms for incorporating genetics into their EHDI program activities. In Virginia, parents are called after a newborn is identified as DHH to verify enrollment in early intervention services and the completion of the diagnostic work-up, including a genetics evaluation. In New Jersey, primary care providers are sent a checklist of necessary post-diagnosis evaluations including genetics after identification. In Delaware, all newborns are referred to audiology at a single site, which allows for tracking of the post-diagnostic work-up. Going forward, HRSA-funded EHDI programs will be held responsible for improving care coordination through the patient/family-centered medical home model. Programs are required to report the number of care coordination plans developed with the parent or family and the number of care coordination plans that are shared across providers. This new, funded activity provides EHDI programs an opportunity to incorporate genetic evaluations into care plans and to include the sharing of care plans with the genetics provider.

Primary care provider education and public health surveillance are key to improving access to genetic services for newborns that are DHH, but based on our results, other barriers exist. Although we found that primary care providers perceive family acceptance as a barrier, a previous survey of parents of children who are DHH indicated that about 96% of parents—of whom none were DHH and about a quarter reported a family member born deaf—had a positive attitude toward genetic evaluation. A broader community of hearing, deaf, and hard-of-hearing adults supported genetic evaluation for newborns and expressed their willingness to engage in genetic counseling. Most families of children who are DHH value the availability of genetic testing as a means of better understanding the cause of the hearing loss and promoting discussion of the condition. (Geelhoed, Harrison, Davey, & Walpole, 2009).

Response to Needs

In response to the identified need for genetics education, NYMAC conducted an educational campaign using professional marketing (Figure 3) from May 1, 2017 through May 29, 2017. The campaign targeted pediatricians from New York, Delaware, Washington DC, Maryland, New Jersey, Pennsylvania, Virginia, and West Virginia. Standard desktop and tablet banners were placed on the American Academy of Pediatrics and AAP Publications websites. The campaign reached 215,949 pediatricians. This campaign represents one approach to provide education on genetics and hearing loss, but novel methods for ongoing education is needed—not only for this topic, but for genetics as a whole. In the survey, providers indicated a preference for handouts (83.9%), a quick reference guide (82.4%), and contact information about genetic professionals (73.7%). Although preparation of these materials is straightforward, incorporating them into practice is likely to be more challenging.

Figure 3. **Ad used for educational campaign on the effect of genetics on being deaf or hard of hearing in the New York–Mid-Atlantic Consortium for Genetic & Newborn Screening Services Region.**

Results also showed that providers perceive appointment logistics as a barrier to families receiving genetics evaluations. NYMAC is piloting several methods to reduce barriers related to appointment logistics. These methods include a phone line for help identifying a genetic service provider, expansion of telegenetics services, primary care provider education, and a formal relationship between primary care providers and a geneticist to review cases. The HRSA-funded Regional Genetics Networks are piloting different approaches to improve access to genetic services. Individuals with a variety of genetic conditions, including children who are DHH, will benefit from these nationwide activities.

Notably, there are a number of limitations to the study. First, the survey response rate was low, with only 266 of more than 24,000 professionals returning the survey. This indicates that the sampling may be biased toward certain groups. It is therefore difficult to assess the generalizability of this single sample. However, this may not be a major limitation given the corroboration between these results and prior results mentioned earlier. Second, there were missing values for many questions. For all tables, the number of participants that answered each question was noted.

Conclusion

Ultimately, this study may highlight the challenges and needs for healthcare professionals in their work with children who are DHH. Results indicate that there is a need for integrated, targeted, and user-friendly genetics education strategies for providers of children who are DHH, to ensure adequate awareness and delivery of genetics services for these children. This could include early intervention providers, as they may be able to encourage families to learn more about the genetic evaluation process if they have not pursued this. With recommendations coming from multiple sources, parents/family members may progressively become more interested in understanding their child's genetic background.

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Appendix

The following is what was presented to the online surveys. The paper surveys are extremely similar in appearance.

Each year, 3 in 1,000 infants are born in the US with moderate, severe, or profound hearing loss (HL). By age 19, 15% of adolescents have HL in one or both ears. Newborn hearing screening (NBHS) is included in the Recommended Universal Screening Panel for newborns. The national and state Early Hearing Detection and Intervention (EHDI) programs funded by the Centers for Disease Control & Prevention (CDC) and the Maternal & Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA), in cooperation with professional societies, support families of children with HL and their providers based on the EHDI 1-3-6 guidelines:

- Screening by 1 month of age
- Diagnosis of HL by 3 months of age
- Entry into early intervention (EI) services by 6 months of age

The objectives of this research survey are to

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

This research survey's long-term goal is to use the findings to improve services for infants and children with HL as well as their families by integrating genetic services into the management of patients and families with HL.

Your completion of this survey or questionnaire will serve as your consent to be in this research study. Please take about 10–20 minutes to tell us about your experiences. Your responses are completely confidential and will be used to improve services for infants and young children with hearing loss. Your participation is greatly appreciated.

1. Please indicate your profession:

- ○ Pediatrician
- ○ Family Medicine Physician
- ○ Otolaryngologist (ENT)
- ○ Neonatalogist
- ○ Nurse Practitioner
- ○ Physician Assistant
- ○ Resident/Fellow (specify) _____

Other _____

2. Practice setting where you spend most of your time:

- ○ Private Package
- ○ Community clinic
- ○ Hospital setting
- ○ Medical school/parent university
- ○ Federally Qualified Health Centers
- ○ Other _____

3. Practice Location:

- ◯ Small metropolitan area
- ◯ Large metropolitan area
- ◯ Small town
- ◯ Rural Areas

4. State/District:

_____

5. Year(s) of practice with pediatric population:

- ◯ Less than a year
- ◯ 1–2
- ◯ 3–5
- ◯ 6–9
- ◯ 10–19
- ◯ More than 20 years

Hearing Screening Genetics
Hearing Loss that is permanent, bilateral or unilateral, sensorineural or conductive, and averaging loss of 30 decibels or more in the frequency range important for speech recognition. The following questions are about children who were identified through newborn hearing screening (NBHS) as having hearing loss.

6. Do you receive reports about children who have failed their newborn hearing screening (NBHS)?

- ◯ Yes
- ◯ No

7. Have you referred parents who have a child with hearing loss identified through NBHS to genetics professionals? (*If you answered "No" to this question, you will skip to Question #11*)

- ◯ Yes
- ◯ No

8. If/When you have a child with hearing loss identified through NBHS, how often do you refer the parents to genetics professionals?

- ◯ Rarely
- ◯ Sometimes
- ◯ Often
- ◯ Unsure
- ◯ Not applicable

9. In the last year, approximately how many patients did you refer for genetic evaluation of hearing loss after an abnormal NBHS result:

- ◯ 0–10
- ◯ 11–20
- ◯ 21–50
- ◯ >50

10. For infants identified through NBHS as having hearing loss, what is your best estimate of the earliest stage at which:

	At the time of screening	At the time of diagnosis	At the time of enrollment in early intervention	Other (please specify)
Parents/family need to be informed about genetic referrals	◯	◯	◯	◯
Genetic referral should be made	◯	◯	◯	◯

Other Identification and Genetics
The following questions are about children who were identified as having hearing loss via means OTHER THAN newborn hearing screening (NBHS).

11. Have you referred parents who have a child with hearing loss identified through means other than NBHS to genetics professionals? (*If you answered "No" to this question, you will skip to Q15*)

- ◯ Yes
- ◯ No

12. If/When you have a child with hearing loss identified through means other than NBHS, how often do you refer the parents to genetics professionals?

- ◯ Rarely
- ◯ Sometimes
- ◯ Often
- ◯ Unsure
- ◯ Not applicable

13. In the last year, approximately how many patients did you refer for genetic evaluation of hearing loss after identification through a means other than NBHS?

- ◯ 0–10
- ◯ 11–20
- ◯ 21–50
- ◯ >50

14. For patients identified through a means other than NBHS as having hearing loss what is your best estimate of the earliest stage at which:

	At the time of screening	At the time of diagnosis	At the time of enrollment in early intervention	Other (please specify)
Parents/family need to be informed about genetic referrals	○	○	○	○
Genetic referral should be made	○	○	○	○

All Children with Hearing Loss and Genetics
The following questions are about ALL children with hearing loss, regardless of how they were identified.

15. Which is the primary reason why you refer families to a genetics professional for hearing loss?

- ○ Parents ask for a referral
- ○ There is a family history of hearing loss
- ○ Parents are discussing another pregnancy
- ○ I have not referred to a genetics professional for hearing loss
- ○ Other_____

16. How often do you consult with (i.e., do not make a formal referral) geneticists and genetic counselors regarding hearing loss?

- ○ Never (1)
- ○ Rarely (2)
- ○ Sometimes (3)
- ○ Often (4)
- ○ Unsure (5)

17. What challenges have you experienced when referring parents/families for a genetic evaluation of the infant/child?

	Not a Challenge	Sometimes a Challenge	Always a Challenge	Not Applicable
Lack of information/ resources to make referrals	○	○	○	○
Lack of insurance	○	○	○	○
Insurance limitations	○	○	○	○
Lack of family support	○	○	○	○
Transient families	○	○	○	○
Parents/families may not consider genetic referral a priority	○	○	○	○
Lack of local genetics provider	○	○	○	○
Lack of telehealth options	○	○	○	○
Other	○	○	○	○

Other _____

18. What reimbursement challenges have your patients encountered regarding hearing loss genetic referrals?

	Not a Challenge	Sometimes a Challenge	Always a Challenge	Not Applicable
Lack of insurance	○	○	○	○
Medicaid constraints and/or cost limitations	○	○	○	○
Preexisting or other regulations and policies	○	○	○	○
Preauthorization	○	○	○	○
Other	○	○	○	○

Other _____

19. For parents/families who already have a diagnosis of hearing loss, do you discuss with them the genetics of hearing loss?

- ○ No
- ○ Yes, sometimes
- ○ Yes, always
- ○ Not applicable

20. It is possible that infants with nonsydromic hearing loss have indentifiable gene changes in Connexin 26 and/or 30 (GJB2/GJB6). At what age (in months) do you thing connexin testing should be offered in a failed newborn hearing screen workup when there are not dysmorphic features/anomalies or a known genetic condition?

- ○ Age (in months) _____
- ○ Don't think connexin testing should be ordered
- ○ Don't know/not familiar

21. Do you order connexin testing in infants with nonsyndromic hearing loss?

- ○ Never
- ○ Rarely
- ○ Yes, sometimes
- ○ Yes, almost always
- ○ Not applicable

22. How often do you test for Cytomegalorvirus (CMV) when HL is identified on NBHS?

- ○ Never
- ○ Rarely
- ○ Sometimes
- ○ Always
- ○ Unsure
- ○ Not applicable

23. Do you have a designated system (i.e., person or computer system/database) for tracking...

	No	Yes	I don't know	Not Applicable
Failed newborn metabolic screening	○	○	○	○
Failed NBHS	○	○	○	○
Failed newborn blood spot screening	○	○	○	○
Delayed developmental milestones	○	○	○	○
Preauthorization for HL genetic testing	○	○	○	○
Follow up after referrals	○	○	○	○

24. Which specialist would you routinely refer the family of a chld with confirmed hearing loss? Please select all that apply.

- ○ Genetic specialist
- ○ Audiologist
- ○ ENT
- ○ Other_____

25. How informed do you think you are about...

	Uninformed	Somewhat Informed	Very Informed
The genetics of HL	○	○	○
The importance of genetic referrals	○	○	○

26. Do you have a designated system (i.e., person or computer system/database) for tracking...

	Uninformed	Somewhat Informed	Very Informed
Genetic causes of HL	○	○	○
The importance of genetic referrals	○	○	○
The logistics of genetic referral for HL	○	○	○
The significance of genetics in HL due to ototoxic medication exposure	○	○	○

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

- ○ Unimportant
- ○ Somewhat important
- ○ Very important

28. Prior to receiving this questionnaire were you familiar with the “EHDI 1-3-6” guidelines?

- ○ No
- ○ Somewhat
- ○ Yes

29. Would you implement the following strategies to faciliate (or ensure) tracking of genetic referrals?

	Yes	No
Implement a system to follow up with patients and families	○	○
Engage case managers in the EHDI programs	○	○
Increase genetics education efforts	○	○
Implement a system to follow up with other providers	○	○
Other	○	○

Other _____

30. Have you used the following strategies to facilitate (or ensure) tracking of genetics referrals?

	Yes	No
Implement a system to follow up with patients and families	○	○
Engage case managers in the EHDI programs	○	○
Increase genetics education efforts	○	○
Implement a system to follow up with other providers	○	○
Other	○	○

Other _____

31. If you have used the following strategies to facilitate (or ensure) tracking of genetic referrals did it work?

	No	Somewhat	Yes	Have Not Used
Implement a system to follow up with patients and families	○	○	○	○
Engage case managers in the EHDI programs	○	○	○	○
Increase genetics education efforts	○	○	○	○
Implement a system to follow up with other providers	○	○	○	○
Other	○	○	○	○

Other _____

32. Would you implement the following strategies to enhance collaborations and communication with EHDI/EI programs and primary care providers regarding genetic referrals and follow-up?

	No	Yes
Genetics of HL Reference guide	○	○
Handout of HL genetic testing	○	○
List of available resources on HL genetics	○	○
State/district-specific contact information on EHDI programs and genetics centers	○	○
Other	○	○

Other _____

33. Have you used the following strategies to enhance collaboration and communication with EHDI/EI programs and primary care providers regarding genetic referrals and follow-up?

	No	Yes
Genetics of HL Reference guide	○	○
Handout of HL genetic testing	○	○
List of available resources on HL genetics	○	○
State/district-specific contact information on EHDI programs and genetics centers	○	○
Other	○	○

Other _____

34. If you have used the following strategies to enhance collaboration and communication with EHID/EI programs and primary care providers regarding genetic referrals and follow-up, did it work?

	No	Somewhat	Yes	Have Not Used
Genetics of HL reference guide	○	○	○	○
Handout of HL genetics testing	○	○	○	○
List of available resources on HL genetics	○	○	○	○
State/district-specific contact information on EHDI programs and genetics centers	○	○	○	○
Other	○	○	○	○

Other _____