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Factors influencing parental decision about genetics evaluation for their deaf or hard-of-hearing child

Christina G.S. Palmer, PhD[1,2], **Jason T. Lueddeke, BA**[1], and **Jin Zhou, MS**[3]

[1] Department of Psychiatry & Biobehavioral Sciences, University of California, Los Angeles

[2] Department of Human Genetics, University of California, Los Angeles

[3] Department of Biomathematics, University of California, Los Angeles

Abstract

PURPOSE—To identify factors that are associated with why parents of deaf children who have had *GJB2/GJB6* testing as part of a genetics research study do or do not take their children for genetics evaluation.

METHODS—Self-administered questionnaire was completed by parents of a deaf child participating in a *GJB2/GJB6* testing study.

RESULTS—30 parents (representing 24 children) completed the questionnaire; 11 of 24 children (46%) underwent a genetics evaluation. Compared to parents who did not take their child for a genetics evaluation, those who did were more likely to 1) have supportive pediatricians, 2) feel it was important or would be helpful to their child, 3) recall the recommendation for evaluation, 4) have family members who wanted the child to have an evaluation, and 5) be Hispanic or Asian. Genetic test results, knowledge of genetics evaluation, psychosocial factors, language concerns, or structural factors were not substantively associated with attending a genetics evaluation.

CONCLUSION—Parental perceptions, family environment, and pediatricians play a role in decisions regarding genetics evaluation. Because genetic testing for deafness likely will occur outside of traditional genetics clinics and without comprehensive genetics evaluation, efforts to increase pediatricians' awareness of the usefulness of genetics evaluation may be essential to ensure appropriate care for deaf and hard of hearing children as recommended by the American College of Medical Genetics.

INTRODUCTION

Congenital deafness is relatively common, present in 1–3 in 1000 newborns [1, 2]. Although etiologically heterogeneous, at least 50% of early onset deafness is genetic, and among genetic cases, most are likely to be autosomal recessive, and non-syndromic [2, 3]. Due to the etiological heterogeneity of deafness, genetics evaluation and genetic counseling for deafness can be challenging, especially in young children [4]. However, with recent identification of deafness-causing variants in the *GJB2* and *GJB6* genes[5–8], which have been found to account for up to

Correspondence should be sent to: Christina G.S. Palmer, Ph.D., UCLA Semel Institute, 760 Westwood Plaza, Room 47-422, Los Angeles, CA 90024, T: (310) 794-4796, F: (310) 206-4446, E*: cpalmer@mednet.ucla.edu.

*may be published with the article

50% of autosomal recessive non-syndromic deafness in some populations [9–11], genetics evaluation and genetic counseling have the potential to offer a great deal more to families compared to the past.

Shortly after *GJB2* was identified, the Joint Committee on Infant Hearing (JCIH) endorsed universal newborn hearing screening and initiation of intervention for deaf infants by 6 months of age [12] because early detection and intervention could offer infants the best opportunity to develop nearly age appropriate language [13]. About the same time, the American College of Medical Genetics (ACMG) published a recommendation that all children with confirmed hearing loss be referred for genetics evaluation and counseling [14], a statement echoed by the JCIH [15, 16]. Newborn hearing screening has since become widely available in the United States and other countries [17–19], however, widespread involvement of genetics service providers within the early hearing detection and intervention programs is lagging [20, 21].

The workup for a deaf infant can include a number of medical tests in order to establish the etiology of the deafness [22]. However, in the absence of syndromic findings, *GJB2* testing currently is recommended as the first test [14], followed by *GJB6* deletion testing for negative or heterozygous *GJB2* results[23], and there is evidence that this genetic testing has become an important tool for non-genetics health providers, such as pediatric otolaryngologists [22], to explain why infants identified through the newborn hearing screening process are deaf or hard of hearing. A potential benefit of *GJB2/GJB6* testing is that in some cases, it will explain why a child is deaf and eliminate the need for additional medical evaluation [24]. A *GJB2/GJB6* result that explains why a child is deaf will also provide mode of inheritance, recurrence chance, and psychological benefit to the parents [24, 25]. However, for a considerable number of cases, *GJB2/GJB6* genetic testing will not explain why a child is deaf. Because inconclusive and negative genetic test results are difficult to explain and understand [26], and because the etiology of the child's deafness is still unknown, additional genetic and medical evaluation may be warranted. Although there are considerable benefits to *GJB2/GJB6* (and other DNA-based) testing, recent empirical data suggests that non-genetics providers, such as pediatric otolaryngologists, do not have an adequate understanding of genetic testing and counseling of test results [22]. A significant issue that may surface in relation to *GJB2/GJB6* testing by non-genetics health providers is the importance of referral for genetics evaluation and genetic counseling for both positive and negative/inconclusive results, particularly in light of a recent report that both deaf and hearing consumers would prefer to discuss genetic information with a professional trained in genetics such as a geneticist or genetic counselor [25].

Despite recommendations by the ACMG and the JCIH regarding genetics evaluation and counseling [14–16], empirical experience suggests that perhaps only ~50% of deaf or hard-of-hearing children are referred for genetics evaluation [27]. Although referral is an important step in the process, parents are the final decision makers about whether or not to pursue genetics evaluation for their child, and there is evidence to suggest that some parents do not pursue an evaluation [27]. To date, there have been no published studies to examine why some parents of a deaf or hard-of-hearing child take their child for a genetics evaluation and others do not. Moreover, there have been no studies that specifically examine why parents of deaf children who have had *GJB2/GJB6* testing outside of a genetics clinic choose or choose not to have their child undergo a genetics evaluation after receiving their results.

There are a variety of factors that may explain variation in seeking healthcare which could apply to parental decisions to seek a genetics evaluation for their deaf or hard-of-hearing child. For instance, financial or structural obstacles can exist [28], such as inadequate transportation to a genetics clinic, locality of the clinic itself, or lack of health insurance or coverage for a genetics evaluation. In addition, cultural hindrances may be present, such as an individual's inability to speak the majority language (e.g., English in the United States) or communicate comfortably

with a physician [29]. Many individuals avoid information that may discomfort them [30], and because learning genetic information can be disturbing and uncomfortable for some people [31, 32], they may view a genetics evaluation as a psychologically harmful process. Some people may fear being told their child's deafness is genetic[33] while some may simply fear the unknown and do not wish to learn about what they do not understand [30]. Others, however, may feel comfortable with their current understanding of why their child is deaf or hard-of-hearing, or believe that discovering the cause of their child's deafness is an irrelevant piece of information.

However, despite the potential barriers to seeking a genetics evaluation, there are parents who choose to learn more about why their child is deaf, and the explanation for this variability is currently unknown. By studying families of deaf and hard-of-hearing children identified through newborn hearing screening who are known to have received written and verbal recommendations for a genetics evaluation, we may be able to identify the factors that influence parents' decisions regarding the evaluation that extend beyond simple lack of awareness. Furthermore, by studying families who underwent *GJB2/GJB6* testing and counseling outside of a traditional genetics clinic, we may better understand the factors involved in parental decisions about genetics evaluation as these factors apply to this context. The purpose of this study is to identify factors that explain why parents of deaf or hard-of-hearing children who have had *GJB2/GJB6* testing outside of a genetics clinic do or do not take their children for genetics evaluation, with the ultimate goal of minimizing these sources of variation in order to optimize healthcare for these children.

MATERIALS AND METHODS

Sample

The sample is composed of parents who participated in the UCLA Genetics of Hearing Loss (GHL) study. The purpose of the GHL study was to examine clinical, educational, and psychosocial outcomes of *GJB2/GJB6* testing in deaf or hard-of-hearing children under the age of 3 years. Participants in the GHL included deaf infants/toddlers and their parents, who were recruited through a variety of venues in the Los Angeles area, including UCLA newborn nursery, genetics, audiology, and otolaryngology clinics, and education intervention programs. Parents could enroll in the GHL study at two different time points: after referral through newborn hearing screening (prior to confirmed audiologic diagnosis) or after identification of the child as deaf/hard-of-hearing. As part of the GHL study, a board-certified genetic counselor provided information about genetics of deafness and *GJB2/GJB6* testing to parents. With parental informed consent, *GJB2/GJB6* testing was performed on the child, and the genetic counselor explained the test results to parents in a face-to-face counseling session. Additional details of the GHL study methodology can be found in references [34–36]. Regardless of the genetic test results, the genetic counselor recommended a genetics evaluation for the child and all parents received written information about local genetics clinics, and copies of the ACMG and American Academy of Pediatrics (a member of JCIH) guidelines for care of a deaf or hard-of-hearing child. The exception to this recommendation occurred for 4 families (6 parents) enrolled at pre-diagnosis whose child subsequently passed outpatient hearing screening. Information regarding follow up on the genetics evaluation recommendation was not systematically collected as part of the GHL. A total of 164 parents from 103 families were enrolled in the GHL study. The current study focuses on the 99 families (158 parents) who received a recommendation for genetics evaluation for their child. Of these 99 families, 94 (151 parents) gave permission to be re-contacted for future studies and were eligible to participate in the current study.

Procedures

Between April and November 2007, 151 eligible parents representing 94 families were mailed a letter of invitation to participate in this study with up to 3 mailings to increase sample size. The study questionnaire was subsequently mailed to parents who consented to participate. This study was approved by the UCLA institutional review board.

Questionnaire Construction

A self-administered mailed questionnaire was developed to identify factors associated with parental decisions about a genetics evaluation. Eight potential factors were selected for investigation from pertinent literature in genetic counseling, health services research, and health psychology (see Table 2). These factors encompassed *structural* aspects (health insurance possession and coverage; general location and proximity of a genetics clinic; and transportation availability), potential concerns about *language* barriers, *perceived importance* of a genetics evaluation, *psychosocial* issues in relation to feelings of discomfort that might arise in a genetics evaluation, *knowledge* and understanding of genetics evaluations, *family's* role, *pediatrician's* role (parental request for a genetics referral; pediatrician's provision of a referral; pediatrician's knowledge of deafness; and pediatrician's interest in a genetics evaluation), and role of *GJB2/GJB6 counseling* session. Basic demographic information and genetic test results were previously collected through the GHL and available for use in the study.

The questionnaire contained 36 multiple option items (primarily yes/no/unsure and Likert scale response categories) and an open-ended item that asked parents to briefly describe why their child has had/has not had a genetics evaluation. The questionnaire was pilot tested for clarity, revised, and translated into Spanish using a translation-back translation procedure [37] to ensure maximum equivalence between the English and Spanish versions.

Analysis Plan

To examine the representativeness of the current sample to the larger GHL sample, the two samples were compared on a variety of demographic, audiologic, and genetic indices using Fisher's exact test and t-test for qualitative and quantitative variables, respectively. Parents in the current sample were then divided into two groups: those who reported taking their child for a genetics evaluation after they learned their child's *GJB2/GJB6* test results and those who reported that they did not take their child for a genetics evaluation. Fisher's exact test and t-test were performed to identify qualitative and quantitative factors, respectively, that were associated with whether or not parents took their child for a genetics evaluation. Because this sample contains six instances where both members of the couple completed a questionnaire, we randomly excluded one member of each couple and conducted preliminary analyses with and without the excluded respondents. Results did not substantively differ, indicating that the responses from the total sample can be treated as independent. Hence, we report the results of analyses from all 30 respondents, except for a subset of variables which can only be counted once per family (whether or not asked pediatrician for referral, whether or not pediatrician gave referral, family income, *GJB2/GJB6* status of child, child's age) where we report results of analyses on the 24 families. Likert scale items were analyzed as quantitative variables for greater power; however, for ease of interpretation, we present the distribution of responses rather than the means. Alpha of 0.05 was used to determine statistical significance.

RESULTS

Sample Characteristics

Of the 151 eligible parents, 28 were unreachable due to address change or unknown address. Of the remaining 123, 30 parents representing 24 families completed the questionnaire for a response rate of 24.3%. Table 1 provides sample demographic information for the current study and compares it to the 158 parents who received a recommendation to take their child for a genetics evaluation in the larger GHL study. As shown in Table 1, the current study sample does not significantly differ from the larger GHL study on any of these demographic factors, suggesting that the study sample is representative of the larger sample on a variety of demographic, audiologic, and genetic indices. In 11 of the 24 families, the deaf child underwent a genetics evaluation (45.8%), for a total of 12 parents (40%) who took their child for a genetics evaluation and 18 parents who did not.

As shown in Table 2, parents who took their child for a genetics evaluation rated the importance of the evaluation as higher than the other parents ($p<0.0001$), were more likely to feel that their family wanted their child to have a genetics evaluation ($p=0.04$), and that an evaluation would help their child ($p=0.056$). Most parents felt that their pediatricians were 'somewhat or very knowledgeable' about their child's deafness, however, those who took their child to a genetics evaluation rated their pediatrician's interest in a genetics evaluation as higher than did the other parents ($p=0.003$), and those families were more likely to ask for (36.4% vs. 0%, $p=0.03$) and receive a referral (36.4% vs. 0%, $p=0.03$) from their pediatrician. These parents also were more likely to recall the recommendation for the evaluation (67% vs. 28%, $p=0.05$). Parents did not differ substantially in their reported knowledge and understanding of a genetics evaluation, however, parents who took their child to genetics evaluation were less likely to respond that recommendations for other tests are made during an evaluation compared to the other parents ($p=0.007$). This result suggests that for nearly 40% of the former group, parents did not recall having additional tests recommended for their child.

The two parental groups differed on two psychosocial concerns related to a genetics evaluation that might be brought out as a result of learning more about their child's deafness. Parents who took their child for genetics evaluation were more likely to have been concerned that an evaluation would not make them feel better ($p=0.02$) and that they would learn information that would make them feel uncomfortable ($p=0.06$) compared to those who did not take their child to genetics evaluation. Nearly all children were covered by health insurance, however, parents who did not take their child to a genetics evaluation were non-significantly more likely to indicate that they were unsure if the insurance covered the visit compared to those who took their child for a genetics evaluation ($p=0.13$).

Ethnic heritage was the only demographic, audiologic, or genetic index associated with parents' response regarding genetics evaluation, where parents of Hispanic or Asian heritage were more likely to take their child for evaluation compared to parents of Caucasian heritage ($p=0.04$). In order to further examine if the Hispanic and Asian children were more likely to have had a genetic evaluation because their *GJB2/GJB6* result was normal, we constructed a 3-way table of ethnic heritage, *GJB2/GJB6* result, and whether or not child had a genetics evaluation for qualitative analysis because the sample is too small to perform a valid statistical analysis While the table reconfirms that more Hispanic (66.7%) and Asian (100%) children than Caucasian (25%) children in our sample had a genetics evaluation, it also illustrates that *GJB2/GJB6* results were less likely to explain the child's deafness in the Caucasian and Hispanic families (75%, 100%) compared to the Asian families (50%) in the sample. Furthermore, while 100% of Caucasian and Hispanic parents who took their child for a genetics evaluation received a negative *GJB2/GJB6* result, 50% of the Asian parents received a positive *GJB2/GJB6* result.

Hence there is no strong evidence for an interaction between ethnic heritage, *GJB2/GJB6* result, and whether or not a child had a genetics evaluation.

Parents' comments to the open-ended question revealed that the most common reason for taking their child to a genetics clinic was because the child had multiple malformations or medical issues (n = 3 children). In contrast, none of the parents in the other group indicated that their child had multiple malformations; instead the most common reason for not taking their child to a genetics clinic was that learning the etiology of their child's deafness was not perceived as important (e.g., "Doesn't really matter to us and is not a concern enough to warrant further investigation," "Our purpose/goal is to love our little guy with all our hearts, and we didn't see how exploring the reasons why this may have happened would be important or matter to us"). In order to further examine if parents of Hispanic or Asian heritage were more likely to have taken their child for a genetics evaluation because of multiple anomalies, we performed a 3-way analysis evaluating ethnic heritage, comments about multiple anomalies, and whether or not child had a genetics evaluation, recognizing that the sample is too small to perform a valid statistical analysis. Although there seems to be an association between ethnic heritage and anomalies in our sample (8.3%, 16.7%, 25%, 0% Caucasian, Hispanic, Asian, and mixed heritage children, respectively), this phenomenon does not appear to explain the relationship between ethnic heritage and whether or not a child had a genetics evaluation as 33%, 25%, and 25% of Caucasian, Hispanic, and Asian children who had a genetics evaluation also had a parental comment about multiple anomalies as the reason for taking their child for the evaluation.

DISCUSSION

Despite recommendations for genetics evaluation and counseling for all children with confirmed deafness [14], a significant percentage of children do not undergo evaluation [22]. The purpose of this study was to identify factors that are associated with why some parents pursue genetics evaluation for their deaf or hard-of-hearing child and others do not. The participants of the UCLA Genetics of Hearing Loss study serve as a model for the increasingly common scenario of parents whose children receive genetic testing for deafness outside of a standard clinical genetic setting, followed by a recommendation for a clinical genetics evaluation and counseling.

The findings from our study suggest that compared to parents who did not take their child for a genetics evaluation, those who did were *more likely to* 1) have supportive pediatricians, 2) feel it was important or would be helpful to their child, 3) recall the recommendation for a genetics evaluation, 4) have family members who wanted the child to have an evaluation, and 5) have Asian or Hispanic background. In addition, qualitative data suggests that parents who took their child for a genetics evaluation were more likely to report that their child has multiple anomalies. Although the survey response rate was low, the study sample was found to be representative of the larger Genetics of Hearing Loss sample from which it was drawn on a variety of demographic, audiologic, and genetic indices, thereby increasing the credibility of these findings.

The importance of parental perceptions and pediatrician support in facilitating genetics evaluation for deaf children was recently reported [27], and our results suggest that this is a robust finding that should be considered when developing strategies to increase uptake of genetics evaluations for deaf children. Although not previously reported, it is not surprising that parents of children with multiple malformations are more likely to seek genetics evaluation as clinical genetics has traditionally evaluated children with anomalies. However, the recommendation for genetics evaluation of deaf children is not limited to those with multiple anomalies, and

perhaps greater awareness of the utility of genetics evaluation for apparently non-syndromic deaf children is needed.

Ethnic differences in parental perceptions of genetic testing for deafness have been observed previously[35], where parents of Asian or Hispanic background were more likely than Caucasian parents to view genetic testing in terms of helping with their child's medical care. In this study, we observed a similar phenomenon in which parents of Asian or Hispanic background were more likely than Caucasian parents to take their child for a genetics evaluation. Further examinations revealed that the ethnic differences were not explained by differences in frequencies of multiple anomalies or *GJB2/GJB6* result, suggesting instead that ethno-cultural differences may play a role in views about medico-genetic information. Similarly, the social and family context has been shown to influence decisions regarding prenatal and presymptomatic genetic testing [38, 39], and this study extends these findings to suggest that the family also plays some role in influencing parents' decisions regarding genetic evaluation for deafness.

Parents in this sample had a generally good understanding of what takes place during a genetics evaluation and their level of understanding was not substantively associated with their decisions to take their child for genetics evaluation. The two parental groups also did not substantively differ on potential psychosocial aspects associated with a genetics evaluation, nor did they have concerns about possible language barriers, suggesting that components of genetic counseling such as communication, risk assessment, and handling potentially uncomfortable information, neither facilitate nor hinder decisions about genetics evaluation for their child. Finally, structural factors such as access to a genetics clinic, a reliable method of transportation, or presence of health insurance were not problematic for parents in this sample and were not significantly associated with parental decision regarding genetics evaluation. Although the two parental groups differed weakly in terms of whether or not the health insurance would cover a visit to a genetics clinic, this difference appears to be explained by a lack of investigation on the part of those who did not take their child for a genetics evaluation rather than denial by the health insurance company.

This study has several limitations and so generalizing our findings should be done with caution. First, this is a very small study, with a relatively low response rate. Although the current study sample is representative of the larger group from which it was drawn, the small sample size yields limited power to identity additional variables associated with whether or not parents take their child to a genetics clinic. Second, although ethnically diverse, the socioeconomic and educational status of the participants was generally high, and this may explain why logistical factors and language did not seem to be particular impediments to attending the genetics clinic. Additional studies with larger and more socioeconomically diverse samples may help to further illuminate barriers to, and facilitators of, genetics evaluation. Third, it is important to bear in mind that all of the parents in this study were able to participate in a larger study on genetic testing and were counseled by a genetic counselor regarding the importance of genetics evaluation.

Algorithms have been proposed that recommend *GJB2* testing as the first step in the evaluation of an apparently nonsyndromic deaf/hard of hearing child because a positive result eliminates the need for other medical tests [14, 40]. It is currently unclear whether *GJB2* testing, along with pre- and post-test counseling, primarily will take place within or outside of a genetics clinic. However, it seems likely that non-genetics professionals such as pediatricians, otolaryngologists, and audiologists will play a large role in first-tier genetic testing due to the limited number of genetics professionals and the fact that parents of a deaf/hard of hearing child will have early and repeated contact with these healthcare professionals. When genetic testing takes place outside of a genetics clinic, it would seem logical that parents who receive

a positive *GJB2* result would remain within their primary care setting while those who receive a negative or inconclusive result would be referred for a genetics evaluation. A potential advantage of this step in the evaluation algorithm is that it would focus the limited genetics workforce on children with possibly less straightforward etiologies of their deafness, including syndromic forms of deafness, where clinical and family history data may be required to determine the best course of action. However, genetic testing is different from other routine testing, and raises a number of complex issues including the technicalities of genetic testing, the language of genetics, the relevance to other family members, and reproductive issues [14, 22]. In order for this triage step to be successful, non-genetics professionals need to understand and convey the complexities and subtleties of genetic information. However, recent studies raise concerns about non-geneticists' understanding of genetic information (positive *and* negative results) and their implications [22, 41], illustrating a current limitation to this step in the algorithm. Possible solutions to this limitation include increasing the level of understanding of genetic information among non-genetics professionals and including genetic counselors within these non-traditional genetics settings, as has happened in other non-traditional genetics settings such as oncology. Given current limitations, empirical data are needed to determine if the potential advantages of focusing genetics referrals only on deaf/hard of hearing children with non-*GJB2* etiologies outweigh the potential benefits of genetics referral for all parents of a deaf/hard of hearing child regardless of *GJB2* results. Interestingly, in the current sample, the ability of prior *GJB2/GJB6* testing to explain their child's deafness did not appear to play a role in subsequent parental decision about genetics evaluation. This result provides empirical evidence that genetics evaluation can be perceived as important to parents regardless of the genetic test result and that genetics clinics should expect to counsel some parents whose children have received a genetic explanation for the deafness outside of traditional genetics clinics.

In closing, the importance of the pediatrician regarding genetics evaluation for deaf children has now been observed in two studies. Although referral is an important factor in facilitating genetics evaluation, the overall pediatrician referral rate appears to be low (4 of 24 families (~17%) received a referral in the current study; 24% in Powell et al[27]), and one contributing factor may be poor understanding of the benefits and purposes of genetic evaluations or the inability to adequately convey this information to parents [27]. Our study suggests that efforts to educate pediatricians about the role of genetics evaluation for deaf children may be essential to ensure appropriate care for deaf or hard-of-hearing children as recommended by the American College of Medical Genetics.

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Table 1

Description of study sample and comparison with sample from which participants were recruited

Descriptor	Current Study	GHL Study[a]	p-value
# Parents (# couples)	30 (6)	158 (59)	-
# Families	24	99	-
% Female - parents	73%	62%	0.30
Ethnicity/Race - parents			
Caucasian	50%	38.0%	0.77
Hispanic	33.3%	41.8%	
Asian	16.7%	16.5%	
African-American	0%	1.9%	
Hawaiian/Other Pacific Islander	0%	1.9%	
Average age at GHL enrollment – parents (SD)	33.2 (5.60)	32.7 (6.4)	0.70
Median family income - parents	$95K	$35K - $50K	0.15
Median education - parents	4 years of college	2–4 years of college	0.72
% deaf or hard of hearing - parents	3.3%	5.7%	1.0
Average age at GHL enrollment – children (SD)	12.4 months (11.1)	13.6 months (11.1)	0.62
% children with *GJB2/GJB6*-related deafness	25%	23.2%	1.0

[a] GHL = Genetics of Hearing Loss study, the study from which the current sample of parents was recruited

Table 2

Survey items and results of analyses to identify factors associated with whether or not parents took their child to a genetics evaluation

Question	Took child for genetics evaluation (n=12 parents; 11 families)	Did not take child for genetics evaluation (n=18 parents; 13 families)	p-value
Structural variables			
Do you have a reliable method of transportation to take your child to see a genetics doctor?	Yes: 100%	Yes: 100%	-
Are there genetics doctors in your city?	Yes: 67%	Yes: 61%	1.0
	No: 8%	No: 6%	
	Unsure: 25%	Unsure: 33%	
Can you get to a genetics doctor easily?	Yes: 83%	Yes: 89%	1.0
	No: 17%	No: 11%	
Does your child have health insurance?	Yes: 92%	Yes: 100%	0.40
	No: 8%	No: 0%	
Does your child's health insurance cover a visit to a genetics doctor?	Yes: 75%	Yes: 39%	0.13
	No: 0%	No: 6%	
	Unsure: 25%	Unsure: 56%	
Potential Language Barriers			
I was (am) worried that I would (will) receive information in a language I am not comfortable with.	SA/A[a] = 8.3%	SA/A = 11.1%	0.95
	N = 8.3%	N = 5.6%	
	SD/D = 83.3%	SD/D = 83.3%	
I was (am) worried that the doctor would (will) communicate with me only in English.	SA/A = 18.2%	SA/A = 11.1%	0.37
	N = 9.1%	N = 5.6%	
	SD/D = 72.7%	SD/D = 83.3%	
Perceived Importance of Genetics Evaluation			
In your opinion, how important is (was) it for your child to be examined by a genetics doctors	Very = 83.3%	Very = 11.8%	<0.0001
	Somewhat = 16.7%	Somewhat = 23.5%	
	Not important = 0%	Not important = 64.7%	
Psychosocial variables			
I was (am) worried that I would (will) learn information that will make me feel uncomfortable.	SA/A = 50%	SA/A = 5.6%	0.02
	N = 8.3%	N = 11.1%	
	SD/D = 41.7%	SD/D = 83.3%	
I was (am) worried that I would (will) be told that I should not have any more children.	SA/A = 25%	SA/A = 11.1%	0.63

Question	Took child for genetics evaluation (n=12 parents; 11 families)	Did not take child for genetics evaluation (n=18 parents; 13 families)	p-value
	N = 8.3%	N = 16.7%	
	SD/D = 66.7%	SD/D = 72.2%	
I was (am) worried that a genetics evaluation would (will) not make me feel better.	SA/A = 25%	SA/A = 11.1%	0.06
	N = 33.3%	N = 0%	
	SD/D = 41.7%	SD/D = 88.9%	
I was (am) worried that I would (will) find out that I caused my child's hearing loss.	SA/A = 8.3%	SA/A = 22.2%	0.79
	N = 16.7%	N = 0%	
	SD/D = 75%	SD/D = 77.8%	
I was (am) worried about learning more about my child's hearing loss.	SA/A = 25%	SA/A = 5.6%	0.42
	N = 0%	N = 16.7%	
	SD/D = 75%	SD/D = 77.8%	
I was (am) comfortable with what I already know about my child's hearing loss.	SA/A = 50%	SA/A = 66.7%	0.73
	N = 16.7%	N = 11.1%	
	SD/D = 33.3%	SD/D = 22.2%	
I was (am) worried that I would (will) not understand what the doctor tells me.	SA/A = 16.7%	SA/A = 16.7%	0.82
	N = 0%	N = 11.1%	
	SD/D = 83.3%	SD/D = 72.2%	
Knowledge and Understanding			
Do you know why your child has hearing loss?	Yes: 33.3%	Yes: 33.3%	1.0
I understand what happens during a genetics evaluation.	SA/A = 83.3%	SA/A = 55.6%	0.15
	N = 8.3%	N = 27.8%	
	SD/D =8.3%	SD/D =16.7%	
During a genetics evaluation of a child with hearing loss…			
The child has a physical exam	True: 66.7%	True: 58.8%	0.72
The family history is discussed	True: 100%	True: 100%	--
The doctor tries to identify the cause of the hearing loss in the child	True: 91.7%	True: 94.4%	1.0
Recommendations for other tests are made	True: 58.3%	True: 100%	0.007
The chance of having another child with hearing loss is discussed	True: 83.3%	True: 100%	0.15
Family's role			
My family wants my child to have a genetics evaluation with a genetics doctor.	SA/A = 41.7%	SA/A = 11.1%	0.04
	N = 25%	N = 27.8%	

Question	Took child for genetics evaluation (n=12 parents; 11 families)	Did not take child for genetics evaluation (n=18 parents; 13 families)	p-value
	SD/D = 33.3%	SD/D = 61.1%	
Pediatrician's role			
How would you rate your pediatrician's knowledge of your child's hearing loss?	Very = 33.3%	Very = 22.2%	0.92
	Somewhat = 41.7%	Somewhat = 61.1%	
	Not knowledgeable = 25%	Not knowledgeable = 16.7%	
How would you rate your pediatrician's interest in your child going to a genetics doctor?	Very = 41.7%	Very = 0%	0.003
	Somewhat = 33.3%	Somewhat = 25%	
	Not interested = 25%	Not interested = 75%	
Did you ask your child's pediatrician for a referral to see a genetics doctor?[b]	Yes: 36.4%	Yes: 0%	0.03
	No: 63.6%	No: 100%	
Did your child's pediatrician give you a referral to see a genetics doctor?[b]	Yes: 36.4%	Yes: 0%	0.03
	No: 63.6%	No: 100%	
Role of GJB2/GJB6 counseling session			
The counseling session made me feel like…			
I learned everything I needed to know about why my child has hearing loss.	SA/A = 16.7%	SA/A = 44.5%	0.29
	N = 8.3%	N = 11.1%	
	SD/D = 75%	SD/D = 44.4%	
A visit to a genetics doctor could help my child.	SA/A = 41.7%	SA/A = 16.7%	0.056
	N = 50%	N = 38.9%	
	SD/D = 8.3%	SD/D = 44.4%	
A visit to a genetics doctor would give me more information about my child's hearing loss.	SA/A = 58.3%	SA/A = 38.9%	0.33
	N = 33.3%	N = 44.4%	
	SD/D = 8.3%	SD/D = 16.7%	
The genetic counselor helped me understand what happens during a genetics evaluation with a genetics doctor.	SA/A = 90.9%	SA/A = 66.7%	0.14
	N = 9.1%	N = 11.1%	
	SD/D = 0%	SD/D = 22.2%	
The genetic counselor recommended that my child go to see a genetics doctor.	Yes: 67%	Yes: 28%	0.05
	No: 8%	No: 44%	
	Unsure: 25%	Unsure: 28%	
Demographic Variables			
Ethnicity/Race - parents			0.04

Question	Took child for genetics evaluation (n=12 parents; 11 families)	Did not take child for genetics evaluation (n=18 parents; 13 families)	p-value
Caucasian	25%	66.7%	
Hispanic	41.7%	27.8%	
Asian	33.3%	5.6%	
Median family income – parents[b]	>$95K	>$80	0.74
Median education level - parents	4 years of college	4 years of college	0.34
Age at current study – parents (SD)	34.9y (5.7)	37.2 (4.3)	0.23
% deaf/hard-of-hearing - parents	8%	0%	0.40
Age at current study – child (SD)[b]	3.1y (1.3)	3.3y (0.9)	0.77
% children with *GJB2/GJB6*-related deafness[b]	18.2%	30.8%	0.65

[a] SA=strongly agree; A=agree; N=neutral; D=disagree; SD=strongly disagree

[b] Analyses based on 24 independent families