Special Section on Deaf Studies

Deaf Adults' Reasons for Genetic Testing Depend on Cultural Affiliation: Results From a Prospective, Longitudinal Genetic Counseling and Testing Study

Patrick Boudreault[1], Erin E. Baldwin[2], Michelle Fox[2], Loriel Dutton[2], LeeElle Tullis[2], Joyce Linden[1], Yoko Kobayashi[1], Jin Zhou[2], Janet S. Sinsheimer[2], Yvonne Sininger[2], Wayne W. Grody[2], Christina G. S. Palmer*[,2]

[1]California State University

[2]University of California, Los Angeles

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This article examines the relationship between cultural affiliation and deaf adults' motivations for genetic testing for deafness in the first prospective, longitudinal study to examine the impact of genetic counseling and genetic testing on deaf adults and the deaf community. Participants ($n = 256$), classified as affiliating with hearing, Deaf, or *both* communities, rated interest in testing for 21 reasons covering 5 life domains. Findings suggest strong interest in testing to learn why they are deaf, but little interest in using it for decisions about a partner or having children. Culturally mediated variation was also demonstrated. Deaf and *both* communities groups viewed testing as useful for more life domains than the hearing community group. Deaf and *both* communities had similar motivations related to further exploration, understanding, or strengthening of deafness. Motivations related to "hearing" were also relevant for *both* communities. We conclude that cultural affiliation is an important factor for constructing motivations for genetic testing.

Over the years, many individuals in the Deaf community[1] have expressed either concerns or a "wait-and-see" attitude about genetic research and genetic information related to deafness (Arnos, 2003; Martinez, Linden, Schimmenti, & Palmer, 2003; Middleton, 2007; Middleton, Emery, & Turner, 2010; Middleton, Hewison, & Mueller, 1998; Stern et al., 2002; Taneja, Pandya, Foley, Nicely, & Arnos, 2004). Concerns have included the fear that genetic research will do more harm than good (Middleton et al., 1998), that genetic testing will devalue deaf people (Martinez et al., 2003; Middleton et al., 1998), that genetic information may be used to eliminate or "cure" deaf people (Lane, 2005; Martinez et al.; Middleton et al., 2010), and that genetic testing could lead to discrimination in health insurance, employment, and education (Martinez et al.); disrupt family relationships (Martinez et al.); and be used for prenatal diagnosis for the purpose of pregnancy termination based on hearing status (Middleton et al., 2010; Stern et al.).

Deaf individuals' concerns about genetic research and genetic information are not without merit as genetics concepts and technologies have been misapplied at times throughout the past 150 years and around the world to justify policies to decrease the number of deaf individuals. Examples of this phenomenon include the promotion of educational policies in the United States in the late 19th and early 20th centuries to decrease the number of deaf marriages (Bell, 1883) and sterilization and death of deaf individuals in Nazi Germany during World War II (Biesold, 1999; Brueggemann, 2009), due to erroneous beliefs about how deafness is inherited; and more recently, the amendment made to the UK Human Fertilisation and Embryology Act 1990, in which the effect of Clause 14, Section 4, Number 9, is to prohibit deaf individuals from using

*Correspondence should be sent to Christina G. S. Palmer, UCLA Semel Institute, 760 Westwood Plaza, Room 47-422, Los Angeles, CA 90024 (e-mail: cpalmer@mednet.ucla.edu).

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prenatal genetic diagnosis to preferentially select embryos with genes for deafness over embryos without genes for deafness (Emery, Burke, Middleton, Belk, & Turner, 2008).

Although Deaf individuals' concerns about how genetic information may be used against them and the Deaf community have been well documented, recent data from the United States and Australia suggest that deaf individuals' attitudes and perceptions of genetic testing for deafness may be shifting toward greater personal interest (Burton, Withrow, Arnos, Kalfoglou, & Pandya, 2006; Guillemin & Gillam, 2006; Withrow et al., 2009a). As one example, Withrow et al. (2009a) in their recently published survey found that the majority of their sample of 156 deaf adults in the United States either had genetic testing or were somewhat or very interested in it. To date, there have been few studies to examine deaf adults' motivations for pursuing genetic testing for deafness for themselves (Burton et al., 2006; Taneja et al., 2004; Withrow, Burton, Arnos, Kalfoglou, & Pandya, 2008; Withrow et al., 2009a). There is a significant dearth of information in light of the fact that (a) deaf individuals potentially make up a large proportion of the consumer group for genetic testing for deafness and (b) genetic testing pursued by deaf individuals raises important questions about the effects of their use of genetic information (in contrast to outsiders' use of genetic information) on deaf individuals and the Deaf community, which only empirical studies can answer.

Based on the few recent empirical studies, there is evidence to suggest that there are a variety of reasons that deaf adults might pursue genetic testing for themselves (or their deaf child). The reasons include the following: to clarify their self-identity (Burton et al., 2006) or to satisfy their curiosity about why they are deaf (Withrow et al., 2008), to acquire information to help them and their family members in the future (Withrow et al., 2008), to learn the chance that they or other family members may have deaf children or grandchildren (Burton et al.; Withrow et al., 2008), to learn why their child(ren) is deaf (Burton et al.), and to prepare for the education and language development of future children (Burton et al.; Withrow et al., 2008), for example, because school placement is an important issue in the Deaf community and some families choose to locate near particular school programs in the event that they have or might have deaf children. It is much less clear if deaf individuals are interested in genetic testing for purposes of partner selection or to ensure the birth of a deaf or a hearing child, as studies have produced conflicting results on this topic (Taneja et al., 2004; Withrow et al., 2009a).

Although it appears that there are a variety of reasons for deaf individuals to pursue genetic testing, it is unlikely that all deaf individuals pursue genetic testing for exactly the same reasons because the deaf community is culturally diverse (Jacobs, 1989; Padden & Humphries, 1988, 2005). This diversity is due to a variety of factors, including language preference, interactions with the social environment, and past and present experiences (Bat-Chava, 1994, 2000; Nikolaraizi & Hadjikakou, 2006). As a result, the deaf community is composed of individuals who differ in their identity as a deaf person, that is, their cultural affiliation (Hole, 2007; Sheridan, 2000).

Some researchers studying the topic of deaf identity or cultural affiliation have identified important individual differences that yield four different deaf identity orientations: hearing, marginal, Deaf (also called immersion), and bicultural (sometimes referred to as equal involvement in deaf and hearing communities; Bat-Chava, 2000; Fischer & McWhirter, 2001; Glickman & Carey, 1993; Holcomb, 1997; Kannapell, 1993; Leigh, Marcus, Dobosh, & Allen, 1998). Descriptions within this literature on deaf identity suggest that individuals hold a hearing identity when the mainstream hearing society is their primary frame of reference. These individuals identify with the hearing society in attitude, behavior, and communication style, and they tend to view deafness as a medical–pathological condition. Individuals who are ambivalent about their deafness and their cultural frame of reference (vacillating between hearing, deaf, or hard of hearing) are considered to have a marginal identity. Individuals with the immersion or culturally Deaf identity engage in a high level of involvement with the Deaf community, feel a strong sense of "Deaf" pride, view deafness as a personal characteristic, and communicate with sign language, for example, American Sign Language (ASL). Finally, individuals with a bicultural identity feel comfortable with both deaf and hearing

people while recognizing the strengths and limitations of both cultures. These individuals may be bilingual in sign and written or spoken languages.

Although these descriptions suggest easy classification of deaf individuals, deaf identity is a much more complex and fluid concept because the development of a deaf individual's deaf identity is a process that evolves over time and is influenced by many factors including interactions with deaf and hearing peers (Leigh, 1999; Ohna, 2004; Padden & Humphries, 2005). As a result of these processes, at any given moment in time it may not be possible to precisely capture an individual's deaf identity because it may be context dependent or in flux (Sheridan, 2000). Moreover, even if it were possible to capture an individual's deaf identity at a single point in time, it might be different at another point in time. Because of these complexities, dimensional measures such as the Deaf Identity Development Scale—Revised (DIDS-R; Fischer & McWhirter, 2001) and the Deaf Acculturation Scale (Leigh et al., 1998; Maxwell-McCaw, 2001) have been developed to provide more nuanced understanding of deaf identity; however, there is considerable discussion about deaf identity and Deaf culture within Deaf studies, with some individuals suggesting alternative models for understanding what it means to be deaf (Bauman, 2008; Brueggemann, 2009).

With those caveats in mind, there is evidence that deaf individuals' motivations for genetic testing for deafness may depend to some extent on their deaf identity or cultural affiliation (Taneja et al., 2004; Withrow et al., 2009a). Specifically, there are data to suggest that individuals who identify as culturally Deaf are more likely to be motivated by genetic testing to learn why they are audiologically deaf, to provide information to their families, and to learn the chance that individuals in future generations will be deaf, and are less likely to be motivated by the idea of using genetic testing for "treatment purposes," compared to those who identify with the hearing community or equally with the Deaf and hearing communities (bicultural) (Withrow et al., 2009a). It is also possible that culturally Deaf individuals are more motivated by the idea of using genetic testing for selecting a partner compared to those who identify with both the Deaf and the hearing communities (Withrow et al., 2009a), but results are conflicting (see Taneja et al., 2004).

In general, the results of previous studies suggest that there are a variety of reasons for deaf individuals' interest in genetic testing for deafness and that their reasons span across the domains of self, partner, children, and extended family. Furthermore, results suggest that cultural affiliation or deaf identity may provide a lens through which to understand and explain differences in deaf individuals' motivations for genetic testing for deafness. However, these previous studies suffer from several methodological limitations, including cross-sectional designs, mixed genetic testing context (resulting in either retrospective reporting when testing occurred in the past or reporting based on hypothetical testing when no testing had occurred), generally small samples (ranging from 30 to 156 deaf adults; three quarters of studies with less than $n = 65$), and inclusion of hearing individuals in analyses of cultural affiliation. Hence, the extent to which the findings of these studies reflect the culturally associated motivations of the diverse community of deaf individuals to pursue genetic testing for themselves when embarking on the genetic counseling and genetic testing process may be limited. To date, there are no empirical data on deaf adults' motivations for pursuing genetic testing for deafness in the context of actual genetic counseling and genetic testing.

Genetic counseling and genetic testing for deafness is now readily available with the identification of two genes, *GJB2* (Denoyelle et al., 1997) and *GJB6* (del Castillo et al., 2002), which produce Connexin 26 and Connexin 30 proteins, respectively, necessary for hearing. We refer to *GJB2* and *GJB6* as Connexin 26 and Connexin 30, respectively, throughout the remainder of the article. Studies of samples of deaf individuals with sensorineural deafness in the United States have found that 22%–40% have Connexin-related deafness (Cohn & Kelley, 1999; Green et al., 1999; Kelley et al., 1998; Kenna, Wu, Cotanche, Korf, & Rehm, 2001; Pandya et al., 2003; Schimmenti et al., 2008), but ethnic background and family history can influence these estimates upward or downward. Because Connexin-related genetic variants are common in the population, it is anticipated that there will be consumer demand for Connexin 26 and Connexin 30 testing. Yet, there are no data on the impact of genetic counseling and genetic testing for deafness on deaf adults and the deaf community.

The purpose of this article is to report the first set of results to examine deaf adults' motivations for genetic testing through the lens of deaf identity or cultural affiliation in the context of a prospective, longitudinal study that involves testing two deaf-causing genes: Connexin 26 and Connexin 30. Because we examine the relationship between cultural identity and reasons for genetic testing at baseline, that is, before deaf individuals met with a genetic counselor, our analyses provide the best comparison with other studies of deaf adults' motivations for genetic testing under hypothetical or unknown testing conditions, while at the same time providing a firmer basis for understanding deaf adults' motivations because the data are collected in anticipation of actual genetic counseling and testing.

Materials and Methods

Research Design

This is a prospective, longitudinal study to examine the impact of genetic counseling and genetic testing (Connexin 26 and Connexin 30 genes) on deaf adults and the deaf community. The core research team is composed of Deaf, hard of hearing, and hearing researchers from a variety of disciplines, including Deaf studies, linguistics, sign language interpreting, genetics, genetic counseling, audiology, and statistics. In addition, there is an advisory board from the Greater Los Angeles deaf community. To facilitate communication among project members, the research team includes two certified sign language interpreters, and multiple technologies to accommodate a variety of modes of communication. By incorporating cultural and linguistic diversity in the research team, the project has taken an important step toward integrating the Deaf cultural perspective, the hearing cultural perspective, the academic cultural perspective, and the community service perspective into the research design and implementation of a genetic testing study.

Sample

Individuals who have been deaf or hard-of-hearing since an early age (defined as birth to 6 years of age) were eligible to participate in this study if they (a) were at least 18 years old, (b) had a sensorineural deafness, and (c) had no prior clear explanation for why they are deaf. Participants were recruited from the Los Angeles, San Francisco Bay, and Riverside areas of California, through a variety of venues including deaf/hard-of-hearing agencies, Deaf expos, Deaf conferences, support groups, and audiology clinics. A study Web site (www.deafgeneticsproject.org), study brochure, postcard mailings, advertisements, and in-person presentations were developed to inform deaf and hard-of-hearing individuals of the project. Through these mechanisms, individuals were informed that the focus of the study was on genetic counseling and genetic testing for two genes called Connexin 26 and Connexin 30, that this testing might explain why they are deaf, and that sharing their experience of genetic counseling and genetic testing will help society understand how genetic testing can impact deaf and hard-of-hearing individuals. Among other things, the materials also provided information that the research team was composed of Deaf, hard-of-hearing, and hearing individuals; that genetic counseling and testing would be provided at no charge; and that the study was not intended to cure deafness or to affect an individual's hearing. All materials were deaf friendly with use of visual aids, laymen's terms, and ASL. Interested individuals were invited to contact our project personnel via point-to-point Webcam communication (e.g., videophone), teletypewriter, e-mail, or voice telephone.

Study Protocol

There were three stages to the study protocol. In the first stage, interested individuals contacted study personnel in the language of their choice and completed a brief screening questionnaire to determine initial eligibility. Individuals determined to be initially eligible were then scheduled for an audiology evaluation to confirm the presence of sensorineural deafness to ensure that Connexin 26 and Connexin 30 genetic testing was offered only to individuals for whom it was potentially relevant, that is, those with early-onset sensorineural deafness. To facilitate participation, individuals could select one of four locations for their participation (University of California, Los Angeles [UCLA]; California State University, Northridge; California School for the Deaf–Fremont; and California School for the Deaf–Riverside). During the

informed consent process, participants were informed that the overall goal of the study was to "learn what deaf/hard-of-hearing individuals think about genetic counseling and testing, what it means for their lives, and what it means for the Deaf community" to allay potential concerns that the study had a medical or disability focus. Among the four audiologists collaborating on this project, one was a certified sign language interpreter, one was familiar with ASL, and two were not familiar with ASL. A certified project staff sign language interpreter interpreted for audiology sessions at the participant's request. Following audiological confirmation of eligibility for the genetic counseling and testing stage of the study, participants completed the first of four questionnaires (called the baseline questionnaire). The baseline questionnaire is the focus of the current article.

For completeness, we describe the rest of the study protocol. Because data collection is still ongoing, results from these later stages will not be presented in this article. The second stage of the study entailed a face-to-face pretest genetic counseling session with a hearing, board-certified genetic counselor and the project staff certified sign language interpreter (some participants chose to communicate orally with our project genetic counselor). During that session, the genetic counselor explained the remaining study protocol, along with general information about genetic epidemiology of deafness, Connexin 26 and Connexin 30 deafness, and genetic testing. If the participant was interested in moving forward with the genetic testing part of the study, they signed a consent form and a buccal (cheek) sample was obtained for genetic analysis. The buccal sample was sent to a Clinical Laboratory Improvement Amendments-approved UCLA molecular testing laboratory, where the gene for Connexin 26 was sequenced and the 309-kb deletion in Connexin 30 (del[*GJB6*-D13S1830]) assayed (del Castillo et al., 2002). Family and personal medical history was also obtained. Immediately following this genetic counseling session, participants completed the second questionnaire (post-counseling questionnaire).

The third stage of the study entailed a face-to-face genetic counseling session with the genetic counselor and sign language interpreter (some participants chose to communicate orally with our project genetic counselor) when the genetic test results were available. The genetic counselor explained the Connexin 26 and Connexin 30 genetic test results, put them in the context of the participant's family and medical history, and answered participants' questions. All participants received a copy of their genetic test report and a genetic counseling summary letter. In some cases, participants received additional information about genetics clinics in their area, either because they were interested in continuing to try to learn why they are deaf or because something of clinical importance with a strong genetic component was noted in the family history, for example, early-onset breast cancer. Genetic counseling and Connexin 26 and Connexin 30 genetic testing were provided at no charge to participants in the study. As part of the informed consent process, potential participants were informed that genetic counseling and genetic testing were also available outside of this study on their own.

About 1 month and 6 months after participants received their genetic test results, they were asked to complete the third questionnaire (1-month posttest questionnaire) and the final questionnaire (6-month posttest questionnaire), respectively. All four study questionnaires assessed nearly identical information to examine the effect of genetic information in a longitudinal framework. This study was approved by the relevant institutional review boards.

Measures

The study questionnaires used a mix of standard and newly developed items to assess demographic factors, reasons for genetic testing, attitudes toward genetic testing, knowledge and understanding of genetics and genetic testing, cultural affiliation and deaf identity, and a variety of psychological and behavior measures. Newly developed questionnaire items were pilot tested with a culturally diverse sample of deaf and hard-of-hearing individuals for clarity and comprehension, and revisions were made as needed. All questionnaire items were translated into ASL and Spanish, and back translated to ensure accuracy and equivalency of meaning (Brislin, 1970). For approximately the first year of participant enrollment, questionnaires were available only in English or Spanish while the

ASL translation process was taking place; hence, enrollment of individuals who preferred the ASL version of study questionnaires was deferred until the ASL translation process was complete. For the remaining period of the study, all questionnaires were available online in ASL only, English only, ASL–English, and Spanish only, and participants were able to complete the questionnaires in the language of their choice. This article focuses on cultural affiliation and reasons for genetic testing assessed from the baseline questionnaire.

Cultural affiliation. Cultural affiliation was assessed at baseline using two methods. One method used a categorical measure in which participants indicated "with which community they identify more" from a set of four response categories (hearing community, Deaf community, *both* communities, and *neither* community). The other method used the DIDS-R (Fischer & McWhirter, 2001), which contains 47 five-point Likert scale items (*strongly agree* to *strongly disagree*) decomposed into four identity subscales (hearing, immersion, bicultural, and marginal). The immersion, bicultural, and marginal identities of the DIDS-R are synonymous with the Deaf community, *both* communities, and *neither* community categories of the categorical measure, respectively. For the purposes of this article, we use the DIDS-R to validate the categorical measure of cultural affiliation. More detailed analyses of the DIDS-R and its relationship to the impact of genetic information are planned for a future publication.

Reasons for genetic testing. The baseline questionnaire contained 21 potential reasons for genetic testing, which were grouped into five general categories (self, family, partner, children, and community). These items were developed or adapted from studies on genetic testing and genetic testing for deafness (Brunger et al., 2000; Dagan, Hochner, Levi, Raas-Rothschild, & Sagi, 2002; Palmer et al., 2008; Taneja et al., 2004) or identified as important through pilot testing. For each item, respondents were asked to rate how strongly they agreed that the item was an important reason for genetic testing using a 5-point Likert scale of *strongly agree* (scored as 5) to *strongly disagree* (scored as 1). These items are treated as quantitative variables in the analyses.

Three items addressed "self": (a) "to learn why I am deaf/hard-of-hearing," (b) "to learn if genetics is the reason that I am deaf/hard-of-hearing," and (c) "to help me make decisions about the use of hearing aids, cochlear implants, or other devices for myself."

Four items addressed "family": (a) "to learn why there are other deaf/hard-of-hearing people in my family," (b) "because my family expects me to find out the reason I am deaf/hard-of-hearing," (c) "to help my deaf family to know why I am deaf/hard-of-hearing," and (d) "to help my hearing family to know why I am deaf/hard-of-hearing."

Five items addressed "partner"—Participants were asked if they have a current partner (yes or no) and those without a current partner rated four reasons for genetic testing: (a) "to make decisions about choosing a deaf/hard-of-hearing partner," (b) "to make decisions about choosing a hearing partner," (c) "to make decisions about choosing a genetically deaf/hard-of-hearing partner," and (d) "to make decisions about choosing a genetically hearing partner." Individuals with a current partner rated one reason for genetic testing: "to make decisions about continuing my relationship with my current partner."

Seven items addressed "children." First, participants were asked if they have children, with four possible response options (yes and planning to have more children, yes and not planning to have more children, no and planning to have children in the future, and no and not planning to have children in the future). Individuals whose response indicated that they have children rated two reasons for genetic testing: (a) "to help me make decisions about the use of hearing aids, cochlear implants, or other devices for my current or future children" and (b) "to explain why my child is deaf/hard-of-hearing." Individuals whose response indicated future family plans rated the following five reasons for genetic testing: (a) "to make decisions about whether or not to have (more) children," (b) "to learn if I can have deaf/hard-of-hearing children," (c) "to learn if I can have hearing children," (d) "to determine whether to have biologically deaf children or to adopt deaf children," and (e) "to determine whether to have biologically hearing children or to adopt hearing children." Individuals whose response indicated that they have no children and have no plans

for future children did not respond to any of these items.

Two items addressed "community": "to strengthen the deaf community" and "to help research." Pilot testing revealed a preference for using lowercase "d" with the former item.

Demographic characteristics, including age, sex, ethnicity/race, income, highest level of education achieved, linguistic preference, family history of deafness, and type of high school setting were assessed. Linguistic preference was defined as a categorical variable with four levels based on the participant's linguistic preference during the audiology and genetic counseling and testing sessions: ASL with interpreter present, ASL and English with interpreter present, English with no interpreter present, and other (signed English or pidgin signed English). Family history was defined as a categorical variable with two levels: no first- or second-degree relatives with early-onset deafness, or at least one first- or second-degree deaf relative. Type of high school program the participant attended was classified into four categories: hearing-based high school, which captures programs with predominantly oral instruction in the classroom, that is, oral school for the deaf, or a public school without interpreter/support services; deaf-based high school, which captures programs that predominantly used sign instruction (ASL or coded communication) in the classroom; mainstream high school, which captures programs in a public school that predominantly provided sign instruction (ASL or coded communication) with interpreter/support services; and mixed, which was defined as attending two or more of the previously described high school programs.

Statistical Analyses

Descriptive statistics were produced and reviewed for the presence of outliers and data blunders. Bivariate analyses were performed to determine if cultural affiliation was associated with demographic factors. Chi-square or Fisher's exact analysis was performed to determine if cultural affiliation was associated with sex, ethnicity/race, type of high school program, percent with bachelor's degree or higher, linguistic preference, family history, current partner status, percent with children, percent who desire future children, and involvement in community activities. Analysis of variance was performed to determine if age at enrollment or income was associated with cultural affiliation.

To examine deaf individuals' motivations for genetic testing through the lens of cultural affiliation or deaf identity, regression analysis was performed, with cultural affiliation as the independent variable and reasons for genetic testing as the outcome variable. A separate analysis was performed for each potential reason for genetic testing. Because participants' age, sex, family history (presence or absence of other deaf/hard-of-hearing family members), and education level may independently influence responses to reasons for genetic testing, these potential confounders were included as covariates in the analyses. Furthermore, because highest attained education level was significantly correlated with income, we included only education in the regression analyses because inclusion of income decreased the sample size due to missing data on this variable. For these analyses, education was treated as a categorical variable with three categories: high school diploma/vocational training or less, some college/2-year college degree, and 4-year college degree or higher. Regression analyses were conducted using the Proc MIXED procedure in SAS version 9.1 (SAS, 2002). Post hoc two-way comparisons were conducted to identify the specific group differences using Tukey's honestly significant difference test (Box, Hunter, & Hunter, 1978), which controls for the Type I experiment-wise error rate. To determine the sensitivity of the results to the relatively small sample size of the hearing community group, all regression analyses were rerun with only those in the Deaf community group and the *both* communities group. Restricting the analyses to the Deaf community and *both* communities groups led to no substantively different conclusions regarding similarities and differences between these two groups. Therefore, we report the results obtained from analyses using all three cultural groups with the caveat that outlier responses in the hearing community group may carry more weight in the analyses than would outlier responses in the other two cultural groups. Statistical significance was set at $\alpha = .05$.

Table 1 Demographics in entire sample and by cultural affiliation

	Entire sample	Hearing community	Deaf community	Both communities	*p* Value[a]
Sample size	256	18	140	90	
DIDS-R subscale mean score (*SD*)					
Hearing	41.7 (6.2)	36.2 (6.0)	43.9 (5.6)	39.3 (5.6)	–
Immersion (Deaf)	38.1 (8.6)	50.6 (7.3)	35.5 (7.2)	39.7 (8.4)	
Bicultural (*both*)	24.9 (6.5)	36.3 (7.5)	24.4 (5.8)	23.8 (5.6)	
Average age (*SD*), in years	46.2 (15.9)	37.5 (13.0)	47.3 (15.8)	45.7 (15.4)	.04*
Minimum	18.1	20.6	18.1	19.7	
Maximum	88.5	73.3	86.6	86.0	
% Female	58.6	61.1	58.6	55.6	.86
Ethnicity/race					
% Non-Hispanic Caucasian	77.7	66.7	80.7	77.8	.25
% Hispanic	11.7	11.1	12.9	10.0	
% Asian	9.0	16.7	5.7	10.0	
% Other	1.6	5.6	0.7	2.2	
Median income category, in thousands of $	35–50	50–65	35–50	35–50	.05*
High school program					
% Hearing based	30.1	82.4	19.3	34.9	<.0001*
% Deaf based	37.0	0.0	54.1	18.6	
% Mainstream	22.4	17.7	17.8	31.4	
% Mixed	10.6	0.0	8.9	15.1	
% With undergraduate bachelor or higher degree	53.9	72.2	58.0	47.8	.10
Linguistic preference during audiology/genetic counseling sessions					
% ASL, interpreter present	66.4	5.6	78.6	60.0	<.0001*
% ASL and English, interpreter present	21.5	5.6	20.7	27.8	
% English, no interpreter present	10.9	88.9	0.0	10.0	
% Other (signed English, PSE)	1.2	0.0	0.7	2.2	
% With deaf first- or second-degree relatives	55.3	44.4	62.9	48.9	.06
% Have a current partner	55.9	44.4	62.9	50.0	.09
% Have children	53.5	22.2	58.6	50.0	.01*
% Want future children	44.9	77.8	39.3	47.8	.007*

ASL, American Sign Language; DIDS-R, Deaf Identity Development Scale—Revised; PSE, Pidgin Signed English.
[a]Significance of association between demographic variable and cultural affiliation.
*Group differences significant at $p < .05$.

Results

Sample Demographics

A total of 271 participants completed the audiology evaluation portion of the study, 263 were determined to be eligible to participate in the genetic counseling and testing part of the study, and 248 of 263 (94.3%) continued on to that stage. Data collected on the baseline questionnaire from 256 of the 263 individuals (97.7%) determined eligible for genetic counseling and testing are reported in this article. Seven individuals were excluded because they (a) withdrew from the study following the audiology evaluation ($n = 3$), (b) did not complete a baseline questionnaire ($n = 3$), or (c) provided unusable questionnaire data ($n = 1$). Prior to the completion of the ASL version of the questionnaires, 88 participants completed a paper-and-pencil English version. After availability of the ASL version of the baseline questionnaire, 26, 79, and 63 participants completed it in ASL only, English only, and ASL–English combination, respectively. No participant selected the Spanish version of the questionnaire. Tables 1 and 2 provide selected demographic information on the sample.

Cultural Affiliation

Responses to the cultural affiliation item indicate that the sample is culturally diverse. As shown in Table 1,

Table 2 Involvement in Deaf community or hearing community activities, by cultural affiliation

	Cultural affiliation			
Activity	Hearing	Deaf	Both	*p* Value[a]
Deaf community involvement				
% Attend formal Deaf gathering events	22.2	92.1	80.0	<.0001*
% Attend informal Deaf gathering events	38.9	90.7	83.3	<.0001*
% Attend captioned movies	50.0	79.3	67.8	.01*
% Attend Deaf performances	16.7	78.6	64.4	<.0001*
% Participate or follow Deaf conferences	11.1	72.1	47.8	<.0001*
% Attend Deaf club activities	5.6	66.4	44.4	<.0001*
% Attend interpreted performances	11.1	63.6	54.4	<.0001*
Hearing community involvement				
% Attend informal hearing gathering events	88.3	36.4	54.4	<.0001*
% Attend hearing organization in neighborhood	66.7	11.4	26.7	<.0001*
% Attend hearing performance without ASL interpreter	72.2	14.3	30.0	<.0001*
% Attend formal hearing events	77.8	42.8	48.9	.02*

ASL, American Sign Language.
[a]Significance of association between involvement in activity and cultural affiliation.
*Group differences at $p < .05$.

7.0% of the sample selected the hearing community as their primary affiliation, 54.6% selected the Deaf community, 35.1% selected affiliation with *both* communities, and 2.3% responded that they do not feel affiliated with either the Deaf or the hearing community. Subsequent analyses exclude the six participants who marked "neither community" due to the small sample size of this group.

To assess the validity of the self-reported measure of cultural affiliation, the three cultural groups were compared on the DIDS-R subscale mean scores, linguistic preference, type of high school program, and involvement in community activities. As shown in Table 1, the hearing community group had the lowest DIDS-R hearing subscale mean score (indicating stronger endorsement of the subscale items), the Deaf community group had the lowest immersion subscale mean score, and the *both* communities group had the lowest bicultural subscale mean score. Thus, the categorical measure of deaf identity/cultural affiliation appears to provide a classification that is generally consistent with responses to the dimensional quantitative measure of deaf identity/cultural affiliation. In addition, consistent with the findings of previous studies (Bat-Chava, 2000; Hintermair, 2008; Nikolaraizi & Hadjikakou, 2006), the three cultural groups differed as expected in terms of linguistic preference and type of high school program they attended (see Table 1). Finally, participation in community activities also differed in expected ways across the three cultural groups (see Table 2). Together, these analyses support the validity of the categorical measure of cultural affiliation.

Although there were no expectations about a relationship between cultural affiliation and remaining demographic variables, we found that age, income, family history, percent with children, and percent desiring future children differed significantly, or nearly so, across the groups (see Table 1).

Examining Relationship Between Reasons for Genetic Testing and Cultural Affiliation. Results of the regression analyses are presented in Table 3, and the estimated mean scores for each cultural group for each reason for genetic testing are graphed in Figure 1.

Self. Results show that respondents are strongly interested in using genetic testing to learn why they are deaf and if it is genetic in origin. Furthermore, interest in using genetic testing for these reasons is not related to cultural affiliation. However, there was a significant association between cultural affiliation and interest in genetic testing to help make decisions regarding the use of hearing aids, cochlear implants, or other devices for themselves, even after accounting for age, sex, family history, and education (Table 3). Post hoc tests

Table 3 Significance (p values) of age, sex, family history, education, and cultural affiliation in explaining variation in responses to reasons for genetic testing: Results of regression analyses

Reasons for genetic testing	n	Age[a]	Sex	Family history	Education	Cultural affiliation	Significant cultural group differences (Tukey's HSD)[b]		
							D–B	D–H	B–H
Reasons related to "self"									
1. To learn why I am deaf/hard-of-hearing	245	.56	.58	.24	.33	.60			
2. To learn if genetics is the reason that I am deaf/hard-of-hearing	246	.75	.96	.99	.025*	.50			
3. To help me make decisions about the use of hearing aids, cochlear implants, or other devices for myself	246	.017*	.21	.12	.0007*	<.0001*	<.0001*	<.0001*	
Reasons related to "family"									
4. To learn why there are other deaf/hard-of-hearing people in my family	246	.031*	.71	<.0001*	.014*	.52			
5. Because my family expects me to find out the reason I am deaf/hard-of-hearing	245	<.0001*	.12	.33	.006*	.025*	.079		.062
6. To help my deaf family to know why I am deaf/hard-of-hearing	241	.08	.20	<.0001*	.058*	.0006*		.005*	.007*
7. To help my hearing family to know why I am deaf/hard-of-hearing	243	.029*	.65	.017*	.057*	.007*	.02*		.039*
Reasons related to "partner": those without a current partner									
8. To make decisions about choosing a deaf/hard-of-hearing partner	101	.40	.28	.23	.009*	.44			
9. To make decisions about choosing a hearing partner	100	.97	.15	.21	.21	.001*	.001*		
10. To make decisions about choosing a genetically deaf/hard-of-hearing partner	100	.26	.015*	.87	.007*	.82			
11. To make decisions about choosing a genetically hearing partner	100	.97	.059*	.74	.029*	.02*	.018*		
Reasons related to "partner": those with a current partner									
12. To make decisions about continuing my relationship with my current partner	140	.003*	.018*	.77	.17	.16			

Table 3 Continued

Reasons for genetic testing	n	Age[a]	Sex	Family history	Education	Cultural affiliation	Significant cultural group differences (Tukey's HSD)[b]		
							D–B	D–H	B–H
Reasons related to "children": those planning future children									
13. To make decisions about whether or not to have (more) children	109	.25	.85	.63	.034*	.98			
14. To learn if I can have deaf/hard-of-hearing children	110	.50	.46	.86	.91	.005*		.02*	.003*
15. To learn if I can have hearing children	108	.81	.19	.40	.49	.002*	.001*		
16. To determine whether to have biologically deaf children or to adopt deaf children	111	.078	.41	.96	.038*	.004*		.002*	.027*
17. To determine whether to have biologically hearing children or to adopt hearing children	108	.007*	.46	.98	.009*	.10			
Reasons related to "children": those with children and/or planning future children									
18. To help me make decisions about the use of hearing aids, cochlear implants, or other devices for my current or future children	204	.53	.31	.72	.25	<.0001*	<.0001*	.002*	
19. To explain why my child is deaf/hard-of-hearing	199	.28	.20	.016*	.98	.95			
Reasons related to community									
20. To strengthen the deaf community	245	.51	.081	.30	.21	<.0001*		<.0001*	.0001*
21. To help research	242	.74	.48	.60	.002*	.97			

HSD, honestly significant difference.

[a]Numbers in age, sex, family history, education, and cultural affiliation columns are the significance (p values) of these variables as predictors of interest in reason for genetic testing.

[b]Significant pairwise comparison between cultural groups where D = Deaf community, B = *both* communities, H = hearing community.

*Statistical significance at $p \leq .05$.

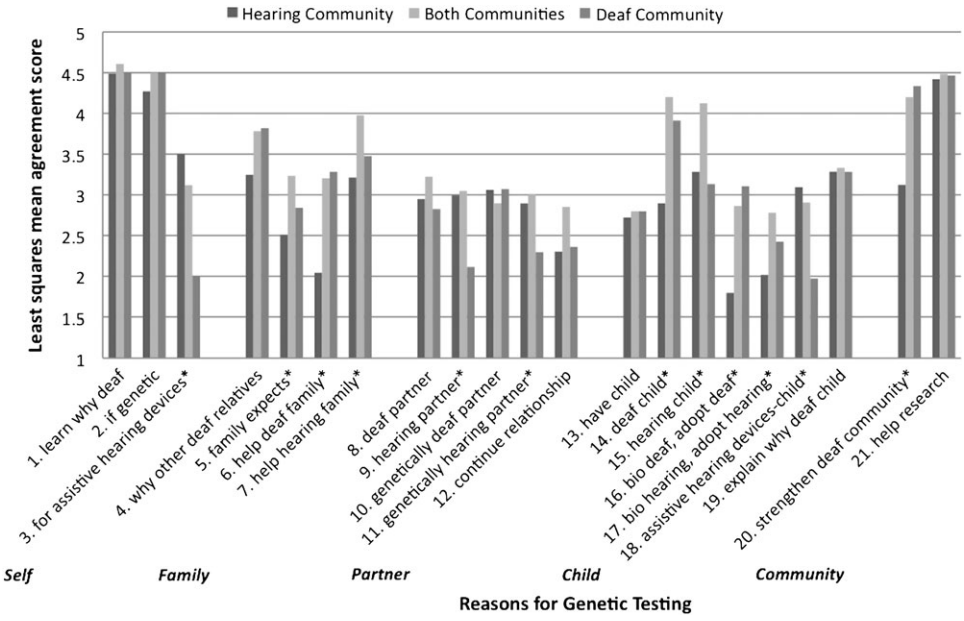

Figure 1 Least squares mean agreement scores for reasons for genetic testing by cultural affiliation (5 = *strongly agree*, 1 = *strongly disagree*). *denotes group differences at $p<.05$.

revealed that participants who marked affiliation with the Deaf community had significantly lower agreement scores with this item ($M = 2.0$), indicating stronger disagreement, than participants who marked cultural affiliation with the hearing community ($M = 3.5$) or with *both* communities ($M = 3.1$). Sex and family history were not significant predictors of responses to reasons for genetic testing related to "self," whereas age and education contributed to variation in responses to one or two of these items, respectively (Figure 1).

Family. Age, family history, and education level appear to contribute to interest in using genetic testing for family-related reasons. For all four items, there was a significant or nearly significant correlation between age and reason for testing, and education and reason for testing. Increasing age was associated with greater interest in using genetic testing for these reasons. In contrast, higher level of education was associated with less interest in using genetic testing for these reasons. Not surprisingly, individuals with at least one deaf first- or second-degree relative responded with greater interest in using genetic testing for reasons related to deaf family members, whereas those with no closely related deaf relatives had greater interest in using genetic testing to help their hearing family members to know why they are deaf (Table 3).

Inspection of Figure 1 reveals a general trend that individuals who marked cultural affiliation with the hearing community have lower mean scores, indicating greater disagreement, for pursuing genetic testing for reasons related to family than the other two cultural groups. Cultural affiliation was significantly associated with responses to three of the four items related to "family" reasons, even after controlling for the effects of age, sex, family history, and education. Individuals who marked affiliation with the Deaf community and those who marked affiliation with the hearing community were more likely to disagree that a reason for genetic testing is "because my family expects testing" (Ms = 2.8 and 2.5, respectively) compared to individuals who marked affiliation with *both* communities ($M = 3.2$). Furthermore, those who affiliate with the Deaf community and those who affiliate with the hearing community were less likely to agree that a reason for genetic testing is "to help my hearing family" (Ms = 3.5 and 3.1, respectively), compared to those who affiliate with *both* communities ($M = 4.0$; Figure 1).

Cultural group differences were also noted for the item "to help deaf family"; but in this case, those who affiliate with the hearing community were more likely

to disagree that this is a reason for genetic testing ($M = 2.3$) compared to those who affiliate with the Deaf community ($M = 3.3$) or *both* communities ($M = 3.3$), even after controlling for presence or absence of deaf family members. However, there was no association between cultural affiliation and interest in testing to learn why there are other deaf/hard-of-hearing people in the family after controlling for age, sex, family history, and education.

Partner. To examine the relationship between cultural affiliation and using genetic testing for partner-related purposes, we divided the sample into those individuals with, and those without, a current partner. Figure 1 reveals that average agreement scores for the four items related to choosing a partner and the one item related to maintaining a current relationship fall into the neutral-to-disagree range. Hence, in this sample, there appears to be little interest in using genetic testing for purposes of selecting a partner, or determining whether or not to continue a current relationship (Table 3).

Although responses generally fell into the neutral to disagree range, there were significant differences in mean agreement scores based on cultural affiliation with the two items about choosing a hearing partner. Individuals who marked affiliation with the Deaf community were more likely to disagree with the idea of pursuing genetic testing to make decisions about choosing a hearing partner ($M = 2.1$) or a genetically hearing partner ($M = 2.3$) compared to those who marked affiliation with *both* communities (Ms = 3.0 and 3.0, respectively; Figure 1). In addition, the data suggest that women were more likely to disagree with using genetic testing to select a partner based on genetic information than men. After accounting for cultural affiliation, age was not a significant predictor of interest in using genetic testing to assist in choosing a partner.

Whereas cultural affiliation was not associated with interest in using genetic testing for making decisions about continuing a current relationship, age and sex were significantly associated with this item. Specifically, level of interest in genetic testing for this purpose increased with age, and men had higher mean scores than women on this item.

Children. To examine the relationship between cultural affiliation and using genetic testing for children-related purposes, only those participants with plans for future children were included in analysis of five items related to future plans and then the larger group of participants with children and/or plans for subsequent children were grouped together for analysis for two additional items related to children (Table 3).

Inspection of Figure 1 demonstrates first that among respondents planning or wanting future children, there is little interest in pursuing genetic testing for the purposes of making decisions about whether or not to have children. Moreover, level of interest is unrelated to cultural affiliation. However, cultural affiliation was significantly associated, or nearly so, with interest in using genetic testing to learn about the chances of having deaf or hearing children and in making decisions about different methods for increasing chances of having deaf or hearing children, that is, biologically or through adoption.

Participants who marked affiliation with the hearing community generally indicated stronger disagreement about pursuing genetic testing to learn about or make decisions about the hearing status of their future children compared to those who marked affiliation with the Deaf community or *both* communities. Specifically, those who marked affiliation with the hearing community were more likely to disagree that a reason for having genetic testing is to learn if they can have deaf children ($M = 2.9$), whereas those who marked affiliation with the Deaf community or with *both* communities were more likely to agree with this reason (Ms = 3.9 and 4.2, respectively). Those who marked affiliation with the hearing community also more strongly disagreed with the idea of having genetic testing to determine whether to have biologically deaf children or to adopt deaf children ($M = 1.8$) compared to those who affiliate with the Deaf community ($M = 3.1$) or *both* communities ($M = 2.8$). Finally, those who marked affiliation with *both* communities were more likely to agree that they were interested in pursuing genetic testing to learn if they can have hearing children ($M = 4.1$) compared to those in the Deaf community ($M = 3.1$) or those in the hearing community ($M = 3.3$). After accounting for cultural

affiliation, family history was not associated with interest in genetic testing for any of these five items. However, age was significantly associated, or nearly so, with interest in determining whether to have biologically deaf or hearing children or to adopt deaf or hearing children, with level of interest increasing with age. Although education was not associated with learning if one could have deaf or hearing children, higher level of education was associated with less interest in using genetic testing to make decisions about future children.

Among participants with children and/or plans for subsequent children, Figure 1 shows that there is mild agreement in all three cultural groups for pursuing genetic testing for themselves to explain why they have deaf children even after controlling for presence or absence of closely related deaf relatives. In addition, family history was associated with level of interest with this item, where individuals with closely related deaf relative(s) had higher agreement scores than those without a closely related deaf relative. Cultural affiliation was also associated with deaf individuals' level of interest in using genetic testing to make decisions about the use of assistive hearing devices for their children (Table 3). Specifically, those who marked affiliation with the Deaf community were more likely to disagree that this was a reason for genetic testing ($M = 2.0$) compared to those who marked affiliation with the hearing community ($M = 3.1$) and with *both* communities ($M = 2.9$).

Community. Participants indicated that one reason they are interested in genetic testing is to help research, regardless of their cultural affiliation ($p = .97$), with mean scores indicating very strong agreement with this item. Cultural affiliation was associated with level of interest in having genetic testing to strengthen the deaf community. For this item, individuals who marked affiliation with the Deaf or *both* communities were more likely to agree with pursuing genetic testing to strengthen the deaf community (Ms = 4.3 and 4.2, respectively) compared to those who marked affiliation with the hearing community ($M = 3.1$; Table 3).

We also identified the levels of agreement or disagreement that most differentiated one cultural group from the other two groups and found that the largest difference occurred for agreement level of 3.75 and disagreement level of 2.5 (Figure 1). Doing so revealed that those who affiliate with *both* communities have mean agreement scores of 3.75 or higher for eight different reasons for testing (covering four domains), those who affiliate with the Deaf community have a score of at least 3.75 for six reasons (covering four domains), and those who affiliate with the hearing community have a score of 3.75 or higher for three reasons (covering two domains). In terms of disagreement, those who affiliate with *both* communities have mean disagreement scores of 2.5 or below for none of the reasons, those who affiliate with the Deaf community have a score of 2.5 or below for six reasons (covering three domains), and those who affiliate with the hearing community have a score of 2.5 or below for five reasons (covering three domains).

Discussion

This is the first study to examine the reasons why deaf individuals may be interested in genetic testing for deafness in the context of a prospective, longitudinal study design involving actual genetic counseling and genetic testing. With a sample size of 256 deaf/hard-of-hearing adults, this is also the largest study to date to examine how genetic testing is viewed and experienced by individuals in the deaf community. We examined participants' motivations for genetic testing across the domains of self, family, partner, children, and community, through the lens of deaf identity/cultural affiliation, to better understand and explain deaf individuals' reasons for pursuing genetic testing for deafness. Results from this study revealed a complex picture of deaf individuals' reasons for genetic testing, where cultural affiliation appears to play an important role in constructing the relevant motivations for testing. Overall, the findings of this empirical study both confirm and extend those of previous studies conducted either in the absence of genetic testing or sometime after genetic testing had occurred.

We found that three of the most important reasons for pursuing genetic testing in this sample of deaf and hard-of-hearing participants, regardless of their cultural affiliation, was to learn why they are deaf, if their

deafness is genetic in origin, and to help research. The first two items fall into the domain characterized as "self," and the third item was classified in the "community" domain. These results confirm and extend the findings of previous studies, in which the importance of genetic testing for clarifying self-identity or to satisfy curiosity (Burton et al., 2006; Withrow et al., 2008) has been observed in focus groups and cross-sectional surveys of deaf individuals, most of whom had not had genetic testing. There was also general consensus, regardless of cultural affiliation, that participants were not motivated to pursue genetic testing for partner-related reasons, as indicated by their neutral-to-disagree mean scores on all five partner-related items. This lack of interest in using genetic testing for partner selection has been noted in Withrow et al. (2008, 2009a) but not in Taneja et al. (2004). There are several possible explanations for the discrepancies across studies, including differences in statistical power; differences in the phrasing of the questions; differences in language of the questionnaire (English, ASL); differences in the genetic testing context in which responses were made, that is, prospectively, retrospectively, or hypothetically; and differences in the composition of the samples. In the current sample, age, sex, and education were significantly associated with partner-related items. Although this finding requires further study, it suggests that sample characteristics other than cultural affiliation may explain conflicting results across studies.

Variation based on cultural affiliation also was demonstrated. There was cultural variation based on the number and type of life domains perceived as relevant for consideration of genetic testing. We found that those who affiliate with the Deaf community and those who affiliate with *both* communities appear to be motivated to pursue genetic testing for reasons that relate to the domains of self, family, children, and community (both research community and deaf community), whereas those who affiliate with the hearing community are motivated by reasons related only to "self" and the research "community." Not only do those in the hearing community perceive little relevance for genetic testing for "family"- or "children"-related reasons, this group stands out with their decreased level of agreement that they are interested in genetic testing to make decisions about whether to have a biologically deaf or hearing child or adopt a deaf or hearing child. Of note, those who marked affiliation with the hearing community are significantly younger and more interested in future children than those who marked affiliation with the Deaf or *both* communities; hence, these findings are likely to be meaningful. Overall, these results suggest that although reasons for genetic testing for deafness can be multifaceted and cover a range of relevant life domains, the extent to which this is the case may be, in part, a function of deaf identity or cultural affiliation.

We found that although those who affiliate with the Deaf community and *both* communities are interested in genetic testing for some family and children-related reasons, those in the *both* communities group appear to be motivated by a larger number of reasons classified in these two domains. Interestingly, the explanation for this finding seems to lie in the relative importance accorded to hearing individuals, for example, family members. We found that individuals in both of the cultural groups were interested in genetic testing to learn why there are other deaf/hard-of-hearing family members. However, even after controlling for family history, those who affiliated with *both* communities were significantly more likely to also agree that they were interested in genetic testing "to help my hearing family to know why I am deaf/hard-of-hearing" than those in the Deaf community group. In a focus group of deaf adults, Withrow et al. (2008) identified "helping family members" as a motivation for genetic testing for deafness. Because our study differentiated between deaf relatives and hearing relatives, our findings suggest that there is greater complexity to family-related reasons than previously recognized. This area warrants further investigation.

There was little interest in this sample to pursue genetic testing for the purposes of making decisions about whether or not to have children, or to make choices between having biological children or to adopt children. However, individuals who affiliated with *both* communities and with the Deaf community were interested in genetic testing to learn if they can have deaf or hard-of-hearing children. Furthermore, those who affiliated with *both* communities were significantly more likely to also agree that they were interested in

genetic testing to learn if they can have a hearing child compared to those who affiliated with the Deaf community. These results both confirm and expand on earlier studies that documented the importance of genetic testing for learning about the chance of having deaf children (Burton et al., 2006; Withrow et al., 2008), as well as the importance of cultural affiliation in responses to reasons for genetic testing in this domain (Withrow et al., 2009a). Overall, the findings of this study suggest that although those who affiliate with the Deaf community and those who affiliate with *both* communities perceive a range of reasons for genetic testing for deafness, the concepts of "deaf" and "hearing" are salient in the motivations of those in *both* communities, whereas the reasons of those in the Deaf community are primarily related to further exploration, understanding, or strengthening of deafness—their own, their family members, their children, and their community.

Further evidence for the salience of "deaf" and lack of salience of "hearing" for those who affiliate with the Deaf community is found with two other reasons for genetic testing. Specifically, individuals who affiliated with the Deaf community were significantly more likely to disagree with the idea of pursuing genetic testing to make decisions about assistive hearing devices, either for themselves or for their children compared to the other two cultural groups. This is the first study to address the question of whether deaf adults consider genetic testing for themselves to make decisions about treating their child's deafness.

The strengths of this study include the large sample size and the prospective, longitudinal study design with actual genetic counseling and genetic testing. Furthermore, our analyses also took into account age, sex, presence or absence of deaf family members, and education, thereby providing the most rigorous analysis of deaf individuals' motivations for genetic testing to date. However, as with all studies, this study has limitations. One limitation is that our measure of cultural affiliation is categorical and may have been too simplistic to capture the nuances of cultural affiliation/deaf identity. To address this potential limitation, we demonstrated that the categorical measure correlated in predictable and expected ways with linguistic preference, type of high school setting, involvement in community activities, and the quantitative dimensional measure of cultural affiliation/deaf identity as measured via the DIDS-R, thereby strengthening the validity of the categorical measure of cultural affiliation/deaf identity. However, it is still likely that a categorical measure does not fully capture the considerable fluidity to deaf identity/cultural affiliation (Breivik, 2005). For that reason, we plan to use data collected on the DIDS-R over the four time points to conduct more detailed analyses to examine potential changes in deaf identity over time, as a function of genetic information.

A second limitation is the lack of ethnic/racial diversity in the composition of the sample (majority were Caucasian individuals), thus generalizing these results to deaf individuals of non-Caucasian backgrounds should be done with caution. Furthermore, because previous research has documented ethnic/racial differences in motivations for genetic testing for deafness in a sample of hearing parents of a deaf child (Palmer et al., 2008), future research is needed to determine if ethnic/racial heritage is a significant factor in motivations for genetic testing among deaf adults. Along the same lines, this sample derives from the United States, and so generalizability of findings to other countries should be done with caution, particularly given cross-national differences in issues raised by genetic testing such as the UK Human Fertilisation and Embryology Act.

It is also possible that the availability of free genetic counseling and genetic testing was an incentive for participation in the study, which might have skewed our results to suggest strong interest in genetic testing. In the United States, many health insurance policies and other forms of health coverage will cover genetic counseling and genetic testing for deaf genes, so in reality, the availability of "free genetic counseling and genetic testing" in our study does not differ substantively from how genetic counseling and genetic testing would normally be available to deaf individuals in the United States. However, for a variety of reasons, deaf individuals currently may be unaware that genetic counseling and genetic testing for deaf genes may be available to them at no cost (or low cost) through their health care provider, or they may be disinclined to pursue these services in environments that are not

culturally sensitive. Consistent with these hypotheses, recent work demonstrates that the majority of a sample of deaf individuals in the United Kingdom (who have access to a national health care service) do not know how to access genetic counseling (Middleton et al., 2010) and that providing deaf awareness training to health providers could facilitate fulfilling the communication preferences of many deaf/hard-of-hearing individuals in a clinic setting (Middleton et al., in press). Thus, rather than limiting the generalizability of our results, the likely effects of providing genetic counseling and genetic testing at no charge in a culturally and linguistically appropriate environment were to increase the number and the diversity of deaf individuals willing to participate in the study, thereby strengthening the validity and generalizability of the results.

Finally, although this study of genetic testing has the largest sample of deaf adults to date, analyses based on cultural affiliation were based on a relatively small number of deaf individuals who identified with the hearing community, and the relatively small sample size of this group might have influenced the results. However, restricting the analyses to focus only on the Deaf community and *both* communities groups did not lead to substantively different conclusions regarding similarities and differences between these two larger groups. It is more likely that we lacked sufficient statistical power to identify all meaningful differences between the hearing community group and the other two groups in motivations for genetic testing for deafness, as indicated by the number of nearly significant p values. Thus, although our results suggest that cultural affiliation influences the number and type of domains for which genetic testing is pursued, replication studies with larger samples and additional potential reasons for testing are needed.

Conclusions

Previous survey studies in the United States have documented that deaf/hard-of-hearing individuals are more hesitant to seek genetic counseling services compared to hearing individuals (Martinez et al., 2003) and that young deaf adults feel that the professional discussing genetic testing should be familiar with Deaf culture (Withrow et al., 2009b). Our experience successfully recruiting this large U.S. sample of culturally diverse deaf and hard-of-hearing participants to participate in a study involving actual genetic counseling and testing provides empirical evidence that deaf and hard-of-hearing adults are interested in genetic counseling and genetic testing for deafness when it is accessible in a culturally and linguistically appropriate manner. Furthermore, the construction of this research project gave deaf individuals an opportunity to empower themselves by expressing their views and beliefs about genetic counseling and testing. As a result, we found strong interest among all participants to use genetic testing to learn why they are deaf and if it is genetic. We also found very little interest among all participants to use genetic testing to make decisions about a current or future partner or decisions about whether to have children. We found that the number of life domains relevant for genetic testing, as well as the number of reasons within a life domain, particularly as they relate to the salience of "deaf" and "hearing," varies as a function of cultural affiliation.

Finally, although this study reinforces the significance attributed to genetic testing for explaining why an individual is deaf, for many deaf people around the world, it is no simple matter to just weigh up whether one should personally avail oneself of genetic testing opportunities as this may be fraught with political and legal implications. Add to this complexity our finding that a variety of motivations for genetic testing are, in part, culturally mediated. The implications of culturally mediated motivations for genetic testing on deaf adults and the deaf/Deaf community are currently unknown, and will be examined in our future publications.

Note

1. In this article, we use the term deaf community to encompass all individuals who are audiologically deaf or hard of hearing, regardless of their deaf identity or cultural affiliation. We use "D" when referring to the subgroup of deaf individuals who are culturally Deaf, and to refer to Deaf culture.

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Conflict of Interest

No conflicts of interest were reported.

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References

Arnos, K. S. (2003). The implications of genetic testing for deafness. *Ear & Hearing, 24,* 324–331.

Bat-Chava, Y. (1994). Group identification and self-esteem of deaf adults. *Personality and Social Psychology Bulletin, 20,* 494–502.

Bat-Chava, Y. (2000). Diversity of deaf identities. *American Annals of the Deaf, 145,* 420–428.

Bauman, H.-D. (Ed.). (2008). *Open your eyes: Deaf studies talking*. Minneapolis: University of Minnesota Press.

Bell, A. G. (1883). Upon the formation of a deaf variety of the human race. *National Academy of Sciences Memoirs, 2,* 177–262.

Biesold, H. (1999). *Crying hands: Eugenics and deaf people in Nazi Germany.* Washington, DC: Gallaudet University Press.

Box, G. E. P., Hunter, W. G., & Hunter, J. S. (1978). *Statistics for experimenters.* New York: John Wiley & Sons.

Breivik, J.-K. (2005). *Deaf identities in the making: Local lives, transnational connections*. Washington, DC: Gallaudet University Press.

Brislin, R. W. (1970). Back-translation for cross-cultural research. *Journal of Cross Cultural Psychology, 1,* 185–216.

Brueggemann, B. J. (2009). *Deaf subjects: Between identities and places.* New York: New York University Press.

Brunger, J. W., Murray, G. S., O'Riordan, M., Matthews, A. L., Smith, R. J. H., & Robin, N. H. (2000). Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Human Genetics, 67,* 1621–1625.

Burton, S. K., Withrow, K., Arnos, K. S., Kalfoglou, A. L., & Pandya, A. (2006). A focus group study of consumer attitudes toward genetic testing and newborn screening for deafness. *Genetics in Medicine, 8,* 779–783.

Cohn, E. S., & Kelley, P. M. (1999). Clinical phenotype and mutations in connexin 26 (DFNB1/GJB2), the most common cause of childhood hearing loss. *American Journal of Medical Genetics, 89,* 130–136.

Dagan, O., Hochner, H., Levi, H., Raas-Rothschild, A., & Sagi, M. (2002). Genetic testing for hearing loss: Different motivations for the same outcome. *American Journal of Medical Genetics, 113,* 137–143.

del Castillo, I., Villamar, M., Moreno-Pelayo, M. A., del Castillo, F. J., Alvarez, A., Telleria, D., et al. (2002). A deletion involving the connexin 30 gene in nonsyndromic hearing impairment. *New England Journal of Medicine, 346,* 243–249.

Denoyelle, F., Weil, D., Maw, M. A., Wilcox, S. A., Lench, N. J., Allen-Powell, D. R., et al. (1997). Prelingual deafness: High prevalence of a 30delG mutation in the connexin 26 gene. *Human Molecular Genetics, 6,* 2173–2177.

Emery, S., Burke, T. B., Middleton, A., Belk, R., & Turner, G. (2008). Reproductive liberty and deafness: Clause 14(4)(9) of embryo bill should be amended or deleted. *BMJ, 336,* 976.

Fischer, L. C., & McWhirter, J. J. (2001). The deaf identity development scale: A revision and validation. *Journal of Clinical Psychology, 48,* 355–358.

Glickman, N. S., & Carey, J. C. (1993). Measuring deaf cultural identities: A preliminary investigation. *Rehabilitation Psychology, 38,* 275–282.

Green, G. E., Scott, D. A., McDonald, J. M., Woodworth, G. G., Sheffield, V. C., & Smith, R. J. (1999). Carrier rates in the midwestern United States for GJB2 mutations causing inherited deafness. *Journal of the American Medical Association, 281,* 2211–2216.

Guillemin, M., & Gillam, L. (2006). Attitudes to genetic testing for deafness: The importance of informed choice. *Journal of Genetic Counseling, 15,* 51–59.

Hintermair, M. (2008). Self-esteem and satisfaction with life of deaf and hard-of-hearing people—A resource-oriented approach to identity work. *Journal of Deaf Studies and Deaf Education, 13,* 278–300.

Holcomb, T. K. (1997). Development of Deaf bicultural identity. *American Annals of the Deaf, 142,* 89–93.

Hole, R. (2007). Narratives of identity: A poststructural analysis of three Deaf women's life stories. *Narrative Inquiry, 17,* 259–278.

Jacobs, L. M. (1989). *A deaf adult speaks out*. Washington, DC: Gallaudet University Press.

Kannapell, B. (1993). *Language choice—Identity choice*. Burtonsville, MD: Linstok Press.

Kelley, P. M., Harris, D. J., Comer, B. C., Askew, J. W., Fowler, T., Smith, S. D., et al. (1998). Novel mutations in the connexin 26 gene (GJB2) that cause autosomal recessive (DFNB1) hearing loss. *American Journal of Human Genetics, 62,* 792–799.

Kenna, M. A., Wu, B. L., Cotanche, D. A., Korf, B. R., & Rehm, H. L. (2001). Connexin 26 studies in patients with sensorineural hearing loss. *Archives of Otolaryngology and Head & Neck Surgery, 127,* 1037–1042.

Lane, H. (2005). Ethnicity, ethics, and the deaf-world. *Journal of Deaf Studies and Deaf Education, 10,* 291–310.

Leigh, I. W. (1999). Inclusive education and personal development. *Journal of Deaf Studies and Deaf Education, 3,* 330–338.

Leigh, I. W., Marcus, A., Dobosh, P., & Allen, T. (1998). Deaf/hearing cultural identity paradigms: Modification of the deaf identity development scale. *Journal of Deaf Studies and Deaf Education, 3,* 329–338.

Martinez, A., Linden, J., Schimmenti, L. A., & Palmer, C. G. S. (2003). Attitudes of the broader hearing, deaf, and hard-of-hearing community toward genetic testing for deafness. *Genetics in Medicine, 5,* 106–112.

Maxwell-McCaw, D. (2001). *Acculturation and psychological well-being in Deaf and hard of hearing people*. Doctoral dissertation. George Washington University: Washington, DC.

Middleton, A. (2007). Attitudes of deaf people and their families towards issues surrounding genetics. In A. Martini, D. Stephens, & A. P. Read (Eds.), *Genes, hearing and deafness: From molecular biology to clinical practice* (pp. 163–172). London: Informa Healthcare.

Middleton, A., Emery, S. D., & Turner, G. H. (2010). Views, knowledge, and beliefs about genetics and genetic counseling among deaf people. *Sign Language Studies, 10,* 170–196.

Middleton, A., Hewison, J., & Mueller, R. F. (1998). Attitudes of deaf adults toward genetic testing for hereditary deafness. *American Journal of Human Genetics, 63,* 1175–1180.

Middleton, A., Turner, G. H., Bitner-Glindzicz, M., Lewis, P., Richards, M., Clarke, A., et al. (in press). Preferences for communication in clinic from deaf people: A cross-sectional study. *Journal of Evaluation in Clinical Practice.*

Nikolaraizi, M., & Hadjikakou, K. (2006). The role of educational experiences in the development of deaf identity. *Journal of Deaf Studies and Deaf Education, 11,* 477–492.

Ohna, S. E. (2004). Deaf in my own way: Identity, learning, and narratives. *Deafness and Education International, 6,* 20–38.

Padden, C., & Humphries, T. (1988). *Deaf in America: Voices from a culture*. Cambridge, MA: Harvard University Press.

Padden, C., & Humphries, T. (2005). *Inside deaf culture*. Cambridge, MA: Harvard University Press.

Palmer, C. G. S., Martinez, A., Fox, M., Sininger, Y., Grody, W. W., & Schimmenti, L. A. (2008). Ethnic differences in parental perceptions of genetic testing for deaf infants. *Journal of Genetic Counseling, 17,* 129–138.

Pandya, A., Arnos, K. S., Xia, X. J., Welch, K. O., Blanton, S. H., Friedman, T. B., et al. (2003). Frequency and distribution of GJB2 (connexin 26) and GJB6 (connexin 30) mutations in a large North American repository of deaf probands. *Genetics in Medicine, 5,* 295–303.

SAS. (2002). *SAS (version 9.1)*. Cary, NC: SAS Institute Inc.

Schimmenti, L. A., Martinez, A., Telatar, M., Lai, C. H., Shapiro, N., Fox, M., et al. (2008). Infant hearing loss and connexin testing in a diverse population. *Genetics in Medicine, 10,* 517–524.

Sheridan, M. (2000). *Inner lives of deaf children*. Washington, DC: Gallaudet University Press.

Stern, S. J., Arnos, K. S., Murrelle, L., Welch, K. O., Nance, W. E., & Pandya, A. (2002). Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss. *Journal of Medical Genetics, 39,* 449–453.

Taneja, P. R., Pandya, A., Foley, D. L., Nicely, L. V., & Arnos, K. S. (2004). Attitudes of deaf individuals towards genetic testing. *American Journal of Medical Genetics, 130A,* 17–21.

Withrow, K. A., Burton, S., Arnos, K. S., Kalfoglou, A., & Pandya, A. (2008). Consumer motivations for pursuing genetic testing and their preferences for the provision of genetic services for hearing loss. *Journal of Genetic Counseling, 17,* 252–260.

Withrow, K. A., Tracy, K. A., Burton, S. K., Norris, V. W., Maes, H. H., Arnos, K. S., et al. (2009a). Impact of genetic advances and testing for hearing loss: Results from a national consumer survey. *American Journal of Medical Genetics Part A, 149A,* 1159–1168.

Withrow, K. A., Tracy, K. A., Burton, S. K., Norris, V. W., Maes, H. H., Arnos, K. S., et al. (2009b). Provision of genetic services for hearing loss: Results from a national survey and comparison to insights obtained from previous focus group discussions. *Journal of Genetic Counseling, 18,* 618–621.