

Consumer Attitudes towards Genetic Testing and Newborn Screening

**K. Arnos, S. Burton, K. Withrow, V. Norris,
S. Blanton, A. Kalfoglou, and A. Pandya**

Gallaudet University, Washington, D.C.

Virginia Commonwealth University, Richmond, VA

University of Miami, Miami, FL, NHGRI, NIH, Bethesda, MD

Background

- NBHS programs along with recent progress in identifying genes for deafness has led to greater utilization of genetic services by parents of children with hearing loss.
- Efforts to assess consumer attitudes have lagged behind.

Long Term Goals

1. To explore knowledge and attitudes of hearing parents of deaf children and deaf adults about emerging ethical dilemmas created by advances in genetics.
2. Study the long term impact of genetic testing on attitudes and behavior of deaf adults.

Objectives

- Explore attitudes towards
 - genetic technologies.
 - motivation for seeking genetic testing for hearing loss.
 - idea of adding universal molecular screening for hearing loss at birth.

Structure for Data Collection

- Phase I – Focus Groups
- Phase II – National Parent Survey

Design and Methods: Phase I

Focus Groups

- 5 Focus Groups
- Groups were conducted by experienced moderators, either deaf or hearing.
- Format of discussion followed detailed focus group moderator guides.
- Sessions lasted about 2 hours, were audio taped, and transcribed.

Focus Groups Composition

Location	Hearing Status	Description	Size	Sex	Race
Virginia Commonwealth University	Hearing	Parents	5	4 F 1 M	5 Caucasian
Gallaudet University	Hearing	Parents	9	8 F 1 M	5 African American 1 Hispanic 3 Caucasian
Gallaudet University	Deaf	Parents	6	6 F	1 African American 1 Pacific Islander 4 Caucasian
Gallaudet University	Deaf	Students	16	7 F 9 M	4 African American 2 Hispanic 3 Asian 7 Caucasian
Gallaudet University	Deaf	Students	8	4 F 4 M	1 African 1 African American 1 Hispanic 5 Caucasian

Methods: Phase I

Focus Groups

- Transcripts were coded independently by 5 investigators into predetermined categories with positive and negative codes for each category.
- Analysis of this coded data was done using qualitative data analysis program NVIVO[©] 2.0.
- Results were used to refine the content of a national parent survey (phase II).

Coding Structure

Broad categories

- Perception of deafness
- Perception of Genetic technology for HL
- Perception of Newborn Hearing Screening/EHDI programs & its Future
- Motivation & Outcome of Genetic Services
- Provision of Genetic Services

Coding Structure

Sub-codes

Motivation & Outcomes

- Diagnosis Confirmation
- Acceptance of Diagnosis
- Self Identity/ Understanding of Self
- Learning about reproductive future
- Information to be used in spouse selection
- Information to be used for family members
- General Curiosity
- Treatment of the Condition

Results: Phase I

Attitudes Towards Genetic Technologies

- Most parents demonstrated a good understanding of advances in genetic technology
- The comments on this issue were equally divided towards positive, negative or indifferent
- Parents also often worried about finding out information they did not wish to know

Results: Phase I

Attitudes Towards Genetic Technologies

- Deaf participants expressed concerned about use of technology leading to elimination of Deaf culture.
- Hearing parents rated technology in tiers, with technologies leading to “solutions” prioritized higher.

Results: Phase I

Attitudes Towards Genetic Technologies

- Negative comments reflected concerns about reliability, cost effectiveness & parental misunderstandings that a test always provides clear answers.

“There should be some sort of caveat saying there is no guarantee that genetic testing will give you answers – That is what happened with my kids. We still don’t know the exact cause of their deafness. There are hundreds of other genes that haven’t been discovered. It would help manage our expectations”

Results: Phase I

Attitudes Towards Genetic Technologies

Deaf participants were concerned that use of technology might lead to elimination of Deaf culture.

“To use [genetic testing] for the purpose of eliminating the deaf gene or preventing the growth of the deaf population, I think that is very negative. It is negative if you abort or terminate the pregnancy to stop the genetic trait of deafness from being passed on. If I, as a deaf person, had a deaf child, I would be proud to pass on my traditions.”

Results: Phase I

Motivations for Seeking Molecular Testing for HL

- Great interest was expressed in using genetic test results to aid in establishing a diagnosis.
- Both deaf & hearing individuals expressed interest in learning about the chance of having deaf children either to satisfy curiosity or for planning purposes.

Results: Phase I

Motivations For Seeking Molecular Testing For HL

- Motivations ranged from assistance with coping process & confirming syndromic forms of HL, to acquiring information to help them, their children & other family members prepare for the future.

**“I think genetic evaluation is part of the healing process ... When your child is first diagnosed, you are in denial, however, if you get enough information, follow-up and resources it helps solidify things. After genetic testing, it was very clear how my baby became deaf.
Ok – we found this out – now what?”**

Results: Phase I

Motivations for Seeking Molecular Testing for HL

Deaf participants expressed interest in learning about the chance of having deaf children either to satisfy curiosity or for planning purposes.

“I think it’s very important for us to know who we are, and how we became deaf. If we didn’t have that opportunity... it would be like part of us is missing. Also, it’s nice to know if my kids are going to be deaf or not.... If we know what to expect, then we are ready to make the right decisions when it comes to schooling or job choices.”

Results: Phase I

Views Towards Newborn Screening

- All participants voiced clear support for the current EHDI programs.
- Concern was expressed about lack of ongoing support and intervention after diagnosis of HL is made.
- Parents were equally divided on need for parental consent prior to doing newborn molecular screening.

Results: Phase I

Views Towards Newborn Hearing Screening

- Parents were divided on the idea of adding universal molecular screening for select mutations. Cost effectiveness was the primary concern.

“H.L. is of fairly low incidence – if you think about the cost of testing 999 that don’t have it vs. the 1 that does – that takes up resources that could be better spent on other things.”

Results: Phase I

Views Towards Newborn Hearing Screening

- Concerns about cost effectiveness were countered with concerns for the overall wellbeing for of the child.

“How much do you have to put into the child who is identified later vs. if you start early. The cost in the long run is way more in terms of education and functionality of that child. ”

Results: Phase I

Views Towards Timing of Referral to Genetic Evaluation and Testing

“It wasn't until my second son was born that genetic testing was available to me... I believe if I had had more information about deafness earlier in my children's lives, I would have been better at advocating for them in the sense that I would have some knowledge, less ambiguity, and therefore more authority where they are concerned. ”

Results: Phase I

Burton et al (Dec 2006). A focus group study of consumer attitudes toward genetic testing and newborn screening for deafness. Genetics in Medicine 8: 779-783.

Please contact the speaker by email for a copy of this publication.

kathleen.arnos@gallaudet.edu

Design and Methods: Phase II National Parent Survey

- Parent survey mailed Fall, 2006 with 36 items designed to assess perceptions about
 - genetic testing for hearing loss
 - audiologic newborn hearing screening
 - addition of molecular screening to newborn screening protocols

Design and Methods: Phase II National Parent Survey

Data from this survey will be published soon.
Please contact the speaker for a copy of
the publication.

Kathleen.arnos@gallaudet.edu

Overall Conclusions

1. Feelings about advances in genetic technology for HL varied based on personal priorities and perception of deafness as a medical problem vs. a cultural identity.
2. The motivations for pursuing genetic testing varied somewhat for the hearing and deaf groups.

Collaborators

- Gallaudet University
 - Kathleen Arnos
 - Ginger Norris
- University of Miami
 - Susan Blanton
- Virginia Commonwealth University
 - Arti Pandya
 - Kara Withrow
- NHGRI/NIH
 - Andrea Kalfaglou
- Focus Group Moderators
 - Alfred Sonnenstrahl
 - Carol Prindle

This study was supported by grant number 1 R01 DC005831 (Arti Pandya, PI) from the National Institute on Deafness and Other Communication Disorders, NIH.