I, Kayla Czape, hereby submit this original work as part of the requirements for the degree of:

Master of Science

in Genetic Counseling

It is entitled:

Parent preferences regarding educational material

and genetic counseling for hearing loss genetic testing

Student Signature: Kayla Czape

This work and its defense approved by:

Committee Chair: John Greinwald, Jr., MD

Erin Acra, MS

John Clark, PhD

Judith A. Johnson
Parent preferences regarding educational material and genetic counseling for hearing loss genetic testing

A thesis submitted to the Graduate School of the University of Cincinnati in partial fulfillment of the requirements for the degree of Master of Science

In the Program of Genetic Counseling of the College of Medicine

By Kayla Czape, B.S., B.A. Indiana University, 2007 on May 26th 2010

Committee Chair: John H. Greinwald, Jr., MD
Abstract

Despite the advances in genetic testing for hearing loss in recent years, testing for genetic causes is not comprehensive and negative genetic test results leave families with uncertainty regarding etiology, progression, associated medical conditions, and recurrence risk. From a genetic counseling standpoint, this gap in patient care should be assessed in order to provide the patient population with adequate information through educational materials and/or genetic counseling. The goal of the current study is to develop a comprehensive review of the services and materials that are provided, needed and preferred for parents whose children have had negative results from genetic testing for hearing loss. We hypothesize that this parent population would prefer some form of educational material or genetic counseling regarding the results of their child’s genetic test. The study was accomplished through qualitative phone interviews of 16 parents and quantitative surveys of 35 healthcare providers involved in hearing loss genetic testing at a large pediatric institution. Both data sets were described individually and compared qualitatively to describe parallels and differences. Our data reveals that parents have a desire for more information regarding the genetic test ordered for their child’s hearing loss and that modification of the current practices and provision of educational materials and genetic counseling for this population may improve their understanding and satisfaction with the hearing loss genetic testing process.

Keywords

genetic testing, hearing loss, genetic counseling, educational materials, client preferences, needs assessment
Acknowledgements

Thank you to those who contributed to and supported this research project during my training in the Genetic Counseling Program at the University of Cincinnati and Cincinnati Children’s Hospital Medical Center. This project was made possible through the guidance of my program director, Dr. Melanie Myers and the endless support of my classmates and supervisors. A special thanks is extended to Bev Lipton, whose tireless recruitment efforts are greatly appreciated! I would like to extend my utmost gratitude to the chair of my Research Advisory Committee, Dr. John Greinwald and the members of the committee: Dr. John Greer Clark, Judith Johnson, and Erin Mundt for their contributions, encouragement, and guidance throughout my learning experience. Thank you all!
# Table of Contents

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>List of Figures and Appendices</td>
<td>vii</td>
</tr>
<tr>
<td>Introduction</td>
<td>1</td>
</tr>
<tr>
<td>Methods</td>
<td>5</td>
</tr>
<tr>
<td>Results</td>
<td>8</td>
</tr>
<tr>
<td>Discussion and Conclusions</td>
<td>16</td>
</tr>
<tr>
<td>Bibliography</td>
<td>22</td>
</tr>
<tr>
<td>Figures</td>
<td>25</td>
</tr>
<tr>
<td>Appendices</td>
<td>27</td>
</tr>
</tbody>
</table>
List of Figures and Appendices

Figure 1: Providers’ reasons for not providing pretest educational materials to the target population

Figure 2: Providers’ selection of best format of educational material for the target population

Appendix A: Parent Interview Guide

Appendix B: Provider Survey Questions
**Introduction**

Hearing loss is the most common congenital sensory deficit found in approximately one in every 500 newborns and 3.5 in every 1000 adolescents (Morton 2006). Approximately 50% of hearing deficits in children are due to a genetic cause. Of these, approximately 70% are not associated with a known syndrome and have a variety of inheritance patterns including autosomal dominant, autosomal recessive, mitochondrial and X-linked (Van Camp et al. 1997). A significant number of those with a non-syndromic genetic etiology can be attributed to a mutation in one of the over 40 genes known to be associated with hearing loss (Hilgert et al. 2009). Although the genetics of hearing loss is extremely heterogeneous, routine screening panels typically target a select number of genes which account for a high frequency of mutations or are associated with well defined clinical or prognostic features (Hilgert et al. 2009). For example *GJB2* mutations have been found to account for 24.3% of autosomal recessive cases (Putcha et al. 2007) and mutations in the *OTOF* and *CDH23* genes have implications for progression and associated conditions (Hilgert et al. 2009). Although current genetic testing accounts for a large number of genetic causes, it is not comprehensive and some patients may have mutations in less prevalent genes or causative genes yet to be identified (Hilgert et al. 2009). Due to these complications, genetic testing may not provide information about the etiology of the hearing loss and many patients and families may be left with few answers regarding progression, etiology, and recurrence risk.

Currently at Cincinnati Children’s Hospital Medical Center (CCHMC), seven common hearing loss genes are tested in a two-tier system ([http://www.cincinnatichildrens.org/svc/alpha/m/molecular-genetics/hearing-loss/](http://www.cincinnatichildrens.org/svc/alpha/m/molecular-genetics/hearing-loss/)). Parents whose child has a genetic test for hearing loss ordered at CCHMC that reveals a genetic cause
(i.e. a positive test result) are typically offered genetic counseling to discuss recurrence risks, etiology, and management. However, when results are negative and no genetic cause for the hearing loss is identified, patients are not typically offered genetic consultations at CCHMC. A negative test result from genetic testing for hearing loss does not rule out genetic etiology of hearing loss and does not eliminate recurrence risk, as these panels test a limited number of genes associated with hearing loss and a genetic etiology may still exist. In fact, even with a negative test result, the recurrence risk may be as high as 25% if the unknown genetic cause is autosomal recessive in inheritance. This concept seems to be misunderstood by many parents of children with hearing loss and negative genetic test results (Brunger et al. 2001; Robin et al. 2005). Recent studies have shown that parents’ knowledge about hearing loss genetics, including knowledge of recurrence risk is poor (Brunger et al. 2001). It has also been shown that genetic counseling increases parents’ knowledge of hearing loss genetics (Parker et al. 2000; Rehm et al. 2004) and a statement by the American College of Medical Genetics calls for pretest and posttest genetic counseling for hearing loss genetic testing regardless of outcome (ACMG 2002).

A negative genetic test result for hearing loss may have psychosocial implications for a family (Palmer et al. 2003). In studies to elicit parents’ motivations for genetic testing for hearing loss, the most important reasons for testing were curiosity of their child’s condition, relief of guilt of causing the hearing loss, coping, and to acquire information to prepare for the future (Brunger et al. 2000; Dagan et al. 2002; Palmer et al. 2003; Withrow et al. 2008; Withrow et al. 2009). For parents whose child’s genetic test is negative, these motivations for testing may not be satisfied and exploration of the residual psychological stress of guilt, inability to cope, and uncertainty about the future is warranted. Additionally, uncertainty of cause may lead parents to inaccurate conclusions about the cause of their child’s hearing loss or lead to anxiety or guilt.
The goals of having genetic counseling regarding a child’s test result are to address these parental feelings of guilt and uncertainty and alleviate misconceptions regarding cause in light of a negative result.

From a genetic counseling standpoint, it is important to provide families who have no known genetic cause for their child’s hearing loss with the proper genetic counseling or appropriate educational material so that they can be more informed about the possible etiology and recurrence risks of the hearing loss in their family. Genetic counseling has been shown to help patients understand their genetic concerns and improve their knowledge about genetic testing (Veach et al. 1999, Palmer et al. 2003). Additionally, the genetic counseling session is the appropriate venue to address any emotional impact of the disorder including feelings of uncertainty or guilt regarding the hearing loss. Genetic counseling strives to provide emotional support for families going through genetic testing and has been shown to improve well-being and reduce anxiety (Bernhardt et al. 2000, Davey et al. 2005). Recent literature reveals that parents would prefer to meet with a genetics professional, such as a genetic counselor or geneticist to discuss the genetic test for hearing loss (Withrow et al. 2008). However, the demand for genetics professionals to provide this counseling may outweigh the supply of qualified professionals available. Additionally, it is anticipated that genetic testing for hearing loss will increase in frequency in the future due to the advances in testing strategies and the implementation of newborn screening programs (Brunger et al. 2001).

Currently, genetic counseling for negative results of hearing loss genetic testing is not typically offered at CCHMC and may not be possible due to the high volume of negative results relative to the number of genetic counselors. Therefore, the needs and preferences in this population for educational materials should be explored (Brunger et al. 2001). Educational
materials have long been used as a supplement to genetic counseling and have been shown to improve patient knowledge and help relieve patient anxiety in genetic counseling and other healthcare settings (Stewart et al. 1993; Ormond et al. 1996; Glazier et al. 1997; Hallowell et al. 1998; Boundouki et al. 2004). Since genetic counseling and educational materials have been found to be beneficial in a variety of healthcare settings, we propose that our patient population will also benefit from the implementation of such services. An exploration of the perspectives, needs and preferences of the families as well as the current state of services provided by the healthcare providers (HCPs) at our institution is the logical first step in providing this population with appropriate counseling or educational materials.

The current study was conducted through a dual interview and survey approach. Qualitative telephone interviews were used to assess the parents’ recent experiences with the genetic testing for hearing loss services at CCHMC as well as their needs and preferences for the timing, content and format of genetic counseling services and educational materials. Quantitative surveys to assess the current services, perceived needs and provider preferences regarding genetic counseling services and educational material were given to the HCPs who are involved in hearing loss genetic testing at the same pediatric institution. This unique methodology was implemented to develop a comprehensive needs assessment of both the provider and the patient population. The purpose of this study is to characterize the needs and preferences of our patient population in order to lay the groundwork for future studies that will further the development of genetic counseling services and educational materials for the target population.
Methods

Parent Telephone Interviews

Participants were recruited for the study during the hearing loss genetic panel results phone call. One specific nurse at CCHMC makes phone calls to parents of children with negative genetic test results. The nurse read a script explaining the current study methods and offering parents the opportunity to participate in phone interview. Parents were eligible for inclusion in the study if they were given their child’s negative genetic test results during the recruitment period of September 1, 2009 to April 27, 2010. If parents agreed to participate, their contact information was recorded by the nurse and given to the primary investigator (PI). Parents were contacted for telephone interviews by the PI within two weeks of their agreement to participate and were excluded from the study if they could not be contacted within four weeks of recruitment.

The Parent Interview Guide (Appendix A) was developed by the research team as an assessment of the parents’ needs and preferences regarding the timing, content and format of material and services provided to them during the hearing loss genetic testing process at CCHMC. It is a structured interview of closed-ended and open-ended questions. The first two recruited participants were considered pilot interviews for the study. However, their data was retained in the final data analysis, as the content of the interview guide was not changed after the piloting interviews were completed.

Once contacted, parents were read an informed consent document. If participants agreed to participate, they were asked the questions listed in the Parent Interview Guide. A skip-pattern was followed based on the answers given to the interview questions. During the interviews, data were collected primarily through audio recording. Detailed written notes were also taken by the
PI to ensure data collection in the event that the recording failed. For those interviews successfully recorded, parents’ answers to the questions were transcribed by playback by the PI into a word document. The parents’ or their child’s identifying information was removed and a unique identifier was assigned to each interview during transcription to protect the privacy of the participant.

Interviews were analyzed using descriptive quantitative and qualitative techniques. Each question of the interview guide was coded separately. Proportions of responses to each question were reported descriptively. Coding was developed concurrently with data collection as patterns in the data became apparent. Themes and sub-themes were developed as patterns from the data emerged. Initial coding and theme development was performed by the PI. Subsequently, codes and themes were reviewed by a co-investigator to check for consistency and validity. Parent narratives were also pulled from the data to support the themes and sub-themes. Additionally, interview responses were compared qualitatively to the responses given in the quantitative provider surveys.

Provider Survey

Healthcare providers were recruited for participation if they were a medical geneticist, genetic counselor, or otolaryngologist at CCHMC during the recruitment period of September 1, 2009 and April 27, 2010. Thirty-five names and email addresses were obtained through the CCHMC directory website, at www.cincinnatichildrens.org. The recruited providers included 15 genetic counselors, 11 geneticists, and 9 otolaryngologists. Providers were recruited for participation through an email containing the web address link for the study survey. The provider survey was developed by the research team as a pilot survey to assess the current services, perceived need, and preferences of the healthcare providers that provide hearing loss genetic
testing services at CCHMC. The questions are predominantly closed-ended. The survey was
developed and distributed through Survey Monkey©. The questionnaire can be viewed in
Appendix B: Provider Survey Questions. Provider survey data was analyzed using descriptive
statistical methods. Responses to questions were reported in proportions. Additionally survey
responses were compared qualitatively to the responses given by parents in concurrent
qualitative telephone interviews.
Results

Parent Interviews

Twenty-six parents of the target population were recruited for participation in the study. Of those recruited, 16 completed telephone interviews (61.5%). Those who did not participate were not able to be contacted within the recruitment period or declined participation once contacted. Length of recorded interviews ranged from 7 minutes, 10 seconds to 20 minutes, 26 seconds, with an average length of 12 minutes, 44 seconds. Two interviews were not recorded due to failure of the recording device and were coded based on PI notes. Fourteen of sixteen parents interviewed were mothers of the child with hearing loss, and 2/16 were fathers. The sample of parents had children ranging from ages 6 months to 16 years, with a median age of 6 years. Five of sixteen participants had children diagnosed with hearing loss at birth, while 11/16 had children diagnosed in childhood with a median age of diagnosis of 4 years. All parents interviewed reported they had received negative test results from their child’s hearing loss genetic test panel offered at CCHMC within the past month.

In the following sections, three main themes that were identified during the parent interviews are identified and discussed. Within these themes, sub themes are also represented. Examples of quotes from the interviews that express these themes are also reported.

Theme #1: Parents seek a cause for their child’s hearing loss

Parent is uncomfortable with uncertainty about unknown cause

To elicit the parents’ feelings about the unknown cause of their child’s hearing loss, we asked parents to describe any explanation given to them by their HCP. Twelve out of sixteen parents report that no explanation has been given by their HCP regarding their child’s hearing
Parents indicated that the uncertainty of not knowing a cause for the hearing loss following the genetic test is confusing or unsettling.

“We were kind of dumbfounded thinking that, ‘Well her ear structure is normal, her nerves are normal, her genes are normal, then what is it?’” (Parent 4, Question 43)

“Right now I have two negatives: no issue with the CT no issues with the genetic test. Now I’m left with not knowing where to go next.” (Parent 10, Question 25)

Since the majority of parents in our sample had no known cause for their child’s hearing loss, parents were asked if they had any beliefs about the cause. Nearly half (7/15) of parents interviewed have their own beliefs about what has caused the hearing loss including non-genetic factors such as medications or illnesses.

“I think that the hearing loss might have been prevented if something was done about the ear infections she was having.” (Parent 4, Question 5)

“Oh I definitely think it was her prematurely and her NICU experience.” (Parent 16, Question 5)

**Parent satisfaction with HCP seeking cause**

We asked how participants found the pretest appointment helpful. Parents found the pretest appointment helpful due to its role in seeking a cause for hearing loss, confirming or ruling out a diagnosis or making plans with the physician for further care. Parents expressed intentions to pursue finding a cause for the hearing loss and expressed their satisfaction with the HCP who shared their desire to pursue a cause.

“It was helpful to know that they are trying to search for a reason. When you have a child who has hearing loss, you want to know why. So just knowing that the doctors are trying to do everything they can to figure out why and how to fix it was very helpful.” (Parent 4, Question 24)

“…[HCP said] ‘We really want to pursue this, we really want to find a cause especially because of her history.’” (Parent 8, Question 24)
Theme #2: Parents’ desire for more information about genetic test

Limited information prior to genetic test

To gain a better understanding of the information parents received before the genetic testing was ordered as well as how they felt about the amount and format of the information, we asked participants to describe the pretest appointment with the ordering HCP. Seven of 16 participants indicated that the information they received about the genetic test during the pretest appointment was limited.

“He [HCP] just told me to go downstairs to get his mouth swabbed.” (Parent 6, Question 8)

“It was a really short appointment. He [HCP] went over everything really briefly. He [HCP] said there were three tests to do and one was a genetic test, a blood test. Since her hearing loss was really mild, he [HCP] didn’t go much more into it. He [HCP] said it was common for results to be negative from the genetic test.” (Parent 7, Question 8)

“I don’t really recall him [HCP] telling me anything specific about the test because I thought she was going to end up getting blood work and it was just a swab. But no I don’t think we went over exactly what the testing entailed.” (Parent 11, Question 8)

Parents reported that some verbal information about the genetic test was provided at the pretest appointment. However, half (8/16) of the parents suggested that more information should be provided during the genetic testing process to better allow understanding. Some parents indicated the information about the hearing loss diagnosis and genetic testing that was provided was novel or surprising.

“This was also a big surprise to have heard the information that I did.” (Parent 1, Question 21)

“Especially when you are a new mom and this is something that blind-sides you and you are grasping to get some answers.” (Parent 10, Question 21)

“I was shocked to confirm that my daughter had hearing loss and it was like now we need to find out why.” (Parent 3, Question 21)
Parents have limited information about or didn’t understand the genetic test result

Parents were asked to describe the genetic test result given to them and asked if they felt well-informed about the results. Parents expressed that they had limited understanding of their child’s genetic test result and 7/16 said they were not well informed about the result.

“Well I don’t know what she is normal for, or what causes she doesn’t have. I couldn’t tell somebody that. So that would be my only complaint...Other than they were normal, I couldn’t explain what they actually tested for.” (Parent 16, Question 60)

“I guess everything was normal. Now what they mean they didn’t go into a great deal.” (Parent 10, Question 3)

“I wouldn’t say I am well informed. I don’t think I was well informed. If there was a grade, I would have given it a C.” (Parent 10, Question 3)

Theme #3: Parent’s Preferences Regarding Content, Format, and Timing of Received Information

Pretest written educational materials would have been helpful to better understand the genetic test

No parents were offered pretest educational materials in our sample (0/16). However, 9/15 parents would have liked educational materials at the pretest appointment and if offered at some point in the process, a majority of parents (13/16) felt educational materials would be best given before the genetic test was done. Additionally, parents felt pretest educational material would allow them to research or formulate questions for the HCP before the test was done.

“Sometimes it might be better to have stuff more in writing in front of you to read, later once the shock of everything wears off.” (Parent 16, Question 25)

“There was no real time to process any of it or think about it to think of any questions. Maybe if there is reading material or something like that offered, maybe the doctor can tell them to be like ok if you want a minute to talk this over or read this brochure and I will come back and answer any questions you have.
Parents were overwhelmed by the verbal information given at the pretest appointment

Parents were asked to describe the pretest appointment and the information provided at that time. Pretest verbal information was perceived by some parents as being overwhelming. Some parents felt this kept them from understanding the genetic test information. Three of 16 participants indicated that they were overwhelmed by the amount of verbal information they received during the pretest appointment. Participants indicated that they were unable to remember what was discussed regarding the genetic test because they were overwhelmed with information.

“Um, you know it is a lot to sit there and listen to all of this stuff and take it all in and depending on what point in the process you are.” (Parent 16, Question 25)

“I really guess I just feel like they just threw us into it.” (Parent 2, Question 67).

“So I was kind of in a state of getting a lot of information and they were saying a lot of things.” (Parent 1, Question 21)

“I think if we slowed down a bit and talked about everything he was telling me piece by piece it would be a little easier for me to take it all in.” (Parent 6, Question 25)

Preferences for a Genetics Appointment

Our sample of parents was asked if and when an appointment with a genetics professional would be helpful for them. Six out of sixteen parents said that an appointment with a genetics professional would be preferred only if there was a positive result. But half (8/16) parents said they would have liked an appointment with a genetics professional at some point, either before the test, after the result was given or at both times. However, of the other half of parents who did
Parents were asked their preferences for the content and format of pretest and posttest materials. Of those parents who wanted pretest educational materials (9/15), the content desired included information on the genes being tested, possible results and implications and any information about the test, and the purpose of the test. For those parents who desired posttest educational materials (7/16), information to be included was the genes that were tested, implications of the results, and a description of the test.

Parents in our sample find written information (14/16) and online/website format (10/16) the most useful for healthcare education. When asked to give suggestions to make the pretest appointment more helpful, half (8/16) of parents suggested to provide written information about the test. More than half of parents (9/16) reported they would find a DVD format to be a helpful format of educational material. Those who did not (7/16) said they prefer reading material or find a DVD inconvenient.

Provider Survey
Twenty-three of the thirty-five healthcare providers (HCPs) surveyed completed the online survey, yielding a response rate of 65.7%. The response rate for otolaryngologists was 7/9 (77.8%), for geneticists was 9/11 (81.8%) and for genetic counselors was 7/15 (46.67%). Of the
HCPs surveyed, 21/23 (91.3%) provide care or counseling for patients with sensorineural hearing loss. Of those who see this patient population, 18/21 (85.7%) order at least one genetic test per month.

Thirteen of twenty-one (61.9%) of the HCPs surveyed report having an appointment with patients before the genetic test is ordered. At the pre-test appointment, 12/13 (92.3%) do not offer educational material for the parents about genetic testing for hearing loss. When asked why they do not provide educational material, 7/12 (58.3%) answered “I do not think that educational material will be beneficial for these parents until they receive the results of the genetic test” while 3/12 (25%) chose “I do not have access to educational material for these parents” (Figure 1).

Of those who order genetic tests, 15/21 (71%) have an appointment with parents who have recently received a positive result, while 10/21 (47.6%) have an appointment with those who have a negative result. Eighteen of 21 (85.7%) report that they do not provide any educational materials to parents who have a negative test result, with the most commonly chosen reason being “I do not have access to educational material that is appropriate for these parents” (8/18, 44.4%).

Of the providers surveyed, 14/22 (63.6%) feel that parents of children with positive genetic test results are given adequate education about the recurrence risk and etiology of their child’s hearing loss, while 9/22 (40.9%) feel that way about parents with negative test results. Over half (54.5%) of the providers feel there is a need to provide more individualized genetic counseling or educational material to parents with a negative result, with a majority saying the best format for counseling would be a session with a genetics professional (15/22, 68.2%) and the best formats for educational material would be a booklet or pamphlet (10/22, 45.5%) or a
website to be accessed at home (9/22, 40.9%). No providers selected a DVD format to be watched at home as the best format for educational materials for this population (Figure 2).
Discussion

The purpose of the current study was to characterize the current services and educational materials provided to parents whose children have received negative results from hearing loss genetic testing as well as to assess the preferences of these parents about the importance of these services and materials. Through in-depth interviews of parents and a survey of the HCPs involved in their child’s care, we found that parents have a desire for more information about the genetic test ordered for their children and that modification of the current practices regarding provision of educational materials and genetic counseling for these parents may improve their understanding and satisfaction with the genetic testing process.

In this study, parents interviewed wanted more information about the genetic test being ordered for their child. Although all of these parents had a pretest appointment with the ordering physician, typically a pediatric otolaryngologist, the information at that appointment was not focused solely on the genetic test and much of the information provided about the test was not well understood by the parents. Some parents expressed they were overwhelmed by the amount of information provided at the pretest appointment. Others indicated that the information was novel or surprising to them, which may inhibit their understanding. Retention of information given in a healthcare setting has been shown to be consistently poor and stress or anxiety at the time of information giving has been found to be a factor in poor retention (Kessels 2003). More specifically regarding hearing loss, it has been shown in a previous study by Brunger et al. (2000) that the level of knowledge that parents have about the genetics and inheritance of hearing loss is poor and that parents who have been through the genetic testing process do not seem to have a much higher level of knowledge than those who have not. Our study in light of
other research elucidates some of the possible underlying factors that contribute to a lack of understanding and knowledge of genetic testing for hearing loss.

A majority of parents (9/16) said they would have wanted pretest educational materials and an even greater majority (13/16) felt that if educational material were provided, they would prefer it be given before the test to facilitate their understanding of the test and to formulate questions for the HCP prior to the test. Results of the concurrent survey of HCPs at our institution showed that a vast majority (92.3%) do not provide any pretest educational material for parents who are getting genetic testing for hearing loss. The prevailing reason (58.3%) for not providing this material is the HCP’s belief that it would not be beneficial for the parents. In the genetic counseling model of genetic testing education, pretest educational material is typically provided to allow for thorough understanding of the genes being tested, the possible results of the test and implications of those results. Educational material provided in a written form was overwhelmingly the most desired format by the parents interviewed, with internet websites being a close second. HCPs agreed with this finding, with a booklet/pamphlet (45.5%) or website (40.9%) being their most popular selections for the best format of educational materials. The content desired by the parents in the pretest information included what genetics were being tests, possible results and implications of the results, and description and purpose of the test.

None of the HCPs (0/22) felt that a DVD format for educational material would be the best format for the target population. However, when parents were asked about the usefulness of DVD about genetic testing for hearing loss, 9/15 said that format would be helpful to them. Video-based information has been shown to be as effective as written information in delivering genetic counseling concepts (Clayton et al. 1995) and can be a useful supplement to genetic
Parents in our sample cited several reasons a DVD format would be preferred over written materials including it being good for their kids to watch and enjoying the audio/visual learning. However, the reviews of a DVD format were mixed among our sample population, with some parents feeling that it would be inconvenient.

Half of the parents (8/16) in our sample population indicated that they would have liked to meet with a genetics professional to discuss the genetic test at some point in the process either before, after, or at both times. In a larger study sample of parents who had children who had genetic testing for hearing loss and were not offered genetic counseling, a similar proportion (51.5%) of parents was reported to have wanted genetic counseling during the testing process (Parker et al. 2000). In a qualitative analysis of focus groups by Withrow et al. (2008) in which parents were asked their preferences on the type of HCP and method provided for genetic counseling for hearing loss genetic testing, parents preferred appointment with an HCP with specific training in genetics. Data from this survey of HCPs shows that a majority (68.2%) of HCPs who are involved in genetic testing for hearing loss think that an appointment with a genetics professional would be the best format of genetic counseling for these patients. The results from this study show that parents and HCPs agree that an appointment with a genetics provider should be offered, yet in light of negative genetic test results, this does not typically occur.

Pretest and posttest genetic counseling for all patients having genetic testing for hearing loss is recommended by the American College of Medical Genetics in their 2002 Guidelines (ACMG 2002) and a proposed method for pretest and posttest genetic counseling for hearing loss genetic testing is provided in an article by Brunger, et al. (2001). A study by Palmer, et al. (2003) shows that pretest genetic counseling for hearing loss genetic testing can increase parental
knowledge and understanding of subsequently presented information. Genetic counseling provided for this population would not only focus on aspects of information given that are desired by the target population and providing informed consent for testing, but would address the psychological of the parents and patients regarding the genetic test and the implications of a negative test result. Parents in our sample population expressed uncertainty regarding the unknown cause of hearing loss in their child and previous inquiries into these issues have revealed feelings of guilt and trouble coping (Brunger et al. 2000; Dagan et al. 2002; Withrow et al. 2008; Withrow et al. 2009). The process of genetic counseling is a forum in which these feelings can be appropriately addressed (Weil 2000). Although genetic counseling may not be desired for all patients, the data indicates that many parents would like to be offered the service to facilitate their understanding and coping with their child’s test and the results.

**Limitations**

The methodology of parent interviews allows for an intimate knowledge of the sentiments, perspectives and preferences of our target population, which is crucial for the development of targeted services and educational materials. As with most qualitative research, this must be balanced with the limitations of sample size and generalizability of the data. Although the number of parents interviewed is small (n=16), the data collected from parents’ responses were consistent, allowing us to draw general conclusions about the sample of parents that may apply to parents at our institution with negative genetic test result. However, we do not have insight into the perspectives of those parents who denied their participation in the study, which may vary from our sample for a variety of reasons including dissatisfaction with services, contentment with services, or disinterest in further information about genetic testing for hearing loss. Additionally, lack of demographic data from the participants does not allow us to make
comparisons of our sample to the general parent population. It should be noted that the median age of diagnosis of hearing loss in our study population is 4 years, which varies markedly from the reference median age of diagnosis, even in populations excluding those diagnosed by newborn screening, of 25 months (Harrison et al. 2003). As a result of this discrepancy, parents in our sample may have a unique perspective on uncertainty, unknown cause, and desire for information due to the extended length of diagnosis. Additionally, the parent population sampled for this study was limited to a single institution, which detracts from the ability to generalize to other institutions which may provide this population with services or materials not offered at our institution.

Future Studies

Due to the exploratory nature of qualitative interviews such as these, this study opens the possibility for many avenues of future research. Our data undoubtedly shows that parents in our sample, and possibly the target population, desire more information about the genetic test offered at our institution. Using the qualitative data from our parent sample, a more powerful quantitative study of the target population’s specific preferences for educational materials and genetic counseling could be developed. This data could then in turn be used for the purpose of developing educational information or a model of genetic counseling for the target population. Studies could be done to evaluate the effectiveness and preference for provision of these educational materials and genetic counseling to best serve this patient population.

Conclusions

Our study has elucidated the perspectives of parents and providers involved in genetic testing for hearing loss at our institution. Although future studies will help to define the provisions of services and materials for this population, the data from this study can be translated
into suggestions for clinical practice for our target population. In general, the data shows that this patient population desires more information about the genetic test being done at our institution and that information is desired in the form of educational materials as well as genetic counseling.

Our data shows that although HCPs feel that pretest educational information would not be beneficial for this patient population, the parents in our sample think pretest educational materials would be helpful to them. Our study also shows parents’ preferences for how this educational material would be provided to them and what content they prefer to be included. Since parents in our sample have a pretest appointment with the ordering physician (typically an otolaryngologist), it may be most appropriate for the ordering physician to provide educational materials for this patient population at the pretest appointment.

A somewhat unexpected finding from our study was that HCPs and many parents in this population agree that genetic counseling services should be offered at some point in the genetic testing for hearing loss process. Although genetic counseling may not be desired or appropriate for all patients in this population, our data indicates the services if offered, may be well received and helpful for these parents. Since many parents in our population showed interest in pretest educational material and genetic counseling for hearing loss, it may be beneficial to develop pretest materials that include information on genetic counseling services and how to make an appointment with a genetics professional.
Bibliography


Figure 1: Providers’ reasons for not providing pretest educational materials to target population.
Figure 2: Providers’ selection of best format of educational material for the target population.

- Website to be accessed at home: 40.9%
- Booklet or pamphlet: 45%
- DVD to be watched at home: 0%
- Other: 14%
Appendix A: Parent Interview Guide

Introduction
1. Please tell me the ages of your children and indicate which of those children are deaf or hard of hearing.

2. At what age was your child’s hearing loss detected?

3. You recently received the results of a genetic test for hearing loss. Can you please describe to me what the results of that genetic test were?

4. What have your child’s physicians or other health care providers told you is or may be the cause of your child’s hearing loss?

5. What do you believe is of the cause of your child’s hearing loss?

Section A—Pre test appointment and educational materials received

6. What type of healthcare provider ordered the genetic test for your child’s hearing loss?

7. Did you meet with the [HCP] before the genetic test was ordered to talk about the genetic test?

___ Yes: 8. Please describe this appointment with the [HCP].

   **Probe:** What did the [HCP] tell you about the genetic test?
   Please, tell me more about what the [HCP] discussed with you during this appointment.

___ No:

9. Did you meet with any healthcare provider to discuss the genetic test before the testing was done?

___ Yes: 10. What provider did you meet with to discuss the genetic test?

___ No or other family member attended pre-test meeting: (Skip to Section B)

11. Please describe this appointment with the [HCP]. **Probe:** What did the [HCP] tell you about the genetic test? Please, tell me more about what the [HCP] discussed with you during this appointment.

12. When meeting with the [HCP] before the genetic test, was any educational material, such as a brochure, website, fact sheet or video about the genetic test offered to you?

___ Yes:

13. What type of material did you get?

14. Did you read/use this material?

___ Yes:

15. What did you think of this [material]?

16. How was the [material] helpful to you?

17. What information do you think was lacking in this [material]?
18. Do you think that the information in the [material] applied to you or your family at that time? **Probe:** Please explain why or why not it was applicable.

___ No:

19. What do you recall was the reason for not reading the [material]?

20. What do you think could be changed about the [material] that would make you or someone else more likely to read/use it?

___ No or I don’t know:

21. Would you have wanted some form of education material (pamphlet, brochure, website etc.) about the genetic test at that time?

___ Yes: 22. What kind of information would you prefer to be included in that material?

___ No: 23. What reasons would you have to not want the educational information at that time?

24. In what ways was this appointment with the [HCP] before the genetic test helpful for you? **Probe:** Please explain those reasons in more detail.

25. In what ways could the appointment with the [HCP] be changed to make it more helpful for you? **Probe:** Please explain those reasons in more detail.

**Section B—Post-test appointment and educational material given after results**

26. You were informed of your child’s genetic test result through a phone call by one of the nurses in the Ear and Hearing Clinic. Was this way of giving the results by phone appropriate for you?

___ Yes

___ No:

27. How would you have preferred to receive those results?

28. Were you aware that the results of the genetic test would be given to you in a phone call?

29. During the results phone conversation with the nurse, did she recommend that you make an appointment with a healthcare provider to discuss the genetic test results?

___ Yes:

30. What type of provider did the nurse recommended you to meet with?

31. Did you follow up with the [HCP]?

___ Yes: 32. Please explain what you discussed during that appointment with the [HCP]. **Probe:** What was helpful during this appointment? What was not helpful during this appointment?

___ No: 33. Please explain what reasons you had for not following up with the appointment that the nurse recommended.

___ No or I don’t know:

34. Would you have liked to have been referred to talk to a healthcare provider at that
time to talk about the genetic test results?
___ Yes:

35. What type of provider would you have liked to meet with?

36. What are some reasons you would have liked to have been referred to meet with the [HCP]?

37. What questions would you have had for that [HCP] at the time?

___ No: 38. What are some reasons you would not have liked to have been referred to a healthcare provider to talk about the genetic test results?

39. After you learned the result of the genetic test, was any educational material, such as a brochure, website, fact sheet or video about the genetic test result offered to you?
___ Yes:

40. Did you read/use this material?
___ Yes:

41. What did you think of this [material]?

42. How was the [material] helpful to you?

43. In what ways could the [material] be improved?

44. In what ways did the information in the [material] apply to you or your family?

45. In what ways did the information in the [material] not apply to you or your family?

___ No:

46. What do you recall was the reason for not reading the [material]?

47. What do you think could be changed about the [material] that would make you or someone else more likely to read/use it?

___ No or I don’t know:

48. Would you have wanted some form of education material (pamphlet, brochure etc.) about the result of the genetic test at that time?

___ Yes:

49. What kind of information would you prefer to be included in that material?

50. What questions would you have like to have had answered at the time?

___ No: 51. What reasons would you have to not want the educational information at that time?

52. After learning the result of the genetic test, did you seek out information about the genetic test results from sources outside of the healthcare providers and the information they provided during medical appointments?

___ Yes: 53. Did you find information about genetic testing for hearing loss that was helpful for you?
___Yes:

54. Where did you find helpful information about the genetic test and the results?

55. What was the format of material that you found to be helpful?

56. What information in that material was most helpful for you?

___No:

57. What was the format of the information you found?

58. For what reason(s) was the material not helpful?

___No. 59. Do you plan to seek out more information about genetic testing for hearing loss or the result of your child’s genetic test?

60. In general, did you feel like you were well informed by the [HCP] or any material that was provided for you about the genetic test results you received?

___Yes:

61. What information provided to was the most useful for you in understanding the information about your child’s genetic test result?

62. What provider appointment or service was most useful for you in understanding information about your child’s genetic test result?

___No:

63. What information do you feel was lacking during the genetic testing process that would have been helpful for you?

64. What services or appointments do you feel was lacking during the genetic testing process that would have been helpful for you?

Section C—Preferences for material format, timing, and content

65. What format of educational material about healthcare is generally the most useful for you? Probe: Please describe why that format is best for you.

66. What are your thoughts on the usefulness of educational material about genetic testing for hearing loss provided in a DVD or video format? Probe: In what ways would this format of material be useful for you?

67. At what point in the genetic testing for hearing loss process do you think would be the best time to provide parents with some type of educational material?

68. At what point in the genetic testing for hearing loss process do you think it would be most helpful to provide parents with an appointment with a genetic counselor or other health professional to discuss the genetic test?

69. In the future, what services or materials can healthcare professionals can provide for parents to help them better understand the results of their child’s genetic test for hearing loss?
Appendix B: Provider Survey Questions

This survey is to be completed by medical professionals who are involved in genetic testing services as genetic counselors or medical geneticists or are involved in hearing loss services as otolaryngologists at Cincinnati Children’s Hospital Medical Center (CCHMC).

Please read and answer the following questions about the educational information provided for parents who have a child undergoing genetic testing for hearing loss at CCHMC. This survey will take approximately 5 minutes to complete.

1. What is your professional title?
   - Otolaryngologist
   - Medical Geneticist
   - Genetic Counselor

2. Approximately how many patients with sensorineural hearing loss do you provide care or counseling for per month?
   - I don’t provide care or counseling for patients with hearing loss (skip to Section D)
   - Less than 10
   - Between 10-20
   - 21 or more

3. Approximately how many genetic tests for hearing loss do you order per month?
   - I don’t order genetic tests for hearing loss (skip to section A)
   - Five or fewer
   - Between 5 and 10
   - 11 or more

4. Of the patients for whom you order genetic testing for hearing loss, approximately what portion have negative results, yielding no genetic cause that can be determined for their hearing loss?
   - Almost none
   - Approximately one quarter
   - Approximately half
   - Approximately three quarters
   - Nearly all

Section A—Pre test appointment and educational materials received

5. Do you typically meet with parents of patients who are going to undergo hearing loss genetic testing before the test is ordered?
   - Yes
   - No (skip to section B)

6. During this pretest appointment, please indicate which topics are discussed in depth with parents/patients. Check all that apply.
   - Possibility of genetic etiology of hearing loss
   - What the genetic test for hearing loss is testing for
   - Inheritance patterns of hearing loss
   - Meaning of positive results with regard to recurrence risk of hearing loss
Meaning of negative results with regard to recurrence risk of hearing loss
Implications of result on management strategy
Other, please specify _____________________________________

7. During the pretest appointment do you provide educational material for the parents about genetic testing for hearing loss?
   Yes (Answer 7a and 7b)
   No (Answer 7c)

7a. Please indicate the format of the educational material you provide for parents in the pre-genetic test session. Check all that apply.
   Internet website
   Book
   Academic journal or articles
   DVD or computer program
   Booklet, pamphlet or fact sheet
   Other: ______________________________

7b. Please indicate what topics are covered in the educational material you provide for parents in the pre-genetic test appointment. Check all that apply.
   Possibility of genetic etiology of hearing loss
   What the genetic test for hearing loss is testing for
   Inheritance patterns of hearing loss
   Meaning of positive results with regard to recurrence risk of hearing loss
   Meaning of negative results with regard to recurrence risk of hearing loss
   Implications of result on management strategy
   I don’t know
   Other, please specify _________________________________

7c. Please indicate what reasons you have for not providing educational material to parents during the pre-genetic test session. Check all that apply.
   Parents do not show interest in learning more about the genetic test that is being ordered
   I do not have access to educational material for these parents
   I do not think that educational material will be beneficial for these parents until they receive the results of the genetic test
   I am not satisfied with the quality, content or format of the educational material I have access to and therefore do not offer this material to parents
   Other: _________________________________

Section B—Post test educational material given

8. Do you typically have a post-test appointment with parents who have recently received a positive genetic test result of their child’s genetic testing for hearing loss?
   Yes
   No
9. Do you typically have a post-test appointment with parents who have recently received a negative genetic test result of their child’s genetic testing for hearing loss?

   Yes (Answer 9a, then move on to Question 10)
   No (Answer 9b, then move on to Question 10)

9a. During the post-test appointment, please indicate which topics are discussed in depth with parents/patients who have a negative genetic test result. Check all that apply.

   Possibility of genetic etiology of hearing loss
   What the genetic test for hearing loss is testing for
   Inheritance patterns of hearing loss
   Meaning of negative results with regard to recurrence risk of hearing loss
   Implications of result on management strategy
   Other, please specify ____________________________

9b. What is your best explanation for the reason you do not have a posttest appointment with these parents?

   Parents are referred to a post-test appointment but don’t show or cancel
   Parents are not referred to a post-test appointment because we do not see a need to have a post-test appointment
   I refer these parents to another healthcare professional for post-test appointments
   Other, please specify: ____________________________

10. Do you provide educational material of any kind to the parents whose children have a negative result of genetic testing after the result is received?

    Yes (Go to Section C)
    No (Answer 10a and move on to Section D)

10a. Please indicate what reasons you have for not providing educational material to parents after they receive a negative test result from their child’s genetic test. Please check all that apply.

    Parents do not show interest in learning more about the genetic test or the results of the genetic test
    I do not have access to educational material that is appropriate for these parents
    I do not think that educational material will be beneficial for these parents
    I am not satisfied with the quality, content or format of the educational material I have access to and therefore do not offer this material to parents
    I don’t meet with these parents, and therefore don’t provide them with educational materials
    Other: ____________________________

Section C—Format, timing, and content of material provided
Please answer the following questions only for materials provided to parents after their child has a negative test result from genetic testing for hearing loss. If you give the same material regardless of test result please answer the following questions about that material.
11. Please indicate what the format is of the educational material you provide parents after their child has a negative test result. Check all that apply:

- Internet website
- Book
- Academic journal or articles
- DVD or computer program
- Booklet, pamphlet or fact sheet
- Other: ______________________________

12. How is this information given to the parents?

- Email
- Mail
- Phone
- In person at post-test appointment
- Other ______________________________

13. How long after the parents test result is this information given or sent?

- Same day as results are given
- Within a week of results
- Within a month
- It varies from family to family

14. Is the information provided for the parents targeted towards negative test results for hearing loss genetic testing?

- Yes
- No
- I don’t know

15. Please indicate which topics are included in the information you provide to these families. Check all that apply.

- Possibility of genetic etiology of hearing loss
- What the genetic test for hearing loss is testing for
- Inheritance patterns of hearing loss
- Meaning of positive results with regard to recurrence risk of hearing loss
- Meaning of negative results with regard to recurrence risk of hearing loss
- Implications of test result on management strategy
- I don’t know
- Other, please specify ______________________________

Section D—Usefulness of DVD or video counseling program

16. Do you believe that parents whose child has a positive test result from hearing loss genetic testing at CCHMC are given adequate educational information about the recurrent risk and etiology of their child’s hearing loss?

- Yes
- No
- I don’t know
17. Do you believe that parents whose child has a **negative** test result from hearing loss genetic testing at CCHMC are given adequate educational information about the residual recurrence risks and possible etiologies of their child’s hearing loss?

   Yes  
   No  
   I don’t know

18. Do you believe that parents whose child has a **negative** test result from hearing loss genetic testing at CCHMC are given adequate genetic counseling about the residual recurrence risks and possible etiologies of their child’s hearing loss?

   Yes  
   No  
   I don’t know

19. Do you think that there is a need to provide more individualized genetic counseling and/or educational material for parents of children who have idiopathic hearing loss after **negative** genetic testing?

   Yes  
   No  
   I don’t know

20. If more individualized genetic counseling were provided for parents whose child has a negative genetic test result for hearing loss, what do you think would be the best format of this counseling? Please select two answers.

   - Counseling session with genetics professional (genetic counselor or geneticist)  
   - Counseling session with other health professional  
   - Phone conversation with genetics professional (genetic counselor or geneticist)  
   - Phone conversation with other health professional  
   - Website to be accessed at home  
   - DVD to be watched at home

21. If more individualized genetic counseling were provided for parents whose child has a negative genetic test result for hearing loss, what do you think would be the most practical format of this counseling in your practice?

   - Counseling session with genetics professional (genetic counselor or geneticist)  
   - Counseling session with other health professional  
   - Phone conversation with genetics professional (genetic counselor or geneticist)  
   - Phone conversation with other health professional  
   - Website to be accessed at home  
   - DVD to be watched at home

22. If more individualized educational material were provided for parents whose child has a negative genetic test result for hearing loss, what do you think would be the best format for this material?

   - Booklet or pamphlet  
   - Website to be accessed at home  
   - DVD to be watched at home  
   - Other