Improving Access to Genetic Counseling Visual Aids for People with Low Vision

Master’s Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences
Brandeis University
Graduate Program in Genetic Counseling
Gretchen Schneider, M.S. LCGC, Advisor

In Partial Fulfillment
of the Requirements for the Degree

Master of Science
in
Genetic Counseling

by
Lindsay Smith

May 2018
Acknowledgments

I would like to thank my thesis Primary Investigator, Gretchen Schneider MS, CGC for the time, effort, and knowledge that she generously provided to this project. I also want to express gratitude to the rest of my committee Emily Place MS, LCGC and Carly Murphy MS, CGC for their support and insight as well as Jared Kitchen from the Carroll Center for the Blind for his unique insights and advice. Additionally, I would like to thank Margarita Corral for reviewing my survey and assisting with data analysis. Thank you to Judith Tsipis, Gayun Chan-Smutko, Cassandra Buck, Missy Goldberg and the entire Brandeis Genetic counseling program for their help and support during this project and my time in the Genetic Counseling Program. Lastly, I would like to thank my classmates, friends, and family for their unwavering encouragement and support.
ABSTRACT

Improving Access to Genetic Counseling Visual Aids for People with Low Vision

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Lindsay Smith

Healthcare disparities occur for individuals with a variety of different disabilities including people with low vision. One of the major obstacles individuals with low vision face when accessing healthcare is limited availability of visually accessible information, which impedes comprehension and affects communication between patients and providers. This can be particularly challenging for genetics providers who often rely on visual aids to help patients understand complex genetic information. Several existing visual aids were modified to make them easier for people with low vision to see, and utilized on an iPad while talking to participants during genetic counseling sessions in an inherited retinal disease clinic. Participants were surveyed after their genetic counseling session to assess if the visual aids were visible, and to investigate the effect they had on comprehension, rapport with the genetic counselor, and informed decision making. Twelve participant surveys were collected, and overall, the participants had a positive reaction to the use of the modified visual aids. On a likert scale of 5, the average participant rating for ‘recommending the routine use of these visual aids in the clinic’ was 4.75. Additionally, the average rating of how visible the images were was 3.75 on a 4 point scale. All participants responded to the survey questions with either neutral or positive
opinions regarding the use of modified visual aids in the clinic. Two participants answered a follow up survey which provided demographic and disease classification information. Given the overall positive response observed in this study, the expanded use of modified visual aids for genetics education may be considered for patients with low vision.

**Keywords:** Low Vision, Visual Aids, Education, Genetic Counseling, Vision Loss, Images
TABLE OF CONTENTS

Acknowledgements........................................................................................................iii

Abstract.........................................................................................................................iv

List of Tables................................................................................................................viii

Introduction...................................................................................................................1

Methods.........................................................................................................................4

   Study Design.............................................................................................................4

   Participants...............................................................................................................4

   Image Design..........................................................................................................5

   Data Collection.......................................................................................................6

   Data Analysis.........................................................................................................7

Results............................................................................................................................8

   Demographics and Disease Information.................................................................8

   Short In-Clinic Survey Responses............................................................................9

   Follow-Up Survey Responses................................................................................10

Discussion....................................................................................................................12

   Visibility................................................................................................................12

   Comprehension.....................................................................................................13
Rapport..................................................................................................................14

Patient Autonomy....................................................................................................15

Healthcare Accessibility..........................................................................................15

Genetic Counseling Implications.............................................................................16

Limitations................................................................................................................16

Future Directions.....................................................................................................17

Conclusion...............................................................................................................19

References...............................................................................................................20

Appendices.............................................................................................................21

  Appendix A: Consent Form.....................................................................................21

  Appendix B: Recruitment Notice............................................................................27

  Appendix C: Study Instrument.............................................................................29

  Appendix D: Images.............................................................................................35
LIST OF TABLES

Table 1: Demographics and Classification Information- Follow-up Survey..........................8

Table 2: Short In-Clinic Survey Responses.................................................................9

Table 3: Follow-up Survey Responses.................................................................11
Introduction

There are an estimated 285 million individuals in the world who are either blind or visually impaired, and many people in this group frequently face several obstacles when trying to access healthcare (World Health Organization, 2012). These include difficulties traveling to and navigating around the clinic, being unable to read written materials provided, communication with medical staff, and lack of autonomy. Not only do these issues have a negative effect on patients’ overall healthcare experience, but the quality of care can suffer as well. (Cuppes, 2012). In a British study of visually impaired patients’ recollection of their clinic visits, 11% of patients did not know the name of their diagnosis, and 17% did not receive any non-medical information such as information about low vision tools and technology, leaflets about visual impairment, contact information for social services, or national registration for services from their healthcare team. Those who reported receiving non-medical information and support were much more satisfied than the group of people that did not (Douglas et. al, 2010). It is clear that there is a gap in the quality of healthcare given to people who are blind or vision impaired, and that accessibility adjustments must be made to reduce this disparity.

The medical community is often criticized for viewing individuals with disabilities through the medical model, focusing primarily on the physical impairment and deficiency. This frame of thinking encourages progress and treatments to minimize the physical impairment, but fails to incorporate the social and emotional aspects of a person with disabilities. In 2001, the World Health Organization approved a model called the International Classification of
Functioning Disability and Health, or ICF, which recognizes physical impairment and biological limitations, while still encouraging participation, environmental adaptations, and social aspects of disability. The advent of this paradigm led to increased research into healthcare disparities and access issues for people with disabilities. Sharby, et.al (2015) outlines several studies which indicate that adopting this model, and being conscious of the disparities that exist for people with disabilities, can improve the quality of their healthcare.

Several studies have looked specifically at people with vision impairment and the obstacles they face when accessing health care. Using the social and ICF models of disability as a foundation, ODay et. al. (2004) conducted a focus group that included people with low vision and blindness. The analysis of these interviews identified four major areas in which people with blindness and low vision face obstacles in accessing healthcare: basic respect, communication, physical access, and giving and receiving information. The Royal National Institute of Blind People published a study confirming many of the same themes, specifically the lack of accessible reference materials (Sibley, 2009). While the study found that 97% of individuals with vision loss agreed that blind and partially sighted people have a right to receive health information in a format that they can read themselves, 72% of the individuals surveyed could not read the personal health information given to them by their General Practitioners. This research further supports that the accessibility of health information for people with low vision is a significant area of disparity for these individuals.

With growing understanding of the genetic causes for vision loss, and education being one of the major areas of disparity for patients with low vision, genetic counselors are challenged with determining how they can provide the best care to their patients (Consugar et. al, 2015). However, there is very limited research regarding the most effective way for genetic counselors
to educate patients with low vision. Genetic counselors commonly use visual aids to facilitate patient learning, but traditional aids may not be accessible to patients with low vision and no adapted aids exist for this population. This study adapted visual aids to be more accessible to patients with low vision, and utilized them during actual genetic counseling sessions to determine their usefulness. A survey was used to assess if the images were visible, and if they affected participant comprehension, rapport with the genetic counselor, and informed decision making. If visual aids can be effectively altered so that they are easily visible to this group of patients, this education tool can become more widely available in this patient population.
Methods

Study Design:

This was an exploratory quantitative study assessing the effectiveness of adapted genetic counseling visual aids. The Massachusetts Eye and Ear Institutional Review Board approved this study through the expedited process. The Brandeis University Institutional Review Board was also consulted and agreed to allow this research to be conducted under the supervision of the Massachusetts Eye and Ear Institutional Review Board.

Participants:

The Inclusion criteria were that participants had to: have an inherited retinal disease; receive genetic counseling at Massachusetts Eye and Ear (MEE); be eighteen years or older; and speak, read, and write English. Patients who had visual acuity at or below counting fingers were excluded due to the likelihood that they would not be able to see the visual aids. We recruited participants in the MEE Inherited Retinal Disease Service at the beginning of their genetic counseling session. Informed consent was obtained at the start of the session, and the images were then used throughout the genetic counseling session to help explain genetic concepts. At the conclusion of the genetic counseling session, participants were given a 6 question survey on an iPad during their check-out procedure. At the end of this initial survey, participants were asked if they would like to complete a longer follow-up survey and, if they agreed to participate, they provided an email address to which an invitation to participate in the follow-up survey was
sent. Participants who provided their email addresses for the follow-up survey were entered into a drawing for one of two $25 gift cards.

Image Design:

The images were designed and modified using the GNU Image Manipulation Program (GIMP). Several design elements were taken into consideration when modifying and creating the visual aids. Highly contrasting colors, including black and white and black and yellow were selected when designing the images, as this is recommended to improve visibility for individuals with low vision (Gwyn, 2014). Light yellow was used in some images in order to reduce the glare that is often seen with pure white colors (Williams, 2012). Each image was designed with four different color combinations to allow patients to select the most visible option depending on their individual needs. These color combinations were as follows: black text/white background, white text/black background, black text/yellow background, and yellow text/black background. The icons used were simple, had thick bold lines, and were placed with plenty of blank uncluttered space between each icon.

The text in the visual aids was in large print at the recommended size of 18 pt. font or larger (Sutton, 2002). The American Printing House for the Blind created a font named APHont in 2004 that is designed specifically to be more comfortable for individuals with low vision. It has elements such as increased spacing between letters, large t and x heights, specially designed letter shapes, and fuller punctuation marks to promote easier visibility (American Printing House for the Blind). This was the font used for all text in the images.

Several of the images were adapted from the Greenwood Genetics Center (2013) electronic counseling aids. These included Autosomal Dominant, Autosomal Recessive, X-linked recessive, and the Chromosome to Gene to Protein images. An image of a sub-retinal
injection from the Genetics & Genomics Wiki was modified to portray this procedure. Other images were created and designed to illustrate the following concepts: DNA sequencing, gene therapy, USH2A being too large to fit within a viral vector, and stem cell therapy.

Data Collection:

Two online surveys through Qualtrics were used to collect data. The initial survey was 6 questions, and the follow-up survey was 18 questions. There were five major categories of questions asked throughout the surveys.

1. Accessibility: These questions assessed how well the adaptations improved the accessibility of the images for participants.

2. Comprehension: These questions evaluated the participant’s confidence with the information provided in the session, and if they believe the images played a role in their comprehension.

3. Rapport: These questions inquired if the visual aids affected the participant’s rapport with the genetic counselor or had an impact on their experience at the clinic.

4. Autonomy: These questions explored if the visual aids had an effect on the participant’s sense of autonomy and allowed them to feel more active in their health care.

5. Demographics and Disease Classification: The questions asked about level of vision loss, genetic diagnosis, and other general demographic information.

The initial survey was primarily composed of Likert scale questions to concisely cover the four main themes of the study. It did not include any demographic information. The Follow-up survey included a mixture of Likert scale, multiple choice, and open ended questions. It inquired about all five categories including demographic and disease classification information.
The Qualtrics Product Accessibility Template was utilized to ensure the Qualtrics survey was compatible with low vision devices and most visible for participants.

Data was collected from December 4, 2017-March 9, 2018.

**Data Analysis:**

The statistical analysis of this data was descriptive in nature. The Likert scale questions were converted to a numerical scale, and averages were taken for each of the short in-clinic survey responses. The standard deviation was also calculated for each question. The follow-up survey responses were described in detail, and linked to their responses to the short-in clinic survey. The two sets of data points were also compared and contrasted for interesting themes and observations.
Results

Demographics and Disease Information:

Twelve individuals seen in the Inherited Retinal Disease clinic, who were eligible to take part in the study, agreed to participate in the study. From these participants, we received a total of 12 responses to the short in-clinic survey, and 2 responses to the follow-up email survey. Four participants, (33%) expressed interest in completing the follow-up survey, and offered their email address. Of the follow-up survey links that were sent out, there was a 50% response rate or 2 participants. No responses were excluded from either survey as all participants answered nearly all of the questions asked. A description of the demographic and disease classification information for the 2 follow-up survey participants is shown in Table 1.

Table 1: Demographics and Classification Information- Follow-up Survey

<table>
<thead>
<tr>
<th>N</th>
<th>Sex</th>
<th>Age</th>
<th>Education</th>
<th>Vision loss</th>
<th>Diagnosis</th>
<th>Genetic diagnosis?</th>
<th>When they lost vision</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>F</td>
<td>36</td>
<td>Professional Degree</td>
<td>Moderate</td>
<td>Retinitis Pigmentosa</td>
<td>Yes</td>
<td>Since Birth</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>53</td>
<td>4 Year Degree</td>
<td>Mild</td>
<td>Macular Degeneration</td>
<td>Unsure</td>
<td>2 years ago</td>
</tr>
</tbody>
</table>

Because the short in-clinic survey did not include demographic questions and there was a small number of responses for the follow-up survey, it is not possible to know if the survey responses collected in this study are representative of the overall patient population of the Massachusetts Eye and Ear Inherited Retinal Disorder Service.
**Short In-Clinic Survey Responses:**

The questions in the short in-clinic survey were in Likert scale format and the responses to each were converted to numerical scales for the purpose of analysis. Question one had a scale from 1 to 4 with 1 being not able to see the images at all, and 4 being able to see the images very well. The other five questions had Likert scales ranging from 1-5. The most positive selection was a 5, and the most negative selection was a 1. After scoring the responses using the numerical scales, averages were taken for each question. Higher averages indicated a more positive overall response, while lower averages indicated a more negative response. The averages for each question are shown in Table 2.

**Table 2: Short In-Clinic Survey Responses**

<table>
<thead>
<tr>
<th>Question</th>
<th>N</th>
<th>Minimum</th>
<th>Maximum</th>
<th>Mean</th>
<th>Std. Deviation</th>
</tr>
</thead>
<tbody>
<tr>
<td>How well were you able to see the images during your genetic counseling session?</td>
<td>12</td>
<td>2</td>
<td>4</td>
<td>3.75</td>
<td>.622</td>
</tr>
<tr>
<td>How would you rate your understanding of the information provided during your genetic counseling session today?</td>
<td>12</td>
<td>4</td>
<td>6</td>
<td>4.92</td>
<td>.289</td>
</tr>
<tr>
<td>How did these images affect your understanding of the information provided in your genetic counseling session today?</td>
<td>12</td>
<td>3</td>
<td>5</td>
<td>4.58</td>
<td>.689</td>
</tr>
<tr>
<td>How did the availability of these images affect the relationship you feel with your genetic counselor?</td>
<td>12</td>
<td>3</td>
<td>5</td>
<td>4.42</td>
<td>.900</td>
</tr>
<tr>
<td>How did the availability of these images affect your ability to make informed decisions about your healthcare?</td>
<td>12</td>
<td>3</td>
<td>5</td>
<td>4.26</td>
<td>.866</td>
</tr>
<tr>
<td>Overall, would you recommend the routine use of these visual aids in genetic counseling sessions at the Massachusetts Eye and Ear Infirmary?</td>
<td>12</td>
<td>3</td>
<td>5</td>
<td>4.76</td>
<td>.622</td>
</tr>
</tbody>
</table>

| Valid N (listwise) | 12 |
Follow-Up Survey Responses:

The follow-up survey explored the participant’s experience with the images in more detail. Analysis of this data was performed by comparing and contrasting the responses from both participants to look for similarities and differences in their opinions. Side by side comparison of responses for each question is shown in Table 3.
Table 3: Follow-up Survey Responses

The final 3 questions listed in the table are these participants’ responses from the short in-clinic survey.

<table>
<thead>
<tr>
<th>Question</th>
<th>1</th>
<th>2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Please select any of the images you recall from your genetic counseling session.</td>
<td>AD, AR, XLR</td>
<td>AD, AR, XLR</td>
</tr>
<tr>
<td>Please Select the Color Combination(s) you recall from your genetic counseling session.</td>
<td>Black text, yellow background</td>
<td>Yellow text, black background</td>
</tr>
<tr>
<td>Which element(s) of the image made it the most visible for you?</td>
<td>Color/Contrast Bold Simple Icons</td>
<td>Color/Contrast Bold Simple Icons*</td>
</tr>
<tr>
<td>Did these images make you feel more confident in describing this information to other family members, friends?</td>
<td>Neither Confident nor Unconfident</td>
<td>Much More confident</td>
</tr>
<tr>
<td>Have you met with a genetic counselor at Mass Eye and Ear before?</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>How often do you face obstacles with written or visual health materials/letters, etc.?</td>
<td>Seldom</td>
<td>Seldom</td>
</tr>
<tr>
<td>How often do healthcare offices offer written materials in accessible format such as large print/audio/braille?</td>
<td>Seldom</td>
<td>Seldom</td>
</tr>
<tr>
<td>How did these images affect your understanding of the information provided in your genetic counseling session today?</td>
<td>Somewhat Better</td>
<td>Much Better</td>
</tr>
<tr>
<td>How did the availability of these images affect your ability to make informed decisions about your healthcare?</td>
<td>About the Same</td>
<td>Much Better</td>
</tr>
<tr>
<td>Overall, would you recommend the routine use of these visual aids in genetic counseling sessions at the Massachusetts Ear and Eye Infirmary?</td>
<td>I Would Recommend</td>
<td>I Would Highly Recommend</td>
</tr>
</tbody>
</table>

* Indicates this participant selected this option as the most helpful
**Discussion**

We used modified visual aids in the genetic counseling sessions of 12 patients, and surveyed them at the end of their visits to determine if the participants felt the images had an impact on their genetic counseling session. The aim of these surveys was to assess the accessibility of these adaptations, comprehension of the genetic information in these visual aids, the effect of these images on rapport, and the impact of these images on the participants’ sense of autonomy. The follow-up survey further explored these elements as well as gathered demographic and disease classification data. Due to a limited sample size available to survey, no statistically significant results were able to be reported, however the data did provide valuable insight into the potential benefits of visual aids for individuals with low vision during genetic counseling sessions.

*Visibility:*

Overall most participants reported that they were able to see the adapted images very well. Participants were selected based on having less severe vision loss, however all participants did have some amount of vision loss. Many were also dilated from their exam during their genetic counseling session, which further reduced their vision. The average of 3.75 on a 4 point Likert scale is a good indication that these images were visible for this study population with low vision. In the follow-up survey both participants indicated that the features that improved visibility the most were ‘Color/Contrast’ and ‘Bold Simple Icons’. While there is not enough
data to make conclusions, these responses point towards these two features being key elements in adapting visual aids for people with low vision. Interestingly, both participants indicated color and contrast were helpful, but recalled viewing two different color combinations. One participant reported seeing images with black text and a yellow background, while the other reported yellow text and black background. This data supports the concept of having images using different color contrasts available to provide individualized options.

Comprehension:

Participants reported a very high level of comprehension, with the question “How would you rate your understanding of the information provided during your genetic counseling session today?” receiving the highest Likert scale score in the survey. The responses to this question also had the least amount of variability in answers with a standard deviation of 0.289. When asked how the images affected their understanding of the information provided during their genetic counseling session, the average Likert scale score was slightly lower at 4.58. While this score is lower than the score for comprehension, a majority of participants reported the images caused their comprehension to be ‘somewhat better’ or ‘much better.’ It seems that, for this sample of people, the images did help with comprehension, but were not the only contributing factor. This is to be expected since the use of visual aids is only one element influencing patient understanding in a genetic counseling session. For example, patients who come in with prior knowledge about genetics may have better comprehension that is due more to prior experience than the visual aids. It is also possible that the genetic counselor’s effectiveness in teaching genetic concepts could have contributed to their level of comprehension.
Despite the overall high comprehension scores, the two participants in the follow-up survey had different responses when asked if the images affected their confidence in the ability to explain this information to other family members, friends, etc. The woman who first experienced vision loss at birth, has a professional degree, and an established genetic diagnosis selected ‘neither confident nor unconfident’, implying that her confidence level did not change with the use of the adapted visual aids. In contrast, the man who started having vision loss 2 years ago, has a bachelor’s degree, and does not have an established genetic diagnosis reported that the images made him ‘much more confident’. This discrepancy is not surprising given that an individual’s familiarity with their condition, the presence of a known diagnosis, and higher level of education may all contribute to both a better understanding and higher comfort level explaining information to others. This finding, while based on limited data, suggests that adapted visual aids may be particularly useful for individuals who have recently begun to experience vision loss and/or have no known underlying genetic diagnosis.

*Rapport:*

Study participants felt that the use of adapted visual aids had a positive effect on their rapport with the genetic counselor, with the average score for the question “How did the availability of these images affect your relationship with your genetic counselor?” being 4.42. Most participants (75%) felt that the images made their relationship ‘somewhat better’ or ‘much better’ while no participants reported any negative impact on rapport. There are many elements that contribute to patient-counselor rapport, so it is not surprising that some patients found the images to improve rapport while others felt neutrally. However, the fact that there were no participants who felt these images had a negative impact suggests that the use of adapted visual aids will cause more good than harm to the patient-counselor relationship.
Patient Autonomy:

Patient Autonomy was another important element assessed with this survey. When asked if the use of these images affected their “ability to make informed decisions about (their) health care”, the average Likert scale score was 4.25. The majority (75%) of participants reported the images made their ability to make these decisions ‘somewhat better’ or ‘much better.’ The remaining 25% had a neutral opinion. Of the three participants who reported neutral feelings, one also reported that the images did not affect their comprehension. The other two had scores of 4 (somewhat better) in regards to the impact of the images on their comprehension. Perhaps participants who did not find the images improved their comprehension also did not find the images to impact their decision making. It is also possible that the participants who reported no impact on their ability to make decisions were not at that time having to make decisions that depended on their comprehension of the genetic information discussed. Overall, most participants reported some positive affect on their ability to make informed decisions, and no participants reported any negative impact.

Healthcare Accessibility

Historically, patients with low vision have been withheld accessibility in the healthcare setting with the greatest reported challenges being mobility and information/communication (ODay et al, 2004). When the participants in the current study were asked, in the follow up survey, how often they are offered written health care materials in accessible formats both said this seldom occurred. This is consistent with past research on the low vision community indicating a lack of accessibility to this type of information (Sibley, 2009). However, these participants also said that they seldom face obstacles associated with written or visual health materials/letters, etc. This may be due to the fact that patients included in this study had mild to moderate vision loss,
therefore they may struggle less with seeing written health materials than people who have more severe vision loss.

**Genetic Counseling Implications**

The results of this study have several implications for the field of genetic counseling and how accessibility is approached. While limited in number, the data indicates that the adaptation of visual aids for people with low vision may improve comprehension, rapport, and decision making abilities for patients. If genetic counselors can continue to address accessibility issues in clinic and in their practice, the disparities caused by lack of access can be mitigated. This study specifically addressed the issue in a clinic specialized to retinal disease, and thus the majority of patients in this clinic have vision loss. However, genetic counselors can see patients with low vision in all areas genetic counseling, and therefore the issue of accessibility is relevant to all genetic counselors and healthcare practices. Using modified images such as these, is one method that can be used to address the gap in information giving and patient comprehension. Other approaches such as providing clinic letters in large print, making audio clips, or offering brochures/documents in accessible formats could also improve accessibility for people with low vision in the practice of genetic counseling. This study begins to explore the concept of accessibility for people with low vision in genetic counseling in a very specific study population, however as advocates for patients, this topic is relevant for all genetic counseling settings.

**Limitations**

The major limitation of this study is the small sample size due, in large part, to challenges in recruiting participants. The images were designed for and used in one specific clinic. The schedule was lighter than usual for a period of time during data collection, and many patients
who did come for appointments did not fit the inclusion criteria. Common reasons for exclusion were patients having vision loss so severe that the images would not be helpful, and not being English speakers. Recruitment during clinic visits, given time constraints of the clinic and the burden on patients, also limited the length of the initial survey we could ask patients to answer. Patients who completed the 5 minute survey were given the option to receive the longer follow-up survey, but interest in this was limited. This is perhaps because it was necessary for patients to provide an email address, and many people have an aversion to providing contact information such as an email address when it is optional. Regardless of the reason, the lack of participants for the longer survey resulted in very limited data from most participants.

Future Directions

This is one of the first studies known to create and assess the effectiveness of visual aids specifically designed for people with low vision. As a pilot study, there are many possible future directions for research in this field. Testing these images in a larger cohort as well as in additional clinics to see if other patient populations have similar responses would be helpful for evaluating their overall effectiveness. Larger studies may also provide additional insight into other ways to edit the images to further improve their visibility. Adapting other patient information such as clinic letters, pamphlets, medical records and prescriptions, and assessing whether this improves access to healthcare for patients with low vision could also provide insight into how to further improve patient experiences in a genetic counseling clinic. Additionally making online resources and websites accessible and compatible with personal visual aid devices could reduce this struggle for patients. Expanding the use of accessible materials, and gaining input from individuals with low vision is an important step towards improving genetic counseling and other healthcare services for these individuals. These improvements could
potentially minimize the healthcare disparities experienced by people with low vision and other
disabilities.
Conclusion

Individuals with low vision can have difficulties accessing many different areas of healthcare. Genetic counseling can be particularly challenging given the complexity of the information and the frequent use of visual aids to reinforce important concepts. This study utilized color, contrast, large print, simple icons, and other methods, to adapt genetic counseling visual aids for people with low vision. Based on the data from this preliminary study, the use of adapted visual aids may improve patient comprehension, rapport, and ability to make informed healthcare decisions. The extent to which the visual aids helped the participants varied, but overall the cohort had a largely positive response to the use of the images in the Inherited Retinal Disorder Service. This study provides the first evidence that the use adapted visual aid genetic counselors may improve the accessibility of their services for people with low vision.
References


APPENDIX A: CONSENT FORM

MASSACHUSETTS EYE AND EAR
GENERAL RESEARCH CONSENT AND AUTHORIZATION FORM
VERSION: August 2016

PROTOCOL TITLE: Improving Access to Genetic Counseling Visual Aids for People with Low Vision

HSC PROTOCOL #:

PRINCIPAL INVESTIGATOR: Emily Place, MS, LCGC

Throughout the consent form, “you” always refers to the person who takes part in the study.

Why is this research study being conducted?

We are asking you to take part in a research study of the use of visual aids during a genetic counseling session for inherited retinal conditions. We are asking you to take part because you have a clinical diagnosis of an inherited retinal condition. During genetic counseling sessions, a genetic counselor uses visual aids to explain genetic topics. Visual aids can improve understanding about genetic topics. The visual aids currently available to counselors are not accessible for patients with visual impairment. The purpose of this study is to use visual aids which have been adapted for individuals with impaired vision during a genetic counseling session to determine if these aids can improve understanding of genetic topics.

We expect to enroll about 100 subjects at Massachusetts Eye and Ear.

Who is doing this research study and where will the study take place?

The principal investigator is Emily Place, who is a certified and licensed genetic counselor. The study will be taking place in the Inherited Retinal Disorder Service at Massachusetts Eye and Ear.
**How long will this study take and what will happen in this study?**

During the genetic counseling session, the genetic counselor will use several visual aids to explain genetic topics. At the end of the counseling session, we will ask you to complete a 6-question survey. This survey will take approximately 5 minutes to complete. You will be asked to rate your experience with the visual aids. At the end of the survey, you will have the opportunity to participate in a longer follow-up survey. This survey contains 19 questions, and will take approximately 10-15 minutes to complete. This survey will be emailed to you to complete at a time that is convenient for you. We ask that you complete this follow-up survey within 2 weeks following your genetic counseling session.

**What are the risks and possible discomforts from being in this research study?**

The physical risks of this study are minimal. There is potentially a risk for psychological discomfort if the survey questions elicit memories or experiences that have been unpleasant for you. There is a risk for loss of anonymity of your responses because your email address will be collected in order to link your initial survey responses to the followup survey responses. If you provide your email address, the researchers will be able to associate your responses with your email address. However, this information will not be shared outside of the research committee and will not be included as part of any report or publication.

There is a federal law called the Genetic Information Nondiscrimination Act (GINA) that, in general, makes it illegal for health insurance companies, group health plans, and most employers, except those with fewer than 15 employees, to discriminate against you based on your genetic information. However, it does not protect you against discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.
Can I decide to stop taking part in this study?

Your taking part in this research study is voluntary, and you may withdraw from the study even after signing this consent. If you decide not to participate or if you withdraw from the study, it will not affect the quality of care you will receive at the Massachusetts Eye and Ear, the payment for your health care, and your health care benefits.

You may request that the genetic counselor stop using the visual aids during your genetic counseling session at any time. You may also discontinue taking your survey at any point in the survey. If you have not submitted your responses to the survey, no data will be collected from your survey response. To withdraw from the study after completing the survey, you can call or email a member of the research team, and tell them that you no longer want to take part. Your survey responses will be deleted from the response database. If data from your survey response has already been published or presented, we cannot withdraw your response after this time.

It is possible that the investigator will stop your participation in the study. If you do not complete the survey responses in entirety, your responses may be discarded from the study.

Will I be paid to take part in this research study?

You will have the opportunity to enter a drawing for a $25 gift card after participating in this study. You will not be otherwise paid or compensated for your participation.

What will I have to pay for if I take part in this research study?

Participation in this study will not incur any additional costs for you. Study funds will pay for certain study-related items and services. We may bill your health insurer for, among other things, routine items and services you would have received even if you did not take part in the research. You will be responsible for payment of any deductibles and
co-payments required by your insurer for this routine care or other billed care. If you have any questions about costs to you that may result from taking part in the research, please speak with the study staff. If necessary, we will arrange for you to speak with someone in Patient Financial Services about these costs.

**How will you protect my confidentiality?**

We cannot guarantee total confidentiality. However, your email address is the only information that will be linked to your response if you choose to provide it in the initial survey. We will keep all data in the secured Qualtrics database. Your email address will be entered into a secure email account in order to send the follow-up survey link and inform gift card drawing winners. Your email address will be deleted from the account when the study is concluded. The consent form will be stored in a locked file cabinet in an area accessible only to study personnel. Your name/birthdate/medical record number or other identifying information will not be collected as with your responses to this research study.

Data from this study will be shared with the collaborating institution, Brandeis University. The only identifier that will be linked to this data is your email address if you choose to provide it.

We may publish the results of this research study in a medical book or journal or use the results to teach others. However, we will not use your name and other identifying information for these purposes without your specific authorization.

**Contact information if you have questions or concerns about this study**

You are free to ask any questions you may have about the study or your rights and treatment as a research subject. Questions may be sent to the genetic counseling research intern email, ljs5783@brandeis.edu or you may contact the Genetic Counselor at 617-573-6902.

If you want to speak with someone not directly involved in this research study, please contact the Human Research Protections Program Office at (617) 573-3446. You can talk to them about:

- Your rights as a research subject
- Your concerns about the research
- A complaint about the research
Your privacy rights:

You have the right **not** to answer any of the survey questions that make you feel uncomfortable. However, if you do not provide enough responses, your response data may be discarded from analysis. You have the right **not** to provide your email address as part of your survey response. If you do not provide your email address, you will not be invited to participate in the follow-up survey.

If you want to withdraw your response data, you must notify the person in charge of this research study by email or over the phone. Once you withdraw your permission to use your responses, you cannot continue to take part in this study.

When you withdraw your permission, we will not gather new information from you after that date, and your responses will be deleted from the Qualtrics survey server and your email address removed from the researcher email account. If you withdraw your permission, we will not be able to take back information that has already been used or shared with others.

Informed consent and authorization

Statement of the Person Giving Informed Consent and Authorization

- I have read this consent form
- This research study has been explained to me, including the risk and possible benefits (if any), other possible treatments or procedures, and other important parts of the study.
- I have had the opportunity to ask questions.
- I understand the information given to me.
- I will be given a signed copy of this form to keep.
Signature of Subject

By signing below, I give my consent to take part in this research study and agree to allow my protected health information to be used and shared as described above.

__________________________
Signature of Adult Subject (18 + years)  ________________ Date  ________________ Time

__________________________
Print Name

Signature of Study Doctor or Person Obtaining Consent:

- I have explained the research to the study subject.
- I have answered all questions about this research study to the best of my ability.

Study Doctor or Person Obtaining Consent  ________________ Date  ________________ Time

__________________________
Print Name
Dear patient and/or family,

Welcome to our clinic. You are here today because you have been diagnosed with an inherited retinal degeneration or this diagnosis is suspected. During your visit today, you may be offered the opportunity to speak with a genetic counselor, as most inherited retinal degenerations are caused by genetic changes or mutations. The genetic counselor can talk about your family history, offer genetic testing, and talk about what this genetic information could mean for you and your family.

We are always looking for ways to improve healthcare for our patients. One project we are piloting is the use of visual aids during genetic counseling to help with understanding complex concepts. These images have been designed to be easily visible for people who have low vision, and are presented on an iPad screen. Our goal is to improve access to medical information for people with low vision and improve their genetic counseling experience.

If you do see a genetic counselor today, they may ask if you would be interested in seeing these images during your genetic counseling discussion, and giving us your feedback in a very brief survey. If you do not want to participate, it will not impact your care in any way. We thank you for any feedback and hope that we can continue to improve care for patients.

If you have any questions about this project, you can ask the genetic counselor or genetic counseling intern.

Sincerely,

Emily Place, MS, CGC (Genetic Counselor)
Lindsay Smith (Genetic Counseling Intern)
Follow-Up Survey Recruitment Email

Dear Study Participant,

Massachusetts Eye and Ear Inherited Retinal Disease Service

Study Number: ##

Thank you for agreeing to be a part of this research study: Improving Access to Genetic Counseling Visual Aids for People with Low Vision

A day or two ago you met with a genetic counselor at Massachusetts Eye and Ear to discuss genetic testing and other genetic topics. Several visual aids were used on an iPad in your genetic counseling session, and you took a brief survey about these visual aids.

You are receiving this email because you indicated that you are interested in participating in a follow-up survey about these visual aids and how they impacted your genetic counseling experience. If you would still like to provide your feedback in a follow-up survey, please click on the link below and answer the survey questions. There are 19 questions, and the survey should take about 10-15 min to complete. You may stop taking the survey at any time. For your participation, you have been entered in a drawing for a $25 gift card.

Your study number is ##. Please provide this number in the first question of the survey.

LINK TO SURVEY

Thank you for your feedback, and for helping us better understand the obstacles people with low vision face when accessing health care. Please feel free to contact me with any questions or concerns.

Best Wishes,

Lindsay Smith
Ljs5783@brandeis.edu
Brandeis Genetic Counseling MS Student
Intern & Researcher at Massachusetts Eye and Ear
APPENDIX C: STUDY INSTRUMENT

Initial In-Clinic Survey

1. How well were you able to see the images during your genetic counseling session?
   Not at all   Not very well   Fairly Well   Very Well

2. How would you rate your understanding of the information provided during your genetic counseling session today?
   Very Bad   Somewhat Bad   Neither Good nor Bad   Somewhat Good   Very Good

3. How did these images affect your understanding of the information provided in your genetic counseling session today?
   Much Worse   Somewhat Worse   No Effect   Somewhat Better   Much Better

4. How did the use of these images affect the relationship you feel with your genetic counselor?
   Much Worse   Somewhat Worse   About the Same   Somewhat Better   Much better

5. How did the use of these images affect your ability to make informed decisions about your healthcare?
   Much Worse   Somewhat Worse   About the Same   Somewhat Better   Much Better

6. Overall, would you recommend the routine use of these visual aids in genetic counseling sessions at the Massachusetts Eye and Ear?
   I Definitely Would Not Recommend   I Would Not Recommend   Neutral   I Would Recommend   I Would Highly Recommend

7. If you would be willing to take a follow-up survey, please provide your email address below. The follow-up survey will be sent to your email for you to do at your convenience. This information will also enter you into a drawing for a $25 gift card. (Optional)
Follow-Up Survey

1. What is your study number? This can be found in your email invitation. __________
2. Please select any of the images you recall from your genetic counseling session. (Select all that apply)
3. Please Select the Color Combination(s) you recall from your genetic counseling session.

<table>
<thead>
<tr>
<th>Color Combination(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Black Text, White Background</td>
</tr>
<tr>
<td>Black Text, Yellow Background</td>
</tr>
<tr>
<td>White Text, Black Background</td>
</tr>
<tr>
<td>Yellow Text, Black Background</td>
</tr>
<tr>
<td>I Do Not Remember</td>
</tr>
</tbody>
</table>

4. Which element(s) of the images improved visibility for you? (Select all that apply)
   - Color/Contrast
   - Ability to Zoom In and Out
   - Bold Simple Icons
   - None of These
   - Large Font
   - Other
   - Uncluttered and a lot of white space

5. Is there one element about the images that was most helpful in making the image visible for you? (Select one)
   - Color/Contrast
   - Ability to Zoom In and Out
   - Bold Simple Icons
   - None of These
   - Large Font
   - Other
   - Uncluttered and a lot of white space
Did these images change how confident you feel about describing this information to other family members, friends, etc.

Much Less confident  Somewhat Less Confident
Neither Confident nor Unconfident
Somewhat More Confident  Much More Confident

6. Prior to this your most recent visit in the IRD Service, had you seen a genetic counselor at Mass Eye and Ear regarding your vision loss? Yes
No
Unsure

*If the participant answers ‘yes’ to question 7, participant is given question 8/

7. Compared to your last meeting with a genetic counselor, how did these images change your overall experience?
Much worse  Somewhat worse  No Change  Somewhat better  Much Better

Consider all of your health care experiences in general for the next 2 questions.

8. How often do you face obstacles with written or visual health materials/letters, etc.?
   Always
   Usually
   About half the time
   Seldom
   Never

9. How often do healthcare offices offer written materials in accessible formats such as large print/audio/braille, etc.
   Always
   Usually
   About half the time
   Seldom
   Never

10. How would you describe your level of vision loss?
    Mild Vision Loss
    Moderate Vision Loss
    Advanced Vision Loss
11. What is the clinical diagnosis of your vision condition?

- Retinitis Pigmentosa (RP)
- Macular degeneration
- Cone-rod degeneration
- Other ______
- Unsure

12. How many years ago did you start losing your vision?

_______

13. Do you have a **genetic** diagnosis for your vision loss?
   
   Note: A genetic diagnosis means that a specific genetic cause for your vision loss has been identified through genetic testing.
   
   - Yes
   - No
   - Unsure

14. What is your gender?

   - Male
   - Female
   - Other ______
   - Prefer Not to Say

15. What is your Age?

   ______

16. What is the highest level of education you have received?

   - Less than high school
   - High school graduate
   - Some college
   - 2 year degree
   - 4 year degree
   - Professional degree
   - Doctorate
18. Do you have any other suggestions for how to improve the educational images used in your genetic counseling session?

19. Please let us know if you have any other additional comments.
Autosomal Dominant

Affected 50% 1/2  

OR

Affected 50% 1/2  

OR

Affected 50% 1/2  

Unaffected 50% 1/2
Autosomal Dominant

Affected 50% 1/2

OR

Unaffected 50% 1/2

OR

Affected 50% 1/2

Unaffected 50% 1/2
Autosomal Recessive

Carrier 100%
Autosomal Recessive

Unaffected 25% 1/4

Carrier 50% 1/2

Affected 25% 1/4
X-Linked Recessive

Carrier Daughter

OR

Unaffected Son
X-Linked Recessive

Carrier Daughter  Unaffected Son

OR
Read the DNA Sequence of the Retinal Genes
DNA Sequencing

Cell → DNA → DNA from retinal genes cut into pieces

Read the DNA Sequence of the Retinal Genes
USH2A

Viral Vector

USH2A is Too Big to Fit
Stem Cell Therapy

Grow Stem Cells

Change into Retina Cells
Stem Cell Therapy

Grow Stem Cells

Change into Retina Cells