Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program

Master’s Thesis

Presented to
The Genetic Counseling Program, Department of Biology
Brandeis University

Gretchen H. Schneider, MS, CGC, Advisor

In Partial Fulfillment of the
Requirements for the Degree

Master of Science

By
Sarah A. Casner
May, 2010

Committee members:

Gretchen Schneider, M.S.
Co-director, Brandeis University Genetic Counseling Program

Linde D’Andrea, M.ED./CAGS Clinical Genetics
School Psychologist, Perkins School for the Blind

Janey L. Wiggs, M.D., Ph.D.
Associate Professor of Ophthalmology, Harvard Medical School, Massachusetts Eye and Ear Infirmary

Paula Labella Belanger
Parent, medical and educational advocate of disabled children
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

ACKNOWLEDGEMENTS:

I would like to sincerely thank the members of my committee, Gretchen Schneider, Linde D’Andrea, Dr. Janey Wiggs, and Paula Labella Belanger, for all of their support, guidance, and feedback throughout the entire thesis process. To Gretchen, for being my primary advisor and always having an open door when new issues arose; to Linde, for being a wonderful liaison between myself and Perkins School for the Blind, and for being a constant source of encouragement; to Janey, for taking time out of her busy schedule to provide much needed feedback and attend both of my presentations; and to Paula, for being as enthusiastic about giving back to the Perkins community as I am. Thank you to Scott Motyka for help in the drafting of my pre-development survey. Many thanks to Betsey Sennott for ensuring that I had my support documents printed in Braille, and for being my IT go-to person at Perkins during my presentation. A special thank you to the staff of Perkins School for the Blind secondary program for participating in the surveys and presentations, as well as their continued enthusiasm for this project. A huge thank you to the entire Brandeis Genetic counseling program, especially Judith Tsipis, Gretchen Schneider, Beth Rosen-Sheidley, Janet Rosenfield, Missy Goldberg, and the class of 2010, for being a great support over the past two years, I couldn’t have done it without you. To my parents and brothers, thank you for everything.
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program  
Principal Investigator: Sarah Casner

ABSTRACT:

Hereditary disease is a leading cause of blindness in children in the United States and other developed countries, where environmental causes of blindness, such as vitamin deficiencies, are less prevalent. Many hereditary causes of blindness are syndromic, and children affected by these diseases have multiple impairments. It is therefore important that those who provide care to these children understand the etiology of their diagnoses. The purpose of our study was to assess interest in a genetics education tool for care providers of children with hereditary blindness, and to develop and pilot test such a tool based on the outcome of the assessment. We surveyed the non-medical staff of the Perkins School for the Blind Secondary Program regarding their interest in and understanding of many different genetic causes of blindness, and their preferences with respect to the format and content of an educational tool. We found that the survey respondents were most interested in learning about Bardet-Biedl syndrome, Batten disease, Leber congenital amaurosis, retinitis pigmentosa, retinoblastoma, and septo-optic dysplasia, all common among students who attend Perkins. The respondents indicated a preference for an in person seminar, rather than online video, book, or fact sheets. We conducted two educational seminars and attendees were asked to complete a survey evaluating the usefulness and effectiveness of the seminars. The respondents indicated that the information they learned was useful to them, and will be a positive influence on the care they provide to students they interact with. Our findings suggest that non-genetics care providers may benefit from educational seminars that focus on genetic aspects of the specific populations they serve. Individual relationships between genetics professionals and care providers in specialty settings such as Perkins may allow for more
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

effective educational interventions than would the use of written materials or recorded media.
TABLE OF CONTENTS:

Title Page........................................................................................................................................i
Acknowledgements.........................................................................................................................ii
Abstract............................................................................................................................................iii
Table of Contents..............................................................................................................................v
List of Tables and Figures.................................................................................................................vi
Introduction......................................................................................................................................1
Methods...........................................................................................................................................7
Results...........................................................................................................................................12
Discussion.........................................................................................................................................20
Conclusion.........................................................................................................................................25
References.........................................................................................................................................27
Appendix A: Recruitment Notice......................................................................................................29
Appendix B: Seminar Recruitment Flyers........................................................................................30
Appendix C: Pre-Development Survey Document..........................................................................31
Appendix D: Post-Development Survey Document 1.................................................................33
Appendix E: Post-Development Survey Document 2.................................................................35
Appendix F: Information Sheet Handouts.....................................................................................37
LIST OF TABLES AND FIGURES:

Table 1. Post-Development Tool Quiz 1 ..............................................................11
Table 2. Post-Development Tool Quiz 2 ..............................................................11
Table 3. Tool Effectiveness Statements .............................................................12
Figure 1. Participant Gender and Age ...............................................................13
Figure 2. Participant Job Role ..........................................................13
Figure 3. Years Worked at Perkins ..........................................................14
Figure 4. Level of Education ..........................................................14
Figure 5. Number of College Level Genetics Courses ..............................15
Figure 6. Participation in Informal Genetics Education ..............................16
Table 4. Diseases of Interest ..........................................................17
Figure 7. Possible Education Tools ..........................................................17
Table 5. Effectiveness of First Seminar ..........................................................19
Table 6. Effectiveness of Second Seminar .......................................................19
Table 7. Correct and Incorrect Quiz Answers ..................................................20
I. INTRODUCTION

Worldwide there are 1.4 million children who are blind. In the United States, the World Health Organization (WHO) estimates that the prevalence of blindness in children is 0.3 per 1000 (Mets, 1999). Environmental or acquired causes of blindness such as infections are becoming less common as interventions and treatments improve, meaning that hereditary causes of blindness are now in the majority. This is mostly true in the more affluent countries where factors leading to blindness such as vitamin A deficiency are not usually observed (Gilbert, 2003; Al-Salem, 1992). In countries with a higher level of socio-economic development, hereditary disease may cause anywhere from 16-51% of childhood blindness (Gilbert, 1995; Mets, 1999). There are a multitude of genetic etiologies that cause isolated or syndromic vision loss in children. The care that a child who experiences vision loss receives is incredibly important, whether that care is vision sparing, or it allows the child to receive a proper education. This has a significant impact on child’s quality of life. The more that the professionals in this field, such as care providers and teachers, understand about a disease, the better the care they can provide.

Bardet-Biedl syndrome (BBS) is an autosomal recessive disorder characterized by retinal degeneration, early-onset obesity, cognitive impairment, genitourinary tract malformations, renal dysfunction, and polydactyly. Mutations in fourteen different genes have been found to be disease causing, and patients with both homozygous and compound heterozygous mutations in this family of fourteen genes will display complete penetrance of the disease. The proteins transcribed by the BBS family of genes have all been found to localize to the basal bodies and cilia, and it is believed that the dysfunction
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

of the basal bodies and cilia play an important role in the pathogenesis of BBS. BBS has been shown to be both a genotypically and phenotypically heterogeneous disease (Beales, 2005). Knowledge of the genotype-phenotype correlations of BBS could give care providers a better understanding of the specific pathogenesis of BBS in each patient, in turn, allowing the care provider to implement the best course of treatment.

Leber congenital amaurosis (LCA) is the most severe cause of congenital vision loss and could account for up to twenty percent of students enrolled in schools for the blind. Four clinical characteristics define LCA, including severe and early vision loss, sensory nystagmus, amaurotic pupils, and absent electroretinogram signals. Additional findings associated with LCA include intellectual disabilities, autism and olfactory dysfunction (den Hollander, 2008). Similar to BBS, LCA is both clinically and molecularly heterogeneous. There are fourteen known genes and loci in which mutations cause disease. Some genotype-phenotype correlations have been established, hopefully allowing care providers to design the most appropriate treatment plan.

Retinitis pigmentosa (RP) is a group of progressive blindness disorders caused by photoreceptor degeneration and can be syndromic or nonsyndromic. RP has been associated with a number of childhood blindness disorders including Bardet-Biedl and Usher syndromes. All forms of RP follow a similar disease course; the first clinical sign of RP is night blindness, followed by a progression of vision loss. The difference between the forms of RP seems to be how quickly the disease progresses. RP follows many different inheritance patterns including autosomal recessive, autosomal dominant and X-linked (Hamel, 2006). X-linked RP has been shown to have a much quicker disease progression than RP inherited in other manners (Sandberg, 2009).
Neuronal ceroid lipofuscinoses (NCL) are the most common progressive neurodegenerative, inherited diseases of childhood and are classified as lysosomal storage diseases. At least ten different loci have been implicated as disease causing, and eight of the genes have been characterized. All appear to be inherited in an autosomal recessive manner and the genes are known as the neuronal ceroid lipofuscinoses (CLN) family. Characteristic symptoms of NCLs include loss of vision, motor abnormalities, seizures and a shortened life span. Some of the associated symptoms include ataxia, spastic limb paresis, osteoporosis, and paralysis. Some genotype-phenotype correlations have been established within the NCLs, and seem to be related to the age of onset, rather than a variation on observed symptoms (Jalanko, 2008). Batten disease is the juvenile form of NCL, and also the most common NCL.

Septo-optic dysplasia (SOD), also known as de Morsier syndrome, is a multifactorial disorder characterized by variable optic nerve hypoplasia (ONH), midline brain abnormalities and pituitary hypoplasia. Some familial cases have been observed, but the majority of cases appear to be sporadic. Two genes have been associated with SOD; HESX1 and SOX2, while the main environmental factor leading to SOD appears to be a variety of viral infections. SOD is a clinically heterogeneous disorder and symptoms may range from epilepsy to global retardation, unilateral ONH to anophthalmia, and isolated growth hormone deficiency to panhypopituitarism (Kelberman, 2008). Genotype-phenotype correlations regarding the known associated genes have not been established.

Retinoblastoma (RB) is the most common childhood intraocular tumor and is caused by mutations in RB1. Tumor formation requires mutation in both copies of the
RB1 gene. In familial cases a child inherits one mutated copy and if a ‘second hit’ (loss of heterozygosity) occurs in the second RB1 copy of a developing retinal cell then a tumor can form. In sporadic cases, two random mutations occur in the same developing retinal cell. Familial retinoblastoma is inherited as an autosomal dominant trait with reduced penetrance (80-90%) and usually is associated with bilateral disease, while sporadic cases are typically unilateral. RB tumors can be identified by an ophthalmic exam (in a child this usually requires general anesthesia). Frequently, the disease is first recognized by a parent or caregiver who notices the characteristic ‘white pupil’ caused by the tumor. If left untreated the tumors will eventually destroy the eye and if the tumor invades the surrounding ocular tissue the disease can be fatal. Unfortunately, the best known treatment for RB is removal of the eye, so children with familial RB frequently are left without any vision (Poulaki, 2009).

Establishing genotype-phenotype correlations for the diseases causing childhood blindness has proven invaluable in determining the course of treatment. Genotype-phenotype correlations help to predict the progression of a disease, and allow for a more accurate prognosis to be established. As the efficiency of mutation discovery increases and the understanding of these diseases improve, more molecular diagnoses can be made, prompting an increase in research into treatments for these diseases (Goodwin, 2008). Treatment for many of the childhood blindness disorders is considered time sensitive, so the earlier a genetic diagnosis is made, the more likely a treatment is to be effective. Much of the currently researched treatment methods for the diseases causing childhood blindness are gene-specific, providing another example as to why a genetic diagnosis is so important (Koenekoop, 2007).
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

There is a lot which needs to be understood about the development of children with vision loss or blindness by those who educate them. As compared to sighted peers, children with vision loss have a delay in the development of fine motor skills. Sight has also been shown to be an important factor in bonding, impacting the relationship a visually impaired child has with his or her family and teachers. A heightened use of other senses, such as hearing and touch, can help compensate for loss of vision; however, it is necessary that educators understand how to teach a child to use these senses effectively (Mets, 1999). Visually impaired children may also feel that they have a poorer quality of life as compared to sighted peers. The more severe the visual loss, the more years of their life patients are willing to “trade” for perfect vision (Brown, 1999). This could be an important concept for counselors and therapists who work with visually impaired children to understand.

Proper nutritional treatment has been shown to significantly modify and improve the course of many genetic disorders, including a few disorders that cause retinal degeneration, which is a major cause of blindness throughout the world (Berson, 1999). In schools such as Perkins School for the Blind, where many students are full time residents and therefore eat all meals on campus, providing a proper diet to these students is imperative. It is also important for all employees who spend time with a student to understand his or her nutritional needs so that non-meal time eating may be closely monitored.

In 2004, the National Coalition for Health Professional Education in Genetics developed core competencies in genetics that health care providers should know. However, simply having knowledge of genetics and genomics is not sufficient; these
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

providers must also be able to integrate this knowledge into how they care for a patient (Kenner, 2005). Unfortunately, even with guidelines, it is possible that health care providers do not receive a proper genetics education. Nursing students have reported that they feel under-educated in the basics of genetics, even though they may feel competent when it comes to the genetics of individual diseases. These students expressed a desire for an improved genetics curriculum so that they could provide the most appropriate care to their patients (Vural, 2009). However, this information is based on a study from Turkey, and may not accurately represent the feelings of nursing students in the United States.

At Perkins School for the Blind many of the students also have some sort of cognitive impairment such as intellectual disabilities and autism. Non-medical staff within this field play an integral role in the lives of those affected by intellectual disabilities, and their families, and often greatly influence the decisions made by the family. If these non-medical workers had a broader knowledge of genetics, more genetic etiologies causing cognitive impairments in those previously undiagnosed could be identified. This would improve both the medical and non-medical care offered to the individual (Finucane, 2003).

Previous studies have focused on the working knowledge of genetics in health care providers, and not on the knowledge of genetics in the multitude of other care providers outside of the medical realm. A broad knowledge of genetics has been shown over and over to be imperative in order for health care professionals to provide the best and most appropriate care to individuals; we believe this to be true for non-medical care workers as well.
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

Previous personal experience with providing a group of non-medical employees at Perkins School for the Blind a brief genetics education yielded positive feedback. Most of the feedback received indicated that these employees found the information interesting and useful in understanding how and why the children they work with behave in certain ways. This was an unscientific look at non-medical employees’ attitudes towards improved genetics education. The consensus opinion suggested that it would be beneficial to analyze, more in the depth, the hypothesis that non-medical employees will benefit from, and will be able to provide, a higher level of care to students, given an improved genetics education.

Perkins School for the Blind provides an excellent environment in which to study this hypothesis. The student body is incredibly diverse; students have been diagnosed with a multitude of different diseases and disorders, many of which are genetic. Perkins School for the Blind also employs a great number of people involved in the day to day care of the students who have not had an in depth medical education, including genetics. Perkins School for the Blind already provides excellent care and education to their students, but there is always room for improvement. As the Perkins motto says “All we see is possibility”.

II. METHODS

Recruitment

A request to participate in the pre-development survey was done by personally e-mailing the recruitment notice to the Secondary Program staff, such as teachers, residential staff and clinicians, at Perkins School for the Blind listserv (Appendix A). The recruitment notice included a link directing participants to an online survey hosted
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

by Survey Monkey (www.surveymonkey.com). We recruited participants to attend the seminars by sending reminder e-mails prior to the events through the listserv, including notifications in the Perkins’ electronic bulletin, and by posting flyers in Secondary Program buildings. Seminar attendees were provided with paper evaluations, and were asked at the beginning and the end of the presentation to fill them out prior to leaving.

To be eligible for participation, a person had to be at least 18 years of age, and to be a member of the non-medical staff for Perkins School for the Blind Secondary Program with daily interaction with students. Support staff, including janitorial and kitchen staff, were excluded from participation. Program nurses were encouraged to participate in the hopes of comparing this cohort to the non-medical staff.

Data Collection

We offered participants an anonymous, online survey (Appendix B) hosted by Survey Monkey (www.surveymonkey.com) in order to collect tool pre-development information. The survey was mostly quantitative, and included a mixture of open ended, Likert scale and multiple choice questions. Participants were asked to list genetic disorders they were aware of, indicate interest in learning about a variety of genetic causes of blindness and how they would prefer to learn about such disorders, as well as provide basic demographic information.

Two seminars were held in order to maximize the amount of information taught. Information about different diseases was presented at each seminar. Post-development data was collected using anonymous, hard copy surveys (Appendices C and D), provided to participants at the time of the seminars. These surveys were mostly quantitative, and included a mixture of open ended, Likert scale and multiple choice questions.
Participants were asked to identify if they felt that they gained knowledge from the presentation, if the information was useful, and whether or not the information would impact the care and/or instruction provided to students. Participants were also asked to take a brief quiz, included in the survey, to evaluate whether or not the information was appropriately conveyed during the seminars.

**Data Analysis**

**Demographics**

Demographic information was collected in the tool pre-development survey in order to assess the makeup of the Perkins School for the Blind Secondary Program staff. Once this information had been collected, we took the total number of responses and calculated the percentages of staff for each demographic question that was posed.

**Genetics Knowledge**

The knowledge of the participants was measured in a number of ways. In the tool pre-development survey participants were asked to list genetic disorders that were known to them. The number and type of diseases were counted for each participant to assess a general knowledge level prior to tool development. Participants were also asked several questions in the tool pre-development survey about their previous genetics education. These responses were analyzed by looking at the average amount of genetics education that the participants indicated they had received prior to taking the survey.

**Disease and Tool Interest**

Using a Likert scale, participants were asked to rank a list of genetic causes of blindness from 1- not at all interested, to 5- very interested, so that we could determine what diseases were of most interest to our audience. Participants were allowed to rank as
many diseases as they desired. Responses were counted and an average score was calculated for each disease. We also looked at the total number of “5” responses for each disease and calculated a percentage. These numbers were used to determine what diseases were included in the genetics education tool.

In order to determine what type of tool would be most useful to the non-medical staff of Perkins School for the Blind Secondary Program, the respondents were asked to answer a multiple choice question that provided them with several tool options. Respondents were only allowed to choose one response. We counted the responses and determined which tool the majority of the respondents were interested in.

Satisfaction

As part of the tool post-development surveys, participants were asked to answer an open ended question about their satisfaction with the tool. Responses were coded and analyzed for themes. We then analyzed each response from the surveys for the frequency of the themes.

Effectiveness

The effectiveness of the seminars (tool) was measured using a brief quiz (Table 1; Table 2) that participants were asked to fill out following the presentation. Answers were scored as correct or incorrect. Unanswered questions were not included in the final tally of correct versus incorrect answers. Quiz scores were analyzed as a group, rather than on an individual basis as some participants did not finish each quiz fully. After being scored, the percentage of correct versus incorrect responses was calculated to analyze tool effectiveness.
Table 1. Post-Development Tool Quiz 1

<table>
<thead>
<tr>
<th>Question</th>
<th>Options</th>
</tr>
</thead>
</table>
| How many pairs of chromosomes are in the human genome?                   | a) 20  
   b) 23  
   c) 40  
   d) 46 |
| Men and women are equally affected in autosomal recessive diseases.       | a) True  
   b) False |
| What is the inheritance pattern of Bardet-Biedl syndrome?                | a) Autosomal dominant  
   b) Autosomal recessive  
   c) X-linked dominant  
   d) X-linked recessive  
   e) Mitochondrial |
| What symptom is characteristic of Batten disease?                        | a) Polydactyly  
   b) Seizures  
   c) Autism  
   d) Keratoconus  
   e) Keratoglobus |
| Mutations in how many genes and/or loci are causative of Leber congenital amaurosis? | a) 1  
   b) 5  
   c) 11  
   d) 14 |

Bold and italicized responses indicate correct answer

Table 2. Post-Development Tool Quiz 2

<table>
<thead>
<tr>
<th>Question</th>
<th>Options</th>
</tr>
</thead>
</table>
| Mutations in how many genes are known to cause retinitis pigmentosa?      | a) 16  
   b) 25  
   c) Over 50  
   d) 2 |
| Which of the following is a common sign of retinoblastoma?                | a) "Red eye" when a picture is taken  
   b) eye rubbing  
   c) "White eye" when a picture is taken  
   d) Nail biting |
| Which of the following is NOT known to be an inheritance pattern for septo-optic dysplasia? | a) Sporadic  
   b) Mitochondrial  
   c) Autosomal dominant  
   e) Autosomal recessive |

Bold and italicized indicate correct answer
Effectiveness was also measured using Likert scale questions (Table 3) included as part of the tool post-development survey. These questions were the same for both post-development surveys. We tabulated the responses from both surveys and found the percentage of each response. We also combined agree and strongly agree, and disagree and strongly disagree, for comparison.

### Table 3. Tool Effectiveness Statements

<table>
<thead>
<tr>
<th>I gained new knowledge from this presentation.</th>
</tr>
</thead>
<tbody>
<tr>
<td>a) Strongly disagree</td>
</tr>
<tr>
<td>b) Disagree</td>
</tr>
<tr>
<td>c) Agree</td>
</tr>
<tr>
<td>d) Strongly agree</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>The information from the presentation was useful to me.</th>
</tr>
</thead>
<tbody>
<tr>
<td>a) Strongly disagree</td>
</tr>
<tr>
<td>b) Disagree</td>
</tr>
<tr>
<td>c) Agree</td>
</tr>
<tr>
<td>d) Strongly agree</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>The information I obtained from the presentation will affect of change the care/instruction I give to my students.</th>
</tr>
</thead>
<tbody>
<tr>
<td>a) Strongly disagree</td>
</tr>
<tr>
<td>b) Disagree</td>
</tr>
<tr>
<td>c) Agree</td>
</tr>
<tr>
<td>d) Strongly agree</td>
</tr>
</tbody>
</table>

### III. RESULTS

#### Demographics

We received a total of 49 responses to the tool pre-development survey, which is approximately half of the eligible non-medical Perkins School for the Blind Secondary program staff. Two program nurses also participated in the tool pre-development survey. Most participants responded to every demographic question. The majority of respondents (almost 80%) were found to be female (Figure 1). Most of the respondents were 55 years of age or younger, however, the respondents 56 years old or older were the single largest group (Figure 1). Over 50% of the respondents indicated that they identify their role in
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

the Perkins School for the Blind Secondary Program as that of a teacher (Figure 2), have been working at Perkins School for the Blind for over 15 years (Figure 3), and their highest level of education is a Master’s degree (Figure 4).
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

Figure 3

Figure 4

Genetics Knowledge
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

A total of 27 participants listed genetic disorders they are aware of in the tool pre-development survey. This is 55% of the total number of respondents who participated in the tool pre-development survey. A total of 34 different disorders were listed by the respondents. The number of known genetic diseases listed ranged from 1 disease to 23 diseases. Most respondents listed knowledge of 6 or fewer genetic diseases.

All 49 of the participants answered the questions in the tool pre-development survey about previous genetics education. Participants were asked how many college level courses they had taken, and if they had ever taken part in any informal genetics education, such as a conference or seminar. Figure 5 shows the number of college level genetics courses participants indicated they have taken. A majority of the respondents indicated that they had participated in some sort of informal genetics education (Figure 6).

Figure 5

College Level Genetics Courses Taken By Staff

Number of People

Number of Courses
Disease and Tool Interest

We asked participants to rank their interest in 24 different genetic diseases that cause blindness. These 24 diseases were known to be diagnoses of both present and past students in Perkins School for the Blind Secondary Program. Seven diseases were ranked “5-very interested” by 40% or more of the respondents, while eight diseases had an average interest score of 3.8 or more (Table 4).

We also asked participants to indicate which of several choices of tool they would find most beneficial. Participants were made aware that the chosen tool would be modified to include accommodations for staff members who are blind or visually impaired. Figure 7 shows the number of responses for each of the tool choices. 49% of participants indicated that their preference for tool would be a live seminar with handouts.
Table 4. Diseases of Interest

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alström syndrome</td>
<td>6</td>
<td>5</td>
<td>7</td>
<td>10</td>
<td>16 (36.4)</td>
</tr>
<tr>
<td>Aniridia (syndromic)</td>
<td>8</td>
<td>4</td>
<td>9</td>
<td>11</td>
<td>11 (25.6)</td>
</tr>
<tr>
<td>Aniridia (non-syndromic)</td>
<td>8</td>
<td>5</td>
<td>10</td>
<td>9</td>
<td>11 (25.6)</td>
</tr>
<tr>
<td>Bardet-Biedl syndrome</td>
<td>3</td>
<td>1</td>
<td>12</td>
<td>11</td>
<td>20 (42.6)</td>
</tr>
<tr>
<td>Batten syndrome (NCLs)</td>
<td>3</td>
<td>4</td>
<td>4</td>
<td>13</td>
<td>22 (47.8)</td>
</tr>
<tr>
<td>CHARGE syndrome</td>
<td>4</td>
<td>2</td>
<td>8</td>
<td>18</td>
<td>13 (28.9)</td>
</tr>
<tr>
<td>Choroideremia</td>
<td>7</td>
<td>6</td>
<td>14</td>
<td>5</td>
<td>10 (23.8)</td>
</tr>
<tr>
<td>Congenital cataracts</td>
<td>2</td>
<td>10</td>
<td>9</td>
<td>11</td>
<td>12 (27.3)</td>
</tr>
<tr>
<td>Leber congenital amaurosis</td>
<td>2</td>
<td>3</td>
<td>10</td>
<td>10</td>
<td>18 (41.9)</td>
</tr>
<tr>
<td>Leber hereditary optic neuropathy (LHON)</td>
<td>4</td>
<td>5</td>
<td>9</td>
<td>10</td>
<td>16 (36.4)</td>
</tr>
<tr>
<td>Lenz microphthalmia syndrome</td>
<td>7</td>
<td>7</td>
<td>9</td>
<td>7</td>
<td>11 (26.4)</td>
</tr>
<tr>
<td>Lowe syndrome</td>
<td>8</td>
<td>8</td>
<td>10</td>
<td>6</td>
<td>9 (22)</td>
</tr>
<tr>
<td>Neurofibromatosis 1 (NF1)</td>
<td>5</td>
<td>4</td>
<td>12</td>
<td>8</td>
<td>13 (31)</td>
</tr>
<tr>
<td>Neuropathy, ataxia and retinitis pigmentosa (NARP)</td>
<td>0</td>
<td>4</td>
<td>11</td>
<td>15</td>
<td>14 (31.8)</td>
</tr>
<tr>
<td>Norrie disease</td>
<td>6</td>
<td>5</td>
<td>10</td>
<td>7</td>
<td>16 (36.4)</td>
</tr>
<tr>
<td>Oculocutaneous albinism</td>
<td>4</td>
<td>8</td>
<td>6</td>
<td>13</td>
<td>11 (26.2)</td>
</tr>
<tr>
<td>Oculofaciocardiodental syndrome (OFCD)</td>
<td>6</td>
<td>7</td>
<td>7</td>
<td>10</td>
<td>8 (21.1)</td>
</tr>
<tr>
<td>Retinitis pigmentosa (syndromic)</td>
<td>1</td>
<td>2</td>
<td>12</td>
<td>11</td>
<td>21 (44.7)</td>
</tr>
<tr>
<td>Retinitis pigmentosa (non-syndromic)</td>
<td>1</td>
<td>3</td>
<td>12</td>
<td>10</td>
<td>18 (40.9)</td>
</tr>
<tr>
<td>Retinoblastoma</td>
<td>0</td>
<td>4</td>
<td>16</td>
<td>5</td>
<td>19 (43.2)</td>
</tr>
<tr>
<td>Septo-optic dysplasia (SOD or de Morsier syndrome)</td>
<td>1</td>
<td>1</td>
<td>7</td>
<td>12</td>
<td>26 (55.3)</td>
</tr>
<tr>
<td>Stargardt disease</td>
<td>4</td>
<td>6</td>
<td>10</td>
<td>8</td>
<td>15 (34.9)</td>
</tr>
<tr>
<td>Stickler syndrome</td>
<td>5</td>
<td>6</td>
<td>12</td>
<td>11</td>
<td>7 (17.1)</td>
</tr>
<tr>
<td>Usher syndrome</td>
<td>3</td>
<td>4</td>
<td>14</td>
<td>8</td>
<td>16 (35.6)</td>
</tr>
</tbody>
</table>

*Number in parentheses indicates the percentage of respondents for that disease who chose "5- very interested."

Figure 7

What education tool would you find most useful?
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

Satisfaction

First Seminar

Several themes were apparent in the satisfaction responses from the first tool post-development survey. Overall, participants indicated a high level of satisfaction with the seminar and the information presented. Participants also indicated a desire for increased audience participation, as well as more information on how the diseases impact the students’ day to day lives. The final theme discovered from participant responses was suggestions for improving the quality of presentation. The most common suggestion was for the presenter to slow down and avoid reading from the screen.

Second Seminar

Different themes were apparent in the satisfaction responses for the second tool post-development survey than those in the first. All responses included a commendation for the work presented. Responses also indicated that any dissatisfaction from the previous presentation had been resolved. A new theme that emerged following the second seminar was the appreciation for the seminars and the information presented, especially as it pertains to the students’ lives.

Effectiveness

A total of 23 participants filled out surveys following the presentation of the first seminar. Table 5 shows the responses to several Likert scale questions assessing the effectiveness of the information presented in the first seminar. These questions were kept the same in both of the tool post-development surveys. Participants were asked to rate each statement from “strongly disagree” to “agree”. The majority of participants strongly agreed that they gained new knowledge and that the information presented was useful to
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

them. The majority of participants also agreed that the information will affect the care or instruction they provide to students.

Table 5. Effectiveness of the first Seminar

<table>
<thead>
<tr>
<th>Statements:</th>
<th>Strongly Disagree</th>
<th>Disagree</th>
<th>Agree</th>
<th>Strongly Agree</th>
</tr>
</thead>
<tbody>
<tr>
<td>I gained new knowledge from this presentation.</td>
<td>3 (13%)</td>
<td>0 (0%)</td>
<td>4 (17%)</td>
<td>16 (70%)</td>
</tr>
<tr>
<td>The information from this presentation was useful to me.</td>
<td>3 (14%)</td>
<td>0 (0%)</td>
<td>5 (23%)</td>
<td>14 (63%)</td>
</tr>
<tr>
<td>The information I obtained from the presentation will affect or change the care/instruction I give to my students.</td>
<td>1 (4.5%)</td>
<td>6 (26%)</td>
<td>15 (65%)</td>
<td>1 (4.5%)</td>
</tr>
</tbody>
</table>

**Bold** Indicates most common response

A total of 20 participants filled out surveys following the presentation of the second seminar. Table 6 shows the responses to the same Likert scale questions, assessing the effectiveness of the information presented in the second seminar. The second seminar results were very similar to the results from the first seminar; the majority of participants strongly agreed that they gained new knowledge and that the information presented was useful to them. The majority of participants also agreed that the information will affect the care or instruction they provide to students.

Table 6. Effectiveness of the second Seminar

<table>
<thead>
<tr>
<th>Statements:</th>
<th>Strongly Disagree</th>
<th>Disagree</th>
<th>Agree</th>
<th>Strongly Agree</th>
</tr>
</thead>
<tbody>
<tr>
<td>I gained new knowledge from this presentation.</td>
<td>1 (5%)</td>
<td>0 (0%)</td>
<td>10 (50%)</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>The information from this presentation was useful to me.</td>
<td>1 (5%)</td>
<td>0 (0%)</td>
<td>10 (50%)</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>The information I obtained from the presentation will affect or change the care/instruction I give to my students.</td>
<td>0 (0%)</td>
<td>1 (5%)</td>
<td>16 (85%)</td>
<td>2 (10%)</td>
</tr>
</tbody>
</table>

**Bold** Indicates most common response
Several quiz questions were included at the end of each seminar post-development survey in order to assess whether or not the information had been properly presented to those in attendance. Table 7 summarizes the number of correct and incorrect question responses for both of the quizzes. For every question posed to the seminar attendees, the majority answered the questions correctly. No question had less than a 70% correct response rate.

Table 7. Correct and Incorrect Quiz Answers

<table>
<thead>
<tr>
<th>Quiz 1</th>
<th>Correct</th>
<th>Incorrect</th>
</tr>
</thead>
<tbody>
<tr>
<td>How many pairs of chromosomes are in the human genome?</td>
<td>19 (83%)</td>
<td>4 (17%)</td>
</tr>
<tr>
<td>Men and women are equally affected in autosomal recessive diseases</td>
<td>15 (71%)</td>
<td>6 (29%)</td>
</tr>
<tr>
<td>What is the inheritance pattern of Bardet-Biedl syndrome?</td>
<td>20 (95%)</td>
<td>1 (5%)</td>
</tr>
<tr>
<td>What symptom is characteristic of Batten disease?</td>
<td>18 (82%)</td>
<td>4 (18%)</td>
</tr>
<tr>
<td>Mutations in how many genes and/or loci are causative of Leber congenital amaurosis?</td>
<td>16 (89%)</td>
<td>2 (11%)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Quiz 2</th>
<th>Correct</th>
<th>Incorrect</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mutations in how many genes are known to cause retinitis pigmentosa?</td>
<td>13 (76%)</td>
<td>4 (26%)</td>
</tr>
<tr>
<td>Which of the following is a common sign of retinoblastoma?</td>
<td>20 (100%)</td>
<td>0 (0%)</td>
</tr>
<tr>
<td>Which of the following is NOT known to be an inheritance pattern in septooptic dysplasia?</td>
<td>14 (82%)</td>
<td>3 (18%)</td>
</tr>
</tbody>
</table>

IV. DISCUSSION

Genetics Knowledge

Overall, the staff of Perkins School for the Blind Secondary Program appear to have been exposed to genetics and received at least a basic genetics education. Close to 60% of the respondents have taken at least one college level genetics course, and over 75% indicated that they had participated in some sort of informal genetics education, such as a seminar or conference. Also, over half of the respondents indicated that they were aware of several genetic disorders.
However, the genetics knowledge of the participants was hard to assess because participants were only asked to indicate whether or not they had participated in genetics education courses. They were not quizzed on genetics information prior to the development of the genetics education tool, or even asked what they felt their level of genetics knowledge is. Asking participants to list the genetic diseases they were aware of also did not necessarily assess their knowledge of genetics. Many of the diseases that were listed included diagnoses of students in the Perkins School for the Blind Secondary Program.

**Disease and Tool Interest**

Responses from the tool pre-development survey were used to assess which diseases the non-medical staff of Perkins School for the Blind Secondary Program was most interested in learning about, as well as determining what the most effective education tool would be. Diseases that were selected to be included in the tool were chosen based on the average interest score the disease received, as well as the percentage of respondents who indicated their level of interest in the disease as “5- very interested”. Eight diseases received an average interest score of 3.8 or higher, and seven of these diseases had 40% or more of respondents indicate an interest of 5. Neuropathy, ataxia and retinitis pigmentosa (NARP) was not included in either seminar even though the interest score was above 3.8 since it did not also have 40% or more of respondents indicating an interest level of 5. Syndromic and non-syndromic retinitis pigmentosa was combined into one category for the seminars in order to present on the most material possible. The final six diseases selected for presentation based on the participants responses were Bardet-Biedl syndrome, Batten syndrome, Leber congenital amaurosis,
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

retinitis pigmentosa (syndromic and non-syndromic), retinoblastoma, and septo-optic dysplasia. These six diseases are common genetic diagnoses in students in the Perkins School for the Blind Secondary Program.

It was important to allow the non-medical staff of Perkins School for the Blind Secondary Program choose the education tool that we developed and presented. Based on the overwhelming number of respondents choosing a live seminar versus any other option proposed to them, this was the tool we developed. Two 50 minute seminars were presented so that there was sufficient time to present information on all six diseases.

Satisfaction

Comments provided by seminar attendees following the first seminar indicated that the participants were pleased with the presentation information, one participant stating that the presentation was “wonderful.” However, most of the comments made suggestions for improving the presentation style, or for other information to be included in the presentation.

Satisfaction following the second seminar was improved over the satisfaction following the first seminar. None of the comments indicated a desire to improve any aspect of the seminar in the future. All of the comments indicated that the seminar attendees were satisfied with the information presented to them. One attendee noted that the seminar “was good, logical, and organized,” and was appreciative that we had developed an education tool for the non-medical staff of Perkins School for the Blind Secondary Program.

Since we had the benefit of seeing the comments from the first seminar prior to presenting the second seminar, this likely had much to do with the increase in
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

satisfaction. Information that was lacking in the first seminar was included in the second. The presenter was also able to improve her presentation style based on the suggestions made by attendees following the first seminar.

Effectiveness

Effectiveness scores from the first post-development survey to the second post-development survey increased. Fewer participants indicated that they “strongly agreed” with the first two effectiveness statements following the second seminar, however the combined number of participants who “agreed” and “strongly agreed” increased nearly 10%. Those who “agreed” or “strongly agreed” with the third effectiveness statement, that the information presented would affect the care or instruction they provided students, increased by over 25% from the first seminar to the second seminar.

Several factors could have caused this change. We were able to edit the information presented in the second seminar based on audience feedback provided following the first seminar. We also presented on different diseases in each seminar, and interest level and relevance to the attendees may have been different for the diseases presented on at each seminar.

Overall, participants scored well on the quizzes. A large majority of participants answered the questions correctly on each quiz. However, only one of eight total questions (from both quizzes) had 100% of respondents answer it correctly. We believe that the high percentage of correctly answered questions is indicative of the tool effectively providing a genetics education to the non-medical staff of Perkins School for the Blind Secondary Program.

Limitations
Two program nurses responded to the tool pre-development survey, however, the tool post-development surveys did not ask about the role in which participants identified. It is unknown if nurses attended the seminars. Based on the number of nurses employed by Perkins School for the Blind Secondary Program, and our lack of knowledge as to whether the nurses attended the seminars, it was impossible to compare their responses to those of the non-medical staff.

The entire tool pre-development survey was visible to participants when they accessed it online through Survey Monkey (www.surveymonkey.com). It is possible that participants looked ahead at the survey before completing it, and therefore were made aware of many genetic diseases before responding to the first question, which may have significantly skewed those results.

49 people chose to participate in the tool pre-development survey, while only 23 and 20 responded to the first and second tool post-development surveys, respectively. It was our hope that all those who participated in the pre-development survey would also attend the seminars. Less than half of those who initially responded were able to attend a seminar/fill out a post-development survey. Seminars were scheduled at 9:45am which is a built in meeting time for non-residential staff. This is a students’ break time and residential staff are assigned to be with students during at this time. Therefore they were not likely able to attend the seminars. The small sample size for all three surveys may also not allow for an accurate representation of the non-medical staff of Perkins School for the Blind Secondary Program. Also, because we presented two seminars, and a different number of people filled out post-development surveys, we can assume that not all of the same people attended each of the seminars.
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

We used the quizzes to evaluate the effectiveness of the seminars as a genetics education tool. As part of the tool, we provided the attendees with fact sheets on each of the diseases, which were handed out at before the presentations began. The fact sheets contained the information needed to correctly answer the quiz questions, so it is possible that participants looked up some or all of the answers.

V. CONCLUSION

The results of this study suggest that the non-medical staff of Perkins School for the Blind Secondary Program have an interest in an increased genetics education. The results also suggest that this cohort of staff members intend to use the knowledge they have gained from the seminars in the care and instruction they provide the students they work with. Participants in the post-development surveys mostly indicated that they believe the knowledge they gained from the seminars is useful and will improve the care they provide to the students they interact with. It also appears that our tool effectively taught the Perkins staff about the genetic causes of the disease presented on, as we intended.

Non-medical staff who work with children in settings similar to Perkins would benefit from a similar education tool if it were available to them. Genetic counselors are in the unique position to provide this education, based on our knowledge base and access to resources.

The development of our genetics education tool and associated surveys was able to provide evidence that the staff feels that genetics education is useful, and may help to improve the care and instruction provided to secondary students. However, it was unable
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program

Principal Investigator: Sarah Casner

to answer definitively that the non-medical staff of Perkins School for the Blind Secondary Program will implement the genetics knowledge they gained into their curricula or care for students. It would be interesting to follow up with the non-medical staff of Perkins School for the Blind in the future to see if not only the intent to use the genetics education was there, but whether or not those who attended the seminars actually use the knowledge to improve the care and instruction of their students.
REFERENCES


Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner


Appendix A: Recruitment Notice

Are you interested in learning more about the genetic diagnoses of the students in Perkins Secondary program?

I am a graduate student in the Genetic Counseling Program at Brandeis University. Last fall, as part of my Master’s work, I had the opportunity to spend 9 weeks shadowing Linde D’Andrea. For my thesis, a requirement for the completion of a Master’s of Science Degree, I am developing a genetics education tool specifically for the non-medical staff of Perkins Secondary Program. The goal of my project is to provide Perkins School for the Blind Secondary Program with a useful tool that can be used year after year and will not only benefit the staff, but also the students they work with.

In order to provide the Secondary Program staff with the most effective genetics education tool I am asking that you participate in a brief online survey. Participation is voluntary, anonymous, and will require about 15 minutes of your time.

If interested please access the following website:

https://www.surveymonkey.com/s/895HYNL

This survey has been approved by the Brandeis University Institutional Review Board (IRB).

If you have any questions or comments, please feel free to contact Sarah Casner at casnersa@brandeis.edu or Linde D’Andrea at linde.d’andrea@perkins.org.

Thank you for your participation!
Appendix B: Seminar Recruitment Flyers

Secondary Program Inservice Training

Introduction to Genetics-Part I

Presented by
Sarah Casner
Genetic Counseling Student
Brandeis University
(Perkins advisor-Linde D'Andrea)
on
Thursday, March 18, 2010
at 9:45 am in Rm 11W

Secondary Program Inservice Training

Introduction to Genetics-Part II

Presented by
Sarah Casner
Genetic Counseling Student
Brandeis University
(Perkins advisor-Linde D'Andrea)
on
Thursday, April 8, 2010
at 9:45 am in Rm 11W
Appendix C: Pre-Development Survey Document

1) Please list any genetic disorders that you are aware of:
   2) Please indicate your level of interest in learning about the following disorders (1- not at all interested; 5- very interested) **Likert Scale Questions**
      a. Alstrom syndrome  1  2  3  4  5
      b. Aniridia (syndromic)  1  2  3  4  5
      c. Aniridia (non-syndromic)  1  2  3  4  5
      d. Bardet-Biedl syndrome  1  2  3  4  5
      e. Batten syndrome (NCLs)  1  2  3  4  5
      f. CHARGE syndrome  1  2  3  4  5
      g. Choroideremia  1  2  3  4  5
      h. Congenital cataracts  1  2  3  4  5
      i. Leber congenital amaurosis  1  2  3  4  5
      j. Leber hereditary optic neuropathy (LHON)  1  2  3  4  5
      k. Lenz microphthalmia syndrome  1  2  3  4  5
      l. Lowe syndrome  1  2  3  4  5
      m. Neurofibromatosis 1 (NF1)  1  2  3  4  5
      n. Neuropathy, ataxia and retinitis pigmentosa (NARP)  1  2  3  4  5
      o. Norrie disease  1  2  3  4  5
      p. Oculocutaneous albinism  1  2  3  4  5
      q. Oculofaciocardioidal syndrome (OFCD)  1  2  3  4  5
      r. Retinitis pigmentosa (syndromic)  1  2  3  4  5
      s. Retinitis pigmentosa (non-syndromic)  1  2  3  4  5
      t. Retinoblastoma  1  2  3  4  5
      u. Septo-optic dysplasia (SOD or Morsier syndrome)  1  2  3  4  5
      v. Stargardt disease  1  2  3  4  5
      w. Stickler syndrome  1  2  3  4  5
      x. Usher syndrome  1  2  3  4  5

3) What education tool would you find most useful?
   a. Online video (with descriptive features for the visually impaired)
   b. Book (made available in Print, Large Print, Braille, and on audio disc compatible with screen reader software and other technology used by blind/VI)
   c. Pamphlets/fact sheets (made available in Print, Large Print, Braille and on audio disc compatible with screen reader software and other technology used by Blind/VI)
   d. Live seminar lecture (with hand-outs available in print, large print and Braille; tactile models if available to represent various visual concepts)
   e. Other (please specify) __________________

4) Please select your gender
   a. Male
   b. Female

5) What age group do you belong to?
   a. 18-25
b. 26-35
c. 36-45
d. 46-55
e. 55+

6) What best describes your role in the Perkins Secondary Program?
   a. Administrative Staff/Administrative Support Staff
   b. Teacher
   c. Clinician
   d. CRL/Assistant CRL
   e. Program Nurse
   f. Volunteer
   g. Other __________

7) How many years have you worked at Perkins?
   a. <1 year
   b. 1-5 years
   c. 6-10 years
   d. 11-15 years
   e. >15 years

8) What is your highest degree?
   a. Some high school
   b. High school diploma
   c. Bachelor’s
   d. Master’s
   e. Master’s plus certification (i.e. CAGS)
   f. M.D. Ed. D., Ph.D., J.D.
   g. Other __________

9) How many college level courses that included genetics (biological information dealing with heredity and variation in organisms) in the curriculum have you taken?
   a. 0
   b. 1
   c. 2
   d. 3
   e. 4+

10) Have you ever taken part in any informal genetics education (i.e. attended an in-service training or conference that taught genetic information, etc.)?
    a. Yes
    b. No
Appendix D: Post-Development Survey Document 1

Introduction to Genetics Part 1
Sarah Casner

Evaluation

Please list your employee ID number ____________________

Please use the following scale to address statements #1, #2, and #3 by circling the letter that represents your response.

1) I gained new knowledge from this presentation.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

2) The information from the presentation was useful to me.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

3) The information I obtained from the presentation will affect or change the care/instruction I give to my students.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

4) What suggestions do you have to improve the presentation?

Quiz
(circle the letter that represents your response)

5) How many pairs of chromosomes are in the human genome?
   a. 20
   b. 23
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

c. 40
d. 46

6) Men and women are equally affected in autosomal recessive diseases.
   a. True
   b. False

7) What is the inheritance pattern of Bardet-Biedl syndrome?
   a. Autosomal dominant
   b. Autosomal recessive
   c. X-linked dominant
   d. X-linked recessive
   e. Mitochondrial

8) What symptom is characteristic of Batten disease?
   a. Polydactyly
   b. Seizures
   c. Autism
   d. Keratoconus
   e. Keratoglobus

9) Mutations in how many genes and/or loci are causative of Leber congenital amaurosis?
   a. 1
   b. 5
   c. 11
   d. 14

Thank you for your time and participation in this presentation! Your feedback is greatly appreciated!
Appendix E: Post-Development Survey Document 2

Introduction to Genetics Part 2
Sarah Casner

Evaluation

Please list your employee ID number ____________________

Please use the following scale to address statements #1, #2, and #3 by circling the letter that represents your response.

1) I gained new knowledge from this presentation.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

2) The information from the presentation was useful to me.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

3) The information I obtained from the presentation will affect or change the care/instruction I give to my students.
   a. Strongly disagree
   b. Disagree
   c. Agree
   d. Strongly agree

4) What suggestions do you have to improve the presentation?

Quiz
(circle the letter that represents your response)

5) Mutations in how many genes are known to cause retinitis pigmentosa?
   a. 16
   b. 25
6) Which of the following is a common sign of retinoblastoma?
   a. “Red eye” when a picture is taken
   b. Eye rubbing
   c. “White eye” when a picture is taken
   d. Nail biting

7) Which of the following is NOT known to be an inheritance pattern in septo-optic dysplasia?
   a. Sporadic
   b. Mitochondrial
   c. Autosomal dominant
   d. Autosomal recessive

Thank you for your time and participation in this presentation! Your feedback is greatly appreciated!
Appendix F: Information Sheet Handouts

Basic Genetics Information Sheet

DNA
- Stores our genetic information
- Made up of four different bases
  - Adenosine pairs with Thymine
  - Cytosine pairs with Guanine
- Forms a double helix
- Replicates itself
- Two different types of DNA in humans
  - Nuclear and mitochondrial

Chromosomes
- DNA in humans is packaged into 23 chromosome pairs for a total of 46
  - 22 pairs of autosomes and 1 pair of sex chromosomes (X and Y)
- Have two arms
  - p arm is the short arm
  - q arm is the long arm

Genes
- There are approximately 25,000-30,000 genes in the human genome
- Come in pairs, called alleles

Inheritance Patterns
- Autosomal Dominant
  - Males and females affected equally
  - Offspring and siblings of a person who is affected have a 50% chance to also be affected
- Autosomal Recessive
  - Males and females affected equally
  - Offspring of two carriers have a 25% chance to be affected, 50% chance to be unaffected carriers, and 25% chance to be unaffected non-carriers
- X-Linked Dominant
  - Affected women have a 50% chance to have affected children
  - Affected men will have 100% affected daughters, and no affected sons
  - Many times lethal in males
- X-Linked Recessive
  - Daughters of unaffected female carriers have a 50% chance to also be a carrier
  - Sons of unaffected female carriers have a 50% chance to be affected
  - Daughters of affected males are obligate carriers
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

- Sons of affected males will not be affected
- Mitochondrial Inheritance
  - Passed from a mother to all of her offspring; No paternal transmission
  - Caused by mutations in mitochondrial DNA
- Multifactorial
  - No true inheritance pattern seen; appears to run in families
  - Believed to be a combination of both environment and genetic predisposition

For additional information see http://ghr.nlm.nih.gov/

Bardet-Biedl Syndrome Information Sheet

Background Information
- Rare, autosomal recessive disorder caused by mutations in multiple genes
- Described independently by Bardet (French) and Biedl (German) in the 1920s
- Used to be considered part of Laurence Moon syndrome
  - Distinctive by presence of polydactyly

Characteristics
- Rod-cone dystrophy (>90%)
- Obesity (72%)
- Postaxial polydactyly
- Cognitive impairment
- Male hypogonadism
- Complex female genitourinary abnormalities
- Renal abnormalities

Gene Information
- BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, TRIM32/BBS11, BBS12, MKS1/BBS13, CEP290/BBS14

Diagnosis
- Molecular diagnosis
  - Molecular genetic testing is only available on a clinical basis for BBS1, BBS2, BBS10, and CEP290 BBS 14.
  - Research testing for the other loci
  - 20% of individuals with a clinical diagnosis of BBS do not have identifiable mutations in any of the 14 identified loci
- Clinical diagnosis
  - Presence of four primary features or three primary features and two secondary features
    - Primary features
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

- Rod-cone dystrophy, Postaxial polydactyly, Obesity, Learning disabilities (2º as well), Genitourinary malformations, Renal anomalies
  - Secondary features
    - Speech delay/disorder, Developmental delay, Behavioral abnormalities, Strabismus/cataracts/astigmatism, Brachydactyly/syndactyly, Ataxia/poor coordination/imbalance, Mild hypertonia, Diabetes mellitus, Dental crowding/hypodontia/small dental-roots/high arched palate, Cardiovascular anomalies, Hepatic involvement, Craniofacial dysmorphism, Hirschsprung disease, Anosmia

Treatment
- Management
  - Visual aids
  - Diet, exercise, behavioral therapies
  - Early intervention; special education
  - Renal transplantation
  - Surgery to correct genitourinary and cardiac malformations as well as to remove accessory digits
  - AVOID things contraindicated in persons with renal disease
- Surveillance
  - Regular eye exams
  - Monitoring of renal function
  - Screening for diabetes mellitus
  - Annual blood pressure screening

For additional information please see

Batten Syndrome Information Sheet

Background Information
- Batten Disease is a rare, hereditary, single-gene disorder
- First described by Batten, a British pediatrician in 1903
- Part of the NCL (neuronal ceroid lipofuscinoses) family
  - 4 main types of NCL; Infantile, Late Infantile, Juvenile, Adult.
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

- Most common form (Juvenile) also known as Spielmeyer-Vogt-Sjogren-Batten Disease.
- Occurs in 2 to 4 of every 100,000 live births in the United States.
- More common in Finland, Sweden, other parts of northern Europe, and Newfoundland, Canada.

Characteristics
- Personality and behavior changes
- Developmental delay
- Clumsiness, stumbling
- Seizures
- Vision loss

Gene Information
- CLN3 is the single gene identified as causing Juvenile Batten Disease.
  - CLN1 causes Infantile; CLN2 causes Late Infantile.
- The protein that CLN3 codes for has not yet been identified.
  - CLN2 is known to code for an acid protease, an enzyme that hydrolyzes (breaks down) proteins.
- CLN3 has been localized to the p arm of chromosome 16

Diagnosis
- Molecular diagnosis
  - In families where the mutation in the gene for CLN3 is known, DNA analysis can be used to confirm the diagnosis or for the prenatal diagnosis of Batten disease.
- Clinical diagnosis
  - Urine tests to detect elevated levels of dolichol
  - Skin and tissue biopsy for NCL deposits
  - Electroencephalogram (EEG) to detect seizures
  - Electrical studies of the eyes to detect characteristic eye changes
  - CT and MRI studies to detect decaying areas of the brain

Treatment
- Seizures controlled by anticonvulsant drugs.
- Physical Therapy
- Occupational Therapy
- Some reports suggest that diets high in vitamins C and E and low in vitamin A slow the progression of the disease, but do not affect the fatal outcome.

For additional information please see

40
Leber Congenital Amaurosis Information Sheet

Background Information
- First described in 1869 by Theodor Leber
- Considered to be the earliest and most severe form of retinal degeneration
- Accounts for at least 5% of inherited retinal degeneration
- Up to 20% of children in schools for the blind are affected with LCA
- Still, it’s rare, affecting approximated 3/100,000 liveborn children

Gene Information
- **CRX, CRB1, GUCY2D, AIPL1, RDH12, RPRG1P1, RPE65, CEP290, SPATA7, LCA5, RD3, TULP1, IMPDH1**
- **LCA9**
  - Locus, no gene has been identified yet

Diagnosis
- Molecular diagnosis
  - Molecular genetic testing available for GUCY2D, RPE65, AIPL1, RPRG1P1, CRB1, CRX
    - ~50% detection rate
  - Molecular genetic testing on a research basis is available for RDH12 (~4% detection rate) and CEP290 (10-20% detection rate).
- Clinical diagnosis
  - Undetectable electroretinograms (ERG) early in the disease,
  - Severe vision loss within the first year of life, sensory nystagmus,
  - Amaurotic pupils (pupils that do not respond to light),
  - Pigmentary retinopathy in the fundal portion of the eye
  - Family history

Secondary characteristics
- Optic disc oedema
- Keratoconus, Keratoglobus
- Photophobia
- Intellectual disabilities
- Autism
- Hypotonia

Treatment
- Visual aids
- Early intervention/special education
- Discourage touching of the eyes
- Gene therapy
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program  
Principal Investigator: Sarah Casner

For additional information please see

Retinitis Pigmentosa Information Sheet

Background Information
- Group of diseases causing retinal degeneration
- Photoreceptor cells (rods and cones) die, which causes the decline in vision

Clinical course of RP
- Typically the first symptom is night blindness
- Decreased peripheral vision
- Loss of ability to discriminate color
- Most people with RP are legally blind by the age of 40, but this varies widely

Gene Information
- Over 50 genes
- Inheritance
  - Autosomal dominant
  - Autosomal recessive
  - X-linked
  - Digenic
  - Mitochondrial

Treatment
- Diets high in vitamin A and DHA
- Gene therapy
- Technologies for delivering therapeutic agents to rod and cone cells are being studied
- An implantable microchip to enhance retinal function is under development

Syndromes that RP is associated with
- Usher syndrome
- Leber congenital amaurosis
- Kearns-Sayre
- Bardet-Biedl

For additional information please see
Retinoblastoma Information Sheet

Background Information
- Malignant tumor of the retina
- Develops in early childhood
- May be unilateral or bilateral
- Retinoblastoma is diagnosed in 250 to 350 children per year in the United States
  - It accounts for about 4 percent of all cancers in children younger than 15 years
- Familial retinoblastoma is inherited in an autosomal dominant pattern

Symptoms
- A white glow in the eye
  - Versus “red eye” when a flash picture is taken
- White spots in the pupil
- Crossed eyes
- A red, painful eye
- Poor vision
- The iris may be a different color in each eye

Gene Information
- RB1
  - Tumor suppressor gene
  - Located on chromosome 13q14.2
  - Mutations within the gene or deletions of chromosome 13 involving RB1 cause retinoblastoma

Diagnosis
- An examination of the eye with dilation of the pupil
- A CT scan or MRI of the head to evaluate tumor and possible spread
- An ultrasound of the eye (head and eye echoencephalogram)

Treatment
- Laser surgery for small tumors
- Radiation and chemotherapy
- Removal of the eye

For additional information please see
Septo-optic Dysplasia Information Sheet

Background Information
- Also known as de Morsier’s syndrome or optic nerve hypoplasia
- First described by Reeves in 1941
  - de Morsier in 1956
- Incidence of 1/10,000 live births
- Associated with younger maternal age

Characteristics and symptoms
- Abnormal development of the optic disk
- Pituitary deficiencies
- Optic nerve hypoplasia
- Midline abnormalities of the brain
- Blindness in one or both eyes
- Pupil dilation in response to light
- Nystagmus
- Inward and outward deviation of the eyes
- Hypotonia
- Hormonal problems
- Seizures
- Jaundice
- Intellectual disabilities
- Extra and underdeveloped digits
- Hypoglycemia
- Short stature
- Diabetes
- Cerebral palsy
- Temperature instability
- Sensorineural hearing loss
- Sleep disturbances
- Precocious puberty

Gene Information
- **HESX1 gene**
  - Located on chromosome 3p21.2-p21.1
  - Autosomal dominant, autosomal recessive and sporadic inheritance
- **SOX2 gene**
  - Located on chromosome 3q26.3-q27
  - Only in sporadic cases
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

Diagnosis
- Clinical diagnosis made when at least 2/3 features present
  o Optic nerve hypoplasia
  o Hypopituitarism with pituitary hypoplasia
  o Midline forebrain defects
- CT or MRI scans
- Genetic diagnosis made in <1% of all cases

Treatment
- Symptom based
  o Hormone replacement therapy for hormone deficiencies
  o Diet control for diabetes
  o Vision, physical and occupational therapies

For additional information please see

Glossary of Terms 1

Affected – Displays symptoms of a disease or syndrome

Brachydactyly – Short fingers or toes

Carrier – An unaffected person who bearing an unexpressed, deleterious gene

Dolichol - refers to any of a group of long-chain mostly unsaturated organic compounds which are made up of varying numbers of isoprene units terminating in an α-saturated isoprenoid group, containing an alcohol functional group

Dysmorphism – an anatomical malformation

Hypodontia – Additional teeth

Hydrolyze – to break apart a substance by the addition of water

Locus (plural Loci) – The location of a gene on a chromosome

Mutation – a permanent, heritable change in a gene or chromosome

Polydactyly – Extra fingers or toes

Surveillance – close observation
Development of a Genetics Education Tool for the Non-medical Staff of Perkins School for the Blind Secondary Program
Principal Investigator: Sarah Casner

Syndactyly – Webbed fingers or toes

http://www.biology-online.org/
http://medical-dictionary.thefreedictionary.com

Glossary of Terms 2

Affected – Displays symptoms of a disease or syndrome
Bilateral – Affecting left and right sides of the body
Carrier – An unaffected person who bearing an unexpressed, deleterious gene
Corpus callosum – structure that connects the left and right hemispheres of the brain
Digenic - A term used for characters or traits controlled by the integrated action of two genes
DHA – Docosohexanoic acid; omega-3 fatty acid
Dysmorphism – An anatomical malformation
Hypoglycemia – low blood sugar
Hypoplasia – Underdevelopment or incomplete development of a tissue or organ
Jaundice – Yellowing of the skin
Mutation – A permanent, heritable change in a gene or chromosome
Nystagmus – Rapid, involuntary to-and-fro movement of the eyes
Septum pellucidum – structure of the brain that separates the lateral ventricles
Sporadic – occurring occasionally, singly, or in irregular or random instances
Unilateral – Affecting one side of the body

http://www.biology-online.org/
http://medical-dictionary.thefreedictionary.com
http://www.merriam-webster.com/dictionary/