ACKNOWLEDGEMENTS

I would like to thank my committee, Judith Tsipis, Tomi Toler and Kelly Jackson for their help and guidance. Thank you to Margarita Corral for her assistance with data analysis. Many thanks to Judith Tsipis, Gretchen Schneider, Gayun Chan-Smutko and Missy Goldberg and the Brandeis University Genetic Counseling Faculty for their guidance and support. Thanks to my classmates for support and encouragement over the last two years, and a huge thank you to my husband, Kevin, without whom becoming a genetic counselor would have remained only a thought.
Parent to Parent, Peer to Peer: An Investigation of Mutual Social Support Groups as a Resource for Genetic Counselors

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Jodie Huff

Finding appropriate support for families facing issues surrounding a child with a genetic condition, whether diagnosed or not, is an important part of what pediatric genetic counselors do. We sought to investigate how pediatric genetic counselors utilize referrals to social support for families with a genetic diagnosis, how counselors determine which groups are most appropriate for a given family and whether counselors refer patients on a diagnostic odyssey to comparable support groups to help these families cope. We distributed an online survey to members of NSGC through an NSGC eblast. We geared our recruitment to current pediatric genetic counselors and those who have practiced within the last 10 years. One hundred thirty six pediatric genetic counselors responded to our survey. We found 95% of our respondents refer families with a genetic diagnosis to parent support groups at least some of the time. Counselors determine which type of support group to recommend to families based largely on the individual family’s preference, the family’s access to the internet, and the availability of a suitable support group near the family’s home. Over half (54%) of the counselors indicate that they do not refer families who are on a diagnostic odyssey to support groups, and a portion of these indicate that
they know of no groups to which to refer these families. Of those who do refer, many refer them to groups for the “the medical problems the child is having… such as autism groups, epilepsy groups, etc.” Referral to a support group for parents/families of children with genetic conditions remains an important way counselors facilitate emotional support for families, putting them in touch with those who can offer them support outside the session.

Keywords: genetic counseling, support groups, pediatric, diagnostic odyssey
# Table of Contents

List of Figures.............................................................................................................. vii

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

Methods....................................................................................................................... 5

  Study Design............................................................................................................. 5
  Recruitment.............................................................................................................. 5
  Data Collection and Analysis..................................................................................... 5

Results.......................................................................................................................... 7

  Demographics.......................................................................................................... 7
  Referral to Support Groups........................................................................................ 8
  Impact of Internet on Referral Patterns to Support Groups........................................ 8
  Factors that Affect Referral Patterns to Support Groups........................................... 11

Discussion................................................................................................................... 17

Future Research......................................................................................................... 20
Limitations........................................................................................................20

Conclusions....................................................................................................22

References.......................................................................................................23

Appendices.....................................................................................................25

Appendix A. Recruitment Notice.................................................................25

Appendix B. Survey........................................................................................26
List of Figures

Figure 1. Respondents by Region.................................................................13

Figure 2. Pediatric Counseling/Years of Experience..................................... 14

Figure 3. Support Group Referral..............................................................15

Figure 4. How Counselors Find Out About Support Groups......................16
Introduction

Referring families to parental social support groups is a fundamental component of the education and support that pediatric genetic counselors can offer to their patients. Indeed, as stated in A Guide to Genetic Counseling (Uhlmann, Schuette, and Yashar 2011),

Information and resources constitute one of the most critical needs individuals, parents, and/or families of a child or family member newly diagnosed with a birth defect or chronic medical or genetic condition. For some individuals and families, the need to seek as much information as possible and network with other families can be intense and immediate……

In fact, referring to support groups may be such an essential part of the genetic counseling profession that few studies, if any, have been done to determine what percentage of genetic counselors actually do refer to support groups on a regular basis and/or how genetic counselors find out about them.

We can define social support as the sum of what Holt-Lunstad and Uchino term the “functional aspects of relationship processes.” In other words, social support is a name for the particular functions served by social relationships (Holt-Lunstad and Uchino 2015). Individuals who are supportive can provide or offer emotional, informational, tangible, and/or belonging support (Holt-Lunstad and Uchino 2015).

The emotional aspect of social support can be an important element for families of children diagnosed with a genetic condition. The period after diagnosis can be a time of
uncertainty as parents may be unsure what the diagnosis means for the family as well as the child’s future (Kerr and McIntosh 2000). Aside from the stress of maximizing the health and wellbeing of the child with the condition, families often face “the deterioration of family structure” (Diehl, Moffitt, and Wade 1991). As emotional support involves expressions of caring (Holt-Lunstad and Uchino 2015), and/or understanding, expression of such support can aid a family as it transitions and accommodates to a “new normal.” In addition, emotional support can facilitate the grieving process after the loss of a child. Kriecbergs et al. reported that parents who had shared their problems with others during their child's illness and who had access to psychological support during the last months of their child's life were more likely to have worked through their grief several years later (Kreicbergs et al. 2007).

The informational aspect of social support is also a prominent component of social support. Parents of children with medical conditions often express concern about the lack of illness-specific information for transitioning from hospital to home. A parent who has been through the same type of transition and experience can offer valuable information and insights about how to coordinate a child’s care as an outpatient. For some, this is a greater need than emotional support. “Although emotional support is important, it is less so than finding out what others say about a medication or treatment, researching others’ knowledge and experiences, and learning how to manage a condition.” (Sarasohn-Kahn 2008)

Parents of newly diagnosed children also express concern about not knowing about all of the important medical equipment available for the child or whether they are using the equipment properly. Parents who have issues and difficulties in daily care for their child, like bathing, feeding or caring for a tracheostomy, may also benefit from sharing and learning from others (Diehl, Moffitt, and Wade 1991).
Tangible support addresses needs that fall under the “instrumental, practical or financial support” category (Holt-Lunstad and Uchino 2015), and may be one of the few types of support for which support groups may only offer minimal assistance. They may be able to share the names of any local groups or agencies that can offer some assistance but in most cases tangible assistance, particularly financial, is more likely to come from family and/or close friends.

A final functional component to social support is belonging support. Belonging support can involve encouraging parents to participate in support groups or participate in associated support group functions (Cohen et al. 1985, Cutrona and Russell 1990). In other words, belonging support involves making families feel a part of a community. Some parents have reported feeling isolated as they dealt with their unique and stressful situation, one to which the parents of their child’s peers could not relate (Kerr and McIntosh 2000). Peer-to-peer, or mutual, social support groups give these parents a place in which they can relate and be related to, a place that lessens their feeling of isolation.

As evidenced by the preceding studies and examples, social support can help alleviate or modulate the stress that arises with the birth of a child with a genetic disorder or the diagnosis of a child with one. Additional research (Holt-Lunstad and Uchino 2015) indicates that mutual social support, or peer-to-peer support, in which parents of children with medical conditions support each other, is particularly beneficial in terms of emotional, informational and belonging supports. This type of peer-to-peer support can often serve as a resource that can directly affect parental mental health, which, in turn, can shape the family’s social and psychological well-being (Ireys et al. 2001).

While there has been a great deal of research on the benefits of support groups for families of children diagnosed with a genetic condition (Ainbinder et al. 1998, Benson et al.
1991, Fidika, Salewski, and Goldbeck 2013, Ireys et al. 2001, Konrad 2007, Solomon, Pistrang, and Barker 2001, Plumridge et al. 2012, Kerr and McIntosh 2000), many children with developmental delay, intellectual disabilities, and/or medical issues do not have a specific diagnosis. They undergo what is known as a diagnostic odyssey in the hope of identifying an underlying cause for the child’s issues. Immediate identification is not always possible. It can be difficult for these families in a different way than for those whose children have a diagnosis. Families with undiagnosed children may also need support and assistance but their lack of a diagnosis can make finding an appropriate peer-to-peer support group more difficult.

The purpose of this study is to investigate the area where genetic counselors and parental social support groups intersect: the referral of support groups to the family. We investigated how pediatric genetic counselors utilize referrals to social support for families with a genetic diagnosis, how counselors determine which groups are most appropriate for a given family, and whether counselors refer patients on the diagnostic odyssey to comparable support groups. In addition, we examined whether counselors are referring more to face-to-face or to online support groups, and whether the rise of online support groups, which “add an element of anonymity, ease of access, and openness” (Mehta and Atreja 2015) are as beneficial for families as face-to-face groups.
Methods

Study Design

We invited pediatric genetic counselors who currently practice pediatrics or who have practiced pediatrics within the last ten years to participate in an anonymous, online survey regarding parent support groups. Counselors were asked to provide information regarding several aspects of support group referral including whether or not the participant regularly offers referrals, the types of groups referred (face-to-face or online), and how, and if, they vetted groups before referring a patient to them. We asked questions to determine the impact of social media on how these groups are referred. In addition, we offered the participants the opportunity to comment on these and other aspects of support group referral.

Recruitment

The Brandeis University Institutional Review Board approved this study. We presented the invitation to participate through an eblast to all NSGC members on the NSGC listserv. The recruitment notice (Appendix A) included a link to the study, and participation in the study constituted consent. We sent a reminder eblast approximately one week after the initial eblast. The survey was available from June 23, 2015 until July 7, 2015. Participation was limited to current pediatric genetic counselors and those who have practiced pediatric genetic counseling within the last 10 years. We invited participants to enter a drawing for one of four $50 Amazon gift cards as an incentive to participants.
Data Collection and Analysis

We developed an online survey instrument using Qualtrics and we distributed the link to the instrument to members of the NSGC listerv. Our survey consisted of three groups of multiple-choice questions including a demographics section, a parent support group/referral section, and a set of questions regarding additional factors that could affect referrals. The parent support group/referral section covered items such as frequency of referral, types of referral, how appropriate support groups are located/learned of, and how the quality of such groups are assessed. The final section addressed potential factors that could alter the way one typically refers, including patients from rural areas or families who are on a “diagnostic odyssey.” A comment section followed several of the multiple-choice questions, and we included one open-ended question.

We analyzed quantitative data using SPSS 22. We analyzed the open-ended responses and comments using thematic analysis.
Results

We received a total of 136 responses. We removed 3 responses from our analysis due to incomplete responses for the majority of the questions in the survey. We based our analysis on responses from 133 genetic counselors who are currently or have practiced pediatric genetic counseling within the last 10 years.

DEMOGRAPHICS

Our demographic is comprised of 133 pediatric genetic counselors representing all of the NSGC regions. The majority of our respondents (74%) are currently pediatric genetic counselors while 26% no longer work in pediatrics but did so within the last 10 years. The largest group of respondents (38%, N=129) practice in NSGC Region 4, followed by 20% in Region 2 (See Figure 1). Over 86% (N=132) of counselors that answered our survey had at least one year of experience (See Figure 2). Almost half (45%, N=132) had 1-5 years’ experience, and 19% (N=132) had over 10 years’ experience.

A majority of the genetic counselors (58%, N=132) taking our survey either did or currently practice in a university medical center setting, followed by another 21% who practice in a private hospital or medical facility. The largest portion of respondents (67%, N=133) saw at least some of their pediatric patients in a specialty clinic; of these, 43% (N=87) reported seeing
1-20% of all pediatric patients in such a clinic while 23% saw 80-100% of their pediatric patients in a specialty clinic.

REFERRAL TO SUPPORT GROUPS

When asked to choose an answer that most closely relates to the statement “I offer/offered parents of children diagnosed with a medical condition a referral to a parent support group either online or face to face,” a majority of those that answered (85%, N=130) chose “almost always,” or “sometimes.” Another 10% indicated that they always refer to support groups, while only 5% indicated that they rarely refer (See Figure 3).

For the 129 counselors who do refer to support groups, the largest number of counselors (88%) indicated that the internet was a major source of information for them about parental support groups. Seventy-seven percent indicated that they hear about support groups through colleagues, and 56% noted that they hear about them from support group outreach through conferences, email blasts, and email (See Figure 4).

IMPACT OF INTERNET ON REFERRAL PATTERN TO SUPPORT GROUPS

The internet is a relatively new technology and provides individuals with new ways to connect with one another. Whether or not a family has internet access will influence and/or limit the types of support groups available to the family. We asked our respondents to indicate whether they referred families more to online support groups or more to face-to-face groups. A number of counselors (44%, N=123) answered that they were referring parents to a combination of both online and face-to-face support groups. Another 36% indicated that how they referred families to support groups depended upon the condition rather than whether a group was face-to-
face or online. “Even in urban areas,” wrote one counselor, “rare diseases do not have a local presence; therefore, online is really the easiest way to connect.”

Other counselors commented that they considered family preferences when they were referring to groups: “We let people know about the in-person opportunities to get together (LPA, OIF), but there are also online support groups that are quite active through social media and some people like to spend time lurking or participating actively in these forums as well.”

When asked to assess which type of group (online or face-to-face) was of better quality, a majority of respondents (77%, N=123) indicated that the quality of a particular support group depended more upon the specific group than on whether a group was face-to-face or online. Only 15% felt there was a difference between face-to-face and online, with 7% of the total indicating that face-to-face had stronger support and higher quality of information.

Online support can be accessed in a number of ways, ranging from disease-specific websites, to Facebook pages, to personal blogs and we asked our respondents to indicate which type(s) of online support they most often used when referring a family to an online support group. We invited them to choose as many answers as applied. Many of the genetic counselors that answered (N=123) referred to a support group’s webpage or social media site. Forty-three respondents referred parents to listservs, and 35 counselors referred to blogs. A few expressed concern about blogs:

“I tend to avoid blogs and small, privately-run support groups because I don’t necessarily feel as confident that they are providing accurate information about the condition. I have certainly come across parent blogs that offer inappropriate advice. If possible, I direct my patients to large, national non-profit groups after screening information myself.”
One hundred twenty-one of the 126 respondents who refer online sites review them before referring them to families. Of these, 119 review the sites for the quality of information available on the site, 85 look for the involvement of parent or family members of those with the condition, and 80 look for the involvement of a medical professional or institution.

Many of our respondents (96%, N=126) prepare patients that not all sites may be appropriate or suitable. A recurrent theme expressed by those who wrote comments was that they prepared families of patients by explaining the variation in phenotypes of many disorders and that the phenotypes shown or discussed on these sites might not represent how the disease or condition presents in the parents’ child. One counselor wrote, I “discuss directly that information online may be inaccurate, and that representation may be skewed toward families with children who are more severely affected, as those families often need the most support.” Another explained that she/he prepares families for internet groups by addressing

“the spectrum of the clinical picture and the fact that the entire spectrum will be represented in information and images on sites, and encourage them to contact us with any questions or for reassurance about what they may have read or seen.”

A second way counselors prepare families is that they remind the parents that much information on the internet can be inaccurate. “Some information on the internet is great but some is misleading,” commented one counselor. Another wrote,

“I warned them to be careful about Googling their condition and emphasized that things like ‘Mommy blogs’ can very dangerous. People with hard/bad stories often write about them, while people not struggling don’t. I encouraged my families to try to find websites with some sort of national affiliation…”

The increasing use of social media, particularly Facebook, adds a newer dimension to the online experience and a new way for people to connect directly. Several genetic counselors (37%, N=122) indicated that social media has not changed the way they refer to support groups,
while 52% (N=122) selected that social media has influenced it. “Facebook has been a great tool for connecting families,” wrote one counselor, “A family in Georgia can talk to a family in Ireland without leaving their living room!” One respondent indicated that she had to vet social media sites whereas she did not have to before the popularity of social media, as she knew that patients would visit the page at some point. Other counselors avoid referring to social media entirely: “I generally don’t refer patients to social media pages because I feel that they often become a place for people to complain about physicians, treatments, etc. and if often not appropriate for families at all”.

FACTORS THAT CAN AFFECT REFERRAL PATTERNS TO SUPPORT GROUPS

In attempting to better understand factors that can affect referral patterns to support groups we asked counselors to indicate whether their geographical catchment area influenced the way they currently refer. Thirty-two percent (N=122) indicated that their catchment area was an urban area and their patients had access to a full range of medical and support groups. Many others (50%, N=122) indicated that a significant percent of their patients reside in a rural area. We asked counselors if it was generally more difficult to refer rural patients to support groups. Slightly more than half (56%, N=117) of our respondents indicated that it was more difficult to find appropriate support group referrals in rural areas because of the lack of nearby face-to-face groups within a reasonable distance and lack of internet. Another 44% indicated that it was no more difficult to refer to support groups because of the existence of online support groups. Families that our respondents work with have internet access but not all; three percent (N=123) respondents said 21-40% of their families did not have access, while another 8% said that 41-60% of their families did not.
Coping with a newly diagnosed genetic condition in a child warrants a great deal of support as a family deals with adapting to a new normal. Having a child with multiple symptoms but no diagnosis makes obtaining support even harder. More than half of the counselors in our survey (54%, N=118) said that they did not refer families on a diagnostic odyssey to a support group. Man (43%, N=118) indicated that they knew of no groups that would be appropriate. Another 25% (N=118) indicated that they do refer to some type of support group most often based on “the medical problems the child is having… such as autism groups, epilepsy groups, etc.” Said another counselor, “There are many groups for broad categories such as developmental delay. It is not always needed to have a specific diagnosis to find people going through the same journey.”
Figure 1:

Respondents by Region

Region

1. CT, MA, NH, RI, CN, MARITIME PROVINCE
2. DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, QUEBEC
3. AL, FL, GA, KY, LA, MS, NC, SC, TN
4. AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI...
5. AZ, CO, MT, NM, TX, UT, WY, ALBERTA, MANITOBA...
6. AK, CA, HI, ID, NV, OR, WA, BRITISH COLUMBIA

Number of Respondents

0  5  10  15  20  25  30  35  40  45  50

N=129
Figure 2:

Pediatric Counseling

Years of Experience

N=132
Figure 3

Support Group Referral

- Always: 40%
- Almost Always: 45%
- Sometimes: 5%
- Rarely Refer: 10%

N=130
Discussion

This study is one of few, if any, studies to examine parental support groups as a resource for pediatric genetic counselors. We queried current and past pediatric genetic counselors on several aspects of support group referral, including whether they refer families of patients diagnosed with a genetic condition to these groups, whether they have a preference for face-to-face or online support groups, what differences, if any, they perceive between online and face-to-face support and if the increased presence of social media has changed the way they refer. We also asked if they made comparable referrals to support groups for families who did not yet have a diagnosis but were still on a diagnostic odyssey.

The most typical respondent to our survey was someone who is a current pediatric genetic counselor with 1-5 years’ experience. He/she works in a university medical setting, is affiliated with a specialty clinic and sees 1-20% of patients in the clinic. Given that referring families to appropriate resources to help them cope with the diagnosis of a genetic condition is embedded in our training as genetic counselors, this study yielded no surprises in that regard: 55% of respondents almost always or always refer, while 39% refer when they think it is appropriate. The most common ways counselors learn of appropriate genetic support groups is by an internet search. The two websites most frequently cited by respondents to locate parental support groups were Genetics Home Reference and Gene Reviews. Others included the websites for NORD (National Organization for Rare Disorders), Genetic Alliance, Disease Infosearch, and searching or posting on NSGC Special Interest Groups. While some national groups are well known throughout the profession (those for Down syndrome support, for example), good support
groups for some conditions may not exist while others vary in levels of visibility. As one counselor put it, “I would refer more frequently if the information about them were more accessible and unified in the GC profession.” Other ways counselors reported learning about support groups were through their colleagues and through patients. I “might find about the existence of a group from the internet,” said one respondent, “but from parents about how helpful it is or what the group’s focus is.”

The internet has made connecting with others easier. Many people and/or groups can and do employ social media or online forums and webpages to communicate, socialize and generally form communities. These online groups have had an impact on the ways in which people interact. Less face-to-face interaction and a greater degree of anonymity has made communicating easier and less intimidating. We wanted to see if the same dynamic applied specifically to parental support groups— and therefore changed the types of groups to which counselors refer. Since a majority of counselors reported that over 60% of their patients’ families had internet access, we anticipated that a significant portion of referrals would be directly to genetic support groups based online. In fact, approximately 44% of counselors said they now referred a combination of online and face-to-face groups, and a 36% indicated that they took the families’ preferences or circumstances into account before referring to an online group. As an example, one counselor worked with an urban population in which many patients had transportation issues and preferred referrals to online sources. Another counselor indicated that she saw families in a rural setting but in some cases, the families preferred face-to-face groups and would “drive 5 hours” to attend a group. Another counselor said, “Most of my patients are diagnosed with a rare condition. It does not matter if they live in a rural or urban setting—there probably is not another family with the same diagnosis close by!” Several respondents indicated that it is as difficult, if not more so,
to refer to support groups to those families who do not have internet access as to refer those who live in areas with no face-to-face support. The most difficult group of patients/families to refer are those with the combination of living in an isolated area and having no access to the internet. As the type of referral many counselors can offer depends upon the needs or circumstance of the family rather than whether it is an online group or face-to-face group, over 75% of our respondents indicated the quality of a support group is dependent on the internal dynamics of the group itself. The internet is a democratic medium and anyone can post online or develop a website. As one counselor indicates, “Caution them that these websites are no different than any other website. There is good/bad information.” Over 95% of the counselors, when they have referred online groups, have prepared patients for what types of information they might find on that website or others like them. This preparation includes explaining the spectrum of a phenotype and/or the various manifestations of a condition followed by an explanation about how each individual with the condition is unique. Many warn their patients that sometime the people who are having the most difficult time relative to the condition are the most likely to seek support and post online and often this is a representation of the most severe form of the condition. One counselor “explains factors to look for: last update on their website, used by other parents, content matching information from clinic, etc.”

A subset of families who likely need support may not be getting it. Families who are ill but are without a genetic diagnosis can be difficult to refer. Over half (54%) of the counselors who took this survey indicated that they do not refer these families, and a portion of those indicated that they knew of no groups to which to refer these families. Of those who do refer, many refer them to groups for the “the medical problems the child is having… such as autism
groups, epilepsy groups, etc.” Others referred to Syndrome Without A Name (SWAN), and one counselor mentioned Parent to Parent as a support group.

Emotional support in the counseling session and a referral to a support group outside the clinic are not the only way counselors facilitate support. Many genetic counselors (41%) themselves have volunteered in or are volunteering in outside support groups for those with genetic conditions. Offering support is not just written in our textbooks and mentioned in our classrooms. It is an important part of what we do—it is part of our professional identity and speaks to how we care about more than just the medical implications of a condition. Genetic counselors know firsthand how hard coping with, living with and adapting to a genetic condition can be for families, and we want to help our patients find appropriate sources of support.

**Future Research**

An in depth survey focusing on genetic counselors’ referral patterns and families undergoing the diagnostic odyssey would be an important first step in determining how often these families are not receiving referral. In addition, studies examining gaps in support/support groups for families who have a child diagnosed with particular genetic conditions relative to specific regions could be done, with the aim of filling these gaps.

**Limitations**

One limitation is that this study reached only those pediatric genetic counselors that subscribed to the NSGC listserv. We do not know if those who subscribe to the listserv have different experiences or opinions than the rest of the pediatric genetic counselor population. Secondly, we had only 133 respondents to our eblast. These represent a subset of all pediatric
genetic counselors on the NSGC listserv, and may be biased towards those most interested in genetic support groups. Finally, some of our respondents may have left the field of pediatrics 8-9 years ago, prior to the rise of the internet and social media. We do not know how many respondents fell into this category but their answers might potentially skew our results. While we believe that our research encompasses the experience of the general pediatric counseling population, we cannot assert as such.
Conclusions

Our study investigated the degree to which pediatric genetic counselors utilize referrals to peer-to-peer support groups for families with a genetic diagnosis, how counselors determine which groups are most appropriate for a given family, and whether counselors refer patients on the diagnostic odyssey to comparable support groups. We found that over 95% of our respondents refer families with a genetic diagnosis to parent support groups at least some of the time. Counselors determine which type of support group to refer to families based largely on the individual family’s preference, the family’s access to the internet, and the availability of a suitable support group near the family’s home. Almost all of the counselors who referred families to online support group’s sites and social media pages vetted these sources before referral. Counselors looked for the quality of information, involvement of parents or family of children with the condition, and the involvement of medical professionals before recommending these sites to families. Another finding is that our respondents were less likely to refer families on a diagnostic odyssey to support groups. Many of those counselors indicated that they were unaware of appropriate support groups for referral and were concerned that the family might receive misinformation.
References


Appendices

Appendix A: Recruitment notice

Are you currently or have you been a pediatric genetic counselor in the last 10 years?

I am a student in Brandeis University’s Genetic Counseling Program. I invite you to participate in a research study investigating the importance of family/parental support groups, otherwise known as mutual social support groups, as a resource for pediatric genetic counselors. One aim will be to examine how frequently pediatric genetic counselors refer these groups to parents of children with a genetic condition as well as to those still on the diagnostic odyssey. Additionally, we will seek to understand the role of these groups as they have evolved from face-to-face groups to those with an ever-increasing online presence.

Participation in this study is open to all pediatric genetic counselors who are currently practicing or who have practiced within the last 10 years in a pediatric genetic counseling setting. The study consists of an anonymous online survey that will take approximately 20-30 minutes to complete. Participation is voluntary, and you may discontinue at any point.

Upon completion of the survey, participants will have the option to have their name placed in a drawing to win one of four $50 Amazon gift cards.

If you are interested in participating in this survey, please follow the link below:

Link to survey

If you have any question or comments, please feel free to contact me by email at jhuff@brandeis.edu, or the Brandeis University faculty sponsor, Judith Tsipis, at tsipis@brandeis.edu. Thank you for your consideration.

Sincerely,

Jodie Huff
Brandeis University Genetic Counseling Program
Appendix B: Survey

Q1 Parent to Parent, Peer to Peer: An Investigation of Mutual Social Support Groups as a Resource for Genetic Counselors. My name is Jodie Huff, and I am a second year genetic counseling student at Brandeis University. Thank you for choosing to participate in my survey. The purpose of this study is to investigate the importance of parent support groups as a resource for pediatric genetic counselors. One aim will be to examine how frequently pediatric genetic counselors refer these groups to parents of children with a genetic condition as well as to those still on the diagnostic odyssey. Additionally, we will seek to understand the role of these groups as they have evolved from face-to-face groups to those with an ever-increasing online presence. Upon completion of the survey, you will have the option to enter a drawing to receive one of four $50 Amazon.com gift cards. This study has been approved by the Brandeis University Institutional Review Board. By clicking the forward button below, you are consenting to participate in this study.

Q2 Do you currently see pediatric patients as part of your clinical practice?

- Yes
- No

If No Is Selected, Then Skip To End of Block

Q3 Which NSGC region do you practice in?

- Region 1: CT, MA, NH, RI, VT, CN, Maritime Province
- Region 2: DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec
- Region 3: AL, FL, GA, KY, LA, MS, NC, SC, TN
- Region 4: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario
- Region 5: AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan
- Region 6: AK, CA, HI, ID, NV, OR, WA, British Columbia

Q4 How long have you been seeing pediatric patients as part of your practice?

- Less than a year
- 1-5 years
- 5-10 years
- Over 10 years
Q5 In which type of setting do you currently see pediatric patients?
- University medical center
- Private hospital or medical facility
- Public hospital or medical facility
- Private hospital or medical facility using telegenetics
- University medical center using telegenetics
- Public hospital or medical facility using telegenetics
- Other ____________________

Q6 Do you see some of your pediatric patients in a specialty clinic?
- Yes
- No

If No Is Selected, Then Skip To End of Block

Q7 What type(s) of specialty clinic(s) are you affiliated with?

Q8 In a typical month, what percentage of your pediatric patients do you see in a specialty clinic?
- 1-20%
- 21-40%
- 41-60%
- 61-80%
- 81-100%

Q9 Did you see pediatric patients as part of your clinical practice?
- Yes
- No

If No Is Selected, Then Skip To End of Survey

Q10 Which NSGC region did you practice in?
- Region 1: CT, MA, NH, RI, VT, CN, Maritime Province
- Region 2: DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec
- Region 3: AL, FL, GA, KY, LA, MS, NC, SC, TN
- Region 4: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario
- Region 5: AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan
- Region 6: AK, CA, HI, ID, NV, OR, WA, British Columbia
Q11 How long did you see pediatric patients as part of your practice?
- Less than a year
- 1-5 years
- 5-10 years
- Over 10 years

Q12 In which type of setting did you see pediatric patients?
- University medical center
- Private hospital or medical facility
- Public hospital or medical facility
- Private hospital or medical facility using telegenetics
- University medical center using telegenetics
- Public hospital or medical facility using telegenetics
- Other ____________________

Q13 Did you see some of your pediatric patients in a specialty clinic?
- Yes
- No
If No Is Selected, Then Skip To End of Block

Q14 What type(s) of specialty clinic(s) were you affiliated with?

Q15 In a typical month, what percentage of your pediatric patients did you see in a specialty clinic?
- 1-20%
- 21-40%
- 41-60%
- 61-80%
- 81-100%
Q16 Please choose the answer that most closely describes how you relate to this statement: I offered parents of children diagnosed with a genetic condition a referral to a parent support group (parent-to-parent or peer-to-peer support group) either online or face-to-face?

- Never
- Rarely
- Sometimes
- Almost always
- Always

Answer If

Q17 Your reasons for not referring parents of children with genetic conditions to a parent support group were: (Select all that apply)

- I don’t believe them to be helpful
- I am concerned that my patients may receive misinformation from them
- It isn’t the job of the genetic counselor
- There aren’t any good ones available for my patients
- Most of my patients; parents aren’t interested in becoming a part of parent support groups
- Other ____________________

If There aren’t any good o... Is Selected, Then Skip To End of Survey

Answer If

Q18 In what ways did you find out about the parent support groups that you referred to parents of children with genetic conditions? (Select all that apply)

- Internet search
- Support group outreach (conferences, email blasts, mail)
- Through colleagues
- I hear from my patients
- My institution runs at least one
- I run my own
- General word-of-mouth
- Other ____________________

Q19 Comment:
Q20 For the past several years, the internet has allowed families of children with genetic conditions to connect electronically. As such, access to the internet has become a factor in which support groups can be referred to. Can you estimate what percentage of your families had internet access?

- 0%
- 1-20%
- 21-40%
- 41-60%
- 61-80%
- 81-100%

Q21 When you referred a parent to a support group, did you refer primarily to face-to-face support groups, online support groups, or a combination of both?

- Primarily to face-to-face
- Primarily online
- A combination, but more face-to-face
- A combination, but more online
- Both equally
- It depends upon the condition. Some have excellent local face-to-face while others do not.
- Depends upon the family’s access to the internet

Q22 Comment:

Q23 Did you feel there was a difference in the quality of support offered by face-to-face parent support groups and the support offered by an online support group?

- Yes
- No
- It depends upon the specific group

If it depends upon the specific... Is Selected, Then Skip To To which types of online support grou...

Q24 Which did you feel offered stronger support/higher quality of information?

- Online support groups
- Face-to-face support groups
- It depends upon the specific group
Q25 To which types of online support groups did you refer parents of children with genetic conditions to? (Select all that apply)

- Blogs
- Listservs
- Support group webpages/social media sites
- Other ____________________

Q26 Comment:

Q27 Did you generally review the sites before you referred your families to them?

- Yes
- No

If No is Selected, Then Skip To Do you prepare your patients and/or...

Q28 What characteristics did you look for in your review of online sites to determine whether or not you would refer a parent to a particular online support site? (Select all that apply)

- Involvement of a medical professional/institution
- Involvement of parents or family members of those with the condition
- Quality of information available on the site
- Other ____________________

Q29 Comment:

Q30 Did you prepare your patients and/or parents that not all sites were appropriate or suitable?

- Yes
- No

If No is Selected, Then Skip To How has social media (e.g., Facebook)...

Q31 How did you prepare your patients and/or parents?
Q32 How did social media (e.g., Facebook) influence the way you refer to parent support groups?

- I referred parents of my patients to social media pages (e.g., Facebook) of particular support groups so that they could have immediate, direct access to other parents of children with the same condition
- Social media didn’t influence the way I referred to parent groups
- Other ____________________

Q33 Comment:

Q34 Did your geographical catchment area influence the way you referred to parent support groups?

- It did not since all of my patients lived in an urban area with a full range of medical and support groups available
- Yes, a significant percent of my patients resided in what was considered to be a rural area
- Other ____________________

If Yes, a significant percent ... Is Selected, Then Skip To What constitutes an area or region as...

Q35 What constituted an area or region as rural to you/your institution?

- Not proximal to a regional medical center
- Relatively isolated area/low population density
- Combination of A and B
- Other ____________________

Q36 Did you find it more difficult to refer to appropriate support groups for parents who lived in rural areas?

- No, most parents in the rural areas around my institution had access to the internet and therefore had access to many support groups
- Yes, most people did not have internet access
- Yes, because the availability of face-to-face support groups was limited, if not non-existent in rural areas, and therefore limited the overall options for those in these areas even if they had internet access

Q37 Comment:

Q38 Many children with developmental delay, intellectual disabilities, and/or medical issues are referred to a pediatric genetics clinic in the hope of identifying an underlying reason for these issues. Immediate identification is not always possible. The ensuing pursuit is known as the diagnostic odyssey and can be
very difficult for families. Did you refer families of children who do not have a diagnosis, or were still on a “diagnostic odyssey” to parent support groups?

- No, I didn’t believe it would be helpful
- No, I didn’t know of any groups that would be appropriate
- No, I worried that they would receive information that was confusing/irrelevant for them
- Yes, I did refer to some type of support group (online or face-to-face) Please provide a brief description of those that you tended to refer to: ____________________
- It depended on the age of the child/severity of condition

Q39 Did you volunteer or participate in (in your role as a professional) any parent support groups in person or online?

- Yes
- No

Q40 Please describe briefly:

Q41 Please choose the answer that most closely describes how you relate to this statement: I offer parents of children diagnosed with a genetic condition a referral to a parent support group (parent-to-parent or peer-to-peer support group) either online or face-to-face?

- Never
- Rarely
- Sometimes
- Almost always
- Always

If Always Is Selected, Then Skip To In what ways do you find out about t...

Answer If Please choose the answer that most closely describes how you relate to this statement: I offer parents of children diagnosed with a genetic condition a referral to a parent support group Is Selected
Or Please choose the answer that most closely describes how you relate to this statement: I offer parents of children diagnosed with a genetic condition a referral to a parent support group Is Selected

Q42 Your reasons for not referring parents of children with genetic conditions to a parent support group are: (Select all that apply)

- I don’t believe them to be helpful
- I am concerned that my patients may receive misinformation from them
- It isn’t the job of the genetic counselor
- There aren’t any good ones available for my patients
- Most of my patients’ parents aren’t interested in becoming a part of parent support groups
- Other ____________________

If There aren’t any good o... Is Selected, Then Skip To End of Survey

Answer If Your reasons for not referring parents of children with genetic conditions to a parent support gr... Is Not Displayed

Q43 In what ways do you find out about the parent support groups that you refer to parents of children with genetic conditions? (Select all that apply)

- Internet search
- Support group outreach (conferences, email blasts, mail)
- Through colleagues
- I hear from my patients
- My institution runs at least one
- I run my own
- General word-of-mouth
- Other ____________________

Q44 Comment:

Q45 For the past several years, the internet has allowed families of children with genetic conditions to connect electronically. As such, access to the internet has become a factor in which support groups can be referred to. Can you estimate what percentage of your families have internet access?

- 0%
- 1-20%
- 21-40%
- 41-60%
- 61-80%
- 81-100%
Q46 When you refer a parent to a support group, do you now refer primarily to face-to-face support groups, online support groups, or a combination of both?

☐ Primarily to face-to-face
☐ Primarily online
☐ A combination, but more face-to-face
☐ A combination, but more online
☐ Both equally
☐ It depends upon the condition. Some have excellent local face-to-face while others do not.
☐ Depends upon family’s access to the internet

Q47 Comment:

Q48 Do you feel there is a difference in the quality of support offered by face-to-face parent support groups and the support offered by an online support group?

☐ Yes
☐ No
☐ It depends upon the specific group

If It depends upon the specific group is Selected, Then Skip To To which types of online support groups do you refer parents of children with genetic conditions to? (Select all that apply)

☐ Blogs
☐ Listservs
☐ Support group webpages/social media sites
☐ Other ____________________

Q49 Which do you feel offers stronger support/higher quality of information?

☐ Online support groups
☐ Face-to-face support groups
☐ It depends upon the specific group

Q50 To which types of online support groups do you refer parents of children with genetic conditions to? (Select all that apply)

☐ Blogs
☐ Listservs
☐ Support group webpages/social media sites
☐ Other ____________________

Q51 Comment:
Q52 Do you generally review the sites before you refer your families to them?

☐ Yes
☐ No

If No Is Selected, Then Skip To Do you prepare your patients and/ or ...

Q53 What characteristics do you look for in your review of online sites to determine whether or not you will refer a parent to a particular online support site? (Select all that apply)

☐ Involvement of a medical professional/institution
☐ Involvement of parents or family members of those with the condition
☐ Quality of information available on the site
☐ Other ____________________

Q54 Comment:

Q55 Do you prepare your patients and/or parents that not all sites are appropriate or suitable?

☐ Yes
☐ No

If No Is Selected, Then Skip To How has social media (e.g., Facebook)...

Q56 How do you prepare your patients and/or parents?

Q57 How has social media (e.g., Facebook) influenced the way you refer to parent support groups?

☐ I refer parents of my patients to social media pages (e.g., Facebook) of particular support groups so that they can have immediate, direct access to other parents of children with the same condition
☐ Social media hasn’t influenced the way I refer to parent groups
☐ Other ____________________

Q58 Comment:

Q59 Does your geographical catchment area influence the way you refer to parent support groups?

☐ It does not since all of my patients live in an urban area with a full range of medical and support groups available
☐ Yes, a significant percent of my patients reside in what is considered to be a rural area
☐ Other ____________________

If Yes, a significant percent ... Is Selected, Then Skip To What constitutes an area or region as...
Q60 What constitutes an area or region as rural to you/your institution?

- Not proximal to a regional medical center
- Relatively isolated area/low population density
- Combination of A and B
- Other ____________________

Q61 Do you find it more difficult to refer to appropriate support groups for parents who live in rural areas?

- No, most parents in the rural areas around my institution have access to the internet and therefore have access to many support groups
- Yes, most people do not have internet access
- Yes, because the availability of face-to-face support groups is limited, if not non-existent in rural areas, and therefore limits the overall options for those in these areas even if they have internet access

Q62 Comment:

Q63 Many children with development delay, intellectual disabilities, and/or medical issues are referred to a pediatric genetics clinic in the hope of identifying an underlying reason for these issues. Immediate identification is not always possible. The ensuing pursuit is known as the diagnostic odyssey and can be
very difficult for families. Do you refer families of children who do not have a diagnosis, or are still on a “diagnostic odyssey” to parent support groups?

- No, I don’t believe it would be helpful
- No, I don’t know of any groups that would be appropriate
- No, I worry that they will receive information that is confusing/irrelevant for them
- Yes, I do refer to some type of support group (online or face-to-face) Please provide a brief description of those that you tend to refer to: __________________ It depends on the age of the child/severity of condition

Q64 Do you volunteer or participate in (in your role as a professional) any parent support groups in person or online?

- Yes
- No
- I have in the past, but am not currently a volunteer

Q65 Please describe briefly: