Now What Happens? An Exploration of the Genetic Counselor’s Role in Transition Planning

Master’s Thesis

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Judith Tsipis, Ph.D., Advisor

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Master of Science
in
Genetic Counseling

by
Danielle Hays

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ABSTRACT

Now What Happens? An Exploration of the Genetic Counselor’s Role in Transition Planning

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Danielle Hays

When children with special healthcare needs approach adulthood, their medical, social, and educational services change, and often these individuals lose their entitlements to special needs education and social services. The time period in which a child undergoes these changes into adulthood is called the transition phase. The goal of our study was to better understand the role that genetic counselors play in helping patients and their families with transition planning. We sent a survey of quantitative and open-ended questions to pediatric and specialty genetic counselors through the NSGC listserv and received 85 responses. We assessed how often genetic counselors were being asked about specific transition topics, how they were responding to these questions, and what challenges they encountered while providing transition planning services. Our results show that the most frequently asked about transition topic was the child’s prognosis and future health care needs; the topic least frequently asked about was guardianship. Respondents were most comfortable answering questions about the child’s future prognosis and health care needs, and were most likely to refer questions on future insurance coverage and guardianship to others. We found that the transition discussion is standard procedure for many pediatric clinics and that some genetic counselors initiate the discussion when the family is unaware that a transition is approaching. Common challenges faced by respondents when
providing transition planning include: a lack of appropriate adult genetics services, parent or patient reluctance to transition from current care, and counselor uncertainty in how to best help the patient. Our research showed that pediatric and specialty genetic counselors are being involved in conversations about transition planning. The development of institutional resources and training to familiarize genetic counselors with local transition services and regulations is recommended to help genetic counselors expand their ability to help their patients and families with transition planning.

Keywords: genetic counselor, transition planning, transition, pediatric, specialty
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Introduction

Due to recent advancements in healthcare, many children who formerly would have died at birth or in infancy due to a genetic condition are now living into adulthood (Hallum, 1995). They often have intellectual, developmental, and/or physical disabilities that result in special health care needs and special education services in school and the community. There are now approximately 750,000 children in the United States with special health care needs transitioning to adult care each year (Davis, 2014). The term “transition phase” is used to describe the time period in which an individual with a genetic disorder and/or disability goes from childhood services to adult care.

In the education system, “transition” is a term used to describe “the progression from youth to adulthood, and the specific individual planning processes available to certain students with disabilities as they shift from life as a student to life as an adult” (Massachusetts Department of Elementary & Secondary Education, 2015). Each state has their own guidelines but, in Massachusetts, students entitled to special education services lose those services when they graduate from high school or reach the age of 22. There are laws in place that stipulate that a “student who may require continued disability-related services upon exiting school (by graduating or turning twenty-two years of age, whichever occurs first), shall be offered transition planning.” (Massachusetts Department of Elementary & Secondary Education, 2015).

“Transition” in the healthcare setting is defined as “a comprehensive set of support processes and care that ideally begin before and extend after transfer of care” (Davis, 2014). There is agreement amongst healthcare professionals that transitioning involves more than
simply a physical “transfer” of care from pediatric to adult medical care. However, according to Davis et al. (2014), fewer than 50% of children with special health care needs receive the support and services they need for their transition to adult care. Further, less than 15 percent of pediatricians reported providing transition education materials to their patients (Davis, 2014).

Considering either the education or health care definition of the term “transition”, it has been suggested that genetic counselors can play a role in helping their patients and their families traverse this stressful time. In regards to educational planning, Simon, et al. (2011) suggest that genetic counselors who specialize in caring for patients with intellectual disabilities could be integral members of an individual’s support team and work with psychologists, behavior specialists, and social workers to educate the team members about a child’s genetic diagnosis and to develop support planning tailored to a person’s syndrome-related strengths and weaknesses. In regards to the medical definition of “transition”, Taylor (2006) suggests that “A transition from a pediatrician to an adult primary care physician can be difficult and genetic counselors can help patients to have their genetic conditions managed by adult treating physicians.”

Previous studies have highlighted why genetic counselors are prime candidates to provide information regarding genetic conditions and disability support services to the patient and their families during transition planning. They are knowledgeable about genetic conditions and their prognosis, often have the opportunity to establish long-term relationships with the patient and their family, and are familiar with community and social resources. Szybowska, et al. (2007) found that genetic counselors are one of the primary sources of genetic information for adolescents with a genetic condition. They determined that seeing a genetic counselor at an early age can help in the process of transitioning to the adult health care setting and that adolescents
with a genetic condition wanted information regarding their genetic diagnosis around the age of 12 years old. Hartley, et al. (2011) described the unique experience of genetic counselors who provided ongoing support in a pediatric metabolic clinic for patients with inborn errors of metabolism. Many of these counselors had long-term relationships with the patients and their families and provided counseling at multiple time points including adolescence. This relationship allowed ample opportunity for the patient and family to ask questions regarding the genetic diagnosis and to discuss transition planning. Hartley, et al. (2011) had similar findings to Szybowska, et al. (2007) in that genetic counseling during adolescence allowed the patients to ask questions and receive formal education about their genetic condition. Ormond, et al. (2003) studied health care trainees’ attitudes toward disability and the perception of what defines the quality of life for people with a disability. They found that, perhaps due to differences in training, genetic counselors were more likely than other medical trainees to consider social issues, in addition to medical issues, an important aspect of quality of life, and they were more likely to provide community and social support resources for individuals with a genetic disability. A health care professional’s knowledge and attitude about a genetic condition will affect how they communicate and share information with the patient; therefore, the genetic counselor’s ability to integrate medical information and social resources make them the ideal healthcare provider to help with transition planning.

The goal of our study was to determine the role that genetic counselors currently play in helping their patients and their families with transition planning. We wanted to determine if genetic counselors are being asked questions in relation to transition planning, what topics are being addressed, and how the genetic counselors are responding to these questions. This study will provide the foundation for future studies to expand the role of the genetic counselor in
transition planning and perhaps encourage genetic counselors in pediatric clinics to re-evaluate how they can better help their patients and families for transitioning.
Methods

Study Design

We used an anonymous online survey to collect information from genetic counselors who worked in a pediatric and/or specialty clinic within the past 5 years. The recruitment notification was sent out through the National Society of Genetic Counselors (NSGC) website via the NSGC listserv. The survey was open for four weeks from January 21, 2016 to February 18, 2016 with a reminder email sent on February 4th. We limited the inclusion criteria to any genetic counselor who received emails from the NSGC listserv, had worked within the past 5 years in a pediatric or specialty clinic, and could speak and understand English.

We provided the link to the online survey in the recruitment notice (Appendix A) and participants acknowledged consent by entering the survey. The Brandeis University Review Board approved the study (Protocol #16067).

Data Collection Tool

We used Qualtrics, an online survey tool, to create the anonymous survey. The survey collected data on the clinical genetic counselor’s role in transition planning for their patients with a genetic condition and/or disability (Appendix B). The survey included the following sections: frequency of involvement with transition planning, experiences with transition planning and demographics. To explore the frequency of involvement with transition planning, we included a likert scale question with 12 transition-specific topics to determine how often the respondent had been asked about the topic. The likert scale options included: ‘never’, ‘once or twice’, ‘often’, and ‘almost always’. If a participant indicated they had been asked about a transition topic, they
would then see relevant follow-up questions asking how they had responded to the specific transition question and what condition/diagnosis the patient asking the question had. Participants who had never been asked about the particular transition topic did not get the follow-up question. Each survey was therefore unique to the participant and the number of follow-up questions they answered depended on their experience. The survey included open-ended questions to learn about any challenges they faced, whether they had ever initiated the transition discussion with a patient, and whether they had had any particularly interesting or challenging cases. We also asked counselors who had not yet been involved with transition planning whether they thought there was a role for genetic counselors in this. If the participant completed the survey, we offered the opportunity to participate in a raffle for three $50 Amazon.com gift cards. This raffle was not linked to the survey so the survey results remained anonymous.

**Data Analysis**

We analyzed the data using IBM Statistical Package for the Social Sciences (SPSS) version 23 software. We excluded the participants who did not complete a majority of the survey questions from the data analysis. We used frequencies and descriptive statistics to analyze the results of the quantitative data collected. We manually coded and analyzed the open-ended responses using Microsoft Excel to identify common themes. Raw and analyzed data was stored on a password-protected personal computer and will be destroyed after submission of the data for publication.
Results

We received 85 completed surveys that were used for analysis, including 10 in which respondents failed to fill out the demographic questions but were otherwise complete. Another three surveys, included in data analysis, lacked a response as to how often they had been asked a specific transition question in their role as genetic counselor.

Demographics

Of the 75 participants who completed demographic information, 66 worked in the pediatric setting when they provided transition planning services for their patients and most (72%) selected academic/university-based clinic or hospital to describe their work setting. Participants were from all NSGC regions, with the highest representation from Region IV (34.7%). There was a range of years of experience as a practicing genetic counselor, with the highest percent of responses from participants with 0-2 years of experience (32%). Full demographic information is included in Table 1.

Forty-two respondents (56%, n=75) had previous experience working or volunteering directly with children or adults with disabilities and 25 respondents (33.8%, n=74) indicated that they currently or previously have volunteered for genetic support groups.
Table 1. Demographic Information

<table>
<thead>
<tr>
<th>Work Setting</th>
<th>Frequency (n)</th>
<th>Percent</th>
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<tbody>
<tr>
<td>Prenatal</td>
<td>4</td>
<td>--</td>
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<tr>
<td>Cancer</td>
<td>6</td>
<td>--</td>
</tr>
<tr>
<td>Pediatric</td>
<td>66</td>
<td>--</td>
</tr>
<tr>
<td>Adult</td>
<td>14</td>
<td>--</td>
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<tr>
<td>Specialty</td>
<td>26</td>
<td>--</td>
</tr>
<tr>
<td>Lab</td>
<td>1</td>
<td>--</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Type of Work Setting</th>
<th>Frequency (n)</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Academic/University-based clinic or hospital</td>
<td>54</td>
<td>72%</td>
</tr>
<tr>
<td>Non-Academic clinic or hospital</td>
<td>15</td>
<td>20%</td>
</tr>
<tr>
<td>Private Practice</td>
<td>2</td>
<td>2.7%</td>
</tr>
<tr>
<td>Non-Profit Organization</td>
<td>3</td>
<td>4%</td>
</tr>
<tr>
<td>Other: Free Standing Children’s Hospital</td>
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<td>1.3%</td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>NSGC Region</th>
<th>Frequency (n)</th>
<th>Percent</th>
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<tbody>
<tr>
<td>Region I</td>
<td>6</td>
<td>8%</td>
</tr>
<tr>
<td>Region II</td>
<td>18</td>
<td>24%</td>
</tr>
<tr>
<td>Region III</td>
<td>6</td>
<td>8%</td>
</tr>
<tr>
<td>Region IV</td>
<td>26</td>
<td>34.7%</td>
</tr>
<tr>
<td>Region V</td>
<td>9</td>
<td>12%</td>
</tr>
<tr>
<td>Region VI</td>
<td>9</td>
<td>12%</td>
</tr>
<tr>
<td>Other: IV and V</td>
<td>1</td>
<td>1.3%</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Years of experience</th>
<th>Frequency (n)</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2 years</td>
<td>24</td>
<td>32%</td>
</tr>
<tr>
<td>3-5 years</td>
<td>18</td>
<td>24%</td>
</tr>
<tr>
<td>6-10 years</td>
<td>19</td>
<td>25.3%</td>
</tr>
<tr>
<td>11-20 years</td>
<td>13</td>
<td>17.3%</td>
</tr>
<tr>
<td>21-30 years</td>
<td>0</td>
<td>--</td>
</tr>
<tr>
<td>31+ years</td>
<td>1</td>
<td>1.3%</td>
</tr>
</tbody>
</table>

Experience with transition planning

Almost all of the 85 respondents (92.9%) indicated that they had been asked questions about a child’s future medical, social, educational, and/or psychosocial needs in anticipation of, or during, the transition into adulthood during a meeting with a patient and/or their family.

We asked the 79 respondents who had been involved with transition planning how often they had been asked about each of 12 specific transition topics (Full transition topic questions...
can be found in Appendix B). The transition question that was most frequently asked was “What is our child’s prognosis and future health care needs?” This was also the only question that every counselor had been asked at least once; no respondents indicated that they had never been asked this question. The transition question that was least frequently asked was “Can you answer questions about guardianship?”, with 50.6% of respondents indicating that they had never been asked this question. (Figure 1).

Figure 1. How often Genetic Counselors were asked about transition topics in the past 5 years (n =79).

* indicates less than 79 responses. Written medical summary (n = 77). Outside Resources (n= 78).
If a participant indicated that they had been asked about a transition topic at least once, they received a set of follow-up questions asking them to indicate how they had responded to the specific transition question at one time or another. For each specific transition question, we analyzed answers only from those respondents who indicated that they had encountered the question ‘often’ or ‘almost all the time’. We categorized the responses into three main categories: I answered the question, I have answered the question and I have referred the patient, or I just referred the patient. Options for referring the patient included referring to someone else, to a specialty clinic, or providing literature and resources on the topic. The question that the genetic counselors were most likely to answer themselves was “What is our child’s prognosis and future health care needs?” The question that the genetic counselors most often provided a referral for was “Will insurance coverage change when my child reaches 22?” (Figure 2). We also asked the participants to list the condition/diagnosis of the patient who asked the specific transition planning question and we received a wide range of responses. The conditions/diagnoses were typical of any pediatric clinic and included birth defects, single gene disorders, chromosome abnormalities, cancer predisposition syndromes, metabolic conditions and more.

When asked whether they had an ongoing relationship with the patients and families that they helped with transition planning, most of those responding (n=73) indicated that they had had an ongoing relationship with at least some of their patients and/or their families. A majority (61.6%) indicated that they sometimes had an ongoing relationship, 24.7% said that they almost always had an ongoing relationship, and 13.7% said they did not have an ongoing relationship. (Figure 3).
Figure 2. Genetic Counselor’s responses to specific transition questions: Answer vs Refer*

*responses shown are only from counselors who indicated that they had encountered the question ‘often’ or ‘almost all the time’

Figure 3. Percent of counselors with ongoing relationships with their transitioning patient (n = 73).
We then asked the respondents to briefly describe times that they have initiated the transition discussion with the patient and their family, the challenges that they have encountered surrounding transition planning, and any particularly interesting or challenging cases that they have seen. Of the 43 responses received regarding initiating the transition discussion: 17 participants indicated that they frequently or regularly initiated the discussion, 7 participants sometimes initiated the discussion, 6 participants rarely initiated the discussion, and 13 participants never initiated. (Figure 4).

Figure 4. How often Genetic Counselors initiate the transition discussion (n = 43).

Most participants who frequently or regularly initiated the discussion indicated that it was standard protocol for their clinic. Participants who sometimes initiated the discussion stated that it was situation dependent and often because the family was unaware that a transition and change in services was going to occur. Two participants said:

“Yes, in our clinic we generally discuss transition with patients and families when the patient reaches the later teenage years if the patient/family does not bring it up. Some patients who have been followed here for many years have some reluctance or aniet (sic) over transitioning to adult providers and so they may not bring it up on their own.”
“I have had multiple families with a child with intellectual disability who are unaware that they must obtain guardianship in order to make medical decisions for their child past the age of 18. In these circumstances, I usually initiate the conversation around (sic) the age of 17 in order for the families to have time to complete paperwork before the child turns 18 years old. I usually refer these families to a social worker.”

Fifty-two respondents (66%, n=79) reported encountering challenges while helping patients and their families with transition planning. We found consistent themes across responses including: a lack of appropriate adult genetics services for the patient to transition into, parent’s or patient’s unwillingness to transition into adult care, a counselor’s uncertainty about how to best help the patient, and a concern for the parent and/or patient being comfortable making the transition. (Table 2).

Table 2. Challenges faced by genetic counselors during transition planning (n= 52).

<table>
<thead>
<tr>
<th>Theme</th>
<th>Responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>No appropriate adult services</td>
<td>24</td>
</tr>
<tr>
<td>Lack of willingness of the parent or patient</td>
<td>15</td>
</tr>
<tr>
<td>Counselor uncertainty</td>
<td>10</td>
</tr>
<tr>
<td>Comfort of the parent or patient</td>
<td>6</td>
</tr>
<tr>
<td>Clinic time/resources</td>
<td>4</td>
</tr>
<tr>
<td>Insurance</td>
<td>4</td>
</tr>
<tr>
<td>Family resources/finances</td>
<td>4</td>
</tr>
<tr>
<td>State laws</td>
<td>3</td>
</tr>
<tr>
<td>Intellectual ability/maturity of child, Lost to follow up, Non-genetics focused transition, Specific answers about prognosis, Language barrier</td>
<td>≤ 2 each</td>
</tr>
</tbody>
</table>

Many responses included multiple themes:

“We have frequently encountered difficulty in identifying adult primary care providers willing to take medically complex patients. Additionally, we have historically not had access to an adult geneticist so we have often cared for these patients into adulthood (sic). Parents may have some difficulty allowing their child to take control of their healthcare in adulthood.”
“There are a lot of assumptions made by the family. It's obvious to the family that their child needs extra help/physicians/schooling but they have no idea that this is something that needs to be ARRANGED, it doesn't just happen automatically. Also, parents are often very anxious about changing to a new non-pediatric doctor because they are so comfortable with their current medical team who knows their child inside and out.”

When asked to describe one of their most interesting or challenging cases that involved transition planning, the most common cases described by the 22 respondents were the ones that involved many logistical hurdles, a lack of willingness to transition, and onset of adult symptoms.

“We had to get an entire family with Fabry transitioned to adult care for management and ERT. This was difficult logistically because there were so many parties involved (our clinic, current hospital where infusions were done, new hospital, new MD, insurance (sic), Genzyme liason (sic)). Additionally, we wanted to transfer care in one sweep, efficiently. However, this was difficult due to ages (some were 16 and 17, so not quite ready for adult care). All had different insurance. Furthermore, the family spoke Spanish (sic), so all correspondence with the family had to be done with translator. The new MD, rightfully so, wanted the patients to have up to date evaluations (cardiac evals, eye evals). This further complicated things due to language, insurance, waiting times, scheduling (sic), etc, etc. I'm breaking out in hives just thinking about this fiasco.”

“We had a 40yo patient with 18p- that we held onto as long as we could out of the goodness of our hearts and the belief that we knew the most about his condition. However, when he began to have symptoms that were not related to his genetic diagnosis and more (sic) related to his age, then WE felt ill-prepared for caring for him. Neither the patient or our service was happy with transferring his care to an adult provider, but we all knew it was for the best. Change is hard for everyone.”

Lastly, of the six (7.1%, n=85) respondents who said that they had not been involved with transition planning, five thought that there was a role for the genetic counselor in transition planning. Two respondents suggested roles for the genetic counselor:

“Recommendations for adult medical specialists. Preconception counseling”

“make sure the family is set up with the appropriate resources (referrals, access, availability)”
Discussion

Our study surveyed pediatric and specialty genetic counselors to determine their role in helping their patients and families with transition planning. We assessed how often genetic counselors were being asked about specific topics in relation to transition planning and how they were responding. We included open-ended questions about times that the counselor has initiated the transition discussion and about challenges that they have faced when counseling about transition.

Of the 85 participants that took the survey, 79 (92.9%) indicated that they had been asked questions about a child’s future medical, social, educational, and/or psychosocial needs in anticipation of, or during, their transitioning into adulthood, by a patient and/or their family. As might be expected, most of the 79 respondents (83.5%) indicated that they are ‘often’ or ‘almost all the time’ asked about the child’s future prognosis and health care needs, and about half (55.7%) are asked ‘often’ or ‘almost all the time’ about how the child’s health care services will be affected by transitioning. Since most patients are seeing a genetic counselor in a healthcare setting, these questions are about the medical care that genetic counselors are already providing (Simon 2011). These are also typical questions asked by patients during any medical visit, not just during the transition phase. The frequency of these questions align with findings by Hallum et al. (1995) that many parents caring for a child with a disability are most concerned about their child’s future. For the other ten transition topics, over half of the respondents indicated that they had ‘never’ been asked about the topic or had only been asked ‘once or twice’. The question that was the least frequently asked about was guardianship. However, many respondents indicated
that the issue of guardianship was one of the topics that they frequently brought up with families when discussing transition planning. This often happened when their patient was approaching age 18 and the parents were unaware that they would not automatically become the legal guardian for their child even when the child was unable to make their own medical decisions. Respondents reiterated throughout the short answer responses that they often felt compelled to discuss transition when it seemed like their patient and family was unaware that medical and educational changes were potentially going to occur and that services needed to be arranged in advance.

For each of the 12 transition topic questions, we created a list of three to five specific responses that we felt were most likely to be utilized by the genetic counselor. We categorized the responses into three main categories: I answered the question, I have answered the question and I have referred the patient, or I referred the patient. We asked participants to select any response that they had done at least once and we only analyzed the responses from participants that had encountered the transition topic ‘often’ or ‘almost all the time’. Our findings show that counselors most often answered the question about prognosis and future health care needs, provided a written medical summary, and answered how health care services would be affected. These results are not surprising as these are questions that genetic counselors would typically be expected to answer given they work in the health care setting. This is consistent with our previous finding that during transition genetic counselors are most often asked medical questions about a child’s future prognosis and health care needs. We found that counselors were most likely to refer their patients when asked about insurance coverage, guardianship, or to create a transition plan. We also found that most respondents (61.6%, n=73) had had an ongoing relationship with at least some of their patients prior to the discussion about transition planning.
This is expected given that many children with a genetic disorder or disability are seen regularly in the clinic for follow-up, although some may change providers, move to new areas or be lost to follow up.

The participants identified that they help many patients with a broad range of diagnoses and conditions with transition planning. Due to the range of responses we were unable to draw any conclusions between the condition/diagnosis and the type of transition question asked.

When asked to describe a case where they initiated the transition discussion, we received a range of responses with 40% (n = 43) regularly initiating the conversation while 30% had never initiated the transition discussion. Many of the genetic counselors who frequently initiated the conversation stated that it was standard practice for their clinic to bring up the topic and several counselors who had sometimes initiated the discussion stated that it was usually when the family was unaware of the impact of transitioning on their child’s care. This statement is consistent with the findings by Davis, et al. (2014) that over half of the children with special health care needs are not getting the support services or transition materials that they need. This confirms a need for the genetic counselor to educate the family about upcoming changes in medical care. It is worth noting, however, that not initiating the discussion does not mean that transition was not discussed during a session. Interestingly, two participants- one who rarely initiated the discussion and one who regularly initiated the discussion- indicated that around the time of this survey their institutions were conducting internal research regarding patient transition services. When asked if they had initiated the transition discussion they said:

“Yes, but we were piloting a study regarding transition services for adolescent genetic patients. Otherwise no, we didn't typically, but for the purposes of our research, we did with each patient that fell between 14-22.”
“As our patients approach adulthood, we routinely bring up the issues of transition planning. Our practice (a multi-specialty pediatric practice), actually recently had a QI project centered around transitioning our patients to adult care.”

Perhaps, due to the increasing number of children with special health care needs that are now surviving into adulthood, institutions have begun to evaluate their transition care programs. As Davis, et al. (2014) describes, there is currently no accepted way to measure transition success, but it is important to assess the established procedures in order to create a more effective standard practice.

In analyzing the challenges described by 52 of the respondents, we found consistent themes across many responses. In agreement with Taylor’s (2006) findings that the continuity of genetics care from the pediatric clinic to adulthood is frequently lacking, most respondents indicated their biggest challenge was that there are no appropriate adult genetics services or resources available. Other frequently cited challenges were lack of willingness to transition by the parent or the patient and counselor uncertainty. One respondent stated:

“The three biggest challenges are: 1. when parents are hesitant about initiating the conversations with their child because they think the child is too young 2. since it is a gradual process, it is problematic if a family stops coming to clinic. you don't now (sic) if someone other (sic) provider has picked up the discussion. 3. Having an adult clinic to refer them to. Often there is not, and you have to rely on a primary or another specialist to be the point person. Even if someone does have a medical home as an adul (sic), I think individuals with genetic disorders/complex needs get lost.”

We looked further at counselor uncertainty and found that all participants who gave this response had ten or less years of experience as a genetic counselor. However, the uncertainty was equally represented from the three different groups that fall within the ten-year range: four respondents had 0-2 years of experience, three had 3-5 years of experience and three had 6-10 years of experience. Some participants who fell into the counselor uncertainty theme noted that challenges they encountered included:
“My own knowledge of what services are available”

“Lack of available resources, uncertainty about who to contact or make referrals to, delayed contact by agency becoming involved with transition”

“I don't know enough about the resources available in my state”

“Lack of knowledge on my part about laws, health care coverage, services, etc, in the "adult" world”

“I feel like I have a lack of depth of knowledge about how to help guide transition planning and I think I would benefit from further education.”

Additionally, genetic counselors may also find the transition process to be challenging for emotional reasons. One respondent summarized their feelings by saying:

“…Neither the patient or our service was happy with transferring his care to an adult provider, but we all knew it was for the best. Change is hard for everyone.”

We also asked participants to describe a particularly interesting or challenging case with which they had been involved. One response highlights the struggle of what to do with adolescents living with a life-limiting condition:

“I don't think there was one in particular. However, the children with DMD were a topic of discussion between the physicians due to the fact that the children were typically not going to live for a long time after transitioning into an adult clinic. Plus, he (sic) adult physicians were not familiar with the condition. We tended to keep those children in the (sic) pediatric clinic.”

Another response reflects the need to introduce the topic of transition at an early age:

“I had a case with a 21 year old who we saw in pediatric clinic as a special exception and while she was fully capable of managing her care, she was resistant to. It brought to light for me the psychological impact of having special healthcare needs. When (sic) parent has been managing care for so long, it is scary for young adults to take on care roles. This was a girl with a connective tissue disorder and who was planning on getting married to her boyfriend. She was emotionally and intellectually mature in other (sic) ways, but when it came to health care she was so dependent on her mother. It made me realize that it's important to start these conversations early and help the family find small ways to start encouraging independence when the child is younger so that hen (sic) they are older, it won't be such a shock.”
This response is congruent with findings by Szybowska et.al. (2007) which noted that seeing a genetic counselor at an early age can help in the process of transitioning to the adult health care setting and that adolescents with a genetic condition wanted information regarding genetics starting around the age of 12 years.

Limitations

A major limitation for this study is the small sample size. Although we had a response rate of about 45% of the pediatric and specialty genetic counselors according to the 2014 NSGC Professional Status Survey (PSS), the sample size was not large enough to draw statistical significance. The regional distribution of our responses was not proportionate with that found by the 2014 NSGC PSS with a higher response rate from those living in Regions II and IV and a lower rate from the others. This was a retrospective survey that asked genetic counselors to recall events from the past five years and we have no way of assessing recall bias or verifying the respondents’ statements. Also, by including “check all that apply” answers to get a broad idea of how a counselor had responded to a transition question, we were unable to conclude how the counselor would typically respond to the question. Lastly, we had to exclude 22 incomplete surveys from our analysis.

Future Research

This study provides insight into the questions that genetic counselors are currently being asked about transition planning, how they are answering the questions, and the challenges that genetic counselors face when discussing transition services with their patients. More studies need to be done to determine the best way to utilize the genetic counselor in helping patients plan for transition. Although it seems some institutions have begun to look at their transition services, perhaps a study similar to Davis, et al. (2014) could be done to compare institutional standards
for transition care and best practices for genetic counselors. Future research can be done to look at the differences in responses from rural vs urban populations and to see if specific transition questions are asked more often by patients with certain conditions/diagnoses. All of this research could result in the development of resources for genetic counselors on transition planning. As the field of genetic counseling continues to grow and develop, this research will allow counselors to expand their skills in helping their patients and families with transition planning.
Conclusion

Our observational study identified how often genetic counselors were being asked about specific transition topics, how they were responding to these questions, and challenges encountered while providing transition planning services for patients and their families. We found that, in general, genetic counselors are being asked medical based questions more than questions about social services and education. We found that genetic counselors are most able to answer medical based questions on their own and tend to refer patients when asked questions about education or social services. We found that there is a need for genetic counselors to initiate the transition discussion with their patients and families when they were unaware that transition changes were approaching. In many cases, this discussion should be started early to help make the patient and their family more comfortable with the transition. Lastly, we found that there is a need for increased training and/or establishment of a standard procedure, where possible, for transition planning. These observations are important for the field of genetic counseling and especially for pediatric and specialty genetic counselors. Having a foundation of what genetic counselors are being asked and what they are doing about it will allow for expanding the role of the genetic counselor in the area of transition planning and services.
References


Appendix A: Recruitment Notice

Seeking Pediatric and Specialty Genetic Counselors to Participate in a Master’s thesis Research Study

You are invited to participate in an anonymous online research survey to investigate genetic counselor’s role in helping patients and their families plan for the transition from pediatric to adult care. When a child with a genetic disorder and/or disability transitions from childhood to adulthood, there could be potential changes in their medical, social, and school needs. Many families begin planning for the transition as early as age 14 and continue past the age of 22 as the individual settles into the new adulthood services. Are genetic counselors being asked questions about this potentially stressful time in their patient’s lives? If so, what questions are being asked and what are genetic counselors doing to help their families?

This study is open to any genetic counselor who has worked in a pediatric or specialty clinic within the past 5 years.

The specific goals of this research study are to:

- Determine if pediatric and specialty genetic counselors are being asked questions by families in anticipation of, or during, their child’s transitioning into adulthood
- Learn what specific questions genetic counselors are being asked about the transition of a child’s care from pediatric to adult services
- Explore the various roles pediatric and specialty genetic counselors can play when one of their patients transitions from pediatric care to adult services
- Gain more insight into potential challenges faced by genetic counselors when providing assistance around transition planning

The survey will take 10-15 minutes of your time. All participants who complete the survey will have the opportunity to enter a drawing for one of three $50 gift cards to Amazon.com. Your survey responses will not be connected to your email address.

This study was reviewed and approved by the Brandeis University Institutional Review Board. If you have any questions, concerns, or comments, please feel free to contact me by email at haysdl1@brandeis.edu, or the Brandeis University faculty sponsor, Judith Tsipis, at tsipis@brandeis.edu.

Click here to take the survey.

Thank you in advance for your time and participation.
Sincerely,
Danielle Hays, BA
Master's Degree Candidate, Class of 2016
Genetic Counseling Program
Brandeis University

Judith E. Tsipis, PhD
Director, Genetic Counseling Program
Professor of Biology
Brandeis University
Appendix B: Survey Instrument

Informed Consent Screen Language (1st page of online survey)
Thank you for participating in this study of the genetic counselors’ role in transition planning. Your participation in this study is voluntary and you may exit the survey at any time. The survey is anonymous and no identifying information will be collected. The study has been reviewed and approved by the Institutional Review Board of Brandeis University in Waltham, MA. All information will remain confidential. By clicking next, you have acknowledged that you have read the above information and that you consent to participating in this study.

Most special childhood disability services, particularly the entitlement to publicly funded special education services, end when an individual turns 22 years old, at which time they are typically eligible for adult services. The term “transition phase” is used to describe the time period in which an individual with a genetic disorder and/or disability goes from childhood to adulthood. During this time, there will likely be changes in the individual’s medical, social, vocational and/or educational needs, resulting in the need for new and different services. Many families begin planning for the transition phase as early as age 14 and continue past the age of 22 as the individual settles into the new adulthood services.¹

Survey Questions

1. In your role as a pediatric or specialty genetic counselor, have you ever been asked questions about a child’s future medical, social, educational, and/or psychosocial needs in anticipation of, or during, their transitioning into adulthood, during a meeting with a patient and/or their family?
   a) Yes
   b) No
   (If the answer is no, the survey taker will see a different set of questions designed to get as much information as possible about whether they think there is a role for the GC in the transition phase.)

2. In the past 5 years, how often have you been asked about the following topics in the context of a child transitioning from pediatric to adult care? The question could have been asked by anyone (healthcare professional, social worker, transition coordinator, parent, etc.) involved in the child’s care, including the child. (This will be a Likert question with the following options: Never, once or twice, often, almost all the time)

   Note: Questions should be broadly interpreted as to the topic highlighted in bold; a positive response does not require that the person asking the question used exactly the language
written. Also questions are written in the third person; if question was asked by the child, then substitute ‘my’ for ‘his/her’.

**Instructions:** When the respondent indicates that they have been asked about a topic (i.e.: once or twice, often, almost all the time) they will then be asked follow up questions. The follow up questions are listed below each topic. If the respondent selects ‘never’, they will not get follow up questions.

a) How will his/her health care services be affected?
   1. How did you respond when asked the question: “How will his/her health care services be affected?” Check all that apply at one time or another.
      a) I answered the question(s) about health care services.
      b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
      c) I referred the patient to a specialty clinic.
      d) I gave them literature/resources on how the health care services will be affected.
      e) Other: Please describe.
   2. What condition(s) or diagnoses did the patient(s) have?
      Please list all conditions if you have assisted multiple patients with different conditions.

b) What is our child’s prognosis and future health care needs?
   1. How did you respond when asked the question: “What is our child’s prognosis and future health care needs?” Check all that apply at one time or another.
      a) I answered the question(s) about the prognosis and future health care needs.
      b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
      c) I referred the patient to a specialty clinic.
      d) I gave them literature/resources about the prognosis and future health care needs.
      e) Other: Please describe.
   2. What condition(s) or diagnoses did the patient have?
      Please list all conditions if you have assisted multiple patients with different conditions.

c) Will insurance coverage change when my child reaches 22?
   1. How did you respond when asked the question: “Will insurance coverage change when my child reaches 22?” Check all that apply at one time or another.
      a) I answered the question(s) about insurance coverage.
      b) I looked up insurance contact information for the patient or the person asking.
      c) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
      d) I gave them literature/resources about insurance coverage.
2. What condition(s) or diagnoses did the patient have?
*Please list all conditions if you have assisted multiple patients with different conditions.*

d) Will you provide a written **medical summary** for our child?
1. How did you respond when asked the question: “Will you provide a written **medical summary** for our child?” Check all that apply at one time or another.
   a) I provided a written medical summary.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) Other: *Please describe.*
2. What condition(s) or diagnoses did the patient have?
*Please list all conditions if you have assisted multiple patients with different conditions.*

e) Will you help our child learn about **medical self-management**?
1. How did you respond when asked the question: “Will you help our child learn about **medical self-management**?” Check all that apply at one time or another.
   a) I helped the child learn about medical self-management.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) I referred the patient to a specialty clinic.
   d) I gave them literature/resources about medical self-management.
   e) Other: *Please describe.*
2. What condition(s) or diagnoses did the patient have?
*Please list all conditions if you have assisted multiple patients with different conditions.*

f) Do you have any materials that would help us plan for transitioning?
1. How did you respond when asked the question: “Do you have any materials that would help us plan for transitioning?” Check all that apply at one time or another.
   a) I gave them literature/resources on the topic of transition.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) I referred the patient to a specialty clinic for materials.
   d) Other: *Please describe.*
2. What condition(s) or diagnoses did the patient(s) have?
*Please list all conditions if you have assisted multiple patients with different conditions.*

g) Will you help us create a transition plan?
1. How did you respond when asked the question: “Will you help us create a transition plan?” Check all that apply at one time or another.
   a) I helped them create a transition plan.
   b) I referred the person to someone else (i.e. another health care provider,
social worker, case manager, etc.)
c) I gave them literature/resources on how to create a transition plan.
d) Other: Please describe.

2. What condition(s) or diagnoses did the patient(s) have?
   Please list all conditions if you have assisted multiple patients with different conditions

h) Will you help me identify outside resources to help with transition planning?
1. How did you respond when asked the question: “Will you help me identify outside resources to help with transition planning?” Check all that apply at one time or another.
   a) I looked up contact information for outside resources to help with transition planning.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) I referred the patient to a specialty clinic.
   d) I gave them literature/resources about outside sources that can help with transition planning.
   e) Other: Please describe.
2. What condition(s) or diagnoses did the patient have?
   Please list all conditions if you have assisted multiple patients with different conditions.

i) Can you answer questions or help me with my child’s IEP (Individualized Education Program)?
1. How did you respond when asked the question: “Can you answer questions or help me with my child’s IEP (Individualized Education Program)” Check all that apply at one time or another.
   a) I answered the question(s) about the child’s IEP.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) I gave them literature/resources about IEP’s.
   d) Other: Please describe.
2. What condition(s) or diagnoses did the patient have?
   Please list all conditions if you have assisted multiple patients with different conditions.

j) Can you answer questions about guardianship?
1. How did you respond when asked the question: “Can you answer questions about guardianship?” Check all that apply at one time or another.
   a) I answered the question(s) about guardianship.
   b) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   c) I gave them literature/resources about guardianship.
   d) Other: Please describe.
2. What condition(s) or diagnoses did the patient have?

Please list all conditions if you have assisted multiple patients with different conditions.

k) Can you answer questions about adult programs for my child?
1. How did you respond when asked the question: “Can you answer questions about adult programs for my child?” Check all that apply at one time or another
   a) I answered the question(s) about adult programs.
   b) I looked up contact information for adult programs.
   c) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   d) I gave them literature/resources about adult programs.
   e) Other: Please describe.

2. What condition(s) or diagnoses did the patient have?

Please list all conditions if you have assisted multiple patients with different conditions.

l) Can you answer questions about long-term care?
1. How did you respond when asked the question: “Can you answer questions about long-term care?” Check all that apply at one time or another.
   a) I answered the question(s) about long-term care.
   b) I coordinated long-term care services for the patient or the person asking.
   c) I looked up contact information for long-term care services.
   d) I referred the person to someone else (i.e. another health care provider, social worker, case manager, etc.)
   e) I gave them literature/resources about long-term care services.
   f) Other: Please describe.

2. What condition(s) or diagnoses did the patient have?

Please list all conditions if you have assisted multiple patients with different conditions.

General Questions:
3. Did you have an ongoing relationship with the patient(s) and their family prior to the discussion about transition planning?
   a) No
   b) Sometimes
   c) Almost always
   d) Varied with different children and their families
   e) Additional: Text box for any comments.

4. Was there ever a case where you initiated the transition discussion with a family? If so, please describe and indicate why you initiated the discussion? Free text box.

5. What were some of the challenges you encountered when providing assistance around transition planning? Free text box.
6. Please describe one case that you found particularly interesting or challenging that involved transition planning? *Free text box.*

*If “NO” is selected in response to the first question. (The GC has not encountered transition phase questions)*

1. Do you think there is a role for genetic counselors in the transition planning?
   a) *Yes.*
   b) *No.*

2. How do you think a GC could assist a child or family planning for the transition from pediatric to adult services? Please describe. *Free text box.*

3. Have you experienced a case where a genetic counselor *was not* involved and you thought they should have been? *Free text box.*

*If “NO” is selected:*

1. Why do you not think there is a role for the genetic counselor in the transition phase? *Free text box.*

**Information about the counselor**

**Demographics**

1. In what setting did you work when you helped a patient and their family with transition planning? Please list current work setting if you still facilitate transition planning OR if you have never facilitated transition planning. (Choose all that apply)
   a) Prenatal
   b) Cancer
   c) Pediatric
   d) Adult
   e) Specialty: *Please list specialty(ies).*
   f) Industry
   g) Lab
   h) Other: *Please list other.*

2. Which of the following best describes the work setting where you helped patients and their families with transition planning? (If you have never facilitated transition, please select the option that best describes your work setting.)
   a) Academic/University-based clinic or hospital
   b) Non-Academic clinic or hospital
   d) Private Practice
   e) Diagnostic Laboratory
   f) Non-Profit organization
   h) Other: *Please specify.*
3. In what NSGC region did you practice when you helped patients and their families with transition planning? (If you have never facilitated transition, please select the region of your work setting.)
   a) Region I (CT, MA, ME, NH, RI, VT, Canadian Maritime Provinces)
   b) Region II (DC, DE, MD, NJ, NY, PA, VA, WV, Quebec, Puerto Rico, Virgin Islands)
   c) Region III (AL, FL, GA, KY, MS, NC, SC, TN)
   d) Region IV (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario)
   e) Region V (AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan)
   f) Region VI (AK, CA, HI, NV, OR, WA, British Columbia)
   e) Other. Please specify.

4. How many years of experience do you have as a practicing genetic counselor? (Please round up to the closest year.)
   a) 0-2 years
   b) 3-5 years
   c) 6-10 years
   d) 11-20 years
   e) 21-30 years
   f) 31+ years

5. Did you have any experience working or volunteering directly with children or adults with disabilities prior to or since getting your Genetic Counseling degree?
   a) Yes. Please describe: Text box
   b) No

6. Have you previously or do you currently volunteer for any genetic support groups?
   a) Yes. Please describe: Text box
   b) No