Genetic Counselors’ role in maternal PKU: Patient and Genetic Counselor perspectives

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Abstract

Phenylketonuria (PKU) is a rare autosomal recessive disorder which causes toxic build up of phenylalanine, leading to microcephaly, seizures, severe intellectual disability and behavioral problems in untreated individuals. Maternal PKU results in the risk to have offspring with microcephaly, congenital heart defects, cognitive disability and low birth weight. The goal of this study was to examine the role that genetic counselors currently have in the care of pregnant women with PKU and how that informs us about the potential opportunities genetic counselors may play in the care of these women. We recruited genetic counselors who have been involved in the care of women with PKU as well as women with PKU who have had a prior pregnancy, to complete two separate anonymous, online surveys, which included both multiple choice and open ended questions. Thirty-five genetic counselors and sixteen women with PKU responded to the surveys. Women with PKU presented to genetic counseling in a variety of ways, most often related to maternal PKU, which was observed in both surveys. During genetic counseling visits for non-maternal PKU related indications, maternal PKU was discussed, indicating that genetic counselors are educated about and currently provide information about PKU and maternal PKU effects. The majority of women with PKU that genetic counselors reported they had seen were seen pre-conception or prenatally and not postnatally. The majority of genetic counselors felt that women with PKU should be seen by genetic counselors to discuss risks related to maternal PKU and the risk of PKU in
offspring. Women with PKU reported positive responses to experiences with genetic counselors. Overall, our study suggests that genetic counselors can fill an important supportive and education role in the care of women with PKU surrounding their pregnancies.
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**Introduction**

*Background on PKU*

Phenylketonuria (PKU) is a rare autosomal recessive disorder which affects the conversion of phenylalanine (phe) to tyrosine (tyr) (de la Cruz & Koch, 2001). Mutations in the PAH gene cause a deficiency of phenylalanine hydroxylase. Deficiency of this enzyme causes toxic build up of phenylalanine, leading to microcephaly, seizures, severe intellectual disability and behavioral problems in untreated individuals. There are different clinical categories of PKU that vary in their underlying mutations, the amount of PAH produced, and the levels of phe that are tolerated in affected individuals (Guttler, Guldberg, Eisensmith, & Woo, 1999). Treatment of PKU involves a diet low in phenylalanine and supplementation with a formula containing the other essential amino acids. Some individuals with PKU benefit from 6R-BH4 therapy as well (J. Mitchell, 2010). The current recommendation is for individuals to continue this diet for life to achieve the best outcome.

*Background on maternal PKU*

Women who have PKU are at risk to have babies with microcephaly, congenital heart defects, cognitive disability and low birth weight if they are not on diet during pregnancy, even when the fetus itself is does not have PKU, due to the amplified amount of phe to which the developing fetus is exposed (Gambol, 2007). This teratogenic effect to the fetus is referred to as maternal PKU. With adequate treatment during pregnancy,
children born to mothers with PKU can have normal cognitive development (Guttler et al., 2003) and maintenance of phenylalanine levels in women with PKU during pregnancy can predict good outcomes (Maillot, Lilburn, Baudin, Morley, & Lee, 2008). Maintaining phenylalanine levels requires a strict low protein diet as well as supplementation with medical formula containing amino acids. Due to the nature of this strict diet and the challenges of a pregnancy complicated by maternal PKU, it is important to provide adequate support to women with PKU during their pregnancies.

**Factors that influence pregnancy outcome**

Obtaining phe control is a crucial factor in the outcomes of pregnancies in women with PKU. In a study of women with PKU, it was found that in mothers with classic PKU and an average phe level between 360 and 750 µmol/L, both maternal IQ and average phe exposure were significant predictors of the child’s IQ. In offspring of mothers with mild PKU, IQ was significantly correlated to maternal IQ but not to average phe level during pregnancy, which in all of the mild cases was <750 µmol/L. Offspring whose mothers had phe levels below 360 µmol/L had normal cognitive development not related to maternal IQ. This study shows the importance of phe level control in women with classic PKU. It also shows the good outcome possible when phe levels are controlled, which emphasizes the importance of support and education of women with PKU before they become pregnant (Guttler, et al., 2003)

In a 2003 study, the authors describe the outcome of 228 children born to mothers with treated PKU or untreated mild hyperphenylalanemia. At 7 years of age, 18% of children born to mothers with treated PKU performed in the range of intellectual disability (IQ<70), while 18% performed in the borderline range (IQ<85). Both cognitive
and behavioral outcomes were negatively correlated with the number of gestational weeks before the mother achieved metabolic control. The children born to mothers who achieved control prior to conception did not differ in scores from the control group. This emphasizes the need for pre-conception counseling, planning and treatment. The authors also suggest the need for more research in the areas of environmental risks to the children associated with the mother’s PKU, which include socioeconomic status and proper amounts of stimulation in the home (Waisbren & Azen, 2003).

**Barriers and predictions of diet adherence**

A retrospective interview based study of 24 women with PKU (Brown et al., 2002) assessed medical adherence to low phe diet during their pregnancies. They found that women who intended to become pregnant were more likely to adhere to the diet recommendations, though one third of these women still did not adhere. They found that the majority of the women received support from friends and family but that fewer than half had attended a support program specific to maternal PKU. Two women reported that they were never referred to a metabolic clinic during their pregnancy and both had infants with microcephaly. In the study, how well women adhere to medical recommendations was addressed, as well as some of the challenges women with PKU face during pregnancy. Women in Brown’s study felt they received adequate support from friends and family members, but it is not clear how often their medical providers discussed this need for support.

The resource mothers study paired pregnant women with PKU with a resource mother trained to provide support and assistance. They found that, of the 44% of women who had not begun the diet prior to becoming pregnant, those with a resource mother
gained phe control earlier, although no significant differences in overall metabolic control were seen between the resource mother group and control group. They emphasized how few women remained on the diet after delivery and suggest the possible need for post partum support given that patients with PKU who are on diet are more able to deal with the stresses of parenting than those off diet. They state that women are well-educated about the risks of maternal PKU but there are still barriers to maintaining diet during pregnancy (Rohr et al., 2004).

In a 2010 review article, the authors address the factors that influence control of phe levels during and after pregnancy, such as poor access to medical care, reimbursement issues, demographics, financial constraints, and practical and psychosocial issues. Many women with PKU have learning disabilities, emotional disorders and low socioeconomic statues, all of which create problems with achieving phe control. The majority of women do not continue the diet after pregnancy even if they are on it during pregnancy. Discontinuation of the diet affects the mother and increases the risk of thought and mood disorders, anxiety, and mental processing abnormalities, which can affect the quality of the home environment (Koch, Trefz, & Waisbren, 2010).

In a 2003 study, it was shown that phe controlled diets started earlier in pregnancy decreased the risk for microcephaly and congenital heart disease. Larger head size is seen when weight gain and protein intake is adequate, even in pregnancies where phe control is not achieved (Matalon, Acosta, & Azen, 2003). This is important to consider given the difficulty of obtaining phe control by 8 weeks in the pregnancy, particularly if pregnancy is not planned. Given the risks, women with PKU should be aware of maternal PKU prior to achieving a pregnancy.
A 2003 review highlights some of the important challenges that still need to be addressed regarding maternal PKU. The author emphasizes that the dietary restriction, while currently the only prevention, is not always successful and children continue to be affected. He suggests that solutions are overly simplistic and additional strategies are necessary. The author addresses the complexity of the disease as well as the importance of acknowledging all the complicating factors (Clarke, 2003).

The American Congress of Obstetricians and Gynecologists recommends that all women with PKU should be encouraged to undergo preconception counseling, begin the restriction before conception, and should be managed in experienced PKU centers (Comm, 2001).

**Possible role of genetic counselors**

There are many health care providers that participate in the care of women with PKU during their pregnancies. Gaps in the healthcare system pertaining to PKU have been identified in the fact that care of women with PKU is not consistent (Gambol, 2007). Genetic counselors provide support, education and information about risks for recurrence in offspring to patients with a genetic disease. Genetic counselors play an important role in the care of patients, and in the setting of women with PKU, this may include discussion about recurrence risk for having offspring with PKU, prenatal issues regarding maternal PKU, support and necessary referral as well as discussions of other genetic counseling issues unrelated to maternal PKU that may come up during pregnancy.

Psychosocial issues regarding difficulties with diet have been studied, as have outcomes of offspring born to women with PKU (Koch, et al., 2010) but no study has
been done to determine how women with PKU view the care they received regarding maternal PKU and whether genetic counselors have or may be able to enhance the care of women with PKU surrounding their pregnancies.

**Purpose of study**

The purpose of this study was to assess the counseling women with PKU received surrounding maternal PKU and the perspectives of genetic counselors who have been involved in this care, in order to provide insight on how to best care for these individuals during their reproductive years. We were interested in what the role of genetic counselors has been in the care of women with PKU as well as perceptions about what the role of genetic counselors should be in the care of these women before, during and after their pregnancies.

From the patient perspective, we wanted to examine the role genetic counselors have had in their care during their pregnancies and the value of genetic counseling for women with phenylketonuria as a function of whether or not they saw a genetic counselor, either before, during or after their first pregnancy. We wanted to look at pregnancy outcome, whether or not the indication for genetic counseling was related to maternal PKU and patient satisfaction of care. We attempted to assess patient perceptions about whether genetic counselors are a helpful resource in addressing their concerns and whether patients who had seen a genetic counselor show any difference in satisfaction of care than those who had not.

From the genetic counselors’ perspective, we wanted to determine how many women were being seen pre-conception and what was being discussed with genetic counselors as part of PKU centers as well as in other settings. We also wanted to assess
genetic counselors’ involvement with women with PKU during pregnancy, and the reasons for the involvement in their care. Lastly, we wanted to determine how often genetic counselors see women with PKU postnatally, particularly when they have already established a relationship with them, due to the importance of women with PKU remaining on diet in the postnatal period.

Obtaining knowledge from women with PKU about their experience with a genetic counselor pertaining to maternal PKU or for unrelated reasons during pregnancy, and gathering the perspective of genetic counselors on their involvement in the care of women with PKU could provide insight as to how to best care for women with PKU and their families.
Methods

Study Design

We utilized online surveys using Qualtrics to conduct this study. The study consisted of two separate surveys, one for women with PKU (Appendix C) and one for genetic counselors who have seen women with PKU in their clinic (Appendix D). The survey for women with PKU consisted of detailed questions about their first pregnancy, including outcomes, and their experience with genetic counseling if they received any. They were also asked if they received genetic counseling during any subsequent pregnancy, and if they have not, they were asked how they felt about what it may have been like to have seen a genetic counselor during their first pregnancy.

The survey for genetic counselors contained questions about whether the women with PKU were seen pre-conception, during pregnancy and/or postnatally and how the women presented to them. They were also asked about what was discussed in the sessions. In addition, we asked genetic counselors about the type of clinic the saw the women with PKU in, and from their perspective, the percentage of all women with PKU who are seen by genetic counselors. There was also space for genetic counselors to answer an open ended question about their experience caring for women with PKU surrounding their pregnancies.

We recruited subjects in two ways. The women with PKU were recruited by emailing a recruitment notice containing a link to the online survey through the PKU listserv. The individuals with PKU who received the recruitment notice were able to pass
the link to the online survey along to any other women with PKU who have had at least one pregnancy who may be interested. The genetic counselors were recruited by emailing a recruitment notice containing the link to the online survey through the general National Society of Genetic Counselors (NSGC) listserv. We sent the recruitment notices through both the listservs multiple times between January 2011 and February 2011.

Data Collection

The data were collected through the use of anonymous, online surveys, using Qualtrics. No names or contact information was collected for either survey, although limited demographic information was collected. Both multiple choice and Likert scale quantitative questions were asked on the survey for women with PKU. The survey for genetic counselors consisted of mainly multiple choice questions. One open ended qualitative question was asked on the survey for genetic counselors.

The women with PKU were asked to give information about their first pregnancy and outcomes of that pregnancy. They were also asked if they received genetic counseling during any of their pregnancies and from whom. They were asked to recount what was discussed during genetic counseling if they received it and how they felt about the genetic counseling experience. If a genetic counselor was not seen, they were given information about genetic counseling and asked about their perceptions of the possible value of genetic counseling had it been available for their first pregnancy.

The genetic counselor survey contained questions about their experiences counseling women with PKU prior to, during or after their pregnancies. They were asked about their role in the women’s care as well as their perceptions about the value of genetic counseling for women with PKU at different times surrounding their pregnancies.
Data Analysis

The data were analyzed using PASW version 18 software. Frequencies of responses for quantitative questions were calculated. When possible, depending on the sample size of the particular question, bivariate analysis methods were used to carry out correlations. Chi square tests were used to test differences between groups and to determine significance of these differences for the survey of genetic counselors. Open ended questions were used to supplement the discussion. Due to the small sample size of the survey of women with PKU, frequencies of responses were calculated but it was not possible to test correlations with this number of respondents. The survey of women with PKU was analyzed qualitatively to examine the range of responses.


Results

Genetic Counselor Survey

Demographics

Thirty-five genetic counselors who have seen at least one woman with PKU before, during, or after pregnancy responded to our survey. The majority of respondents were women (33/35) between the ages of 25 and 44. Most respondents reside throughout the United States and four respondents reside outside the United States. Ten respondents have been in practice less than 5 years (29%), 9 have been in practice between 5 and 10 years (26%) and 16 respondents have been in practice greater than 10 years (46%). Other than those who had graduated more than 10 years ago this sample is comparable to the 2010 professional status survey of genetic counselors, in which about 33% had graduated from their graduate program less than 5 years ago, 32% had graduated 5-10 years ago and 35% had graduated greater than 10 years ago (www.nsgc.org).

Description of clinical time for genetic counselors

Thirteen respondents spend 100 percent of their clinical time in prenatal genetic counseling and 3 respondents spend 100 percent of their clinical time in metabolism. All other respondents split their clinical time between different areas. Seventeen (46%) respondents spent 50% or more clinical time in prenatal, 10 (27%) spent 50% or more time in pediatrics, and 8 (22%) spent 50% or more time in metabolism. In comparison
from the professional status survey, 39% of genetic counselors spend fifty percent or more time in prenatal, 17% in pediatrics and 2% in metabolism (www.nsgc.org). Twenty one respondents spend at least some of their clinical time in prenatal counseling, 14 spend at least some clinical time in pediatrics and 14 spend at least some clinical time in metabolism.

Providers who have been a part of the team while the genetic counselors have been involved in care of women with PKU include, clinical geneticist (15/35), obstetrician (24/35), metabolic specialist (25/35), nutritionist (24/35), and other healthcare professionals (11/35); including maternal fetal medicine specialists, dieticians, nurse practitioners and social workers.

Twelve respondents had seen one woman with PKU in the past 5 years, 14 had seen between 2 and 5, 2 had seen 5-10 and 7 respondents had seen more than 10 women with PKU in the past 5 years (Figure 1). Genetic counselors who spent clinical time in metabolism were more likely to see more than 10 women with PKU ($\chi^2 = 13.463$, $p=.004$), genetic counselors who spend some time in prenatal genetic counseling were more likely to see 1 woman with PKU ($\chi^2 = 7.675$, $p=.053$).
The majority of genetic counselors who only saw one woman with PKU saw her for the first time during her pregnancy as opposed to pre-conception. The women with PKU presented to the genetic counselors in various ways, including being seen in the metabolism clinic that the genetic counselors were a part, of as well as for non-maternal PKU related issues, most of which were for other pregnancy risks for which genetic counseling was indicated.

Figure 2. Responses from individuals who answered “other” included preconception referral for carrier testing (for PKU) for the woman’s partner as well as for maternal serum screening (to discuss the option of testing, those whose tests were positive are included below in Figure 2). The majority of all women seen by genetic counselors (23/32, response of “other” taken out) before, during or after pregnancy, were seen for maternal PKU, as opposed to for other unrelated indications (Figure 3).
Figure 2. (n=32), How women with PKU present to genetic counseling

![Bar chart showing how women with PKU present to genetic counseling.](chart)

- PKU related: 72%
- Not PKU related: 28%

Figure 3. (n=32), Reasons women with PKU are seen, PKU related or not.

![Pie chart showing reasons women with PKU are seen.](chart)

- PKU related: 72%
- Not PKU related: 28%
Women seen pre-conception

Some of the respondents had seen no women with PKU pre-conception, while others saw women with PKU only in the pre-conception period and not at other times during pregnancy. However, less than half of the respondents (15/32) saw a percentage of the women with PKU pre-conception. The majority of genetic counselors (10/15) who saw women preconception spent at least some of their clinical time in prenatal genetic counseling. For women who were seen pre-conception, all of the genetic counselors (15/15) discussed risks due to uncontrolled phe levels, 14/15 discussed risk to have a child with PKU, 7/15 discussed referrals to other health care providers, 5/15 percent discussed insurance coverage and 9/15 percent discussed social support (Figure 4).
Figure 4. (n=15). What was discussed in genetic counseling sessions. All genetic counselors discussed risks due to uncontrolled phe levels, and most discussed risk to have a child with PKU. The majority also discussed social support.

Care continued postnatally

Nine respondents (9/35) saw at least a percentage of the women with PKU postnatally. The majority of women that were seen at other times, pre-conception or during pregnancy, were not also seen postnatally. For those respondents who saw some women with PKU postnatally, 1 respondent had also seen women with PKU prenatally, 7 respondents had seen them both preconception and prenatally and 1 respondent saw the woman with PKU only postnatally. Six of the 7 respondents who saw women with PKU in all three time frames, preconception, prenatally, and postnatally, spend at least some of their time in metabolism clinic. The percentage of time spent in pediatric genetic counseling was positively correlated with the percentage of women seen postnatally (p= .015, r= .425).
Open ended responses about what was discussed when women were seen postnatally included continued PKU care and risk reduction for future pregnancies. One responded stated that they discussed “Concern for future children, need for good control of phe values; risks associated with uncontrolled phe levels. Give them encouragement and praise for the levels they were able to accomplish during a past pregnancy.”

**Perspectives on when genetic counselors should be involved in care**

Genetic counselors felt that, from their perspective, a genetic counselor should be involved in the care of women with PKU for a variety of reasons (Figure 5). All of the genetic counselors who answered this question (n=31) felt that genetic counselors should be involved pre-conception to discuss maternal PKU, twenty-nine felt that they should discuss recurrence risk pre-conception, and 25 felt they should be involved prenatally to discuss maternal PKU. Six genetic counselors felt that genetic counselors should be involved in the care of women with PKU prenatally only for reasons not related to maternal PKU, but these respondents also felt that genetic counselors should be involved in maternal PKU care for other reasons, which was difficult to interpret, but may involve confusion with the way the question was asked. Twenty-seven genetic counselors felt that genetic counselors should be a part of the team caring for women with PKU during their pregnancies.
Additional comments about what the genetic counselors role has been or should be in caring for women with PKU

“I function as the clinic coordinator for the PKU clinic, which at our institution is a multidisciplinary team with metabolic geneticist, nutritionists, and social workers. We work together with the common goals of helping the patient keep her PKU well controlled prior to conception and throughout pregnancy. I provide education to the patient regarding the risks of maternal PKU, and answer genetic counseling questions about recurrence risk and carrier testing. I write the visit reports, which are send to the obstetrician caring for the woman. We discuss maternal PKU with all women with PKU at their regular follow-up visits starting in the teenage years.” [sic] (Genetic counselor with more than 10 years experience who
spends 80 percent of clinical time in metabolism and 20 percent in pediatrics and had seen more than 10 women with PKU in the past 5 years.

“We do not have a nutritionist on staff so diet follow-up was done long distance. I served as liaison, support person for patient, monitor of levels, etc as well as initial counselor about risk for PKU in baby. Both our patients were non-compliant so served as counselor re: findings on U/S of microcephaly and IUGR” (Genetic counselor with >10 years experience, who had seen between 2 and 5 women with PKU, all prenatally).

“education plays a big role, patients know about diet/diet resources, but often are not prepared for the future, such as dealing with diet with morning sickness, managing work, diet, fatigue during pregnancy. Many are not sure about the postnatal testing process, whether or not to have newborn screening as the definitive test or a serum phenylalanine and when. much supportive counseling is needed to help women (especially those who were not on diet/controlled diet prior to pregnancy), as executive functions are likely impaired.” (Genetic counselor with >10 years experience, who had seen >10 women with PKU and spends 100% of clinical time in metabolism. Eighty-five percent of women with PKU seen were preconception.)

“The majority of women were seen due to their PKU status, however I have had a patient who presented due to AMA and had not told her OB she has PKU.” (Genetic counselor with <5 years experience, saw 2-5 women with PKU and spends 100% of clinical time in prenatal).

“I think all of the answers depend on the patient. My patient was working closely with the metabolic clinic prior to the pregnancy in order to reach optimal status in the preconception period. Due to her close connections with the metabolic team she had genetic counseling regarding maternal PKU and recurrence risks prior to the pregnancy so the only issue that she felt needed to be addressed with me was her age. All other issues were being handled by the metabolic clinic. So as a prenatal counselor in an MFM clinic, for that type of patient I really only needed to address her issues that were not related to PKU once I learned that she was fully educated about PKU and had no
further questions. However, if a pt were not working with her local metabolic clinic and had not been in touch with them since childhood, I would be obligated to address risks of maternal PKU, recurrence risks, as well as any other non-PKU issues. In addition I would need to connect that person with the local metabolic clinic to hopefully get back on track. So I do believe the answers really depend on the particular patient's needs.” (Genetic counselor with >10 years experience who had seen 1 woman with PKU and spends 100% of clinical time in prenatal).

“As a prenatal counselor, I believe that my role should be to discuss the diagnosis, inheritance pattern of PKU, clinical testing that is available to the patient and her partner, what the population carrier frequency is in order to determine the actual risk for a pregnancy to be affected, the risk to be a carrier for each of her pregnancies, and testing that can be offered prenatally to determine if a baby is affected or unaffected.” (Prenatal genetic counselor with <5 years experience who saw 1 woman with PKU.)

“I hope that the women in my area with PKU are being seen by a metabolic specialist, because I have only seen 1 in the past 12 years.” (Genetic counselor with >10 years experience, saw 1 woman with PKU and spends 90% of clinical time in prenatal and 10% in pediatrics.)

Survey of Women with PKU

Demographics

Sixteen women with PKU who have had at least one pregnancy completed our survey. All of the respondents identified their race as White/Caucasian. Eleven respondents live in the United States and four live outside the United States. All of those who responded to the question about education level had completed at least some college. Seven respondents are employed full time, 6 are employed part time, 1 is a student, and 2 do not work outside the home.
Information about the respondent’s first pregnancies

Fourteen respondents were married during their first pregnancy. Seven respondents had had one pregnancy, seven had had 2 pregnancies, one respondent had had 3 pregnancies and one respondent had had 6 pregnancies. Ten respondents had planned their first pregnancy, 5 had not and one did not respond to this question. Twelve of the pregnancies were continued, 2 miscarried, 1 was terminated due to personal reasons, and one individual did not report pregnancy outcome. Eleven (n=16) respondents reported that they used formula and were on a low phe diet by 8 weeks of the pregnancy and ten reported that they continued this throughout the pregnancy. All but one respondent who had planned her pregnancy reported that they used formula and were on a low phe diet by 8 weeks of the pregnancy. Two respondents had children that were born premature, none had children born with medical problems that seemed to be related to maternal PKU in their first year and none of the infants required surgery.

Experiences with Genetic counseling before first pregnancy

Nine of sixteen respondents had genetic counseling before their first pregnancy. Five respondents had genetic counseling with a genetic counselor and four had genetic counseling with a doctor. Eight of the 9 respondents who had genetic counseling before their first pregnancy had genetic counseling about their PKU, and 1 had genetic counseling about something unrelated. Seven of 9 respondents discussed both risks due to uncontrolled diet during pregnancy and chance to have a child with PKU, while 2 of the 9 women discussed referrals to other health care providers and social support (Figure 6).
Experiences with genetic counseling during pregnancy

Six respondents had genetic counseling during their first pregnancy. Two individuals had genetic counseling with a genetic counselor, 1 had genetic counseling with a doctor, 1 individual had genetic counseling with a dietician, 1 had genetic counseling in a PKU clinic and one did not specify. Five of the 6 individuals who had genetic counseling during their first pregnancy had genetic counseling about PKU and one individual did not specify what her genetic counseling was about. All 5 of these individuals discussed risks due to uncontrolled diet, 4 discussed the chance to have a child with PKU, 4 discussed referrals to other healthcare providers, and 4 discussed social support (Figure 7).
One individual who had genetic counseling with a genetic counselor felt that the involvement of the genetic counselor was very appropriate and that the whole experience was valuable. The other individual who had genetic counseling with a genetic counselor did not comment on the appropriateness of the genetic counselors involvement and felt that she was already educated about her PKU prior to the genetic counseling.

*Satisfaction with genetic counseling*

Eight individuals answered questions about their satisfaction with genetic counseling before their first pregnancy. Five of these individuals had their genetic counseling with a genetic counselor, while the other 3 had genetic counseling with a physician. Examining frequencies of answers to these questions showed that all 5 of the individuals who had genetic counseling with a genetic counselor either strongly agreed or agreed that he/she took their problems seriously, that he/she helped as much as they could, that he/she explained things carefully, and that they could tell the genetic
counselor the truth without worrying. All 5 of the individuals who had genetic counseling with a genetic counselor either strongly disagreed or disagreed that the genetic counselor could never understand them or that he/she was always in a hurry.

Six respondents had had genetic counseling during their first pregnancy and two of these were with a genetic counselor. This did not provide enough data to look at responses from questions pertaining to satisfaction of genetic counseling.

*Perspective of those who had not had genetic counseling*

Four respondents had not had seen a genetic counselor in any of their pregnancies. Of these respondents, 3 felt that a genetic counselor may have explained things carefully, 3 felt they could tell a genetic counselor the truth without worrying, and 3 thought they might have felt nervous when they talked to a genetic counselor.
Discussion

The purpose of this study was to examine the role of genetic counselors in the care of women with PKU surrounding their pregnancies from both the patient and genetic counselor’s perspectives. It is important to determine these perspectives in order to assess how genetic counselors are currently involved and if they should be more involved in the care of women with PKU in the future. Genetic counselors are educated about PKU, recurrence risks, teratogens and supportive counseling, all of which are necessary in caring for women with PKU. Due to this, our hypothesis was that genetic counselors play a valuable role and can enhance overall care of women with PKU surrounding their pregnancies. Our study shows that genetic counselors believe they should be involved in care of women with PKU and the majority felt that genetic counselors should be involved in PKU clinics. In addition, women with PKU indicated that they have had satisfactory experiences with genetic counseling.

Where are women with PKU being seen by genetic counselors?

The major practice areas reported by genetic counselors who responded to the survey were prenatal, pediatric, and metabolic genetics. In particular, almost half of the respondents spent more than 50% of their time in pediatrics or metabolism which greatly exceeds the proportion of all genetic counselors in these two practice areas according to the NSGC Professional Status Survey. This is not surprising however because, while these may not be typical settings in which genetic counselors see patients about
pregnancy related risks, these are the areas where individuals with PKU are most likely to be seen for long term management.

Genetic counselors reported seeing women with PKU in a variety of clinical settings and in conjunction with varying other healthcare providers depending on where they were seen. The majority of genetic counselors who did not report that a nutritionist or dietician were involved in the care of women with PKU spent 100 percent of their time in prenatal genetic counseling. This was expected and is likely due to the fact that they are not directly in PKU clinics. These women seen by primarily prenatal genetic counselors may also be cared for in a PKU clinic in which they are being followed by a nutritionist or dietician during pregnancy. For those that are not involved in a PKU clinic, it may be that prenatal genetic counselors are referring them to one for their dietary management.

Genetic counselors who spent time in metabolism were more likely to have seen more than five woman with PKU in the past 5 years, indicating that at least some women with PKU are likely being seen by genetic counselors in PKU clinics. It is apparent that the majority of genetic counselors do not see women as part of a PKU clinic and it is unclear from the scope of this study whether the majority of women with PKU seen by genetic counselors in other settings are also seen in PKU clinics.

*How many women with PKU do genetic counselors see?*

The majority of genetic counselors in this study had seen 1-5 women with PKU in the past 5 years. Only 20 percent of respondents had seen more than 10 women with PKU in the past 5 years and these genetic counselors were more likely to have spent at least
some time in a metabolism clinic. This is as we had anticipated given that genetic
counselors who are involved in metabolism, by definition, would see more patients with
PKU. Being seen by a genetic counselor in a PKU clinic may offer continuity to these
patients if they see the same genetic counselor prior to and throughout pregnancy or even
from childhood, providing better care for these women. It is likely, however, that many
women with PKU are not seen by a genetic counselor for pregnancy related risks and
while not all PKU clinics have genetic counselors as part of their team, the majority of
respondents felt that genetic counselors should be involved in the team providing care to
women with PKU during their pregnancies.

What is discussed in genetic counseling sessions and what role do genetic counselors
currently provide?

The indication that genetic counselors reported for seeing women with PKU was
most often related to maternal PKU but a fairly large proportion of the women (9/32)
were not seen specifically for maternal PKU. All genetic counselors who saw women
with PKU pre-conception discussed risks due to uncontrolled phe levels, all but one
 genetic counselor discussed risks to have a child with PKU, and majority of genetic
counselors (9/15) discussed social support, which may be helpful for gaining phe control
earlier in pregnancy (Rohr, et al., 2004). In addition, the results from the survey of
women with PKU showed that five women with PKU had genetic counseling with a
genetic counselor before their first pregnancy. Four of these individuals discussed risks
due to maternal PKU and the one respondent who did not discuss this had genetic
counseling about something unrelated to her PKU. The results from both surveys
demonstrate that genetic counselors are educated about maternal PKU and are discussing
maternal PKU risks with these women as the primary topic of discussion during their genetic counseling sessions.

One genetic counselor commented on the importance of education for women with PKU, particularly for the preparation of pregnancy and post natal testing of the infant for PKU. Another genetic counselor saw a woman with PKU who was followed in a PKU clinic and felt that the topics related to maternal PKU had already been discussed prior to the genetic counseling session. This demonstrates that the genetic counseling for women with PKU may vary based on their prior care and the genetic counselor involved may need to assess what should be discussed and tailor the content to the needs of each patient. Even if women with PKU have already discussed risks associated with maternal PKU, it may be helpful for the genetic counselor to assess whether the women with PKU are receiving enough support during her pregnancy and to determine which aspects of maternal PKU have previously been discussed.

Referrals to other health care providers and coordination of care are another role that can be filled by genetic counselors. One genetic counselor stated, “We do not have a nutritionist on staff so diet follow-up was done long distance. I served as liaison, support person for patient, monitor of levels, etc. as well as initial counselor about risk for PKU in baby.” This is important due to the complexity of pregnancy for women with PKU and it is clear that some genetic counselors are currently filling this role. Another genetic counselor had an experience where a patient had not told her obstetrician about her PKU and had presented to the genetic counselor because of her age. In this situation, the genetic counselor was educated about maternal PKU and would have been able to contact the obstetrician to discuss it and make him aware of the risks so the woman could be
properly care for from that point forward. If genetic counselors were involved with care of women with PKU prior to pregnancy this situation would not have occurred and the outcome may have been better for this woman.

*Postnatal care of women with PKU*

Given that individuals with PKU may have executive dysfunction (Azadi, Seddigh, Tehrani-Doost, Alaghband-Rad, & Ashrafi, 2009) and children prenatally exposed to increased levels of phenylalanine may have difficulties with behavioral regulation and are susceptible to having ADHD (Antshel & Waisbren, 2003) it would be beneficial to continue care postnatally for women with PKU. Only 9/33 genetic counselors in our study had seen women with PKU postnatally. This may be because the involvement of the genetic counselors in the care of these women were in a prenatal genetic counseling setting, but even the genetic counselors in a metabolic clinic saw only up to 50% of women with PKU postnatally. The highest percentage of women with PKU seen postnatally by any of the genetic counselors was 50 percent of the women with PKU they had seen. Seven respondents saw women with PKU prenatally, pre-conception and postnatally and six of these respondents spent clinical time in metabolism, which is as we would have expected in that women seen in PKU clinics would be seen more continuously.

Care of women with PKU seems to be the most inconsistent in the postnatal period, and it has been shown that postnatal phe regulation may be important in the outcome of the children born to mothers with PKU (Rohr, et al., 2004). This care, although not involving counseling about a genetic risk to the child, could be given by a
because they are involved in providing supportive counseling for their patients as well (www.nsgc.org). This may be particularly applicable if the genetic counselor had seen the women pre-conception, during pregnancy or both. This expanded potential role for genetic counselors in PKU clinics is already being filled by some of the genetic counselors who were surveyed as evidenced by the respondent who commented on this. … “much supportive counseling is needed to help women (especially those who were not on diet/controlled diet prior to pregnancy), as executive functions are likely impaired.”

*Genetic Counselor’s perceptions of how genetic counselors should be involved in maternal PKU care.*

It is clear both from the patient perspective and the survey of genetic counselors that not all women with PKU are being seen by a genetic counselor. However, the responses from the genetic counselors surveyed indicate that there is a need for genetic counselors to be involved in the care of women with PKU.

Twenty-seven (27/31) genetic counselors felt that counselors should be involved in the team providing care to women with PKU during pregnancy. It is clear that the majority of respondents felt that genetic counselors should be involved in care of women with PKU pre-conception and prenatally to discuss maternal PKU risks and to discuss risks to have a child with PKU. One respondent stated that her role of a prenatal counselor involved mainly discussing risk to a child to have PKU, but it is clear that most respondents feel that maternal PKU risks should also be discussed by genetic counselors.
The majority of genetic counselors (27/31) thought that genetic counselors should be involved in the care of women with PKU as part of the clinic that provides longitudinal care, which could potentially include pre-conception, prenatal, and possibly postnatal counseling. This supports the idea that genetic counselors perceive that the best patient care is not a one time only experience to discuss genetic risks to offspring.

Six respondents felt that genetic counselors should only be involved in care of women with PKU prenatally for reasons not related to maternal PKU. Interestingly, these respondents also felt that genetic counselors should be involved in maternal PKU for other reasons, so it is difficult to tell if the question was unclear and the respondents felt that genetic counselors should be involved in care for reasons unrelated to maternal PKU as well as for reasons related to maternal PKU.

**Women with PKU demographics and pregnancy outcome**

It was surprising that the majority of women with PKU had a college education as this is not representative of education level of the general population. It was also surprising that only 11/16 women were on a therapeutic diet by eight weeks of pregnancy and 10/16 continued the diet throughout the pregnancy, but none of the infants had any medical problems. This may be due to the very small sample or to recall bias of the women taking the survey. The majority of women who planned their pregnancy as well as 2 who had not were on a PKU diet by 8 weeks in pregnancy, but not all of the women were. This may be due to the multiple barriers to diet control and challenges of treatment which includes social issues, possible behavioral concerns, and access to care and diet
(Clarke, 2003). In this study we did not examine specific barriers to beginning or remaining on diet.

*Are women with PKU satisfied with their genetic counseling experience?*

Although the sample size was small, the majority of women with PKU responded positively about their genetic counseling experience. We were particularly interested in the satisfaction of genetic counseling with those who had seen a genetic counselor. Responses of those individuals who had genetic counseling with a genetic counselor before their first pregnancy showed that all 5 of these individuals had positive experiences in that they felt the genetic counselor took their problems seriously, helped as much as he/she could and explained things carefully. None of these respondents felt the genetic counselor could never understand them or was always in a hurry. All of these responses portray a good experience with genetic counseling and a sense of satisfaction with their interaction with a genetic counselor. All genetic counselors discussed risks due to uncontrolled diet, and although we are not aware from this survey if this is the first time they had received this information, we can see that genetic counseling is one context in which this information can be provided. In addition, the few women who had not seen a genetic counselor indicated in their responses that they felt it would have been useful. This illustrates the fact that genetic counseling was a valuable experience for the women who received it, and was perceived as potentially valuable by those who had not.

*Limitations*

The greatest limitation of the study was the small sample size for both the genetic counselors and women with PKU surveys. The nature of the study did not allow us to
analyze the two surveys together because the women with PKU surveyed were not seen
by the same genetic counselors who had been surveyed. The retrospective nature of the
survey may also result in limited recollection or recall bias of the experiences and
feelings of women with PKU as well as the involvement of genetic counselors in care of
women with PKU.

Genetic counselors were recruited through the general National Society of
Genetic Counselor’s (NSGC) listserv. Not all genetic counselors participate in the NSGC
listserv, which was a limitation for recruitment. NSGC also has a special interest group
(SIG) for genetic counselors who practice in the area of metabolism and this group was
not specifically targeted for recruitment. The results of a survey of this group of
counselors, who are more likely to care for women with PKU, could be helpful in more
fully understanding the genetic counselor’s scope of practice. At the same time, our
sample did have a larger percentage of genetic counselors involved in metabolism than
the percentage of these individuals in the field as a whole, so we know we were able to
reach some of these respondents.

For the survey of women with PKU, there may have been an ascertainment bias
given that the recruitment notice explained that this is a survey about genetic counseling.
Individuals who have not had genetic counseling may have seen this and assumed they
were not eligible for the survey. In addition, online recruitment methods may have
limited the participants to those of a certain socioeconomic status or with access to the
survey. Women with PKU were also recruited through the PKU listserv, which may have
created an ascertainment bias based on the population that may be taking part in that
listserv to begin with.
Future Directions

Future directions in this area could include a more extensive look into patient perceptions of the role of the genetic counselor either with a larger scale survey or a qualitative based approach involving interviews of women with PKU who have had genetic counseling surrounding a previous pregnancy. More data is needed in this area to determine the extent of involvement of genetic counselors and the patient’s perspective on the satisfaction of their care as well as the potential for additional genetic counselor involvement.

One genetic counselor commented on how she has only seen one woman with PKU in the past 12 years and that she hoped they were being seen in PKU clinics. It is unclear how many women with PKU of childbearing age continue care in PKU clinics. Future research could include determining what percentage of these women have ongoing care. This can help to determine strategies for extending care to those women either through PKU clinics or alternate education, such as prenatal or pediatric genetic counselors.

In order to further the examination of the role for genetic counselors in maternal PKU, we propose a direct comparison follow up study. The study format that would best help us to further assess of the role of genetic counselor in maternal PKU would be to interview or survey women with PKU and their genetic counselors, so that the interview information would be linked. This would allow us to examine the role of genetic counselors in maternal PKU with direct comparisons of outcomes of the pregnancies and patient satisfaction.
Conclusions

In this study we examined the role of genetic counselors in the care of women with PKU before, during, and after their pregnancies from the perspective of the genetic counselor and from women with PKU. This was the first study to look at genetic counselor’s role in maternal PKU and provides a description of when women with PKU are seen by genetic counselors, what is discussed, what the role of genetic counselors has been and perceptions about what it should be in the future.

The reasons for which women with PKU are seen by genetic counselors are variable, but maternal PKU is discussed even when it is not the indication for the visit. Of women with PKU who are seen by genetic counselors, the majority are seen by a counselor who spends most of their clinical time in a prenatal setting and are seen only during pregnancy. The majority of genetic counselors felt that genetic counselors should be involved in the care of women with PKU preconception, prenatally and as part of the team involved in care of women with PKU during their pregnancies. Genetic counselors as part of metabolism clinics more often saw women before and after pregnancy, which provides women with more continuous and better care than those seen only during pregnancy. Yet, only a small percentage of all practicing genetic counselors spend time in this clinical setting. We feel this is an important role that can be filled by genetic counselors and women with PKU benefit from care of genetic counselors preconception, prenatally and postnally.
References


Appendix A: Women with PKU Recruitment Notice

Are you a woman with PKU who has had at least one pregnancy? If so, I would like to invite you to participate in a survey involving your experiences with genetic counseling.

I am a graduate student in the genetic counseling program at Brandeis University. I am seeking volunteers to participate in a study about women with PKU who have been pregnant and their experiences with genetic counseling. If you have no experience with genetic counseling your input is still valuable and all women with PKU who have at least one pregnancy are invited to participate. The goal of this study is to determine if and how genetic counselors should be involved in the care of women with PKU during their pregnancies in order to provide the best care for women with PKU.

The study consists of an online survey which should take approximately 20 minutes to complete. You will be asked questions about your first pregnancy. You will also be asked if you have had any genetic counseling and what your feelings about the experience were. If you have not had genetic counseling, a brief explanation of what genetic counselors do will be provided and you will have the opportunity to share whether you feel it would have been a valuable experience during your pregnancy.

Participation in this study is completely voluntary and confidential. You may discontinue the survey at any time for any reason.

If you are interested in sharing your experiences and feelings about genetic counseling during your pregnancies please follow the link below to access the online survey.

http://atrial.qualtrics.com/SE/?SID=SV_4PARU4fnTucbzCs

If you have any questions of comments or would like more information please contact me by email at hml30@brandeis.edu or the Brandeis University Faculty Sponsor Gretchen Schneider at gretchen@brandeis.edu.

Thank you for your consideration!

Sincerely,

Holly Bellerose, Master’s Candidate

Genetic Counseling Program, Brandeis University
Appendix B: Genetic Counselor Recruitment Notice

Have you been involved in the care of a woman or women with Maternal PKU? If so, I invite you to participate in a research study regarding the role of genetic counselors in Maternal PKU.

I am a graduate student in the genetic counseling program at Brandeis University. I am seeking volunteers to participate in a study regarding genetic counselor’s role in the care of women with PKU during their pregnancies. The goal of this project is to determine the role of genetic counselor’s in Maternal PKU care from the genetic counselor’s perspective as well as the patient perspective. Participation is open to all genetic counselors who have been involved in the care of at least one woman with PKU during her pregnancy.

The study will consist of one online survey questionnaire which should take approximately 20 minutes to complete. Participation in the study is completely voluntary and confidential. You may discontinue the survey at any time for any reason.

If you are interested in sharing your experiences with your care of women with PKU during their pregnancy, please follow the link below to access the online survey.

http://atrial.qualtrics.com/SE/?SID=SV_0vMADBXnTc7zEBm

If you have any questions of comments or would like more information please contact me by email at hml30@brandeis.edu or the Brandeis University Faculty Sponsor Gretchen Schneider at gretchen@brandeis.edu.

Thank you for your consideration!

Sincerely,

Holly Lovendale, Master’s Candidate
Genetic Counseling Program, Brandeis University
Appendix C: Survey for Women with PKU

Thank you for participating in this study. In this study you will be asked questions about your first pregnancy and about your feelings about genetic counseling. All information will remain anonymous and confidential. All information will be added together with answers from other participants. Please note that participation is voluntary and you may exit the survey at any time. By clicking next you have acknowledged that you have read the above information and that you wish to participate in the survey.

Q1 Have you ever been pregnant?
○ Yes (1)
○ No (2)

Q2 Do you have PKU?
○ Yes (1)
○ No (2)

Q3 What is your race?
○ White/Caucasian (1)
○ African American (2)
○ Hispanic (3)
○ Asian (4)
○ Native American (5)
○ Pacific Islander (6)
○ Other (7)

Q4 Your Age:
If Age: Is Less Than 18, Then Skip To End of Survey
Q5 What is the highest level of education you have completed?

☐ Less than High School (1)
☐ High School / GED (2)
☐ Some College (3)
☐ 2-year College Degree (4)
☐ 4-year College Degree (5)
☐ Master's Degree (6)
☐ Doctoral Degree (7)
☐ Professional Degree (JD, MD) (8)

Q6 Employment status:

☐ employed full time (1)
☐ employed part time (2)
☐ not employed outside the home (3)
☐ student (4)
☐ other please describe (5) ____________________

Q7 Number of pregnancies you have had
Q8 In what state do you currently reside?

- Alabama (1)
- Arizona (2)
- Arkansas (3)
- California (4)
- Colorado (5)
- Connecticut (6)
- Delaware (7)
- District of Columbia (8)
- Florida (9)
- Georgia (10)
- Idaho (11)
- Illinois (12)
- Indiana (13)
- Iowa (14)
- Kansas (15)
- Kentucky (16)
- Louisiana (17)
- Maine (18)
- Maryland (19)
- Massachusetts (20)
- Michigan (21)
- Minnesota (22)
- Mississippi (23)
- Missouri (24)
- Montana (25)
- Nebraska (26)
- Nevada (27)
- New Hampshire (28)
- New Jersey (29)
- New Mexico (30)
- New York (31)
- North Carolina (32)
- North Dakota (33)
- Ohio (34)
- Oklahoma (35)
- Oregon (36)
- Pennsylvania (37)
- Rhode Island (38)
- South Carolina (39)
Q9 What was your marital status during your first pregnancy?

- Single, never married (1)
- Married without children (2)
- Married with children (3)
- Divorced (4)
- Separated (5)
- Widowed (6)
- Living w/ partner (7)

Answer Invalid Logic Click Here to Edit Logic

Q10 Please answer the following questions about your FIRST pregnancy.

Q11 Was your first pregnancy planned?

- Yes (1)
- No (2)
Q12 What was the outcome of your first pregnancy?

- I chose to continue the pregnancy and gave birth (1)
- I chose to terminate the pregnancy (2)
- I lost the pregnancy (miscarriage) (3)

If I chose to continue the pregnancy, then skip to Were you on a therapeutic PKU diet by... If I chose to terminate the pregnancy, then skip to What was the reason you chose to term... If I lost the pregnancy, then skip to Were you on a therapeutic PKU diet by...

Q13 What was the reason you chose to terminate the pregnancy?

Answer If What was the outcome of your first pregnancy? I chose to continue the pregnancy and gave birth Is Selected

Q14 Were you on a therapeutic PKU diet by 8 weeks of your pregnancy?

- I was not on a PKU diet (1)
- No PKU diet/ but did use formula (2)
- I used formula and was also on a diet with low PHE foods (3)
- Other - please explain (4) ____________________

Answer If What was the outcome of your first pregnancy? I chose to continue the pregnancy and gave birth Is Selected

Q15 Were you able to continue a therapeutic PKU diet throughout the pregnancy?

- I was not on a PKU diet but I did use formula (1)
- I was not on a PKU diet (2)
- I used formula and was also on a diet with low PHE foods (3)
- Other - please explain (4) ____________________

Answer If Were you able to continue a therapeutic PKU diet throughout the pregnancy? I was not on a PKU diet but I did use formula Is Selected

Q16 How old is your child now?
Q17 What is your child's gender?

- Male (1)
- Female (2)

Q18 Was your child born premature?

- Yes (1)
- No (2)

Q19 Did your baby have any medical problems within the first year after he or she was born?

- Yes- please explain the type of medical problems (1) ____________________
- No (2)

Q20 Did your child require surgery within the first year after he or she was born?

- Yes- what was the reason for surgery? (1) ____________________
- No (2)

Q21 Please read the following.

Definition of Genetic Counseling: Genetic counseling is used to help people understand genetic disorders. It may include:

- Talking about family and medical histories and the chance for other people in the family to have the same genetic disorder.
- Talking about how genetic disorders are passed on, testing, management, prevention, resources and research.
- Counseling to help people make informed choices and to deal with the genetic disorder.
Q22 Genetic counseling can be provided by a variety of people, but there are some specially trained individuals with a Master’s degree in genetic counseling, who are called genetic counselors.

Q23 Who did you have genetic counseling with?
- a genetic counselor (1)
- a doctor (2)
- a nurse (3)
- other- please explain (4) ____________________

Q24 Did you have genetic counseling BEFORE your first pregnancy?
- Yes (1)
- No (2)
If No Is Selected, Then Skip To End of Block

Q25 Did you have genetic counseling about PKU or for another reason?
- about PKU (1)
- another reason (2) ____________________
If about PKU Is Selected, Then Skip To What did you discuss during your gene...If another reason Is Selected, Then Skip To Please check how much you agree or di...

Q26 What did you discuss during your genetic counseling? Please check all that apply.
- Risks due to uncontrolled diet during pregnancy (1)
- chance to have a child with PKU (2)
- referrals to other health care providers (3)
- social support (4)
- other- please explain (5) ____________________
Q27 Please check how much you agree or disagree to the following statements about the person you had genetic counseling with.

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<tr>
<th>Statement</th>
<th>Strongly Disagree (1)</th>
<th>Disagree (2)</th>
<th>Neither Agree nor Disagree (3)</th>
<th>Agree (4)</th>
<th>Strongly Agree (5)</th>
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<td>He/She took my problems seriously (1)</td>
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<td>He/She was always in a hurry when I saw him/her (5)</td>
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<td>He/She explained things carefully (6)</td>
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<td>He/She sometimes said things I did not understand (7)</td>
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<td>I could tell him/her the truth without worrying (8)</td>
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Q28 Did you have genetic counseling DURING your first pregnancy?
- Yes (1)
- No (2)
If No Is Selected, Then Skip To End of Block

Q29 Who did you have genetic counseling with?
- a genetic counselor (1)
- a doctor (2)
- a nurse (3)
- other- please explain (4) ____________________

Q30 What was the reason you had genetic counseling? Please check all you apply.

- Something was seen on an ultrasound (1)
- I was over 35 years old (2)
- I have a family history of another genetic condition (3)
- I was exposed to something during pregnancy not related to PKU (4)
- other-please explain (5) ____________________
- I had genetic counseling about my PKU (6)

Q31 Did you discuss PKU during your genetic counseling visit?
- Yes- please explain what about PKU was discussed (1) ____________________
- No (2)
Q32 What did you discuss during your genetic counseling? Please check all that apply.

- Risks due to uncontrolled diet during pregnancy (1)
- chance to have a child with PKU (2)
- referrals to other health care providers (3)
- social support (4)
- other- please explain (5) ____________________
Q33 Please check how much you agree or disagree to the following statements about the person you had genetic counseling with.

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I felt nervous when I talked to him/her (9)
I always felt well treated when I left his/her office (10)

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Answer If Who did you have genetic counseling with? a genetic counselor Is Selected

Q34 How would you describe the involvement of the genetic counselor during your FIRST pregnancy?

- Very Inappropriate (1)
- Inappropriate (2)
- Neutral (3)
- Appropriate (4)
- Very Appropriate (5)

Q35 What do you think was the most valuable part of your genetic counseling experience?

Q36 Did you have genetic counseling during ANY of your pregnancies?

- Yes- during my first pregnancy only (1)
- Yes- during a pregnancy other than my first pregnancy (2)
- Yes- during my first pregnancy and another pregnancy (3)
- No- I have never seen a genetic counselor (4)
Q37 Having seen a genetic counselor in a later pregnancy, how helpful do you think it would have been to see a genetic counselor during your FIRST pregnancy?

- Not at all helpful (1)
- Neutral (2)
- Very helpful (3)

Q38 Before this survey, had you heard of genetic counseling?

- Yes (1)
- No (2)

Q39 Do you know anyone who has had genetic counseling?

- Yes (1)
- No (2)
Q40 Please check how you feel about the following statements.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly Disagree (1)</th>
<th>Disagree (2)</th>
<th>Neither Agree nor Disagree (3)</th>
<th>Agree (4)</th>
<th>Strongly Agree (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A genetic counselor could never understand anyone like me (1)</td>
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<tr>
<td>A genetic counselor could have helped me (2)</td>
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<td>A genetic counselor may have explained things carefully to me (3)</td>
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<tr>
<td>A genetic counselor may have said things I did not understand (4)</td>
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<tr>
<td>I could tell a genetic counselor the truth without worrying (5)</td>
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<tr>
<td>I might have felt nervous when I talked to a genetic counselor (6)</td>
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</tbody>
</table>
Appendix D: Survey for Genetic Counselors

During this survey you will be asked questions about your role in the care of women with PKU before, during, and/or after their pregnancies. The goal of this survey is to gain knowledge about the genetic counselors’ role in the care of women with PKU surrounding their pregnancies. Your participation is greatly appreciated. By clicking next, you agree that you have read the above information and that you would like to participate in the survey. Thank you for your time.

Q1 What is your current age? (U.S. Census)

☐ Less than 16 (1)
☐ 16 to 19 (2)
☐ 20 to 24 (3)
☐ 25 to 34 (4)
☐ 35 to 44 (5)
☐ 45 to 54 (6)
☐ 55 to 64 (7)
☐ 65 or over (8)

Q2 What is your gender?

☐ Male (1)
☐ Female (2)
Q3 In what state do you currently reside?

- Alabama (1)
- Arizona (2)
- Arkansas (3)
- California (4)
- Colorado (5)
- Connecticut (6)
- Delaware (7)
- District of Columbia (8)
- Florida (9)
- Georgia (10)
- Idaho (11)
- Illinois (12)
- Indiana (13)
- Iowa (14)
- Kansas (15)
- Kentucky (16)
- Louisiana (17)
- Maine (18)
- Maryland (19)
- Massachusetts (20)
- Michigan (21)
- Minnesota (22)
- Mississippi (23)
- Missouri (24)
- Montana (25)
- Nebraska (26)
- Nevada (27)
- New Hampshire (28)
- New Jersey (29)
- New Mexico (30)
- New York (31)
- North Carolina (32)
- North Dakota (33)
- Ohio (34)
- Oklahoma (35)
- Oregon (36)
- Pennsylvania (37)
- Rhode Island (38)
- South Carolina (39)
☐ South Dakota (40)
☐ Tennessee (41)
☐ Texas (42)
☐ Utah (43)
☐ Vermont (44)
☐ Virginia (45)
☐ Washington (46)
☐ West Virginia (47)
☐ Wisconsin (48)
☐ Wyoming (49)
☐ Puerto Rico (50)
☐ Alaska (51)
☐ Hawaii (52)
☐ I do not reside in the United States (53)

Q4 How many years have you been in practice?
☐ Less than 5 (1)
☐ between 5 and 10 (2)
☐ more than 10 (3)

Q5 What percentage of your clinical time is spent in the following areas?
   ______ Prenatal (1)
   ______ Pediatrics (2)
   ______ Cancer (3)
   ______ Metabolism (4)
   ______ General genetics (5)
   ______ Other (6)

Q6 Who else was a part of the treatment team when you cared for a woman with PKU? Check all that apply for any of the pregnancies you treated.
☐ clinical geneticist (1)
☐ obstetrician (2)
☐ metabolic specialist (3)
☐ nutritionist (4)
☐ other, please explain (5) ____________________
Q7 How many women with PKU have you seen in clinic in the past 5 years?

- 1 (1)
- 2 - 5 (2)
- 5 - 10 (3)
- more than 10 (4)

If 1 Is Not Selected, Then Skip To End of Block

Q8 Was the woman you saw with PKU pregnant at your first encounter?

- Yes (1)
- No (2)

Q9 How did the woman present to you? Please check all that apply

- anomaly suspected to be caused by uncontrolled diet (1)
- Advanced maternal age (2)
- she was screen positive (3)
- she was part of the clinic I work in (4)
- other, please explain (5) ____________________

Q10 At the initial visit, was the pregnant patient accompanied by? Check all that apply.

- partner (1)
- parent (2)
- child (3)
- other (4)

Q11 At the initial visit, was the patient with PKU on a therapeutic diet?

- Yes (1)
- unknown (2)
- No (3)
Answer If At the initial visit, was the patient with PKU on a ther... Yes Is Selected

Q12 How long did they report being off a therapeutic PKU diet for?

- more than one month (1)
- more than six months (2)
- six months- one year (3)
- over one year (4)
- one to five years (5)
- over five years (6)

Q13 What did you discuss with the patient who had PKU? Please check all that apply.

- risks due to uncontrolled phenylalanine levels (1)
- risk to have a child with PKU (2)
- referrals to other health care providers (3)
- insurance coverage (4)
- social support (5)
- other, please explain (6) ____________________

Q14 In the past 5 years, what percentage of the women with PKU you saw were …

- pre-conception (1)
- currently pregnant (2)
- post natal (3)

Answer If In the past 5 years, what percentage of the women with PK... pre-conception Is Greater Than 0

Q15 For those women you saw pre-conception: What did you discuss with them? Please check all that apply.

- risks due to uncontrolled phenylalanine levels (1)
- risk to have a child with PKU (2)
- referrals to other health care providers (3)
- insurance coverage (4)
- social support (5)
- other, please explain (6) ____________________
Q16 Did you see them during their pregnancy as well?

- Yes (1)
- No (2)

Q17 For those who were pregnant at your first encounter, how did they present to you? Check all that apply.

- anomaly suspected to be caused by uncontrolled phenylalanine levels (1)
- anomaly not suspected to be related to PKU (2)
- advanced maternal age (3)
- she was screen positive (4)
- she was part of the clinic I work in (5)
- other, please explain (6) ________________

Q18 For those women with PKU who you saw post natal, what did you discuss with them?

Q19 Had you seen these women preconception or prenatally as well?

- preconception only (1)
- prenatally only (2)
- both preconception and prenatally (3)
- neither preconception nor prenatally (4)
Q20 From your perspective, when should genetic counselors be involved in the care of women with PKU with regards to their pregnancies? Please check all that apply.

- pre-conception to discuss maternal PKU (1)
- pre-conception to discuss recurrence risk of PKU (2)
- prenatal to discuss maternal PKU (3)
- prenatal only for another indication not related to maternal PKU (4)
- genetic counselors should be a part of the team providing care for women with PKU during their pregnancies (5)
- other, please explain (6) ____________________

Q21 From your perspective, in your clinical practice, what proportion of women with PKU have genetic counseling regarding their pregnancies?

- 0-20% (1)
- 20-40% (2)
- 40-60% (3)
- 60-80% (4)
- 80-100% (5)

Q22 Please include any other comments from your perspective about what your role has been or what you feel your role should be in caring for women with PKU?