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The Basis and Implications of Genetic Counseling in Patient Testing Choice

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The Basis and Implications of Genetic Counseling in Patient Testing Choice

A Capstone Project Submitted in Partial Fulfillment of the Requirements of the Renée Crown University Honors Program at Syracuse University

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Perinatal genetic counseling is a health service provided to patients who carry risk factors for genetic abnormalities for their offspring. There are several non-invasive and invasive tests offered to patients in order to provide genetic information about possible disorders. The non-invasive screens calculate a percentage of risk relative to average population risk. If the non-invasive screens predict an elevated risk, a more invasive test can be offered to obtain definitive results about possible disorders in the pregnancy. The most commonly used invasive test is called amniocentesis and it carries a risk for pregnancy complications. After receiving abnormal results on the non-invasive screens, patients have the option to continue with invasive testing to obtain definitive results.

This project sought to discern factors predictive of testing choice after abnormal screen results. From 134 patient charts that fit certain criteria, several factors such as screen-calculated risk were analyzed and evaluated using a statistical analysis program.

Of the factors analyzed, a trend in the data can be seen. The higher a woman’s calculated risk of genetic abnormality in the fetus, the more likely she was to continue onto invasive testing.

This research has future implications for the field of genetic counseling. The more information there is concerning testing implications and testing choice, the better the care that health professionals will be able to provide.
Table of Contents

Acknowledgements..............................................i

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

Methods and Materials.........................................8

Results................................................................10

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

Works Cited........................................................22

Capstone Summary..............................................23
Acknowledgements

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I would also like to thank Karen Fay, M.S. for allowing me to sit in on many genetic counseling sessions and for explaining many facets of the genetic counseling profession to me.

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The Basis and Implications of Genetic Counseling in Patient Testing Choice

Introduction

Genetic counseling can be defined as a “process in which a genetic counselor educates families or individuals about their risk of passing on a genetic predisposition for certain disorders to future generations” (National Society of Genetic Counselors 2011). To achieve this goal, a genetic counselor will explain and discuss the patient’s current medical situation with the patient in order to inform and educate. During this counseling, the patients will be presented with the medical facts as well as a possible diagnosis. They will be taken through the likely progression of the disorder as well as possible treatments or cures. Another goal of the genetic counseling session is for the patient to understand the inheritance of their possible affliction and how their medical history has contributed to the possible occurrence. With clear explanation of this information, the genetic counselor will also provide future options, such as testing and screens or support and referrals to other specialists. Overall, the goal of the genetic counselor is to inform the patients of how genetic history can affect medical future.

Although genetic counseling can be provided to any patient for any medical situation, genetic counseling is especially commonly offered to pregnant women. There are many medical factors that might cause a woman to have a “high-risk pregnancy,” and if she does, she will likely be referred to a genetic counselor. The possible factors that can denote a possible high-risk
pregnancy are many, each with a medical basis that might affect the pregnancy adversely. The first is “advanced maternal age”, where the mother will be 35 years or more at the age of delivery. This is a contributor to a high-risk pregnancy, as many studies have shown that women over 35 have “an increase in intercurrent illness and pregnancy complications” (Jacobsson, Ladfors, and Milsom 2004).

Women over the age of 35 also have “an increased risk for miscarriage and for chromosomal abnormalities…and fetal/neonatal congenital anomalies” (Cleary-Goldman et al. 2005). As the mother ages, so do the viable cells that might be fertilized to produce a pregnancy. This aging in cells is related to cellular division, and cells that do not divide properly may result in chromosomal nondisjunction (Dailey et al. 1996). The normal number of chromosomes in a human is 46: 23 from the mother and 23 from the father. During meiosis, germ cells divide from these 46 chromosomes to haploid cells containing 23 chromosomes, and at fertilization, return to the diploid state of 46 chromosomes. If one chromosome pair does not divide properly, this results in nondisjunction, and the fetus resulting from fertilization will have an abnormal number of chromosomes. This abnormal number typically means that the fetus will have three or only one of a certain chromosome instead of two, and this results in fetal disorders, which denote a high-risk pregnancy.

Another factor indicative of a high-risk pregnancy is fetal abnormalities that might be observed on an ultrasound. “Routine ultrasound screening…has the potential advantage of detecting most major fetal
malformations” (Milunsky 2004). If such an anomaly is noted, a woman will be referred to a genetic counselor because of the high congruency of fetal abnormalities and genetic disease.

Previous pregnancy complications of the patient or the patient’s family can be indicative of a future high-risk pregnancy. If a woman has had one or more miscarriages or stillbirth or neonatal death in a previous pregnancy, this will be a cause for concern, and lead to a referral to a genetic counselor. Also, if there is a clear indication in the patient’s family medical history of a heritable genetic disorder, this will raise alarm in the possibility of a high-risk pregnancy with this same genetic disorder. Similarly, if there is a family history of structural anomalies, again there may be potential for a high-risk pregnancy. If the family medical history contains individuals that have presented with neural tube defects, congenital heart defects, or cleft lip and palate, there is potential for recurrence in a future pregnancy, indicating the need for discussion of risk with a genetic counselor.

Consultation is also recommended to the patient if the patient has an ethnic background with a disorder that occurs more commonly than in the general population. Examples of this include cystic fibrosis in Europeans, Tay-Sachs disease in Orthodox Jews and Eastern Europeans, and sickle-cell anemia in African Americans. If the patient is a member of such an ethnic group, a referral to a genetic counselor is recommended in that there can be prenatal diagnosis of a disorder or carrier testing for early detection of the disorder. A less likely family-related risk factor is consanguinity, where the
parents are related, for example if the parents are first cousins. Consanguinity produces an increased risk of fetal disorders, because the parents may both carry the same defective gene and can pass it on to the fetus. If the pregnancy is the result of a consanguineous union, then genetic counseling is recommended. Moreover, there are many other less common factors that might indicate high-risk pregnancies such as history of or suspected metabolism abnormality, history of or suspected chromosomal rearrangement, or a family history of mental retardation or developmental disabilities.

Currently it is routine to offer pregnant women serum screens that can test the mother’s blood for indicators of abnormalities in the pregnancy. Abnormal maternal serum marker screening results will cause a woman to be referred to a genetic counselor for discussion of risk in the pregnancy. After an abnormal result and consultation with the genetic counselor, the patient has the option to continue on to more invasive testing. Although there are many maternal serum marker screens, two of the most common are the Maternal Serum Alpha-Fetoprotein (MSAFP) and the Triple Screen.

The MSAFP test is a blood test that evaluates the levels of alpha-protein in the mother’s serum. This test is usually given between the 14th and 22nd weeks of pregnancy. The test recognizes abnormal levels of alpha-fetoprotein relative to a normal range and out-of-range values can predict the risk of a disorder in the fetus. This test has utility as a single analyte screen because if the alpha-fetoprotein level is high relative to the normal, this can
indicate the possible presence of a neural tube defect. Conversely, low levels of alpha-fetoprotein can indicate Down Syndrome.

The Triple Screen expands analysis from the MSAFP and measures two other analytes. This screen is primarily given to women who are between their 15th and 21st week of pregnancy, where it evaluates the levels of three analytes from the placenta and the fetus in order to predict elevated risk for the following fetal abnormalities; Down Syndrome (Trisomy 21), Edwards Syndrome (Trisomy 18), and Neural Tube Defects (NTD). Alpha-fetoprotein (AFP) is found in amniotic fluid and abnormal levels of this may indicate the presence of a fetal disorder. Human chorionic gonadotopin (hCG) is a protein produced by the placenta. Unconjugated estriol (uE3) is made in the fetus and placenta. Based on extensive studies of levels of these hormones, a prediction can be made as to the possible abnormality in a fetus. The screen can detect “Down Syndrome in 69% of cases and…neural tube defects in 80% of the cases” (American College of Obstetrics and Gynecology 2007). If the levels of all three analytes are low, this is an indication for Trisomy 18. Also, the test is dependant on the predicted age of the fetus, so an incorrectly assessed age could result in perceived abnormal protein levels.

It is a common misconception that, since these screens do not detect all cases of fetal abnormalities, these screens are not accurate. The screens accurately test analyte levels and return a calculated risk. Also, an elevated calculated risk does not absolutely mean that there is an abnormality in the fetus. The elevated calculated risk is the analyte level relative to baseline
population risk. Although they do not detect all cases of fetal abnormalities, their ability to detect possible fetal disorders makes them very useful to prenatal genetic screening.

Once either of these screens has established an abnormal level of analytes, possibly indicating a disorder in the fetus, the patient has the option to continue on to an ultrasound or a more invasive form of testing. Ultrasound uses sound waves to visualize the fetus and its body structures. Ultrasound is non-invasive and commonly used to detect abnormalities that have physical manifestations. Also, ultrasound allows for “the detection of not only major malformations but also subtle markers of chromosomal abnormalities and genetic syndromes” (Milunsky 2004). For example, fetuses with Down Syndrome often have thick skin at the posterior section of the neck (nuchal fold) and the size of this can be measured on an ultrasound. This procedure assists in diagnosis of possible genetic disorders in the fetus.

The most common invasive test performed in this situation is amniocentesis. Amniocentesis is considered invasive because the test entails the use of a needle to enter the amniotic sac and withdraw approximately 15 – 30 mL of amniotic fluid for testing. Under ultrasound guidance, the physician will find the precise location of the fetus, and point the needle towards an area of the amniotic sac that only contains fluid. Then the fluid is extracted and the needle is removed. The entire procedure takes about two minutes. The amniotic fluid that is withdrawn carries valuable information about the fetus, including the possible occurrence of Down Syndrome. Also, the test provides
a full karyotype of the chromosomes of the fetus, enabling chromosomal studies and prediction of possible genetic disorders. Although this test provides extremely useful diagnostic information, it carries risk factors for the pregnancy because of its invasive nature. These risk factors “can include rupture of the membranes and subsequent miscarriage” (Sloane 144). The estimated increase risk of miscarriage as a result of amniocentesis is approximately 0.2% (R. Lebel, personal communication). As amniocentesis carries the risk of pregnancy complications, the decision to continue from non-invasive to invasive testing is one that often takes much consideration on the part of the patient.

This decision to move from non-invasive testing to more definitive invasive testing in the case of a pregnancy with genetic disorder risk is one that entails many different factors. The various factors that might affect this decision are of particular interest to me, in that they may be predictive of testing choice and they can show the effect of testing on patient choice. While the actual factors that can affect this decision are numerous, the ones that are readily evaluated are age of the patient, the patient’s population risk for a disorder of the fetus, the MSAFP or Triple Screen screen-calculated risk, and whether previous children of the patient have genetic disorders. Another factor that might affect the decision to continue onto invasive testing is the income of the patient. There is a high correlation between income and education level (Day & Newburger 2002). Income is often an indicator of education level, and education level is often an indicator of the patient’s
ability to perceive statistics and calculated risk. As each of these affects patient outlook, they may influence a patient’s choice to continue with invasive testing after a non-invasive screen produces abnormal results.

**Methods and Materials**

In order to conduct a study that evaluates the possible factors that might affect patient testing decisions, subjects that fit certain criteria were obtained. Dr. Robert Roger Lebel provided his patient files that ranged from 1997 to 2003. Because they had reached the statutory limit for medical records after closing of a physician’s practice, these charts were all in the process of being destroyed. This rendered them beyond any possible contact to the patients, and thus satisfied the criteria of the Institutional Review Board of the SUNY Upstate Medical University, to allow use of identifying information. The protocol was approved.

These patients were seen in Dr. Lebel’s practice in Glen Ellyn, Illinois. From these patients, I selected those that fit specific criteria. Patients had elected to have either the Triple Screen or MSAFP test performed by their obstetricians. After having the test performed, they received an abnormal result and were referred to Dr. Lebel to discuss their result. After they had the consultation, they made the decision whether to have no more procedures at all, an ultrasound only, or ultrasound with amniocentesis. From the patient charts that fit these criteria, I recorded additional information on a sheet specific to each chart. I recorded the patient’s age at delivery and the
population risk that was told to the patient. Dr. Lebel told the population risk to the patient as based on Hook 1981. This value was given as a number denoting the possible occurrence of a genetic disorder at that age out of 1000 women of that age. I also recorded the patient’s decision after the discussion of their screen result. I recorded the result of the ultrasound if the patient chose to have one as well as the result of the amniocentesis if the patient chose to have one. Also I recorded the screen-calculated risk, given as 1 out of denominator, denoting the chance of a genetic disorder occurring in the fetus relative to baseline population risks for pregnant women of that age. I recorded whether the women had children previous to the pregnancy in question, and whether those children were healthy or not. Also recorded were the patient’s marital status, previous miscarriages, a family history of genetic disorders, a family history of miscarriage or stillbirth, and the patient’s ethnicity but preliminary analysis did not yield any results with statistical significance.

In total, 134 charts fit the selection criteria. Data were entered into a Microsoft Excel spreadsheet for the purpose of organization. Zip codes were used to estimate the median income of the patient’s neighborhood based on the 2000 Census data of average income (US Census Bureau 2000). The calculated risk and population risks were converted into percentages. For statistical analysis, IBM® SPSS® Statistics V. 18 was used. A one-way analysis of variance (ANOVA), was used to determine whether decision was affected by calculated risk or income. A chi-square analysis was done to
determine whether decision to choose an invasive test was dependent on the presence of previous unhealthy children. A regression analysis was done to examine the relationship between the age of patient and their screen-calculated risk.

**Results**

Figure 1 is a graph of the population risk depending on the age of the patient at delivery. The data are based on Hook’s values as told to the patients. Hook’s data shows that women over the age of 35 are at higher risk of a chromosomal abnormality occurring in their pregnancy. Before the age of 35, the risk of a disorder in pregnancy is relatively low and has a very gradual slope of increase until the age of 35. However, after the age of 35, the slope and risk of disorder in pregnancy increased exponentially. This explains the concern for pregnancies in women who are older and supports genetic counseling as an option for women who have pregnancies later in life. The population risk is calculated as a percentage, such as 0.1% chance of chromosome abnormality in the fetus, which corresponds to a graph value of 0.001.
Figure 1. Population risk of chromosomal abnormalities in the fetus by age of the mother at delivery. These values are based on Hook’s data (Hook 1981). Population risk is given as a percentage, where .001 means 0.1% chance of an abnormality.

Figure 2 depicts the distribution of patients making each possible decision. Out of 134 patients, 12 patients made the decision to not continue to any procedure at all and this is denoted by decision = 0. Twenty-nine patients made the decision to have ultrasound only and this is denoted by decision = 1. Ninety-three patients made the decision to have both an amniocentesis and an ultrasound and this is denoted by decision = 2. This shows that 9% of the patients surveyed chose no procedure, 22% chose to have ultrasound only, and 69% chose to have ultrasound and amniocentesis. Overall, 91% of the patients
chose to have some procedure, whether it was ultrasound only or both ultrasound and amniocentesis. Of those who did choose to continue on to a procedure, 24% chose to have ultrasound only and 76% chose to have an ultrasound and amniocentesis.

Figure 2. Graph of the number of decisions made in each decision category. 134 total patients, 12 chose no procedure (decision = 0). 29 chose ultrasound only (decision = 1). 93 chose ultrasound and amniocentesis (decision = 2).

Figure 3 shows the decision made when previous children were healthy versus unhealthy. Here, only those patients with previous children were considered. The plot shows that some patients made each decision option if their previous children were healthy. However, while the sample size is small, this also shows that none of the patients who had unhealthy children
chose to go without any procedure. This could indicate that the presence of a previously unhealthy child might influence the patient’s decision towards having some procedure rather than no procedure at all, but the chi-square test showed that these two factors were independent ($X^2=0.64; \text{df}=2; p=0.73$).

Figure 3. Previous children healthy or not versus decision made. For previous children healthy 0=healthy, 1=at least one unhealthy child. For decision, 0=no procedure, 1=ultrasound only, 2=ultrasound and amniocentesis. The numbers adjacent to the points indicate the number of patients in each category.

Figure 4 is a scatter plot of the patient’s age versus the screen-calculated risk is defined by personal medical history. The calculated risk is calculated as a percentage, such as 0.1% chance of genetic abnormality in the fetus, which corresponds to a graph value of 0.01. There was no relationship
between patient age and the calculated risk ($F=0.008; \text{df}=1; p=0.929$).

Indicating that the two factors are independent of each other

Figure 4. Age v. calculated risk. Calculated risk is given as a percentage, where .01 means 1% chance of an abnormality occurring in the fetus.

Figure 5 shows the calculated risk versus the decisions made.

Regardless of risk, most patients chose to have ultrasound with amniocentesis. Those who chose no procedure at all had lower risks, although these were not statistically significant ($F=0.391; \text{df}=2; p=0.677$). The calculated risk is calculated as a percentage, such as 0.1% chance of genetic abnormality in the fetus, which corresponds to a graph value of 0.01.
Figure 5. Calculated risk v. decision made. For decision, 0=no procedure, 1=ultrasound only, 2=ultrasound and amniocentesis. Calculated risk is given as a percentage, where .01 means 1% chance of an abnormality occurring in the fetus.

Figure 6 is a scatter plot of average income of zip code versus decision made. In this analysis, income had no influence on the decision made by the patient (F=0.461; df=2, 131; p=0.631).

Figure 7. Average income of each decision option. Income is in dollars. For decision, 0=no procedure, 1=ultrasound only, 2=ultrasound and amniocentesis.
Figure 8 is the average calculated risk of each decision option. Of each decision option, the average calculated risk was calculated and plotted. The calculated risk is calculated as a percentage, such as 0.1% chance of genetic abnormality in the fetus, which corresponds to a graph value of 0.01. The results indicate that there are no significant differences in calculated risk for the three decision categories (F=0.391; df=2,113; p=0.677). Although the data are not statistically significant, a trend can be seen where those with lower percentages choose no procedure. A Power Test was done which indicated that this trend would become statistically significant with a sample size of over 890.

Figure 8. Average calculated risk of each decision. For decision, 0=no procedure, 1=ultrasound only, 2=ultrasound and amniocentesis. Calculated risk is given as a percentage, where .01 means 1% chance of an abnormality occurring in the fetus.
Discussion

The previous results were organized by the logical progression of questions asked about the data and they will be discussed here in the same order. The goal of the study was to examine the factors affecting patient choice of non-invasive screens versus invasive testing. Of the factors analyzed, the calculated risk is the best predictor of testing choice. The higher a woman’s calculated risk of genetic abnormality in the fetus, the more likely she was to continue onto invasive testing. This is especially interesting because it raises the question of understanding values of risk presented by a health professional.

Figure 1 is a graph based on Hook’s values of population risk of genetic disorders at a certain age. This supports the premise that as women increase in age, the risk of a chromosomal abnormality occurring in their pregnancy also increases. Before the age of 35, the risk of a disorder in pregnancy is relatively low and has a very gradual slope of increase until the age of 35. However, after the age of 35, the slope and risk of disorder in pregnancy increase exponentially. This explains the concern for pregnancies in women who are older and supports genetic counseling as an option for women who have pregnancies later in life. The data are a reference to compare population risk to screen calculated risk.

Figure 2 provided the number of decisions made in each category, whether it was no further procedures, ultrasound only, or amniocentesis and ultrasound. These data showed that most women chose to proceed on to at
least some procedure rather than none. Also, the data show a three to one ratio
of women who chose amniocentesis and ultrasound to ultrasound only. From
this, it can be determined that women who are presented with an elevated risk
are nine times more likely to chose some sort of procedure to obtain further
information about their possible high risk pregnancy. This can be explained by
a need for more information in the event of a possible complication in
pregnancy.

There are also readily apparent ascertainment biases here. In order for
a chart to qualify for selection, the patients first had to see an obstetrician and
agree to have one of the prenatal screens. They had to return to their
obstetrician to receive the abnormal results and then agree to see a genetic
counselor to discuss their results. Since all of these levels give the patient the
option to eschew information, those who passed all these levels of selection
clearly wanted information about their pregnancy. Therefore, they are already
more likely to seek more information about their pregnancy in the form of
another procedure, which explains the high occurrence of the choice to at least
have an ultrasound after an abnormal result.

Figure 3 shows the analysis of whether having a previous child with a
genetic abnormality influenced the choice of testing in the current pregnancy.
Statistical analysis showed that the data were not statistically significant, but a
trend can be seen. If the patients had previous children that were healthy, this
did not influence testing choice. If the patients had previous children who
were unhealthy, they chose at least some procedure. This also shows that none
of the patients who had unhealthy children chose to go without any procedure. However, it should be noted that the sample size is small. This could indicate that the presence of a previously unhealthy child might influence the patient’s decision towards having some procedure rather than no procedure at all. The possibility of recurrence of a genetic abnormality seems to predict that women will chose to have at least some procedure to seek more information about their possible high risk pregnancy.

Figure 4 provided an analysis of a patient’s age and her screen calculated risk. The data were not statistically significant, so there was no clear correlation in the data. This indicates that calculated risk is dependant on personal medical history. This is interesting because population risk is influenced by age whereas calculated risk is not influenced by age.

Figure 5 shows the analysis of the screen-calculated risk and the decision made. Overall, these data show that most patients chose to have ultrasound and amniocentesis, which was observed previously as well. A trend can be seen where those with higher screen-calculated risks mostly chose at least an ultrasound over no procedure at all. However, since the data were not statistically significant, screen calculated risk does not affect the decision that is made.

Figures 6 and 7 examine whether income had an effect on the patient’s testing choice. This was analyzed because income is often a predictor of level or education, and level of education is often a predictor of a person’s ability to perceive statistics. Unfortunately, the education level of the patient was not
available, so income had to be used as a best approximation. Income was not listed on the patient chart, so instead the median income of the patient’s zip code was used. The data were not statistically significant, so the analysis of the data did not clearly show that income influenced patient decision. This is understandable because the data for income were not as accurate as desired for each patient. In a future study, it might be interesting to consider the decision made in light of the patient’s education level, to see whether this had an effect on patient testing choice.

Figure 8 is support for the hypothesis that increased screen-calculated risk is a predictor of testing choice. A clear trend can be seen, where the higher a patient’s calculated risk, the more likely she was to choose to have a procedure that would provide more information about her possible high-risk pregnancy. Also, it is interesting to note the difference in the average risk of each choice. The average calculated risk of those who chose to not have any procedure is half of the average calculated risk of those who chose further procedures. However, the trend is not statistically significant at this sample size.

The influence of elevated calculated risk is interesting because it raises the question of patient understanding of values of risk presented by a genetic counselor. Further studies involving surveys of patient understanding of statistics and education levels would be helpful in furthering the understanding of factors that may influence patient testing choice. This research has future implications for the genetic counseling field. The more
information there is concerning testing implications and testing choice, the better the care that health professionals will be able to provide.

Works Cited


Capstone Summary
Genetic counseling can be defined as a “process in which a genetic counselor educates families or individuals about their risk of passing on a genetic predisposition for certain disorders to future generations” (“National Society of Genetic Counselors”). To achieve this goal, a genetic counselor will explain and discuss the patient’s current medical situation with the patient in order to inform and educate. During this counseling, the patients will be presented with the medical facts as well as a possible diagnosis. They will be taken through the likely progression of the disorder as well as possible treatments or cures. Another goal of the genetic counseling session is for the patient to understand the inheritance of their possible affliction and how personal medical history has contributed to the possible occurrence. With clear explanation of this information, the genetic counselor will also provide options, such as testing and screens or support and referrals to other specialists. Overall, the goal of the genetic counselor is to inform patients of how genetic history can affect medical future.

Although genetic counseling can ideally be provided to any patient for any medical situation, genetic counseling is especially commonly offered to pregnant women. There are many medical factors that might cause a woman to have a “high-risk pregnancy,” and if she does, she will likely be referred to a genetic counselor. The possible factors that can denote a possible high-risk pregnancy are many, each with a medical basis that might affect the pregnancy adversely.
Currently it is routine to offer pregnant women serum screens that can test the mother’s blood for indicators of abnormalities in the pregnancy. Abnormal maternal serum marker screening results will cause a woman to be referred to a genetic counselor for discussion of risk in the pregnancy. After an abnormal result and consultation with a genetic counselor, the patient has the option to continue onto more invasive testing. Although there are many maternal serum marker screens, two of the most common are the Triple Screen and the Maternal Serum Alpha-Fetoprotein (MSAFP).

Once either of these screens has established an abnormal level of analytes, possibly indicating a disorder in the fetus, the patient has the option to continue on to an ultrasound or a more invasive form of testing. The most common invasive test performed in this situation is amniocentesis, which carries a low risk of pregnancy complications.

This decision to move from non-invasive testing to more definitive invasive testing in the case of a pregnancy with genetic disorder risk is one that probably entails many different factors. The various factors that might affect this decision are of particular interest to me, in that they may be predictive of testing choice and they can show the effect of testing on patient choice. While the actual factors that can affect this decision are numerous, the ones that can be evaluated are age of the patient, the patient’s population risk for a disorder of the fetus, the patient’s screen calculated risk of disorder in the fetus, previous children and whether those previous children are healthy or not. Marital status, previous miscarriages, a family history of genetic
disorders, a family history of miscarriage or stillbirth, and the patient’s ethnicity were also extracted from the charts, but preliminary analysis did not yield any results with statistical significance. Another factor that might affect the decision to continue onto invasive testing is the income of the patient, as it might denote the education level of the patient and her ability to perceive statistics and calculated risk. As each of these affects the patient’s outlook, they can be factors that affect the patient’s choice to continue with invasive testing after a non-invasive screen produces abnormal results.

In order to conduct a study that evaluates the possible factors that might affect a patient’s testing decision, subjects that fit certain criteria were obtained. These patients were seen in Dr. Robert Roger Lebel’s practice in Glen Ellyn, Illinois. From these patients, I selected those that fit specific criteria. Patient records were available for women who elected to have one of the two screens and received consultation from a genetic counselor. Afterwards they had the option to have no further procedures, ultrasound only, or ultrasound with amniocentesis. The data was recorded and analyzed.

One of the limiting factors of the data used can be attributed to ascertainment bias. Since there are many levels of health professional visits that the patient must attend before the patient qualified for my criteria, these limiting factors must be discussed. In order for a chart to qualify for selection, the patients first had to see an obstetrician and agree to have one of the prenatal screens. They had to return to their obstetrician to receive the abnormal results and then agree to see a genetic counselor to discuss their
results. Since all of these levels give the patient the option to eschew
information, those who passed all these levels of selection clearly wanted
information about their pregnancy. Therefore, they are already more likely to
seek more information about their pregnancy in the form of another
procedure, which explains the high occurrence of the choice to at least have an
ultrasound after an abnormal result.

Of the factors analyzed, a trend in the data can be seen. The higher a
woman’s calculated risk of genetic abnormality in the fetus, the more likely
she was to continue on to invasive testing. The average calculated risk of the
choice to not have any procedure is approximately two times lower than the
average calculated risk of the further procedure choices. This is especially
interesting because it raises the question of understanding values of risk
presented by a health professional. However, this trend is not statistically
significant because of the small sample size.

Further studies involving surveys of patient understanding of statistics
and education levels would be helpful in furthering the understanding of
factors that may influence patient testing choice. This research has future
implications for the field of genetic counseling. The more information there is
concerning testing implications and testing choice, the better the care that
health professionals will be able to provide.