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NORTHERN ILLINOIS UNIVERSITY
A Study of Genetic Counseling in Illinois

A Thesis Submitted to the
University Honors Program
In Partial Fulfillment of the
Requirements of the Baccalaureate Degree

With University Honors
Department of Biology


by

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Genetic counseling is a relatively young discipline in the medical sciences. Each year around 140 students are admitted to one of 25 Master's-Level Training Programs in the world, of which one is located in Illinois at Northwestern University in Chicago. On average, 20 well qualified candidates apply for each position available, causing genetic counseling to be an extremely competitive field. Before a person can seriously consider applying to genetic counseling programs, she must undergo the task of learning as much as possible about the field. Unfortunately, a person may have a hard time obtaining all the information needed to make an informed decision about a possible career in genetic counseling. It is very common for a student to apply to programs, go through interviews, become accepted to a program and still not know everything she should about genetic counseling. This is why it is extremely important for a genetic counseling candidate to know exactly what genetic counseling entails.

Who can become a Genetic Counselor? Genetic counselors usually have a background in biology or psychology. To be accepted into a genetic counseling program, a candidate must have a bachelor's degree, including coursework in genetics, chemistry, biochemistry and psychology or sociology. It is also recommended that a candidate volunteer at a crisis hotline, genetic or pregnancy center. There are minimum scores required on the GRE or MCAT, as well as a minimum undergraduate GPA. What is often unknown is that unlike other fields in which you may have a good chance at being accepted to a program if you meet only the minimum requirements, genetic counseling is such that you must surpass all requirements and recommended criteria in order to compete with the other candidates. For example, if the minimum GRE score is 1700 and you score a 2000, there will be candidates who scored 2200 and higher. If you volunteered one year at planned parenthood, there will be candidates who volunteered 3 years at a genetics center. If you graduated in the upper ten percent of your class at a state university, there will be candidates with master's degrees from Ivy League schools. One interesting fact, which could be seen as an advantage or disadvantage for women, is that a good majority (over 90%) of all genetic counselors are female.

This is not to mean that a student should not apply to schools if they are not the ideal candidate, only that each candidate should be aware of the stature of students they will be competing against. It is quite easy to be let down when you believed you were an exceptional candidate, only to discover that you

were merely average. Many candidates are unprepared for the competition and often fail to be accepted to a program the first year they apply. To ensure success at the first attempt to be admitted, a student should volunteer as much as possible, retake the GRE if necessary to achieve the highest score she is able to and learn as much as possible about genetic counseling before her first interview. The following is to inform genetic counseling candidates and others interested in genetic counseling of the information I have gathered after going to interviews for genetic counseling programs, researching genetic counseling as a career and speaking with seventeen genetic counselors in Illinois.

A student who would like to be employed as a genetic counselor must usually be very flexible as far as job location. Many areas, especially in the midwest, have only a handful of jobs and an abundance of genetic counselors. This renders many genetic counselors unemployed. Most unemployed genetic counselors are not willing to relocate to obtain a job, while some have found that it is too difficult to work the demanding on-call hours of a genetic counselor and raise a family simultaneously. For this reason, some graduate programs, such as at the University of Colorado at Denver, have, at one time or another, actually refused admission to in-state students in order to graduate genetic counselors who would not want to stay in Colorado. Preparing for the possibility of being required to relocate in order to be employed is one thing a genetic counseling student must do.

It is often recommended that a candidate for a genetic counseling program seek out and visit a genetic counselor on the job. Unfortunately, this may be more difficult than it seems. In Illinois, only eight out of about 50 employed genetic counselors work outside the Chicago area. Of these, four are located at Rockford Memorial Hospital, directly West of Chicago, leaving four genetic counselors servicing about 3/4 of the state of Illinois. There are two in Peoria associated with the University of Illinois at Chicago Hospitals, one in Urbana associated with the University of Illinois and one in Springfield associated with Southern Illinois University Medical School. Fortunately, two genetic counselors from St. Louis, MO visit southern Illinois once a month to help out. From these numbers it is obvious that if you are located south of Chicago, you may have difficulty locating a genetic counselor you can conveniently call on. If you somehow do manage to visit a genetic counseling clinic, most counselors will allow you to confidentially observe a genetic counseling session.

After graduation, a genetic counselor has the option to become board certified through the National Society of Genetic Counselors or NSGC. The word option is used due to the fact that many positions do not require genetic counselors to be board certified. In fact, many organizations hiring genetic counselors do not even realize the existence of a board examination. However, all the genetic counselors I have spoken with are certified or plan to become so as soon as possible. The national examination is offered every three years and was recently offered in May of 1996. A genetic counselor must have graduated the master's-level program at least three months prior to sitting for the boards. This causes some recent graduates, who must wait until 1999 to take the exam, to seek jobs which offer a wide range of counseling experiences in stead of a position which specializes in cancer, perhaps. This is another obstacle which must be planned for by the genetic counseling student.

Interestingly, one of the largest obstacles a genetic counselor must overcome is that many physicians do not understand or appreciate the role of a genetic counselor. Any pregnant woman who has an abnormal blood screen or is over the age of 35 must, by Illinois state law, be offered an amniocentesis, but not genetic counseling. Usually, doctors may spend 5 minutes explaining what would take a genetic counselor at least a half hour to explain. Also, the physician is usually not prepared to discuss the results of the test in genetic terms and may not be able to offer the time and support needed when a disorder is encountered. Also, many physicians are afraid that the genetic counselor will take away the patients he refers, which is not true. The genetic counselor is only a small side track in the prevention or treatment of a disease to insure the patients' thorough knowledge of their situation, rites, responsibilities and options. After the counseling is done, the patient will return to her doctor with the knowledge required to make informed decisions about her health care.

A few genetic counselors are lucky enough to be in the position to establish policies for the way in which a physician must deal with patients who may need genetic counseling services. One such counselor works at Smith-Kline & Beecham Laboratories in Illinois and spends the majority of her time explaining test results to physicians. These genetic counselors do not see patients of there own, but are on a mission to see that physicians are aware of the expertise they lack in the area of genetic counseling. Hopefully, these doctor's will realize they cannot handle the delicate matter of counseling patients in this

way. Another problem with doctor's attempting to counsel themselves is that patients tend to have formed a trusting relationship with their doctors and will not be able to make important decisions without some direction from their doctor. A genetic counselor knows she must refrain from influencing the decision of a patient, since often the result of that decision could mean life or death.

A genetic counselor's primary role is to provide information to families about risks for genetic diseases and also to counsel people who had tested positive for a genetic disease either themselves or in their unborn child. The genetic counselor must enable patients to make informed decisions regarding prevention or treatment of genetic diseases. Often, having to inform someone of the presence of a disease can be difficult on the genetic counselor and, for most, fighting depression is an ongoing battle.

One problem genetic counselor's have in common with some physicians is obtaining insurance. As in any medical field, there is the possibility of malpractice. Possible causes for the filing of a malpractice suit could be if a fetus was harmed in an amniocentesis procedure and the parents felt the genetic counselor pressured them to have the procedure done. For this reason the genetic counselor must be even more aware of her responsibility to present the facts in an unbiased manner. Each patient must realize and accept the responsibility of making the important choices surrounding genetic counseling.

Most patients go into a genetic counseling session knowing very little about genetics and often do not understand why they are there. A genetic counselor must present a brief lesson on genetics and then talk to them about their specific disease or risks. The most common case a genetic counselor sees is called AMA or advanced maternal age. When a pregnant woman will give birth to a child after her 35th birthday, she is at a greatly increased risk of having a child with a chromosome disorder. Usually these occur in the form of mutations, deletions or multiple chromosomes. The chromosome abnormality encountered most often in these fetuses is an extra chromosome #21, which causes Down's syndrome. The risks for Down's and other disorders will be calculated according to the mother's age and the counselor will then explain the risks incurred with amniocentesis, the procedure used to test for many chromosomal disorders. For instance, a 42 year old pregnant woman has a 1 in 41 chance of having a child with a chromosomal disorder, while the chance for miscarriage after an amniocentesis is 1 in 300. Although many people choose to have the amniocentesis done, many will opt not to. Often, this is due to

their feelings about abortion, but sometimes it is because they still do not understand what is going on. A genetic counselors most awesome responsibility is to see that each and every patient understands every important detail presented to them.

Among the Genetic Counselors I have spoken with in Illinois, six specialize in pediatrics, seven in prenatal care and six have no specialty (they classify themselves as “general” genetic counselors). Among those with more specific areas of specialization, there were two cancer genetic counselors and one genetic counselor for each of the following specialties: blood disorders, ocular disorders, preconception cases and teratogen exposure cases. In addition, two genetic counselors were in the category mentioned before in which they advise physicians and set standards instead of seeing patients. Each of the genetic counselors with a specialty other than pediatrics or prenatal care were located in Chicago. Therefore, genetic counselors who service a large population are able to specialize only somewhat (pediatrics or prenatal), if at all. This is significant in that it means a genetic counselor who wishes to specialize considerably, will most likely be forced to work in a relatively large city.

A prenatal genetic counselor concentrates on patients who are pregnant and those fetuses at risk for a genetic disease. These cases usually involve patients with an increased risk for giving birth to a child with Down’s Syndrome and other chromosome disorders based on the age of the mother, as well as those with an increased risk of transmitting any genetic disorder due a family history of genetic disease. This type of genetic counselor sees numerous advanced maternal age cases (AMA) and may only be able to offer probabilities as to the fetus’s risk for having a genetic disorder, even after testing has taken place. Often, the prenatal genetic counselor must endure the painful reality of the possibility of abortion of healthy fetuses based on a mere probability of risk. Another situation a prenatal genetic counselor is often faced with is when a couple is tested for a disorder, as well as the fetus or child, and it is evident that the man is not the father of the child. This is one of the many cases in which the genetic counselor must know the legal rights of all parties involved and be able to use discretion in revealing only that information which affects the counselors ability to counsel and present probabilities accurately.

A pediatrics genetic counselor specializes in children who were born with a genetic disorder. Often, patients come to a genetic counselor to determine how the disease was transmitted to the child.

Again, there is a possibility that a genetic test may reveal that the man assumed to be the father is not. Once a disorder is confirmed, the patients will require the knowledge of the options available to them as well as possible treatment. A genetic counselor will encourage the patients to inform their family members of the risk that they might also carry or be at risk for developing the disease. This is especially important when the trait is carried on the X chromosome, as the disorder may be hidden for many generations and then suddenly appear in male members of the family. Even in cases where the family members are at a considerable risk for passing a disorder to their children, many patients have a difficult time deciding whether or not they should inform their family of the problem.

Pediatrics genetic counselors see such diseases as Neurofibromatosis (NF), Gouache's disease, Down's Syndrome, Muscular Dystrophy, Tay Sach's, Fragile X and Cystic Fibrosis. The occurrence of Tay Sach's disease and other disorders prevalent in people of Jewish ancestry is significantly seen less by genetic counselors in Central Illinois due to the small Jewish population. A pediatrics genetic counselor will also see patients who tested positive for six tests which are administered to all newborns in Illinois. These tests are for phenylketoneuria (PKU), hemoglobinopathies, hypoglycemia, hypothyroidism, galactosemia deficiency and biotinidase deficiency. It has been determined that by diagnosing these diseases at birth, these children are able to live normal lives with proper treatment. The cost of testing all Illinois newborns for these diseases is less than the cost of supporting those individuals who would otherwise develop symptoms of these diseases, including retardation which would possibly require a lifetime of family and/or government support. In addition, such diseases as PKU, if not discovered shortly after birth, can lead not only to severe retardation, but death as well.

A common genetic counseling specialty other than prenatal or pediatrics is cancer. These genetic counselors usually concentrate on breast, colon, prostate or ovarian cancers. Breast Cancer has been seen more commonly by genetic counselors since the discovery of the gene associated with breast cancer--BRCA1. One very interesting practice among cancer genetic counselors is bloodbanking. This is when members of a family with a history of cancer have a blood sample taken which can then be used in the future as tests to detect other types of cancer genes become available. This is to enable future generations

to have access to an accurate assessment of where all the alleles for cancer came from in their family, even though many of these relatives may no longer be living.

Another genetic counseling specialty is Hematology. This type of genetic counseling concentrates on patients with a blood disease. The most prevalent blood disease encountered by this genetic counselor is sickle cell anemia. Numerous blood diseases are among those genetic tests administered to all newborn babies in Illinois. This group of blood diseases is labeled "hemoglobinopathies". Sickle cell anemia is tested by use of DNA probe Mst II endonuclease. The other hemoglobinopathies are also tested using recombinant DNA, hybridization and DNA restriction fragment polymorphism technology. Most other tests for genetic disorders are accomplished with one of these methods.

Another genetic counseling specialty I encountered was ocular disorders. There are three main types of eye disorders seen often by this type of genetic counselor: optic nerve hyperplasia, septooptic dysplasia and retinoblastoma. Dysplasia is the term for an abnormal organization of cells into tissues and its morphologic result. Hyperplasia is any developmental excess, such as polydactyly. There is a molecular test for detecting retinoblastoma, whereas the other two disorders have not yet been proven to be genetic in nature, though they seem to follow an autosomal recessive pattern of inheritance.

The last genetic counseling specialty involved a counselor who was working with the effects of teratogen exposure on fetuses during pregnancy. A teratogen is something a fetus can be exposed to during critical stages of development which may cause the fetus to be malformed. Examples of teratogens are the Rubella virus, male hormones, radiation, tetracycline, other drugs and alcohol. Similarly, one genetic counselor in Chicago works at a free clinic to aid and educate people on welfare. In this setting, a majority of cases involved exposures to drugs and other harmful substances during and before pregnancy. This is a situation in which people who are in need of genetic counseling and pregnancy education would otherwise not have access to it.

Most disorders which are tested for use cultured amniocytes. Tay Sachs disease can be detected by a test for hexosaminidase A deficiency, PKU by testing for phenylalanine hydroxylase deficiency, Galactosemia by Galactose-1-phosphate uridyl transferase deficiency and Gaucher disease by Beta-

glucosidase deficiency. Some disorders can be detected through ultrasound, such as Spina Bifida, Neuroblastoma and Achondroplasia. Spina Bifida can also be detected by a blood test for Alpha-fetoprotein.

It is obvious that through the newly developing DNA testing technologies, genetic counseling will become more accurate with time. The only obvious problem is making genetic counseling available to all people. Illinois is fortunate to have many genetic counselors and even a genetic counseling program. Another need is for more jobs to accommodate the unemployed genetic counselors who desire to work in Illinois. However, great steps must be taken to more evenly distribute the counselors across the state.

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ABSTRACT:

A study of genetic counseling in Illinois. Phone interviews with 17 Illinois genetic counselors were conducted as well as one visit to a genetic counseling clinic and observation of one genetic counseling session. Focus on genetic counseling master's programs and individual counselor specialties. Also includes advice on applying to genetic counseling programs from author's personal experience. Emphasis was placed on discovering facts about genetic counseling which the author had not been aware of previously. These facts include how often master's program graduates may sit for the NSGC Board Examination, problems obtaining genetic counseling positions in the Midwest, the geographical unbalance of counselors in Illinois, problems earning respect throughout the medical field, and the numerous genetic counseling specialties besides prenatal and pediatrics.