Department of Regulatory Agencies

Office of Policy, Research and Regulatory Reform

2013 Sunrise Review: Genetic Counselors

October 15, 2013

Consumer Protection

Department of Regulatory Agencies

Executive Director's Office
Barbara J. Kelley
Executive Director

John W. Hickenlooper
Governor

October 15, 2013

Members of the Colorado General Assembly
c/o the Office of Legislative Legal Services
State Capitol Building
Denver, Colorado 80203

Dear Members of the General Assembly:

The mission of the Department of Regulatory Agencies (DORA) is consumer protection. As a part of the Executive Director's Office within DORA, the Office of Policy, Research and Regulatory Reform seeks to fulfill its statutorily mandated responsibility to conduct sunrise reviews with a focus on protecting the health, safety and welfare of all Coloradans.

DORA has completed its evaluation of the sunrise application for regulation of genetic counselors and is pleased to submit this written report. The report is submitted pursuant to section 24-34-104.1, Colorado Revised Statutes, which provides that DORA shall conduct an analysis and evaluation of proposed regulation to determine whether the public needs, and would benefit from, the regulation.

The report discusses the question of whether there is a need for regulation in order to protect the public from potential harm, whether regulation would serve to mitigate the potential harm, and whether the public can be adequately protected by other means in a more cost-effective manner.

Sincerely,

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Executive Director

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Consumer Protection

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Background

Consistent, flexible, and fair regulatory oversight assures consumers, professionals and businesses an equitable playing field. All Coloradans share a long-term, common interest in a fair marketplace where consumers are protected. Regulation, if done appropriately, should protect consumers. If consumers are not better protected and competition is hindered, then regulation may not be the answer.

As regulatory programs relate to individual professionals, such programs typically entail the establishment of minimum standards for initial entry and continued participation in a given profession or occupation. This serves to protect the public from incompetent practitioners. Similarly, such programs provide a vehicle for limiting or removing from practice those practitioners deemed to have harmed the public.

From a practitioner perspective, regulation can lead to increased prestige and higher income. Accordingly, regulatory programs are often championed by those who will be the subject of regulation.

On the other hand, by erecting barriers to entry into a given profession or occupation, even when justified, regulation can serve to restrict the supply of practitioners. This not only limits consumer choice, but can also lead to an increase in the cost of services.

There are also several levels of regulation.

Licensure

Licensure is the most restrictive form of regulation, yet it provides the greatest level of public protection. Licensing programs typically involve the completion of a prescribed educational program (usually college level or higher) and the passage of an examination that is designed to measure a minimal level of competency. These types of programs usually entail title protection – only those individuals who are properly licensed may use a particular title(s) – and practice exclusivity – only those individuals who are properly licensed may engage in the particular practice. While these requirements can be viewed as barriers to entry, they also afford the highest level of consumer protection in that they ensure that only those who are deemed competent may practice and the public is alerted to those who may practice by the title(s) used.

Certification

Certification programs offer a level of consumer protection similar to licensing programs, but the barriers to entry are generally lower. The required educational program may be more vocational in nature, but the required examination should still measure a minimal level of competency. Additionally, certification programs typically involve a non-governmental entity that establishes the training requirements and owns and administers the examination. State certification is made conditional upon the individual practitioner obtaining and maintaining the relevant private credential. These types of programs also usually entail title protection and practice exclusivity.

While the aforementioned requirements can still be viewed as barriers to entry, they afford a level of consumer protection that is lower than a licensing program. They ensure that only those who are deemed competent may practice and the public is alerted to those who may practice by the title(s) used.

Registration

Registration programs can serve to protect the public with minimal barriers to entry. A typical registration program involves an individual satisfying certain prescribed requirements – typically non-practice related items, such as insurance or the use of a disclosure form – and the state, in turn, placing that individual on the pertinent registry. These types of programs can entail title protection and practice exclusivity. Since the barriers to entry in registration programs are relatively low, registration programs are generally best suited to those professions and occupations where the risk of public harm is relatively low, but nevertheless present. In short, registration programs serve to notify the state of which individuals are engaging in the relevant practice and to notify the public of those who may practice by the title(s) used.

Title Protection

Finally, title protection programs represent one of the lowest levels of regulation. Only those who satisfy certain prescribed requirements may use the relevant prescribed title(s). Practitioners need not register or otherwise notify the state that they are engaging in the relevant practice, and practice exclusivity does not attach. In other words, anyone may engage in the particular practice, but only those who satisfy the prescribed requirements may use the enumerated title(s). This serves to indirectly ensure a minimal level of competency – depending upon the prescribed preconditions for use of the protected title(s) – and the public is alerted to the qualifications of those who may use the particular title(s).

Licensing, certification and registration programs also typically involve some kind of mechanism for removing individuals from practice when such individuals engage in enumerated proscribed activities. This is generally not the case with title protection programs.

Regulation of Businesses

Regulatory programs involving businesses are typically in place to enhance public safety, as with a salon or pharmacy. These programs also help to ensure financial solvency and reliability of continued service for consumers, such as with a public utility, a bank or an insurance company.

Activities can involve auditing of certain capital, bookkeeping and other recordkeeping requirements, such as filing quarterly financial statements with the regulator. Other programs may require onsite examinations of financial records, safety features or service records.

Although these programs are intended to enhance public protection and reliability of service for consumers, costs of compliance are a factor. These administrative costs, if too burdensome, may be passed on to consumers.

Sunrise Process

Colorado law, section 24-34-104.1, Colorado Revised Statutes (C.R.S.), requires that individuals or groups proposing legislation to regulate any occupation or profession first submit information to the Department of Regulatory Agencies (DORA) for the purposes of a sunrise review. The intent of the law is to impose regulation on occupations and professions only when it is necessary to protect the public health, safety or welfare. DORA must prepare a report evaluating the justification for regulation based upon the criteria contained in the sunrise statute:[1]

> (I) Whether the unregulated practice of the occupation or profession clearly harms or endangers the health, safety, or welfare of the public, and whether the potential for the harm is easily recognizable and not remote or dependent upon tenuous argument;
>
> (II) Whether the public needs, and can reasonably be expected to benefit from, an assurance of initial and continuing professional or occupational competence;
>
> (III) Whether the public can be adequately protected by other means in a more cost-effective manner; and
>
> (IV) Whether the imposition of any disqualifications on applicants for licensure, certification, relicensure, or recertification based on criminal history serves public safety or commercial or consumer protection interests.

Any professional or occupational group or organization, any individual, or any other interested party may submit an application for the regulation of an unregulated occupation or profession. Applications must be accompanied by supporting signatures and must include a description of the proposed regulation and justification for such regulation.

[1] § 24-34-104.1(4)(b), C.R.S.

Methodology

DORA has completed its evaluation of the proposal for the regulation of genetic counselors. During the sunrise review process, DORA performed a literature search, contacted and interviewed the applicant, reviewed licensure laws in other states, conducted interviews of administrators of those programs, interviewed healthcare associations such as the Colorado Hospital Association and the Colorado Medical Society and contacted the American Board of Genetic Counseling. In order to determine the number and types of complaints filed against genetic counselors in Colorado, DORA contacted representatives of the State Board of Nursing and the Colorado Medical Board.

Profile of the Profession

Genes are made up of deoxyribonucleic acid (DNA) molecules, which are the simplest building blocks of heredity.[2] They are grouped together in specific patterns within a person's chromosomes, forming a unique "blueprint" for every physical and biological characteristic of a person.[3]

Humans have 46 chromosomes, arranged in pairs in every living cell of the body.[4] Current science suggests that human chromosomes carry from 25,000 to 35,000 genes.[5] An error in just one gene (and in some instances, even the alteration of a single piece of DNA) can sometimes be the cause of a serious medical condition.[6]

There are myriad genetic diseases, including but not limited to:[7]

- Chromosomal abnormalities, which cause birth defects, mental retardation and/or produce reproductive problems;
- Single gene disorders such as cystic fibrosis, muscular dystrophy, Huntington's disease (a degenerative nerve disease) and sickle cell disease;
- Familial cancer and cancer-prone syndromes such as inherited breast or colorectal cancer and neurofibromatosis; and
- Birth defects with a genetic component such as neural tube defects and cleft lip and palate.

Genetic Counselors are healthcare professionals who help people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.[8] Genetic counselors are instrumental in the healthcare setting in assisting in determining whether a patient (or a family) is at risk of contracting a genetic disorder. This is done through a variety of procedures, including but not limited to genetic testing, reviewing a patient's medical records and family history.

Genetic testing is done by analyzing small samples of blood or body tissues to determine whether a patient carries genes for certain inherited disorders.[9]

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[2] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

[3] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

[4] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

[5] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

[6] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

[7] Clinical Genetics Society. *What is Clinical Genetics?* Retrieved July 9, 2013, from http://www.clingensoc.org/what-is-clinical-genetics/

[8] National Society of Genetic Counselors. *FAQs About Genetic Counselors and the NSGC.* Retrieved July 9, 2013, from http://www.nsgc.org/About/FAQsaboutGeneticCounselorsandtheNSGC/tabid/143/Default.aspx

[9] Genetic Counseling. *What is Genetic Counseling?* Retrieved July 9, 2013, from http://kidshealth.org/parent/system/medical/genetic_counseling.html

There are several types of specializations within the genetic counseling profession, including but not limited to:

- Cancer;
- General;
- Pediatric;
- Prenatal; and
- Psychiatric.

Cancer genetic counselors evaluate family history and talk about risks for inherited cancer, as well as screening and management for those at increased risk.[10] Cancer genetic counselors may also inform persons about genetic testing and assist persons in determining whether the testing would be useful.[11] In Colorado, the greatest percentage of genetic counselors (36 percent) work as cancer genetic counselors.[12]

General genetic counselors serve children, adults and families with known or suspected genetic conditions and birth defects.[13] In certain instances, families start out in general genetic counselors' clinics and, if a diagnosis can be made, they may then be referred to a specialty clinic.[14]

Pediatric genetic counselors focus on genetic conditions affecting the pediatric and adolescent population.[15] In Colorado, the second highest percentage of genetic counselors (29 percent) work as pediatric generic counselors.[16]

Prenatal genetic counselors work with individuals, couples or families who have an increased chance of having a child with a birth defect or genetic condition.[17] Those who are pregnant or considering having a child can meet with a prenatal genetic counselor to learn more about a condition, understand their risks more clearly and discuss options for prenatal screening, testing and/or assisted reproduction techniques.[18] According to the sunrise application, in Colorado, 19 percent of genetic counselors practice as prenatal genetic counselors.

[10] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[11] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[12] 2012 Sunrise Application. Submitted by the Colorado Genetic Counselors Network. p.6.
[13] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[14] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[15] National Society of Genetic Counselors. *Pediatric & Clinical Genetics SIG – Leadership & Details.* Retrieved July 11, 2013, from http://www.nsgc.org/MemberCenter/SIGs/PediatricClinicalGenetics/tabid/258/Default.aspx
[16] 2012 Sunrise Application. Submitted by the Colorado Genetic Counselors Network. p.6. and 7.
[17] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[18] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf

Psychiatric genetic counselors are utilized when a person has a personal or family history of a psychiatric or mental health condition.[19] In this case, the psychiatric genetic counselor will help answer questions about the cause of the condition and the chance it will happen again in the person's family.[20] The types of psychiatric conditions include:

- Autism spectrum disorders;
- Depression;
- Bipolar disorder; and
- Schizophrenia.

Additionally, the American Board of Genetic Counselors (ABGC), which was created in 1993,[21] offers a certification for genetic counselors for candidates who have completed the required education (a master's level degree in genetic counseling). Prior to 1993, the American Board of Medical Genetics (ABMG) was the sole source of private certification for genetic counselors.

The purpose of the ABGC is to establish standards of competence to protect the public and promote ongoing growth and development of practitioners.[22] To achieve standards of competence, the ABGC established a credentialing process, where applicants meet certain minimum standards and pass an examination to become certified.

The first step in the certification process is for a candidate to achieve active candidate status (ACS). ACS signifies that a candidate has met all eligibility requirements to apply for ABGC certification, including submitting all application materials (including transcripts from an accredited genetic counseling education program) and paying applicable fees.[23]

Once ACS is achieved, a candidate for certification is eligible to take the certification examination. There are several examination categories, which take into consideration a variety of factors such as whether the examination is being taken for the first time for initial certification or recertification or whether it is the first attempt or repeat attempt.[24] However, the vast majority of candidates fall in the first two categories.

[19] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[20] National Society of Genetic Counselors. *Making Sense of Your Genes. A Guide to Genetic Counseling.* Retrieved June 6, 2013, from http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf
[21] American Board of Genetic Counseling. *About ABGC.* Retrieved July 23, 2013, from http://www.abgc.net/About_ABGC/GeneticCounselors.asp
[22] American Board of Genetic Counseling. *About ABGC.* Retrieved July 23, 2013, from http://www.abgc.net/About_ABGC/GeneticCounselors.asp
[23] American Board of Genetic Counseling. *2013 Bulletin of Information for the American Board of Genetic Counseling Certification Examination.* Retrieved July 1, 2013, from http://www.abgc.net/Resources_Links/documents/ABGChandbook2013_Final.pdf
[24] American Board of Genetic Counseling. *2013 Bulletin of Information for the American Board of Genetic Counseling Certification Examination.* Retrieved July 1, 2013, from http://www.abgc.net/Resources_Links/documents/ABGChandbook2013_Final.pdf

Category one is for first time examinees, and candidates are required to submit an official transcript from an ABGC-accredited genetic counseling education program (Colorado has one accredited program – University of Colorado Denver Graduate Program of Genetic Counseling). The examination is administered in two locations in Colorado – Denver (Aurora) and Grand Junction. The fee to take the examination is $1,300.[25]

Category two is for candidates who did not pass their prior examinations. The fee to retake the examination is $1,200.[26] Candidates are eligible to take the certification examination up to three times within five years of graduation.[27]

Once certified, the ABGC certification is valid for five years.[28]

[25] American Board of Genetic Counseling. *Examination Fees and Important Dates.* Retrieved July 26, 2013, from http://www.abgc.net/ABGC/documents/2013-2014ExamInfoforWebsite.pdf
[26] American Board of Genetic Counseling. *Examination Fees and Important Dates.* Retrieved July 26, 2013, from http://www.abgc.net/ABGC/documents/2013-2014ExamInfoforWebsite.pdf
[27] American Board of Genetic Counseling. *2013 Bulletin of Information for the American Board of Genetic Counseling Certification Examination.* Retrieved July 1, 2013, from http://www.abgc.net/Resources_Links/documents/ABGChandbook2013_Final.pdf
[28] American Board of Genetic Counseling. *2013 Bulletin of Information for the American Board of Genetic Counseling Certification Examination.* Retrieved July 1, 2013, from http://www.abgc.net/Resources_Links/documents/ABGChandbook2013_Final.pdf

Proposal for Regulation

The Colorado Genetic Counselors Network (Applicant) has submitted a sunrise application to the Department of Regulatory Agencies for review in accordance with the provisions of section 24-34-104.1, Colorado Revised Statutes. The application identifies state licensure of genetic counselors as the appropriate level of regulation to protect the public.

The sunrise application states that licensure of genetic counselors is the necessary and appropriate approach to regulation because the activities of the genetic counseling profession are complex, requiring specialized knowledge and unique skills.

Additionally, the sunrise application requests that a requirement for licensure be board certification as a genetic counselor by the American Board of Genetic Counseling (ABGC) and/or the American Board of Medical Genetics (ABMG). Recall that prior to 1993, the ABMG was the only certification available, so the proposal submitted in the sunrise application would allow practitioners who obtained certification through the ABMG, before the creation of the ABGC certification, to qualify for state licensure.

The Applicant also recommends the creation of a temporary license if/when a candidate achieves active candidate status for the ABGC certification examination. Active candidate status signifies that an individual has met all of the eligibility requirements to apply for certification, including:[29]

- Submitted all required application materials;
- Paid applicable fees; and
- Been approved to take the examination.

The Applicant, in its sunrise application, asserts that licensure of genetic counselors would:

- Increase the public's awareness of what constitutes quality genetic counseling services and who is appropriately trained to provide these services;
- Improve the public's access to quality genetic counseling services; and
- Allow the public to have a method of recourse for unprofessional genetic counseling services without relying upon malpractice litigation, which is costly and time consuming.

[29] American Board of Certified Genetic Counselors. *2013 Bulletin of Information for the American Board of Genetic Counseling Certification Examination.* Retrieved July 1, 2013, from http://www.abgc.net/Resources_Links/documents/ABGChandbook2013_Final.pdf

Summary of Current Regulation

The Colorado Regulatory Environment

Currently, genetic counselors are an unregulated profession in Colorado, and as such, there are no laws that specifically provide regulatory oversight. However, there are a variety of laws in Colorado that govern various healthcare professions (e.g., physicians and registered nurses) who may provide various types of genetic consultation. These healthcare professionals are under the jurisdiction of their respective regulatory boards, and the boards investigate complaints filed against practitioners for instances where healthcare professionals are accused of harming the public by practicing outside their respective scopes of practice.

Additionally, there are several Colorado laws that are relevant to genetic counseling, including but not limited to the Newborn Screening and Genetic Counseling and Education Act and the Healthcare Availability Act.

The Newborn Screening and Genetic Counseling and Education Act, in section 25-4-1002, Colorado Revised Statutes (C.R.S.), states,

> newborn screening and genetic counseling and education should be made with full public knowledge, in light of expert opinion, and should be constantly reviewed to consider changing medical knowledge and ensure full public protection.

Section 25-4-1002, C.R.S., also states that participation of persons in genetic counseling programs should be voluntary and that all information obtained from persons involved is confidential.

The Healthcare Availability Act, in section 13-64-502, C.R.S., among other things, addresses limitations on actions,

> where no claimant, including an infant or his personal representative, parents or next of kin, may recover for any damage or injury arising from genetic counseling and screening and prenatal care or arising from or during the course of labor, delivery or the period of postnatal care in a healthcare institution, where such damage or injury was the result of genetic disease or disorder or other natural causes, unless the claimant can establish that damage or injury could have been prevented or avoided by ordinary standard of care by the physician or other healthcare professional or healthcare institution.

Regulation in Other States

According to the sunrise application submitted by the Colorado Genetic Counselors Network, there are 16 states that regulate (license) genetic counselors: California, Delaware, Hawaii, Illinois, Indiana, Massachusetts, Nebraska, New Jersey, New Mexico, Ohio, Oklahoma, Pennsylvania, South Dakota, Tennessee, Utah and Washington.

All of the aforementioned states require genetic counselors to obtain certification through the American Board of Genetic Counselors (ABGC) prior to obtaining a license to practice genetic counseling.

In order to discern the number of genetic counselors regulated in other states, and identify the types of complaints and disciplinary actions imposed on licensees, the Department of Regulatory Agencies staff conducted a random sample (half) of the states that currently regulate genetic counselors.

California

California established licensure of genetic counselors in 2011.

To date, there are 435 licensed genetic counselors practicing in California.

Staff stated that there have not been any complaints or disciplinary actions imposed on licensees.

Delaware

In Delaware, the General Assembly established the licensing of genetic counselors in 2010.

Currently, there are 25 licensed genetic counselors. There have not been any complaints filed or disciplinary actions imposed on licensees.

Nebraska

In Nebraska, the legislature established the licensing of genetic counselors in 2012.

As of this writing, staff recently completed information on the licensing process, and they are currently in the process of receiving licensing applications. As a result, there are no licensed genetic counselors, no complaints and no disciplinary actions were imposed.

New Mexico

In 2009, the New Mexico General Assembly established licensure of genetic counselors.

Currently, there are 26 licensed genetic counselors. There have not been any complaints or disciplinary actions imposed on licensees.

South Dakota

South Dakota began regulating genetic counselors in 2009.

Currently, there are 18 licensed genetic counselors and one temporary genetic counselor licensee.

Since the establishment of licensing of genetic counselors in 2009, there have not been any complaints or disciplinary actions imposed.

Tennessee

Tennessee began regulating genetic counselors in 2007.

There are currently 44 licensed genetic counselors in Tennessee.

Staff was unable to disclose whether there were any complaints filed against licensed genetic counselors since 2007. However, staff stated that there have not been any disciplinary actions imposed.

Utah

Utah began regulating genetic counselors in 2001.

Currently, there are 64 licensed genetic counselors and two temporary genetic counselors.

In the past five years, there have been minimal complaints filed against genetic counselors addressing issues such as scope of practice issues and inadequate supervision. However, the complaints, after review by the Genetic Counselors Board, were dismissed with no formal disciplinary actions imposed on licensees.

Washington

In Washington, the regulation of genetic counselors was established by the General Assembly in 2009.

To date, there are 95 active licensed genetic counselors, 5 provisionally licensed (these candidates are waiting to pass the examination) and 8 pending applications.

Since 2009, there have not been any complaints filed against licensed genetic counselors, and therefore no disciplinary actions imposed.

Analysis and Recommendations

Public Harm

The first sunrise criterion asks:

> Whether the unregulated practice of the occupation or profession clearly harms or endangers the health, safety or welfare of the public, and whether the potential for harm is easily recognizable and not remote or dependent on tenuous argument.

Before moving forward with the analysis of harm concerning genetic counselors, it is important to identify what constitutes harm to the public. The improper actions of genetic counselors could result in psychological/emotional, financial and physical harm. There are numerous scenarios where consumers may be harmed, including psychological/emotional harm. For example, a genetic counselor may inaccurately interpret genetic testing results, informing a patient that she is at an elevated risk of breast cancer, when in fact she may not be at an elevated risk.

In order to determine whether regulation of genetic counselors is necessary, the Department of Regulatory Agencies (DORA) requested that the sunrise applicant provide specific examples of harm to the public.

It is important to note that the vast majority of examples of harm provided to DORA concerning genetic counselors were the same examples provided for the 2004 sunrise review. As such, DORA analysis of these examples of harm remains the same. Generally, DORA's analysis of harm in the 2004 sunrise review concluded that non-genetic counselors were harming consumers, such as providing substandard pre-test counseling to patients and inaccurately interpreting genetic test results. The examples of harm provided are included in Appendix A on page 25.

The examples of harm provided in the sunrise application can be broken down into specific content areas, including:

- Incomplete risk assessment;
- Inaccurate test interpretation;
- Psychological and financial issues;
- Inadequate training specializing in genetics;
- Title misuse;
- Medical malpractice;
- Alleged wrongful birth; and
- Inadequate training.

The current sunrise application included a few new examples of harm in the areas of incomplete risk assessment, inaccurate test interpretation and title misuse. Each such example is summarized below along with DORA's analysis.

Incomplete Risk Assessment

A pregnant woman had an increased risk of her baby having a chromosome anomaly. She received prenatal genetic counseling from an unlicensed genetic counselor in another state. The patient chose to have chorionic villus sampling (CVS) for the risk of aneuploidy (abnormal chromosome number). The baby was chromosomally normal on the CVS, but when the baby was born it had a transverse limb defect resulting in severe malformation of the hand and lower arm. The family was emotionally devastated. Theoretically, the transverse limb defect could have been a consequence of the CVS procedure.

The family did not recall receiving pre-test counseling from the genetic counselor concerning the potential risks of having a CVS test performed. As a result, the family sued the hospital employing the genetic counselor for inadequate pre-test counseling.

Analysis

Clearly, the family suffered emotional distress due to their newborn child having a severe malformation of the hand and lower arm. However, it is unclear if the actions of the genetic counselor caused the emotional distress. Since the family could not recall receiving information related to the risks associated with the CVS procedure, it is unclear if the genetic counselor was deficient in performing pre-test counseling to the family.

Since there is no clear link that the genetic counselor failed to perform his/her duties correctly concerning pre-test counseling, it is reasonable to conclude that regulation would not have prevented this situation or if regulation did exist, that disciplinary action would have been appropriate in this instance.

Inaccurate Test Interpretation

A recent publication looked at common themes of errors in cancer genetics. These include cases in which the wrong test was ordered, resulting in inaccurate medical management recommendations, unnecessary testing and misuse of healthcare dollars, as well as the misinterpretation or results leading to an inaccurate diagnosis of advanced cancer or unnecessary preventative surgery.

Analysis

This publication does not highlight any specific instances where genetic counselors are harming consumers. As a result, it is impossible to provide an analysis of harm.

It is important to note, however, that in Colorado, licensed physicians order genetic tests, not genetic counselors.

Title Misuse

In Wisconsin (where licensure of genetic counselors is currently being pursued), a physician assistant was advertising himself as a genetic counselor. Although some discussion of genetic information by a physician assistant may be considered within that professional's scope of practice, physician assistants' training in genetics is quite limited compared to that of genetic counselors and does not qualify them to practice as genetic counselors or misrepresent themselves to the public in this way.

Analysis

The example implies that the physician assistant is soliciting business as a genetic counselor when he may not have the appropriate qualifications to provide genetic counseling services. This case, although not in Colorado, illustrates an example where title protection may be appropriate.

However, there are many areas that are unclear, such as whether the physician assistant actually functioned as a genetic counselor. It is also unclear whether the physician assistant lacked the appropriate training and skills to provide genetic counseling services to consumers. Likewise, the example does not state whether he provided misinformation or otherwise harmed consumers.

Medical Malpractice Case

A Massachusetts couple was expecting a baby in 2007. The couple alleged that several medical staff members, including an obstetrician, a geneticist, a nurse and a genetic counselor were negligent in their duties. Specifically, the couple claimed that they were not offered amniocentesis to determine whether their unborn child was at risk for physical and/or mental disabilities. The defendants in the case, however, maintained that the couple was, in fact, offered amniocentesis.

The pregnant woman was originally from China and did not speak English. The husband served as the interpreter between the medical staff and the woman, which was a violation of medical protocol.

The case reached a $7 million settlement.

Analysis

The information provided in this case indicates that this is a situation where the medical staff did not follow the proper protocol when interacting with a patient who did not speak English. Consequently, the woman was not aware that she was offered amniocentesis, which would have been able to determine if there were issues associated with her unborn child.

Although unfortunate, this case does not illustrate clear harm imposed by the genetic counselor on the patient. Rather, this case highlights the fact that medical protocol was violated in working with non-English speaking patients.

Alleged Wrongful Birth

A New Hampshire couple sued a medical center for wrongful birth of their child. The couple stated that they would have terminated their pregnancy if the hospital's genetic counselors and doctors fully informed them of the extent of their child's disabilities stemming from a rare chromosomal disorder.

A jury awarded the couple $2.3 million, but the New Hampshire Supreme Court reversed the decision, ruling that the hospital sufficiently informed the couple about the potential disabilities for their unborn son.

Analysis

Although it is alleged that the physician and genetic counselor failed to provide the couple with complete and accurate information concerning the extent of their unborn child's disabilities, the New Hampshire Supreme Court disagreed. As a result, it appears that the genetic counselor, at least in part, did not harm the consumers.

Since clear identifiable harm to the consumer was not established related to the actions of the genetic counselor, it is unclear how regulation would have addressed this situation.

Inadequate Training

In Virginia, a genetic counselor specialized in pediatrics but occasionally worked with patients for cancer genetic counseling. The genetic counselor was working with an 86 year old patient who was diagnosed with colorectal cancer at age 74 followed by a recurrence at age 76.

The genetic counselor recommended a variety of genetic tests to determine whether the patient had Lynch syndrome. Lynch syndrome is an inherited disorder that increases the risk of many types of cancer, particularly cancers of the colon, and rectum, which are collectively referred to as colorectal cancer.[30]

The test results were not informative, meaning no mutations were found in any of the genes analyzed. As a result, the genetic counselor recommended that no additional testing or surveillance was necessary for the patient or relatives.

Additionally, several other tests and preventative options could have been discussed with the patient, including but not limited to additional testing for an additional hereditary colorectal cancer gene and recommending more frequent colonoscopies at earlier ages for all members of the family.

Analysis

Presumably, the patient utilized genetic counseling services to determine whether he carried a hereditary gene that would make family members more susceptible to colorectal cancer. Importantly, the case provided to DORA does not indicate whether the genetic counselor harmed the 86 year old patient. Instead, the case focuses on a variety of tests and other preventative options that that genetic counselor failed to recommend. Again, based on the information provided in the case, the testing and preventative options appear to be for the benefit of relatives.

Since the information in the case mentions that the genetic counselor did not recommend additional tests nor did he/she discuss other preventative options available, the relatives of the 86 year old could have been harmed by contracting colorectal cancer in the future. If the genetic counselor was practicing beyond his/her scope of practice, then regulation may serve to enhance consumer protection by authorizing the state to potentially discipline this practitioner.

[30] Genetics Home Reference. *Lynch Syndrome.* Retrieved August 14, 2013, from http://ghr.nlm.nih.gov/condition/lynch-syndrome

Additionally, Centura Health Cancer Network provided examples of harm to the public caused by healthcare professionals without education and experience in genetics. The examples provided are as follows:

- Wrong genetic test ordered and diagnosis missed;
- Wrong genetic test ordered, increasing patient's out-of-pocket cost;
- Inadequate pre-test counseling;
- Inadequate post-test counseling about family risks;
- Incorrect interpretation of results as negative;
- Incorrect interpretation of results as mutation positive;
- Insufficient healthcare provider familiarity with complex ethical, legal and psychosocial issues in genetic testing;
- Inadequate information supplied to testing lab; and
- Inadequate post-testing results notification.

The examples highlighted above do not delineate whether licensed healthcare professionals are providing genetic counseling services without the proper education and experience. If, for example, a licensed physician is attempting to provide genetic counseling services and he/she does not possess the necessary training to do so, then he/she would be practicing outside the scope of his/her practice and be subject to discipline by the Colorado Medical Board.

Centura Health Cancer Network's submissions of harm also stated that in some non-genetic practices, non-medical professionals, such as secretaries, fill out test requisition forms and provide genetic test results to patients.

Secretaries complete many forms at the request of physicians, nurses and others. It is unclear how this qualifies as harming consumers.

Also, providing test results to consumers does not necessarily constitute providing genetic counseling services nor does it indicate that the public was harmed. Therefore, it is unclear if a secretary simply handed a patient a copy of the test results after he/she received genetic counseling from a qualified, competent professional or actually dispensed genetic counseling services.

Additionally, DORA staff contacted the American Board of Genetic Counseling (ABGC) staff to discern the number and types of complaints and disciplinary actions imposed on genetic counselors who possess a certification. ABGC staff stated that one certification had been revoked in the past three to four years. This revocation, however, was not related to the certified genetic counselor harming a consumer(s) while acting as a genetic counselor. Rather, the certified genetic counselor, who was a licensed physician, had his physician's license revoked, so the ABGC also chose to revoke the physician's ABGC certification.

In an attempt to identify harm to consumers, DORA staff also contacted representatives of the Colorado Medical Board and the State Board of Nursing. Staff from both boards did not recall receiving any complaints concerning genetic counseling in the past four or five years.

In sum, DORA utilized a variety of sources in an attempt to identify instances where unregulated genetic counselors were harming consumers. A comprehensive review of the information provided by several sources did not highlight areas where harm was occurring, which calls into question the need for government regulation.

Need for Regulation

The second sunrise criterion asks:

> Whether the public needs and can reasonably be expected to benefit from an assurance of initial and continuing professional or occupational competence.

This criterion addresses the proposition of whether the state should require a certain level of education and/or impose a requirement that genetic counselors pass an examination before being licensed in Colorado. As highlighted earlier in this report, all of the states (16) that license genetic counselors require practitioners to obtain certification from ABGC prior to securing a license to practice.

During this sunrise review, very little evidence was presented via the examples of harm in the sunrise application, or identified in interviews with other states that regulate genetic counselors, that practitioners do not possess adequate skills, education or competence to practice safely. Due to the limited instances of harm to consumers, at least anecdotally, it is conceivable that initial certification by ABGC has insulated consumers from harm. This is evidenced by the fact that this sunrise review uncovered limited instances of harm to the public.

Instead, the issues identified in this sunrise review stemmed from either healthcare professionals or medical office personnel failing to clearly understand the role and expertise of genetic counselors. More specifically, the vast majority of examples of harm were related to individuals without the proper credentials (education and training) providing genetic counseling services to patients, but who are otherwise licensed healthcare professionals.

As a result, the initial certification of genetic counselors by the private sector appears to be an adequate level of protection to protect consumers.

Alternatives to Regulation

The third sunrise criterion asks:

> Whether the public can be adequately protected by other means in a more cost-effective manner.

Public protection for consumers who utilize genetic counseling services could potentially be realized in a cost-effective manner in a variety of ways. First, the ABGC currently offers certification for genetic counseling. The ABGC requires the completion of an accredited master's degree program and the passage of an examination to become certified. According to the sunrise application, the vast majority of the genetic counselors practicing in Colorado already possess the certification offered by the ABGC.

Further, all of the 16 states that currently license genetic counselors require candidates to secure an ABGC certification prior to becoming licensed. Doing so ensures that genetic counselors are uniquely qualified to practice as genetic counselors.

As such, the certification offered by ABGC appears to insulate consumers from incompetent practitioners.

The certification offered by ABGC is well established and utilized by many practitioners throughout the country, including Colorado. Consequently, the certification is a viable option and alternative to state regulation.

Another consumer protection alternative that is cost-effective and provides consumer protection is to protect the title "genetic counselor" in Colorado's Consumer Protection Act (Act). The Act, among other things, protects professional titles to ensure only practitioners who have achieved a certain level of education may call themselves "genetic counselors."

Including genetic counselors in the Act would ensure that only genetic counselors who have the appropriate certification, abilities and degree use the term "genetic counselor." Doing so may serve to heighten consumer protection for consumers who utilize genetic counselors' services.

Collateral Consequences

The fourth sunrise criterion asks:

> Whether the imposition of any disqualifications on applicants for licensure, certification, relicensure, or recertification based on criminal history serves public safety or commercial or consumer protection interests.

The sunrise application does not propose using an applicant's criminal history as a disqualifier.

Conclusion

The sunrise application requested licensure (the most restrictive type of regulation) of genetic counselors. More specifically, the requirement for licensure is board certification as a genetic counselor by the American Board of Genetic Counseling (ABGC) and/or the American Board of Medical Genetics (ABMG). Prior to 1993, the ABMG was the sole source of private certification as a genetic counselor. Consequently, there are a few remaining practitioners in Colorado who are certified by the ABMG.

The examples of harm provided for this sunrise review were largely resubmissions from the previous sunrise review DORA conducted in 2004. Most of the examples, which are located in Appendix A on page 25, focused on non-genetic counselors (healthcare professionals) harming consumers. Harm to consumers can be divided into several categories, including but not limited to:

- Psychological/emotional;
- Financial; and
- Physical.

In an attempt to further uncover harm to the public, DORA staff also contacted a variety of other sources, including the Colorado Medical Board and the State Board of Nursing.

There is also the potential harm that the unregulated genetic counseling profession may cause to consumers. For example, there is always the possibility that a genetic counselor provides misinformation, misinterprets test results, conducts an incomplete pre-test screening or post-test screening. Doing so could have severe negative consequences to consumers. Although the potential for harm exists, there were very few new examples of harm provided for this sunrise review.

As such, it is reasonable to conclude that the actual instances or the potential for harm by genetic counselors is remote.

Additionally, the examples of harm provided highlight two issues related to regulation. First, the vast majority of healthcare professionals (e.g., licensed physicians and licensed professional nurses) are already regulated by the State of Colorado, and if they are practicing beyond their respective scopes of practice, they are subject to disciplinary action by their respective boards. Therefore, the State of Colorado already provides protection to consumers for the majority of examples provided to DORA for this sunrise review. This calls into question the need for additional regulation in the healthcare arena.

This sunrise review also revealed that genetic counselors are very knowledgeable as well as demonstrate a great deal of competence. To illustrate this, DORA contacted a variety of states that regulate genetic counselors, the ABGC and regulatory boards such as the Colorado Medical Board and the State Board of Nursing. There were very few complaints (two in Utah – neither of which resulted in formal disciplinary action) and even fewer disciplinary actions (one by the ABGC, which was not related to a violation for genetic counseling) in the past five years. Also, both the Colorado Medical Board and the State Board of Nursing have not received complaints concerning genetic counseling.

It appears that the private certification offered by the ABGC is adequate to provide a baseline of competency for genetic counselors, including in Colorado. The ABGC illustrates that, in this instance, the private sector is effective in ensuring genetic counselors are competent practitioners.

According to the sunrise applicant, in Colorado the vast majority of genetic counselors are either board-certified or board-eligible by the ABGC. This may explain the small number of instances of harm to consumers.

The effectiveness of the private certification mentioned above calls into question the need to create an additional regulatory model, especially given the fact that the private sector is fulfilling the regulatory need. Thus, government intervention (regulation) may not be necessary.

Another factor that may explain the low instances of harm is the fact that genetic counselors provide interpretive services and counseling related to genetic tests and other issues such as family history and medical records. As such, genetic counselors are required to possess a great deal of skill and knowledge to perform their job satisfactorily.

However, patients who utilize genetic counselors are still able, in most instances, to ultimately decide treatment options for potentially life-threatening or life-altering circumstances. Unlike licensed physicians, who typically assist in decision making with patients, they also perform the procedures. This increases their chances of harming consumers. Whereas genetic counselors counsel patients, but do not perform any medical procedures, so they do not have that additional opportunity to harm consumers.

Further, during the course of this sunrise review, there were concerns about non-genetic counselors utilizing the term “genetic counselor” without completing the proper or adequate training to provide genetic counseling services to consumers. One possible solution is to protect the term “genetic counselor” or “genetic counseling” by including it in the Colorado Consumer Protection Act (Act). Doing so would ensure that only persons who have satisfied the necessary requirements could use the term “genetic counselor” or “genetic counseling.”

However, there are limitations to the effectiveness of the Act such as it does not cover other terms that may be utilized. That is, persons could use terms such as “genetics specialist” to essentially assume the same duties as a genetic counselor.

In sum, the limited instances of harm to the public identified during this sunrise review, as well as the fact the private credentialing appears to be working well, illustrate that the need for government regulation in the marketplace for genetic counselors is unwarranted.

Recommendation – Do not regulate genetic counselors.

Appendix A – Examples of Harm Submitted by Applicant

The following examples of harm were taken from the Department of Regulatory Agencies' 2004 genetic counselors sunrise report.

Case 1

Source: Letter from patient.

Patient: Eight young adults who are members of an extended family.

Genetic Disorder: Retinitis Pigmentosa, an incurable hereditary disease that causes blindness.

Practitioner and Response: Eye doctor informed several patients in their youth that they would become blind.

Result: None of the eight young adults have had children, although three are married and five are 28 years of age or older, due to fear of transmitting the disorder.

Genetic Counselor's Response: Although the eight young adults are carriers of the defect and show slight abnormalities, there is evidence that they do not suffer themselves from the disorder and will not become blind. The counselor explained childbearing options and presented information on ongoing research studies.

Case Notes: The harm to these patients was not because of misinformation given by a genetic counselor but by incorrect information given by an eye doctor. Subsequently the patients chose not to have children and lived with fear of developing the disease.

Case 2

Source: Letter from patient.

Patient: Mother and two biological sons ages 13 and 11.

Genetic Disorder: Marfan syndrome is an inheritable condition that affects the connective tissue. The primary purpose of connective tissue is to hold the body together and provide a framework for growth and development. In those afflicted with Marfan syndrome, the connective tissue is defective and does not function as it should. Because connective tissue is found throughout the body, Marfan syndrome can affect many body systems including the skeleton, eyes, heart and blood vessels, nervous system, skin, and lungs.

Practitioner and Response: Before bearing children, the patient received information that she may have the syndrome. Acting on this information, she had her primary care physician perform some tests. The tests (not identified) did not identify any major findings of the syndrome but did identify many minor findings. She was informed that if she were to bear children, they would not be affected.

Result: She had two sons and they are both affected with the syndrome. Had she been correctly counseled initially, her decisions could have been based on accurate information.

Genetic Counselor's Response: Normally, a first generation person with Marfan syndrome is much more affected than later generations.

Case Notes: The harm to these patients was not because of misinformation given by a genetic counselor but by incorrect information given by their primary care physician.

Case 3

Source: Letter from patient.

Patient: Adult female.

Genetic Disorder: Marfan syndrome.

Practitioner and Response: When first diagnosing the patient with Marfan syndrome, the physician informed her that a pregnancy posed a high-risk for transmitting the disorder and should be discouraged.

Result: Decided to seek further advice from professional genetic counselors.

Genetic Counselor's Response: There is a 50 percent chance of passing the condition on to a child. In addition, there is increased cardiovascular risk associated with pregnancy in women with Marfan syndrome. However, there is an in-vitro fertilization process with gene replacement wherein the mutated gene that causes a form of Marfan syndrome could be identified before the fertilized egg is implanted.

Case Notes: There was potential for harm because of misinformation given to the patient by her physician. In this case, the harm was avoided when the patient sought genetic counseling.

Case 4

Source: Giardiello, Francis M., et al. "The Use and Interpretation of Commercial APC Gene Testing for Familial Adenomatous Polyposis." *The New England Journal of Medicine*. March 20, 1977 336:12, p. 823-828.

Patient: 177 patients from 125 families underwent predisposition genetic testing for a study performed at the Johns Hopkins University School of Medicine.

Genetic Disorder: Familial adenomatous polyposis, an inherited condition that leads to the development of colon cancer at an unusually early age.

Practitioner and Response: In almost one third (31.6 percent) of the cases, the physicians' interpretations of the test results were incorrect and would have resulted in misinformation given to the patients. The physicians did not know that a test in which no mutation was detected could represent a false negative result.

Result: Individuals would have thought that they were no longer at risk for colon cancer.

Genetic Counselor's Response: Eighteen percent of the study group received genetic counseling prior to genetic testing and received an accurate interpretation of their genetic test results. The study supports the concept that physicians who order this test must be prepared to offer their patients genetic counseling, either personally or through referral.

Case Notes: Recommending genetic counseling and obtaining informed consent prior to testing are considered essentials, but neither was done in over 80 percent of the cases.

Case 5

Source: Cohn, Gabriel, et al. "The Importance of Genetic Counseling Before Amniocentesis." *Journal of Perinatology*. 1996 16:5, p. 352-357.

Patient: A retrospective study was undertaken. Charts of 275 consecutive patients referred for genetic counseling and amniocentesis on the basis of advanced maternal age (AMA) were compared with charts of 103 consecutive patients referred for an abnormal maternal serum alpha-fetoprotein (MSAFP).

Genetic Disorder: MSAFP screening is a blood test that looks at specific proteins to screen for certain types of birth defects like neural tube defects (such as spinal bifida) and Down syndrome in pregnancy.

Practitioner and Response: Pedigree information obtained during counseling of these patients was compared with the family histories charted by the referring physician. In 35.6 percent of pedigrees evaluated, a significant genetic risk was discovered during genetic consultation that had not been noted by the referring physician.

Genetic Counselor's Response: 9.8 percent of AMA patients and 10.7 percent of patients with abnormal MSAFP results underwent additional genetic testing or screening on the basis of genetic counseling.

Case Notes: The findings support the relevancy and usefulness of genetic counseling in more accurately ascertaining genetic risk and in maximizing the benefits of genetic evaluation of patients seemingly at low risk for other genetic diseases. However, the physician in these cases, not the genetic counselor, failed to note the genetic risk.

Case 6

Source: Submitted by Applicant.

Patient: Adult female.

Genetic Disorder: Huntington's Disease (HD), an adult onset neurological condition that affects movement, behavior, and thinking.

Practitioner and Response: Primary care physician ordered the HD genetic test for the patient when she mentioned that there was a family history. However, the physician did not offer appropriate pretest counseling or obtain adequate informed consent. The physician was unaware of the well-established HD pre-symptomatic testing protocol, which included not only genetic counseling, but also evaluation by a psychiatrist and neurologist.

Result: The genetic test results were indeterminate, which the primary care physician had not discussed as a possibility with the patient, and the physician could not interpret the results. The patient experienced heightened anxiety and regret that she had genetic testing.

Genetic Counselor's Response: Subsequently, the patient was referred to a genetic counselor.

Case Notes: The harm to this patient was not because of misinformation given by a genetic counselor but by the primary care physician.

Case 7

Source: Submitted by Applicant.

Patient: Adult male.

Genetic Disorder: Dominant vision condition.

Practitioner and Response: A non-genetic healthcare provider diagnosed a patient with vision loss and muscle problems as having a mitochondrial disorder. The provider did not document a complete family history, which is provided as part of routine genetic counseling services. This diagnosis remained with the patient for years.

Result: The diagnosis lead to incorrect risk assessment for the patient's children, causing unnecessary emotional stress.

Genetic Counselor's Response: When the patient presented for genetic counseling, the counselor determined from the detailed family history intake that this was an incorrect diagnosis. Rather, the patient had a dominant vision condition and his muscle problems were not believed to be of genetic etiology.

Case Notes: The harm to this patient was not because of misinformation given by a genetic counselor but by a non-genetic healthcare provider.

Case 8

Source: Submitted by Applicant.

Patient: Adult couple.

Genetic Disorder: Infertility.

Practitioner and Response: A couple underwent infertility treatment for 10 years. During this period, a family history was never taken.

Result: The female underwent unnecessary surgical and hormonal treatments in an attempt to remedy the infertility.

Genetic Counselor's Response: A genetic test determined that the husband carried a genetic rearrangement, called a balanced translocation, which explained their infertility.

Case Notes: The harm to this patient was not because of misinformation given by a genetic counselor but because a family history was never taken during the infertility treatments.

Case 9

Source: Submitted by Applicant.

Patient: Adult female.

Genetic Disorder: Patient's family had a strong family history of Duchenne muscular dystrophy, an X-linked condition, in which affected boys die in their teens. Carrier females are usually unaffected, but have a 50 percent chance of transmitting the condition to their sons.

Practitioner and Response: The patient was never informed of her reproductive risks nor was she offered preconception genetic counseling.

Result: She became pregnant and terminated the pregnancy upon learning she was carrying a boy. She was under the impression that a boy would have a 100 percent chance of being affected.

Genetic Counselor's Response: Did not visit with a genetic counselor.

Case Notes: There was no certified genetic counselor involved in this case. The patient was not given the correct information to make an informed decision.

Case 10

Source: Submitted by Applicant.

Patient: Adult female.

Genetic Disorder: Turner syndrome, a chromosomal condition caused by a missing sex chromosome.

Practitioner and Response: A physician had ordered chromosomal studies for both the patient and her husband. However, in this situation, chromosome studies of the parents were not indicated and would not provide information regarding risks for future pregnancies.

Result: Patient had been falsely reassured that she had no risk of a chromosomally abnormal pregnancy occurring since her and her husband's test results were normal.

Genetic Counselor's Response: Patient was referred to a certified genetic counselor to discuss prenatal testing because she was over 35 years of age. The patient's history revealed a prior miscarriage, whereby the fetus was diagnosed as having Turner syndrome. She was given correct information regarding her risks to allow her to make an informed decision about prenatal testing options.

Case Notes: The harm to this patient was not because of misinformation given by a genetic counselor but by incorrect information given to her by her physician.

Case 11

Source: Letter from patient.

Patient: Adult female.

Genetic Disorder: Gaucher Disease, a genetic condition that affects many parts of the body, including the bones, blood, organs, and energy level.

Practitioner and Response: Despite years of having chronic health problems and having a brother with Gaucher Disease, none of the specialists that treated the patient helped her condition or even identified it.

Result: Only recently did a physician identify that the patient had Gaucher Disease. She was subsequently referred to a genetics counselor.

Genetic Counselor's Response: The genetics counselor performed a risk assessment to determine whether the patient's daughter might have inherited this disease and the pros and cons of having her daughter tested. Additionally, the counselor recommended enzyme replacement therapy, which is a treatment for Gaucher Disease.

Case Notes: The harm to this patient was not because of misinformation given by a genetic counselor but by the lack of proper diagnosis by many physician specialists.

Case 12

Source: Letter from patient.

Patient: Adult female.

Genetic Disorder: 5-T allele carrier. This disorder is a problem if the individual also carries the cystic fibrosis transmembrane conductance regulator (CFTR) gene that together may result in a child born with Cystic Fibrosis.

Practitioner and Response: Patient was referred to the University of Colorado Health Sciences Center Adult Medical Genetics Program because of concerns that she might be a 5-T allele carrier. The patient's sister, who resides in Tennessee, was pregnant and a genetics test identified her as a 5-T allele carrier. The implications of this condition were not adequately explained to the patient's sister by her obstetrician and as a result minor panic ensued. As a result of the test on the patient's sister, the patient visited her family practitioner in order to obtain further information. The family practitioner referred the patient to a genetic counselor at the University of Colorado Health Sciences Center.

Genetic Counselor's Response: The counselor explained that the 5-T allele alone is not enough to cause medical problems. Although it is carried on the Cystic Fibrosis gene, unless there is another mutation, the 5-T allele is non-expressive. The patient was informed of a genetic test to determine whether she had other CFTR mutations.

Result: After gaining the information from the genetic counselor, the patient conveyed the information to her extended family.

Case Notes: There was no harm imposed upon this patient by a genetic counselor. She was given appropriate information by the counselor concerning the various mutations that must occur for Cystic Fibrosis to occur.