DOWNLOAD PDF STRATEGIES IN GENETIC COUNSELING

Chapter 1 : Counseling About Cancer: Strategies for Genetic Counseling - Katherine A. Schneider - Google

Counseling About Cancer: Strategies for Genetic Counseling has a clear language and points out the main topics in hereditary cancer and genetic counseling.

Be sure to check with each program for the most up-to-date application prerequisites. What classes should I take before applying? Program prerequisites vary somewhat; some schools require more prerequisite classes than others, but they most commonly include coursework in genetics, biochemistry, psychology and statistics. For the genetics requirement, most schools require that there is a human genetics component to the class you have taken. Be sure to plan ahead! Other classes that may be helpful or required in some cases include embryology, anatomy, statistics, and cell biology. Check the website of the programs to which you are applying for the most up to date information on prerequisites. Where can I find a ranking of genetic counseling programs? Which is the best? Genetic counseling programs have collectively decided against rankings. The underlying philosophy uniting the genetic counseling programs is that each program has its own relative strengths, but that overall the programs consider themselves equals. It is recommended that prospective students choose programs based on their own intellectual, philosophical, and geographical preferences. All programs have clinical, research, and coursework components, but many programs handle these components differently. Students can research individual programs further in order to decide which programs would suit their needs best. Application requirements and timelines differ slightly between schools, but generally schools require a personal statement, letters of recommendation, and GRE scores. Applications typically open in the fall and close in early winter December-February. Schools then invite qualified applicants for interviews, which typically take place February-April. Applicants are usually notified of application decisions by the end of April. Check the website of the programs to which you are applying for the most up to date information on application requirements and deadlines. How many schools should I apply to? The more schools you apply to, the better your chances are of receiving an interview and ultimately being accepted. However, many applicants are limited by finances, geographic preferences, and time in considering how many schools to which they can reasonably apply. On average, students apply to schools, however, some students choose to apply to only a few, while others apply to many more. It is important to apply to schools at which you could ultimately envision yourself. What experiences should you have before applying to graduate school? There is no one right or wrong set of credentials to have before applying. Many schools require you to have counseling or crisis intervention experience as well as experience shadowing a genetic counselor. Having telephone conversations or interviewing genetic counselors about their job may be ways to bolster your experience to the field if shadowing is difficult. Beyond those, experiences that students have before applying are varied and many. Advocacy work or working with people with disabilities, leadership experience, volunteering, tutoring, lab work and research, working in the healthcare field, holding employment, working with children, or just being involved with things you are passionate about all can be valuable experiences to have. Every applicant is unique and has a different story! Can I shadow or otherwise observe a genetic counselor? Are there other ways to get first-hand experience with genetic counseling before applying to programs? Sometimes it is possible to shadow a genetic counselor. Shadowing a genetic counselor is a great way to become more familiar with the profession and to gain practical experience. You can find a local genetic counselor through the NSGC website http: Some genetic counseling programs and labs have information sessions about genetic counseling that might be helpful as well. Can I still get into a genetic counseling program? Applicants are assessed as a whole, and it is entirely possible to have a strong application in spite of a weak area. That being said, areas of weakness should be addressed in your application and you should ideally show evidence of improvement. However, it is important to note that some genetic counseling programs have minimum GRE or GPA requirements so it is best to check with individual program requirements if there are any concerns. Do I need to complete a subject GRE exam to apply for a genetic counseling program? Some programs will accept subject GRE scores. However, at the time of this guide being written, no programs require it. Check with the individual programs to which you are applying for the most up to date information on application

requirements and deadlines. What are some tips for the application process? Applying takes a lot of organization and you have to send many different pieces of information to multiple schools. The writers of your letters of recommendation need plenty of time to write and mail their letters. Documents may get lost in the mail or end up in the wrong location at the university, so you should consider sending in documents well before the deadline in case something gets lost. What are some tips for writing my personal statement? Giving yourself plenty of time for revisions, and ask your friends and colleagues to give you feedback. Do not simply regurgitate your resume; this is your chance to tell your story and show your passion for genetic counseling. Use your experiences as anecdotes to reinforce what you are saying, but concentrate more on telling your unique story. Put your personality into it and really let the programs know why you are passionate about the field. Elaborate on how your personal background makes you a good fit not only for the school, but for the field as a whole. Check with individual programs to get the most up to date information about what points to address in your personal statement. Preparing for the Interview Are interviews required for acceptance into a program? Yes, interviews are required for acceptance into a program. The interview process varies by program, but typically involves discussions with the program director, assistant director, or other program faculty regarding your experiences and goals. Many programs also provide an opportunity to meet with current students during your scheduled interview date. Do programs allow for Skype or other virtual interviewing so I can avoid needing to travel? In-person interviews are typically required for all applicants. Certain programs have been able to arrange virtual interviews for international applicants or special circumstances, but this is not common. Please inquire with the program directly about the possibility of virtual interviews. What should I wear to interviews? When in doubt, it is better to be overdressed than underdressed. Typically interviews require business professional dress unless specifically stated otherwise. For women, it is best to wear a suit either pants or skirt , a professional dress, or something of a similar caliber. For men, this means a suit or dress pants with button-up shirt with a tie. Any other interviewing tips? Programs want to know that you will fit in with the culture of their program. Be passionate about your strengths and interests, and honest about your weaknesses. If you were invited for an interview, the program thinks you are a qualified candidate. Ask questions, and write thank you notes or emails afterwards. Some people find it helpful to make a list of questions before the interview and take it with you. Having copies of your CV can be helpful as well. This will be helpful when it comes to deciding where you want to go. Life as a Genetic Counseling Student What kind of classes do you take in genetic counseling graduate programs? The classes focus on the skills and topics that you will need for your job as genetic counselor. Of course, you will have to learn basic scientific and genetic principles to build your foundation for a career in this field. What kind of clinical training is involved? Programs all require that you are trained in the three core genetics specialties: Most, if not all, programs offer the opportunity to integrate or participate in specialty clinics outside of or within those three domains, such as neurology, cardiology, or pediatrics specialty clinics. As genetic counselors become more involved in a diverse range settings outside the clinic, programs are offering more opportunities to rotate or train in non-clinical roles such as industry and research. Can you tell me more about the research requirements in genetic counseling programs? Some programs require a thesis in which the student designs a research project, carries it out, and writes up the results. Some programs ask that each student do a thesis, while others allow students to work on a thesis project as a group. Other programs have a capstone project, where the student writes a research paper or develops a project related to genetic counseling, but do not necessarily do a formal research project. What is the search for jobs like? The job search is extremely variable between people. Being open to multiple specialties and locations throughout the country will certainly make the job hunt easier. Many graduates have jobs lined up before they leave school. If the job hunt is something that is already on your mind, make sure you find a school that provides ample professional development opportunities and mentorship throughout the job hunt process! Can I have a life in grad school? Genetic counseling graduate programs demand lots of time and will challenge you. However, many students hold jobs, volunteer, participate in hobbies, and have time for friends and family outside of school. Grad school is what you make it - if you want to get extra involved, that is absolutely an option, but if you want to have balance, that is an option, too. Making sure that you attend a program that feels right, supports you as an individual, and makes

you feel comfortable is a big part of achieving that balance.

Chapter 2 : Effectiveness of the Interventions Utilized in Genetic Counseling

The study offered genetic counseling free of charge to all study participants with a family history of breast cancer, removing the potential barriers of cost, the need for a physician referral.

Frequently Asked Questions by Robin L. Bennett, MS, CGC, Senior Genetic Counselor, University of Washington Medical Center After a diagnosis of cancer is made, it is normal to wonder if you are at risk to develop another cancer or if people in your family have an increased risk of developing cancer. Overall, about 5 percent of cancer has a strong genetic component and therefore there can be a significant risk for other relatives to develop cancer. Persons with an inherited cancer condition may also be at risk to develop another primary cancer. Knowing the medical history of your close relatives siblings, parents, aunts and uncles, grandparents, and first cousins can help to determine if your relatives have a higher risk for cancer. Clues in a medical family history suggestive of an inherited cancer genetic condition include: There are many genealogy software programs that help you trace your family history. The Surgeon General has a free Web based tool that you can even share with your family to record your family medical history. There are also family history tools on the website of the National Society of Genetic Counselors. Genetic counselors can assist you and your health professionals to determine if there is any genetic testing that might help assess your lifetime risk of developing cancer. Usually the best person to have tested for a genetic link to cancer in a family is the relative who has had cancer at the youngest age, or who has had multiple primary cancers. If a gene change mutation is identified, then other relatives can benefit from genetic counseling and testing. However, even if no genetic testing is available, genetic counselors can often help provide recommendations for cancer screening and prevention. A referral is usually required. To schedule an appointment, call Frequently Asked Questions Q. What can happen when genetic test results are placed in medical records? Consequently, individuals considering genetic testing must understand that their results might not be kept private. The Privacy Rule requires that health care providers and others protect the privacy of health information, set boundaries on the use and release of health records, and empower individuals to control disclosure of their health-related information. Many states also have laws to protect the privacy and limit the release of genetic and other health information. GINA prohibits discrimination based on genetic information in relation to health insurance and employment, but the law does not cover life insurance, disability insurance, and long-term care insurance. When applying for these types of insurance, people may be asked to sign forms that give an insurance company permission to access their medical records. The insurance company may take genetic test results into account when making decisions about coverage. Some physicians keep genetic test results out of medical records. National Cancer Institute Fact Sheet: Cancer Risk and Genetic Testing.

Chapter 3 : Genetic Counseling

Important scientific discoveries and ever-changing guidelines for how to identify and manage patients with hereditary cancer syndromes are constantly evolving. This Third Edition of Counseling About Cancer is completely updated and expanded to feature five entirely new chapters on breast cancer, colon cancer, other solid tumors, clients and families, and genetic test results and follow-up.

They determine whether you, your partner, or your baby carry genes for some inherited disorders. Genes are made up of DNA molecules, which are the building blocks of heredity. Humans have 46 chromosomes, arranged in pairs in every living cell of our bodies. When the egg and sperm join at conception, half of each chromosomal pair is inherited from each parent. This newly formed combination of chromosomes then copies itself again and again during fetal growth and development, passing identical genetic information to each new cell in the growing fetus. Current science suggests that every human has about 25, genes per cell. An error in just one gene and in some instances, even a change in a single piece of DNA can sometimes be the cause for a serious medical condition. What Do Genetic Counselors Do? Genetic tests yield complex results. Understanding what they mean is where a genetic counselor comes in. They then pass a certification exam administered by the American Board of Genetic Counseling. Genetic counselors can help: Who Should See a Genetic Counselor? Many problems that happen are treatable. Cleft palate and clubfoot, two of the more common birth defects, can be surgically repaired, as can many heart defects. The best time for genetic counseling is before a woman becomes pregnant. The counselor can help her understand any risk factors. But even during pregnancy, a meeting with a genetic counselor can be helpful. Experts recommend that all pregnant women, regardless of age or circumstance, be offered genetic counseling and testing to screen for Down syndrome. The counselor will want to know of any relatives with genetic disorders, multiple miscarriages, and early or unexplained deaths. The counselor will also want to look over your medical records, including any ultrasounds , prenatal test results, past pregnancies, and medicines you took before or during pregnancy. The counselor can help you understand the inheritance patterns of disorders and help assess your chances of having a child with those disorders. The counselor will talk about risks that every pregnancy faces and risks that you personally face. For instance, a child with cystic fibrosis can have debilitating lung problems or, less commonly, milder respiratory symptoms. If more tests are needed, the counselor will help you set up those appointments and track the paperwork. When the results come in, the counselor will call you with the news and may ask you to come in for another discussion. What Happens After Counseling? Genetic counselors can help you understand your options and adjust to any uncertainties you face. But you and your family will decide what to do next. This is when eggs that have been fertilized in vitro in a laboratory, outside of the womb are tested for defects at the 8-cell blastocyst stage. Only nonaffected blastocysts are implanted in the uterus to establish a pregnancy. Surgery can only be used to treat some defects, such as spina bifida or congenital diaphragmatic hernia, a hole in the diaphragm that can cause underdeveloped lungs. Most defects cannot be surgically repaired. But they will not suggest a particular course of action. A genetic counselor understands that what is right for one family may not be right for another. Genetic counselors can, however, refer you to specialists for further help. Genetic counselors can also refer you to social workers, support groups, or mental health professionals to help you adjust to and prepare for your complex new reality. What Else Should I Know? Working with a genetic counselor can be reassuring and informative, especially if you or your partner have known risk factors. Talk to your doctor if you feel you would benefit from genetic counseling. Many doctors have a list of local genetic counselors they work with. You also can contact the National Society of Genetic Counselors for more information.

Chapter 4 : National Society of Genetic Counselors : Policy & Publications

The editors have solicited the opinions of two end users, Kevin Sweet, a certified genetic counsellor practising in the United States, and Gareth Evans, a physician clinical geneticist practising in the United Kingdom.

Examples of roles include: Laboratory â€" Utilization management, provider and patient support, variant classification, and reporting Research â€" Coordinating research studies, patient recruitment, data collection and interpretation, manuscript preparation and grant writing Education â€" Professors, Directors of genetic counseling training programs Public Health â€" Newborn screening programs, population screening programs Non-profit â€" Patient support and advocacy organizations Corporate â€" Dedicated services for employees and their families Education[edit] A genetic counselor is an expert with a Master of Science degree in genetic counseling. There are currently 41 accredited programs in the United States, four accredited programs in Canada, and four programs with the intent to become accredited. Clinical training including supervised rotations in prenatal, pediatric, adult, cancer, and other subspecialty clinics, as well as non-patient facing rotations in laboratories. Research training typically culminates in a capstone or thesis project. Although not every company requires its counselors to possess a certification, the certification shows that the practitioner has met the standards "necessary to provide competent genetic counseling services". Almost every other state in the US is in the process of obtaining genetic counseling licensure. Utah was the first state to do so. Laws requiring licensure ensure that "professionals who call themselves genetic counselors are able to properly explain complicated test results that could confuse patients and families making important health decisions". Patients who may benefit from genetic counseling may not be able to afford the service due to the expensive out-of-pocket cost. Seymour Kessler, in , first categorized sessions in five phases: The initial contact phase is when the counselor and families meet and build rapport. The encounter phase includes dialogue between the counselor and the client about the nature of screening and diagnostic tests. The summary phase provides all the options and decisions available for the next step. If patients wish to go ahead with testing, an appointment is organized and the genetic counselor acts as the person to communicate the results. Result delivery can happen both in person or via phone. Often counselors will call out results to avoid patients having to come back in as results can take weeks to be processed. If further counseling is needed in a more personal setting, or it is determined that additional family members should be tested, a secondary appointment can be made. Reasons for testing and sub-specialties[edit] Detecting conditions[edit] Diagnostic testing occurs when an individual is showing signs or symptoms associated with a specific condition. Testing can reveal conditions thatâ€"while debilitating without treatmentâ€"can be mild or asymptomatic with early treatment such as phenylketonuria. Genetic tests are available for a number of genetic conditions, including but not limited to: Down syndrome , Sickle cell disease , Tay-Sachs disease , Muscular dystrophy. Establishing a genetic diagnosis alerts any other at-risk individuals in the family to consider testing, if appropriate. Any reproductive risks e. Many disorders cannot occur unless both the mother and father pass on their genes, such as cystic fibrosis ; this is known as autosomal recessive inheritance. Other autosomal dominant diseases can be inherited from one parent, such as Huntington disease and DiGeorge syndrome. Yet other genetic disorders are caused by an error or mutation occurring during the cell division process e. Screening tests are often used prior to diagnostic testing, designed to separate people according to a fixed characteristic or property, with the intention of detecting early evidence of disease. For example, if a screening test during a pregnancy such as maternal blood screening or ultrasound reveals a risk of a health issue or genetic condition, patients are encouraged to receive genetic counseling to learn additional information regarding the suspected condition and about their options with discussion of management, therapy and treatments available for the conditions, including those after delivery, and those during pregnancy, such as the option to terminate the pregnancy. Patients may decline additional screening and testing, elect to proceed to diagnostic testing, or pursue further screening tests to refine the risk during the pregnancy. Presymptomatic or predictive testing occurs when an individual knows of a specific diagnosis typically adult onset in their family and has other affected relatives, but they themselves do not manifest any clinical findings at the time when they seek testing. Availability of

treatment and medical management options for each specific diagnosis, as well as the genetics and inheritance pattern of the particular condition should be reviewed as inherited conditions can have reduced penetrance. Insurance and legal issues should also be discussed during genetic counseling. Adult genetics[edit] Adult or general genetics clinics serve patients who are diagnosed with genetic conditions that begin to show signs or symptoms in adulthood. Many genetic conditions have varying ages of onset, ranging from an infantile form to an adult form. Adult-onset disorders may overlap multiple specialties. PGT-SR, for structural rearrangements, involves testing embryos to establish a pregnancy unaffected by a structural chromosomal abnormality translocation. PGT-A, for aneuploidy, was formerly called preimplantation genetic screening, and involved testing embryos to identify any de novo aneuploidy. Cardiovascular genetics[edit] A rapidly expanding field in genetic counseling is cardiovascular genetics. More than 1 in people have an inherited cardiovascular disease. Hereditary cardiac conditions range from common diseases, such as high cholesterol and coronary artery disease, to rare diseases like Long QT Syndrome , hypertrophic cardiomyopathy , aortic and vascular diseases. Genetic counselors who specialize in cardiovascular disease have developed skills specific to the management of and counseling for cardiovascular disorders. Cardiovascular genetic counselors are also integral in local and national efforts to prevent sudden cardiac death the leading cause of sudden death in young people by identifying patients with known or suspected heritable cardiovascular diseases and promoting cascade family screening or testing of at-risk relatives. Common referral reasons include:

Chapter 5 : Counseling about Cancer: Strategies for Genetic Counseling by Katherine Schneider

Katherine Schneider received her Master's Degree in Public Health from Yale University School of Medicine in and was certified as a genetic counselor by the American Board of Medical Genetics in

Blood samples from numerous members of my family had to be collected and analyzed. I underwent several months of genetic counseling to determine my ability to cope with any possible outcome. After a period of months, nothing remained but the nerve-racking wait for the results Finally, the wait was over: When I learned the results I cried and laughed. It took months for the news to sink in. I am still adjusting. The incomparable relief I felt at finally being free of the fear and uncertainty. Hayes, It is but sorrow to be wise when wisdom profits not. Sophocles, Oedipus Rex Genetic testing raises a broad range of questions and issues for those considering testing and for those offering the test: How great are the risks of the test? How reliable is the test? What does this information mean for me, for my children, for my family, for future generations? What is the nature of the disorder? What is its severity? What options are available? How will we choose? What medical and support services will be needed? What resources are available? What does the future hold for health, longevity, quality of life? Implications for Health and Social Policy. The National Academies Press. Along with their questions, people bring a wide variety of values and personal health beliefs about the central issues raised by genetic testing to the genetic testing and counseling experience. Genetic counseling is the context for helping people address such issues. This chapter includes background information on the nature and basic components of genetic counseling in various settings newborn screening, carrier detection, prenatal diagnosis, and screening for late-onset disorders. It also reviews critical issues facing genetic counseling today and for the future. Among these critical issues are nondirectiveness; informed consent; confidentiality; multiplex testing; recognizing social and cultural differences; and the need for a genetically literate public. Many people who undergo genetic testing receive "good news" and reassurance with their genetic test results. They may learn definitively, or with a high probability, that neither they nor their children have a specific genetic disease. Many other people also learn that they and their children do not carry the gene s for that disorder. Other people who undergo genetic testing will be informed that a genetic disorder or genetic susceptibility has been identified in their fetus, their children, or themselves. Test results may be deeply troubling for those who receive a diagnosis of a genetic disorder or carrier status, raising fundamental questions of medical vulnerability, as well as personal and social image and identity. The counselor acts as a resource in dealing with the sadness, loss, anger, guilt, or anxiety that genetic information can bring Kessler et al. In educating and counseling about genetics, the counselor must convey the varying nature of genetic risk and our varying ability to predict such risks. The prediction of genetic risk also depends on the sensitivity and specificity of the test itself and the quality of laboratory procedures see Chapter 3. The immediacy of decision making is another key variable in genetic testing and counseling. The time pressure surrounding genetic testing varies by circum- Page Share Cite Suggested Citation: For treatable disorders such as phenylketonuria PKU , early identification of the disorder in newborns is critical so that dietary modification can be started early enough to prevent severe mental retardation. For many late-onset disorders and for preconceptional reproductive planning, genetic testing and counseling may occur in adulthood when the information will be of practical use; in these instances, the process of deciding whether to be tested need not be rushed. In other circumstances, it will be necessary to have information from genetic tests much more quickly so that decisions can be made. Although carrier testing is optimally performed before pregnancy, the most time-urgent of such decisions often surround reproduction, especially in prenatal diagnosis where safety dictates only a limited time during pregnancy in which to decide whether to be tested and to make decisions about whether to terminate or to carry to term a pregnancy if a genetic disorder is identified in the fetus. There is tremendous variability in genetic counseling as provided today and envisioned for the future. As genetic testing expands with the growth of new genetic tests, genetic counseling and education will need to adapt to new modes and settings for the delivery of genetics services, without sacrificing quality. Health care providers will require an enhanced appreciation of the contribution of genetics to health, as well as an understanding of the complexities

of genetic testing and decision making. This chapter reviews the basic tenets of genetic counseling as it has been defined and practiced in a variety of situations, and examines issues facing genetic counseling for the future. Page Share Cite Suggested Citation: Unquestionably, there are both educational and psychological support components to all levels of genetic counseling. In the "classic" model of genetics services, genetic counseling is provided by a specialized team of professionals, including a clinical geneticist and genetic counselor, and is often provided in a genetics center see Chapter 6 for discussion of personnel training and certification. The team might be much broader, particularly in specialty clinics where different disciplines are represented. For example, parents of children with cystic fibrosis CF often learn about recessive inheritance from pulmonologists, nurses, or social workers who provide care for their children in a CF clinic. Increasingly, however, genetics services are being provided by primary care providers, who are not necessarily trained in human genetics. Primary care practitioners are also less likely to endorse an important principle of classical genetic counselingâ€"that is, autonomous patient decision making e. The movement of genetics services into primary care is likely to increase as the number of genetic tests expands. Even if specialized genetics professionals are considered the best providers of genetic counseling services, there will simply be too few genetics professionals to meet the growing demand for services. However, traditional genetic counseling services, provided by specialized genetics professionals, are expected to remain a critical resource when test results reveal risks. Once genetic tests are judged to be "standard of care" for routine use, primary care practitioners are likely to be the ones to offer such testing and obtain informed consent. When risks are revealed, especially for nontreatable disorders including late-onset disorders and for those identified with carrier status, referral to specialized genetic counselors will usually be desirable because of the complexity of the issues in counseling for identified risk. Specialized genetics professionals will also increasingly need to train other personnel to provide genetic testing and counseling services as part of their professional activities. In many genetic counseling situations, a client must decide whether to seek diagnosis and, if so, must then decide how to use the information resulting from the test. To date, genetic testing and genetic counseling lead to few opportunities for curative treatment of genetic conditions see Chapter 2 ; thus, the primary emphasis in genetic counseling has been on facilitating autonomous decision making about receiving information on conditions for which treatment may not exist. In a few conditions, accurate diagnosis can lead to medical interventions, such as newborn screening and follow-up treatment for PKU. Prenatal diagnosis may provide reassurance as well as information for decisions on selective abortion if the fetus is determined to be at high risk for a diagnosable genetic disorder or in preparing for the birth of an affected child. Gene therapy has entered early clinical trials but is barely on the horizon for wide clinical use. This means that in many cases, the only intervention to be offered for a genetic disorder is communication about diagnosis, natural history, and information about available options, including a variety of reproductive options. In the absence of treatment, the psychological impact of this genetic information can be tremendous. Awareness of the Impediments to Effective Genetic Counseling Beyond the psychological consequences of receiving genetic testing information are the potential impacts on the familyâ€"not only the individual, but also the partner, parents, grandparents, siblings, and children of the individual being tested or screened. The diagnosis of a genetic condition or the results of a genetic test often have repercussions for future childbearing decisions as well, although this is only one of many components of genetic counseling. Social and psychological stress introduced by genetic diagnosis, as well as future financial and emotional burdens, can severely impact family functioning Schild, The provider of genetics services needs to be sensitive to the concept of the "teachable moment," that is, the point s at which an individual, couple, or family is most able to comprehend and absorb the information being given. The genetic counselor may not have the opportunity to counsel clients at more advantageous teachable momentsâ€"after some of the early shock and denial that often accompany genetic diagnosis have abated. Limited contact with the genetic counselor may often result from restrictions on insurance reimbursement see Chapter 7 and other administrative impediments, such as the practice of scheduling counseling on the day of testing. These and other factors may help explain why certain studies show limited retention and understanding of the genetic information conveyed during counseling interactions e. Potentially, one benefit of having primary care practitioners provide genetic counseling will be more

continuity of care, since continuity of care provides more opportunities over time for teachable moments. The psychological impact of a genetic diagnosis varies with its severity, treatability, and with the unique responses of different individuals and families e. Support, counseling, and follow-up can assist individuals and their families in coping with positive test results. The knowledge and skills of a properly trained counselor can help an individual understand the diagnosis, risk of recurrence, prognosis, Page Share Cite Suggested Citation: There is general agreement that certain issues raised by testing should be discussed before an individual decides to be tested. Education and counseling include providing information and supportive counseling to people considering testing about what they need to know to decide whether to be testedâ€"risk status, the benefits and burdens of testing, the limitations of available testing methods, and the implications of the test results, including the psychosocial consequences of such testing. Education and counseling are particularly important for genetic screening procedures, such as prenatal diagnosis for advanced maternal age or carrier screening for CF of an individual with no previous family history of the disease see Chapter 2. In probabilistic terms, such individuals have a high likelihood of receiving good newsâ€"that their fetus is unaffected or that they are not carriers. Education and counseling following testing include interpretation of test results, discussion of the implications of that information, answering all questions in a language and manner understandable to the person being counseled , providing supportive counseling, and offering information about community support groups and other follow-up resources. For some conditions, one visit might be sufficient to conduct posttest counseling, for example, after determination of TaySachs carrier status in a nonpregnant female. Other disorders might require several visits or, rarely, long-term supportive care may be needed. The variability of genetic disorders and of their impact demands flexibility in the delivery of services, both diagnostic and psychological. After genetic counseling, clients should have enough information at least to attempt to deal with the complex interaction among the risks and benefits of various courses of action and with their own values and personal choices Quaid, Since genetic diagnosis can sometimes present more uncertainty than certainty, it is important to communicate both information and empathy, because information exchange may be taking place in an atmosphere that is filled with anxiety and unfamiliarity Biesecker, a. And while the content and nature of genetic counseling may vary, certain basic tenets will almost always apply: Nondirectiveness Carl Rogers, a clinical psychologist, coined the term nondirectiveness in to describe his psychotherapeutic approach of not advising, interpreting, or guiding his clients. Eventually, Rogers came to recognize that his very presence in a counseling relationship had directive components. Concern about early abuses in the eugenics movement helped to make the principle of nondirectiveness, and the corollary of respect for client autonomy, key concepts of genetic counseling today. Nevertheless, the issue of nondirectiveness in genetic counseling has led to controversy and confusion Kessler, ; Biesecker, a. The desirability as well as the practicability of nondirectiveness in genetic counseling has been challenged e. This controversy reflects an inherent tension in genetic counseling that arises from the complex functions of genetic counseling for different purposes in various settings. The continuum of genetic counseling includes, at a minimum, providing genetic information and education, as well as providing genetic counseling to explore the implications of the information; but it may also include providing specific medical advice for treatable conditions e. Critics have challenged the ability of clinical geneticists and genetic counselors to practice nondirective counseling, and have raised concerns about the training and practice of primary care providers, which tends to encourage directive behavior Epstein et al. Primary care practitioners were found to be more directive in dealing with genetic situations than geneticists Holmes-Seidle et al. Women who were counseled by a general obstetrician were more likely to terminate a pregnancy in which a sex chromosome abnormality had been diagnosed than if they were counseled by a geneticist Holmes-Seidle et al. The variation in approach among practitioners is part of the reason why patients must have the final decision about whether to be tested, even for disorders that are treatable. Biases are inherent to human nature and are often projected by health care providers in less than subtle ways.

Chapter 6 : Prenatal Genetic Counseling

But in implementing new strategies for genetic counseling, questions need to be discussed that are specific to the settings in which genetic counseling is performed, argue two researchers from Indiana University School of Medicine.

This is an open access article distributed under the Creative Commons Attribution License , which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. Advances in genetic science and biotechnology accumulated huge knowledge of genes and various genetic tests and diagnostic tools for healthcare providers including nurses. Genetic counseling became important to assist patients making decisions about obtaining genetic testing or preventive measures. This review was conducted to describe the counseling topics, various interventions adopted in genetic counseling, and their effectiveness. Experimental studies published between and were synthesized. The most frequently covered topic was benefits and limitations of genetic testing on breast cancer ovarian and colorectal cancers. Most of researchers focused on evaluating cognitive aspect and psychological well-being. No single intervention was consistently reported to be effective. Decision aids enhanced with information technologies have potential to improve the outcomes of genetic counseling by providing tailored information and facilitating active engagement of patients in information uptake. Introduction As advances in genetic science and biotechnology have led to discovery of the genes that increase the risk for certain common diseases, various genetic tests and other diagnostic tools have become available to healthcare providers including nurses. As a result, the knowledge base for genetic counseling has become important to all healthcare professionals in terms of assisting patients with accessing timely screening, adopting healthy behaviors, and making decisions about obtaining genetic testing or aggressive preventive measures such as mastectomy, oophorectomy, or chemotherapy [1]. Counseling for genetic risk deals with multilayered complicated information such as the risk for developing certain diseases and benefits and limitations of various options for predicting or preventing the diseases. Uncertainty is inherent in the information communicated during counseling, making it difficult for patients to fully understand the information provided and causing inconsistency in the perceived meaning of the provided information among patients [4 â€” 6]. Effective communication of the information on risk levels and potential benefits of preventive measures to individuals and their family members is one key element of realizing the benefits of genetic counseling [7 â€” 9]. Various interventions designed to facilitate the delivery of complex information to patients have been adopted in genetic counseling and these strategies and effects have been summarized in several review papers [10 â€” 12]. We conducted a literature review on the studies that investigated the effects of various interventions applied to genetic counseling. Specifically, we intended to describe 1 the specific content delivered during the risk counseling, 2 the specific methods used to communicate the content to the patients, and 3 the effectiveness of the methods on the main outcomes that the studies intended to measure. Background A primary focus of genetic counseling in the s and 70s was to assist patients with reproductive decision makings by educating them about etiology and recurrence risks of certain congenital conditions [14]. The central component of genetic counseling is providing information on the possibility of developing a hereditary disease and the risk management options to promote informed decision making [7 , 15]. Therefore, effective delivery of the information on the genetic risk and the various risk mitigation options to the patients is the goal of genetic counseling. Despite its importance, methods of communicating genetic risk between genetic counselors and patients are relatively understudied and underdeveloped [17]. This is partly due to the fact that the genetic risk itself is a complex concept encompassing multiple related factors. Misattributed parentage occurs around 0. Also most importantly, the information keeps changing. According to the Center for Disease Control and Prevention, different genes and mutations are identified on almost a daily basis http: In a more recent review, Meilleuer and Littleton-Kearney [10] synthesized thirteen published papers that described various methods of educating patients on multifactorial genetic risks and their effectiveness. This review describes the effectiveness of each method according to the detailed outcome categories concordant to the multilayered goals of genetic counseling that we previously described. According to this review, the use of computerized aids yielded more positive

outcomes than other types of methods such as the use of video tapes and group counseling. In addition, it was not possible to discern what aspects of computerized decision aids led to the positive outcomes in this review as the interventions used in each study were described by high level categories rather than by specific functionalities of each computerized decision aid. To address the limitations noted in the recent reviews, we conducted a literature review on the studies that reported the effectiveness of various interventions adopted in genetic counseling. We included only the experimental studies conducted as a randomized controlled trial or other types of clinical trial i. We intentionally included journals published from to broaden our review. The same limits on publication dates, the study design, language, and abstract availability were applied. Papers that do not fit the inclusion criteria by both reviewers were excluded from this review. Papers that met the inclusion criteria by only one reviewer but not the other remained in the paper pool and were determined relevancy after reading the full article. The two authors read the full text of the 60 papers. After reading the full texts, additional 17 papers and 4 redundant papers were excluded from the analysis because topics were not relevant, simulated genetic counseling, were assessing genetic risks only, or were a combination of genetic counseling and testing. Finally, a total of 39 articles were reviewed. This paper selection process was summarized in Figure 1. The details on the studies such as related study subjects, interventions, evaluation methods, findings, and any limitations posed in the studies were recorded into a table format during the review Table 1. Results Majority of the studies 35 of 39 described the interventions related to genetic counseling on breast, ovarian, or colorectal cancers [19 â€” 25 , 27 â€” 38 , 40 , 41 , 43 â€” 56]. This is not surprising given that genetic associations were established mostly for these three types of cancers. Only one study dealt with cystic fibrosis [26] and three studies dealt with prenatal counseling [39 , 42 , 57]. Thirty-one of the thirty-nine studies were randomized controlled trials. The remaining eight studies evaluated the intervention effects by measuring and comparing the outcome variables at preintervention and postintervention time points without a control group [19 â€” 23 , 32 , 35 , 42]. Most of the interventions were employed during individual counseling sessions, but a few studies evaluated the effectiveness of community-based group counseling itself without an additional intervention [35 , 36]. Information Topics Covered in the Studies Counseling topics were similar across studies. The most frequently covered topic was the benefits and limitations of genetic testing found in twenty one studies [30 â€” 32 , 35 , 36 , 39 â€” 48 , 50 , 52 , 54 â€” 57] and the second most frequently covered topic was general knowledge of hereditary conditions found in eighteen studies [27 , 30 , 31 , 34 , 35 , 38 , 40 , 42 , 43 , 45 , 46 , 48 , 50 , 53 â€” 57]. Cancer prevention options and screening tests were found in five studies [33 , 36 , 38 , 40 , 41] and preventive surgery such as risk reduction mastectomy was found in three studies [28 , 40 , 50]. Cultural beliefs and values on genetic testing were discussed in one study which tested culturally tailored genetic counseling as an intervention [37]. Main Interventions Evaluated We grouped the key interventions evaluated in the reviewed studies into three general categories: Decision Aids Eight studies 8 out of 31 have tested the effectiveness of decision aids in genetic counseling. Ten studies evaluated paper-based decision aids, which are in the forms of booklets, paper handouts with or without illustrations , flip charts, or brochure [27 , 29 â€” 31 , 33 , 35 , 38 , 44 , 54 , 57]. Paper-based decision aids were usually used during the counseling and also sent with the patients after the counseling as a future reference. Nonpaper-based decision aids were evaluated in ten other studies [28 , 34 , 42 , 45 , 46 , 48 â€” 50 , 55 , 56]. Some of these decision aides were interactive and provided tailored information according to the specific information needs of a patient. Specific Techniques for Stress Coping and Decision Making Six studies have evaluated the effectiveness of the techniques employed to improve psychocognitive well-being when dealing with stress related to positive genetic test results or positive family history of hereditary cancer or to facilitate decision making on uptake of a test or an aggressive preventive measure. The techniques employed in these studies are follow-up reinforcement, psychosocial group counseling, prelive of stressful situation, use of a decision guide tool, and problem-solving training [36 , 39 , 40 , 43 , 47 , 53]. Other Interventions Five studies evaluated various other approaches to genetic counseling such as providing culturally or personally tailored information to individual patients and multidisciplinary team counseling [32 , 37 , 40 , 51 , 52]. Evaluation Methods The most frequently used evaluation method was the self-reported questionnaire. The next most frequently used evaluation method was interview , either face-to-face or over the

phone. Four studies used both interview and survey for evaluation. Chart review was used in two studies as a way to check the office visit appointment made for genetic testing after counseling. Fifteen studies evaluated the outcomes at multiple time points. Twenty-five studies evaluated the intervention effects comparing the postintervention assessments to the baseline assessments [29 , 32 â€” 38 , 40 â€” 48 , 50 â€” 57]. Eleven studies evaluated outcomes immediately after providing interventions [27 , 32 , 39 , 42 â€” 46 , 48 , 51 , 57], but five studies did so 1 or 2 week s after the interventions [30 , 31 , 35 , 40 , 41]. Long term effects were evaluated in many studies but adopted time lapses varied from 1 month to 12 months. Overall, there was no notable pattern of changes in outcomes by the follow-up time points. The interventions designed for improving psychological well-being related to genetic testing were evaluated specifically around the related events. For example, the level of worry and anxiety related to genetic test results were assessed before and after receiving the test results [32 , 40 , 47] and the decision conflict on uptake of genetic testing was assessed after making the decision [31 , 43 , 45 , 48 , 51]. Discussion Genetic counseling is an important clinical practice since it assists patients in choosing preventive screening test, adopting healthy behaviors, and making decisions about having genetic tests and preventive measures. Communicating accurate genetic risk levels effectively is the core process of genetic counseling. However, due to the complicated nature of the genetic risk, conducting such communication is challenging in clinical settings. The studies that we reviewed were less diverse in terms of the target diseases and the population thus may not represent the genetic counseling domain in general. Among the various information topics covered by the studies, detailed knowledge on hereditary disease was the most frequently appearing topic. Considering that people responded more proactively to the risks that they can mitigate [58 â€” 60], specific risk mitigation options should be the topic regularly covered during the counseling. Findings also varied across studies. Similar interventions did not necessarily yield similar outcomes. As noted in previous reviews, the intervention effects were less prominent in many studies and even contradictory across certain studies. One plausible reason might be the fact that personalized risk information was not communicated routinely during counseling even if communicating risk information was the central part of intervention [12]. When an intervention is constructed in a way that personalized risk information can be delivered to the patient effectively, the intervention effects may increase consistently. More studies evaluated the intervention effects on the intention related to health behavior but only one of them followed up on the actual behavior change. Therefore, it is not clear if the intention can be a reliable proxy for behavior change. Although longer term follow-up can be a challenge, more evaluation is needed to better understand the effects of various genetic counseling interventions on the actual behavior outcome. The studies investigated in this review show that a large amount of information is communicated with patients during limited counseling time. Also, the study findings suggest that any formats of decision aids can benefit both counselors and counselees in terms of time saving, streamlining the counseling process by providing readily available comprehensive information. Regardless of its format, however, the studies that used decision aids commonly showed increased knowledge, decreased decision conflicts, and increased satisfaction with counseling interventions. This implies that decision aids could serve as an essential tool in genetic counseling. Studies that utilized decision aids provide canned information to patients. It is clear that information technologies can provide additional benefits by supporting information tailoring and allowing users to take an active role in information seeking and decision making. This study reported improvement in knowledge, risk perception, psychological well-being, and satisfaction with the use of this tool. However, intention or behavioral outcomes were not measured in this study. Although intervention effect of using a computerized decision aid has not been proved widely, a computerized tool has potential to improve outcomes of genetic counseling because it provides a systematic way to customize risk and its mitigation information according to saliency of a patient. Such computerized decision aids can also present various educational topics such as risk level, disease related knowledge, family heredity, genetic testing, and screening options according to literacy of a patient, incorporating various presentation formats such as graphics and numbers. Conclusion The primary objective of this review was to describe the various methods employed to improve patient outcomes including health behaviors, psychological well-being, and enhanced decision makings by genetic counseling and their effects. The methods and outcome variables

evaluated varied across studies. We found that no single method was consistently reported bringing positive outcomes, partly due to the relatively small scale of the review. Various risk communication methods should be developed and outcomes of each method are measured in randomized clinical trial studies. As more such experimental studies accumulate in this domain, a more comprehensive review needs to follow in the future.

Chapter 7 : Genetic counseling - Wikipedia

This Third Edition of Counseling About Cancer is completely updated and expanded to feature five entirely new chapters on breast cancer, colon cancer, other solid tumors, clients and families, and genetic test results and follow-up. This is the only reference and clinical book on the market for cancer genetics counselors and other healthcare providers who must quickly assimilate complex and ever-changing data on the hereditary risk for cancer.

Chapter 8 : Genetic Counseling Strategies for Gaucher-Parkinson's Link Are Needed

Genetic counseling can be helpful in helping patients assess genetic risk factors that could lead to an initial cancer, or its recurrence, and developing a plan for screening and prevention.

Chapter 9 : National Society of Genetic Counselors : Frequently Asked Questions - Students

Genetic counseling is the process of advising individuals and families affected by or at risk of genetic disorders to help them understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.