



# Models of Genetic Counseling Services and Quality Assurance: A Theoretical Inquiry\*

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Since the 1980s, researchers report patient responses before, during, and after genetic consultation services to demonstrate cost effectiveness, patient satisfaction, knowledge gained, preferences for genetic counseling, psychological effects, family communication patterns, effect on physician referral patterns, and disclosure of risks to relatives.

Most researchers describe a model of risk assessment and communication rather than their organizational model of service delivery (Greco, 2003). Consequently, without a clear understanding of the organizational structure, professional roles and collaborations, funding sources, service goals, and quality assurance criteria inherent in each model, it is impossible to know whether one model is better than another, or whether certain tools or professional collaborations might lead to better services in certain settings or populations. The best quality assurance measures of models of services and their relationship to patient outcomes have yet to be well investigated (Roesser & Tatum, 2004; Williams, 2009; Chitayat, Langlois, Wilson, 2011).

By model of service, we mean the organizational structure, professional roles, types of patients/counselees, goals of the service, process of delivering the service, or educational tools used. We discovered that very few authors actually describe their model of service in any detail (Peters & Rubinstein,

2000). These traditional models of service describe the goal(s) of service, the target population, personnel, setting, operations/protocol/tools, and expected outcomes. More recently, researchers have investigated patient expectations (preferences), perceived experience during the service, and patient satisfaction have been used as indicators of quality (DeMarco, Peshkin, Mars, & Tercyak, 2004; Gustafson, Pfeiffer, & Eng, 2011; Holloway et al., 2004; Hopper, Buckman, & Edwards, 2011; Kausmeyer et al., 2006). Women who have the highest risk based on family history and who have had a recent cancer diagnosis in a relative are most likely to desire genetic risk information and preparation for testing (Apicella et al., 2006), while low risk women are most satisfied if they obtain a physical checkup and mammography services along with risk information and standard surveillance recommendations (Holloway et al., 2004).

Other researchers conclude that finding high-risk cases and increasing numbers of referrals to genetic services (Holloway et al., 2004; Washburn et al., 2005), decision making to undergo testing among high risk patients or increase in screening practices (Metcalf et al., 2000) are all indicators of success. Few researchers addressed the cost of consultations.

Despite a voluminous body of literature on patient outcomes, it is impossible to compare between and across different

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models of service to assess the effects of different models on patient outcomes because researchers describe very little detail about the features and characteristics of the models from where these patient outcomes were derived.

The purposes of this paper are to present a state of the science synthesis of published literature on the following topics: 1) the features of gold standard educational programs designed to teach delivery of cancer genetics services; 2) the roles and collaboration dynamics of health care professionals who are the experts; 3) differences between traditional and innovative models of delivering cancer genetic services; and 4) current quality assurance measures used to evaluate cancer genetics counseling services. In addition, we de-construct and describe in great-detail three exemplary cases of models of delivering cancer genetic services. The goal is to draw attention to a variety of characteristics of each model as a way to identify seldom recognized quality assurance measures and to raise questions about how these might influence patient outcomes.

## Method

The research literature on delivering genetic services emphasizes standardization of technical and procedural knowledge and basic ethical principles of autonomy and privacy. What tends to be ill-described is the variation in how models of delivering services are designed and implemented in diverse settings. Nor is there adequate thought about quality assurance measures of the model of delivery in different settings or in different patient populations.

In an effort to better understand the constituent elements of how different models of service are structured and function, the authors gained access to three American and one Korean genetic service models to conduct educational observations as a means to understand different structural and operational characteristics of models of genetic services. This project was never intended or designed as a research study. Consequently, the project was determined by the Institutional Review Boards at all four locations, exempt from Institutional Review Board approval.

The models described below are not intended to represent all possible models. This theoretical analysis is intended to raise attention to quality assurance measures for models of service, in addition to evidence-based patient reactions to the standard

process of delivering risk information.

Based on 114 educational field visits in five genetic service programs in the U.S and Korea from March 2004 to October 2011, each model is deconstructed to understand structure, function, and possible quality assurance criteria. The three models are titled as follows: 1) Medical Geneticist Expert, 2) Generalist Expert. 3) Clinical Research Expert. For each of the models, we describe: a) the goal of the service; b) the organizational system in the clinical setting including the structure and function(s) of key health professionals and allied personnel; c) the type of patient/counselee served; d) the process/events that make up the cancer genetic counseling sessions; e) the role of the physician(s); f) the role of the genetic counselor (nurse/genetics counselor); g) how the patient/counselee interacted with each professional; and h) advantages and disadvantages of the model.

## Result

### The Medical Geneticist Expert Model

The major goals of this model of service were to: 1) apply specialized expert knowledge about cancer genetics to conduct an accurate genetic risk assessment for women at highest risk for developing breast cancer; 2) to determine the best course of medical intervention for each patient; 3) to conduct clinical trials research among this vulnerable population. The main referral criterion was a strong family history of breast cancer or a personal diagnosis of early-age onset breast cancer. Referrals came from physicians in the community, an affiliated hospital, or self-referral.

The organizational system included a medical genetic expert-genetic counselors, advanced practice nurses in genetics (APNG), administrative assistants, and referral physicians. The experts were dressed in formal professional attire such as a dark colored suite and a white coat. The goal was to achieve the highest quality evidenced genetic counseling service, outreach education, telemedicine service, and referral outreach system without losing the art of clinical practice. There were departmental administrative assistants who sent mail correspondence to referred patients/ counselees including a form cover letter and a family cancer history survey tool that was to be completed and returned prior to the face to face counseling session. That information was input by one of the

administrative assistants into a computer data system and printed as a medical genetics pedigree by the time the patient arrived for an appointment. Additional tasks included scheduling patient visits, obtaining insurance authorizations where possible, and inputting any additional patient information into the computer database after each patient visit. Other tasks include transcribing audio-recorded clinical reports from the physician and genetics counselors to create follow up letters which were edited by the assigned genetics counselor/APNG. In this model, all reports and documents were created using templates that were computerized for easy printing, storage and retrieval for research and clinical data.

The counseling process and practice protocols were standardized and every role was delineated. The physician designed the organizational system in such a way to divide the workload and delegate parts of the service to different employees. Like a modern factory, the flow of work was divided, delegated and systematized to run smoothly. The work was segmented into tasks that were automated so that they could be done repetitively, quickly and efficiently. This model required a great deal of funding to build capacity for an acute care high risk service, clinical research, interprofessional education, and an outreach clinical service network including satellite clinics and telemedicine services.

The patients for this genetic service came from a well-established, cooperative referral system from local and distant affiliated hospitals across the state. As a federally funded project over a period of years, the physician established and sustains the referral network system. Weekly, the oncologist and the other genetic experts conducted clinical outreach service using an audiovisual computer assisted telecommunication system to discuss cases of patients in affiliate rural and small urban medical clinics or hospitals. The purpose was to provide education and support for other professionals who are learning to deliver these specialized services and to ensure appropriate recommendations for patients and at risk families.

The average number of counseling sessions for each patient was two face-to-face visits. Usually, the process consisted of a pre-test session, where only the patient and relative and the genetic counselor/APNG attended. The post-test session was attended by both the counselor/APNG and the oncologist together. The duration of each counseling session was over 2 hours.

In the counselor role, there was an exchange of information with the patient in the following ways. The counselor used three tools, one was a standardized questionnaire to obtain personalized patient and family medical history information input into a computer that created a patient database and produced the medical genetics pedigree. The counselor used a standard commercial teaching tool (a flip chart style spiral bound book) that helped her explain cancer genetic information to the patient. The book was used to show and explain the scientific story about genetics and cancer causation. Every patient heard the same story. The counselor's experience in clinical cancer genetics included familiarity with common questions about cancer treatment that were handled with ease, such as hormone replacement therapy. The third tool was a variety of risk calculation formulas using a person's and their family's medical history. Numerical risk figures were reported and described as low, moderate or high and described to the patient in terms of rational for recommendations for genetic testing, treatments including prophylactic options, and future surveillance. The knowledge was presented as if it could apply to any patient.

The genetics counselor and APNG role was virtually the same; they had varying years of experience in providing cancer genetics risk assessments, education, genetic testing information, and surveillance recommendations. Their role emerged from and was defined by the physician's ideal model of providing this medical service. Despite an advanced level of practice with a master's degree, the organizational structure of the delivery model required this role to be a physician assistant position. The genetic counselor/APNG was responsible for establishing a relationship with the patient and family to obtain individualized and contextual knowledge in order to help the physician individualize the medical genetic service. The primary relationship focused on and emphasized the physician patient relationship and the transfer of specialized medical genetics knowledge.

In this medical geneticist model, the medical geneticist had a high level of expertise in genetics. He divided his time between four roles: a clinician, a faculty member, a clinician, and a researcher. This physician was a nationally renowned expert in genetics in both clinical and molecular genetics. His ability to function at an optimal level in so many different roles required the admin assistants, APNGs and genetics counselors to function as assistants to the physician role and

to the medical service. The counselors/APNGs gave up some degree of autonomy and accepted the hierarchical model in exchange for mentorship by the physician and an opportunity to work along side a renowned and well-funded medical oncology service.

The relationship between the APNGs, genetics counselors, and the medical geneticist expert was grounded in respect, mutual goals, friendship, and a shared altruistic value to provide the best possible service to patients, support interprofessional education and conduct clinical research. In exchange for freeing the physician from the time-intensive tasks of the intake interview, documentation of genetic risk, and producing the final consultation letter, the APNGs and counselors gained the responsibility and autonomy of coordinating clinical research projects related to the work of the team. One APNG was encouraged and supported to conduct investigator-initiated nationally funded research projects in this clinical setting.

After being presented with the patient's and family's medical history information and a computer generated three-generation medical genetics pedigree, it was possible for this oncologist expert to provide the entire genetic consultation without input from any other expert. The physician was responsible for completing a physical assessment on each patient and providing an individualized interpretation of the patient's hereditary cancer risk, identifying the recommend genetic test, providing an interpretation of the genetic test results, and explaining the medical, surgical, and surveillance recommendations depending on each woman's unique situation. He was capable of providing all technical assessments, personal, emotional, and ethical information required for a session, as well as individualizing it for each patient. He subjugated some dimensions of his expertise and his expert knowledge in front of the patient and behind the scenes to the genetics counselor role. Doing this, enabled the medical geneticist to delegate functions of his role such as the initial patient intake interview, creating and verifying the three-generation medical genetics pedigree, obtaining the medical history information from various sources for each patient, and authoring the final consultation letter sent to a referring physician and to the counselee.

This model of service appeared expensive because the genetic counselor/APNG and the oncologist provided an overlapping and in some areas a duplicative service.

### The Medical Geneticist Expert Model

This model was targeted toward women who are at low risk for developing breast cancer but they were referred from a physician in the community or the hospital where the service was provided. He met women if patients wanted even though who were not strongly necessary for genetic tests. So in this model, the major goal was to give regular breast health surveillance for low risk women and distinguish genetic risk factors from environmental risk factors.

In this model, counselor was a Certified Genetic Counselor (CGC) . He gave his services independently but directed by the physician's distanced oversight. Physician genetic expert was a medical geneticist. Physically physician did not attend counseling session. Only counselor met the patient. Relationship between counselor and physician was not close in terms of counseling service; he kept distances from the physician's control. This let him be autonomous when he counseled the patient. He did not have special room to provide genetic counseling. Prior to every meeting with patient, he had to looking for and made reservation for available vacant room. He did not wear formal clothing, always he wore casual shirts and cotton pants, brought back pack in which he carried and presented all the documents needed to provide the genetic counseling session. Counselor had not the adequate room for counseling. 'No show up' or appointment cancellation were relatively often occurred. One of reason was that he did not have a secretary, systematic contact system neither a designated special counseling room nor office. He relied mainly on e-mail or telephone for appointment arrangement.

The patients for this genetic service came from self referred or cooperative referral system from local and distant affiliated hospitals where service was provided across the state and the nation. Some women self motivated to take genetic test because of fear of the breast cancer risk due to strong family history. Sometimes this motivation was influenced by the mass media.

This model consisted of only one face to face counseling session. The counselor gave test results over the telephone even for positive tests. The reason was because the fee for service contract paid for only one visit. Only when patients were upset after receiving positive test results did the counselor agree to have them return for a second face to face counseling session. The other two models consisted of two

face to face sessions per patient.

In this model, counselor did not spend more than 2 hours for each counseling session. He/she did not give well documented forms of information or data collection tools. Using pen and paper he applied technical information. In this model, there was likely not to provide well structured guidelines, documenting or recording system. They did not put in computerized data system. They drew pedigree with hand without Pedigree software. They did not send out counseling report to the referral doctors. This model supported multidisciplinary symposium and discussion group.

The information counselor gave was unlikely to be strong technical knowledge. One way of becoming indispensable was to be very popular with physicians. He/she had a great deal of knowledge about general genetics. He/she was not expert in cancer genetics. His/her expertise was in genetic diseases in. He/she did brief physical assessment by visual to document dysmorphology. He/she had no expertise in physical assessment but still included this in his assessments. He/she asked questions about to discern dysmorphology.

He/she had to assess whether the patient needed to see another genetic expert. The counseling session was focused on whether or not the patient will take a test. Following a protocol in that he/she relied on the medical pedigree to determine his/her action. Technically he/she was not strong. He did not provide written literature to the patient. His/her role description was made up based on his/her clinical experience. Even though he/she has low level of knowledge, he/she relies on the resources such as website and journal articles. He/she used an evidence base practice framework. He/she continually tried to expand his knowledge by himself using a variety of resources he/she found on his/her own. He/she already had confidence from clinicians in his community. He/she consulted with physicians about their patients. He/she had a strong personal relationship with these physicians and tried to help solving patient situations. He/she attended many different types interdisciplinary knowledge. He/she worked part time in an oncology department and the other 50 % of the time he/she provided genetic services. In the U.S, NCI (National Cancer Institute) designation required another genetic counselor so with a PhD. His/her salary was paid both from the cancer center and from the radiology department.

The physician was responsible for the predescription of the genetic test, also he/she administratively took the representative

of the department. He/she mainly worked at the laboratory research and epidemiology, and did not show up at the clinic. Shadowing administratively the physician had the role. He/she advised genetic counselor at the genetic cancer conference.

In this model counselor's role was like a patient management role. The role of the counselor was not well defined nor does the role highly specialized or expertise but he/she kept high level of interdisciplinary responsibilities and had good relationship and interacts with other expertise from other disciplines in the community. He/She could easily get support from them and discuss with them autonomously. He/She did not do physical examination because they were not permitted to do that. His/Her service tended to be superficial and was not likely to give profound pathophysiologic information related to breast cancer or surgery nor emotional support effectively for example rarely discussed about the postmenopausal sexual life or symptom management.

Besides genetic counseling service, he/she attended the weekly cancer cases meeting where all doctors and psychologically experts talk about patients. The main purpose of this meeting was to facilitate research activities. He/she was able to initiate his/her own research projects and sometimes served as the coordinators for the physicians. The physician's research was based in laboratory research and epidemiology so he/she looked at tissue. He/she did not need clinical patients to do his/her research.

He/she constructed a rough drawing of the medical genetics pedigree or a risk score to the patient. He/she interpreted the family history as drawn into the pedigree, he/she did not provide a risk calculation. He/she did not have a formal consistent written method of collecting or documenting the family history or calculating a BRCA.

The patient saw less strong representative tools of the science; most of time they hard his interpretations and recommendations. The patients did not use conclusive statistical or probabilistic knowledge to evidence their risk. Counselor used no technical tools he/she used only white paper and pen. He/she was able to describe his interpretive, intuitive knowledge and relate that to the patients' story of the family history. He/she used his experience, and interpretive knowledge. What to recommend to the patient might depend on many factors. His/her collection and representation of the data did not go deep enough to show a holistic view of the patient. The patient could sense there was no depth of scientific

knowledge.

Advantage of this model was that counselor can get maximum autonomy independent from the physician's control. He/She had high level of accountability about their practice. He/she affected the patient's making decision about whether or not take genetic tests. Sometimes he/she could recommend not to take genetic tests even though the referral physician believed the patients have the high risk for hereditary disease. Most of the title of the counselor is Certified Genetic Counselor (CGC).

The relationship between the counselor and physician was friendly cooperated, they respected each other. But patient information exchange between them was not practically done at the scene. Each counseling session, physician did not consistently affect counselor's decision making.

Quality assurance was measured by the patient implicit knowledge rather than explicit knowledge. The patient did not show their recognition of the quality of the genetics counseling process. The best quality service was when the counselor drew a detailed pedigree and showed this to the patient to explain risk and also using risk calculation to the patient to explain the rational for why testing might be necessary or not. So despite the fact that scientific assessment could not resolve patients' uncertainty. How the counseling service was based on a sound scientific knowledge, and how its recommendation or guideline was considered or showed up scientific, would affect the satisfaction of the patient.

### Clinical Research Expert Mode

The goal of these genetic counselling services which was the first program in the country was for clinical genetic epidemiology research, which required collecting blood samples from the client. Patients who were under the service of the physician or who were recommended by any physician in the hospital were advised to undergo genetic testing during their hospitalization.

Under the umbrella of government funding for clinical research, the hospital-based physician free genetic testing for eligible patients and any at risk relatives which enabled start up of the genetic counseling. Fiscal constraints shaped the organizational structure and the physical space in which the program was confined. Genetic risk counseling services were offered in the context of a very busy clinic. The physician's

role was primarily as a chief administrator of the clinical service and the Principal Investigator of a clinical research project aimed at gathering blood specimens and documenting mutation status.

This model of service fit into a busy clinical setting. There were no specifically designated private counseling rooms, there were no teaching tools or computers for storing family history data or programs to calculate risks of getting genetic disease or inheriting a deleterious gene mutation(s). There were no administrative assistants and the Clinical Nurse Specialist (CNS) had no specific knowledge or training in genetic counseling except mentoring by the physician and self-taught learning by reading the literature. However, the CNS holds an advanced degree in nursing and is recognized as an expert clinician in her designated area. Across time, the nurse becomes a self taught expert in genetic counseling services.

The role of the physician was as a clinical expert in disease diagnosis, treatment and follow up care for patients. As a clinician, the physician must concentrate his knowledge and skill to become an expert in certain disease area like breast cancer surgery to gain national reputation and maximize the volume of patients to guarantee their income and prestige in the health care system and the community. The cultural norm was for patient to seek the most expert and renown physician to oversee their care. The physician conducted physical exams as part of his clinical services and he/she checked-ups routinely. The physician breast who was a frontier in new technologies and treatments obtains recognition and greater notoriety and this is motivation for initiating genetic services despite his lack of expertise in genetics.

Administratively, and legally he/she was responsible for ordering the genetic tests. The physician's expertise and knowledge of genetics was limited. He/she must rely on the nurse specialist to provide genetic information and on the laboratory experts who report and interpret the genetic test results.

In this model, the physician must hire clinical nurse specialists (CNSs) to work under his direction and supervision in a hierarchical position where they serve in a variety of roles. The position was a joint combination of clinical care of patients with certain disease during hospitalization for diagnosis, pre- and post-operative management, and post surgery follow up care (including dressing changes and symptom management) and provider of genetic information.

The physician has to depend on the nurse specialist to assist with genetic services when a patient who fits the inclusion criteria is identified as eligible to be in the study and to take the genetic test.

As an assistant to the physician and to the clinical service, the nurse conducted patient assessments, clinical breast exams, prescribed medications and tests according to protocol guidelines. One function was to give direct nursing care to patients. She provided clinical services such as dressing changes, management of symptoms and breast prosthesis. She conducted telephone counseling for patients at home following discharge from the hospital. She organized self-help groups for patients diagnosed with same disease. As a clinical service coordinator, she provided education to students, and other health care professionals, as well as organizing any educational conferences sponsored by the clinical service. As a researcher, she collected data for clinical trials conducted in the clinic.

In terms of genetic services the nurse identified eligible patients by conducting a brief intake interview about personal and family history of the certain disease by reviewing patient charts and by referrals from other physicians. When a patient was admitted for the clinic, the CNS took a family history briefly in order to identify the degree of risk for the genetic testing. She briefly introduced basic information about inherited disease and the idea of genetic testing. The pre-testing session took about one hour and was conducted at a table behind the nursing station of a busy clinical unit or any place available in the ward. One genetic counseling session was held before the patients' blood was drawn for the genetic test. When the patient wanted her husband or her family member(s) they sat by her during the counseling session. After obtaining informed consent, the CNS drew blood for genetic testing. She has no tools, she does no calculation of risk for the disease and mutation.

The test results became available about two months later. If the genetic test result was negative, the CNS contacted the patient by telephone to give this result and to listen to the patient's response. No additional counseling was offered. In the case of a positive test result, the physician told the patients their test results over the telephone or face to face during a brief patient visit. The physician advised the patient to adhere to a vigilant surveillance program consistent with known recommendations and to inform other family members about their risk and the possibility of taking a genetic test. The CNS

reinforced that relatives should be told of the importance of attending an vigilant surveillance program.

Most patients sought well-recognized experts for their service, even though they have common sense knowledge, they wanted to make sure their understanding was confirmed by a famous or renowned expert physician. In addition, they sought reassurance about their fear of the genetic disease. They wanted to legitimate their beliefs or understandings. However, when the patients left the clinic after hearing the test results, their facial expressions remained puzzled and confused. They expressed feeling concern, fear, and uncertainty about their future.

After a brief conversation with the physician, the patient wanted to talk more about their fear of occurrence and they wanted more reassurance from the physician. However, the physician did not have enough time because many other patients were waiting to see him. The surgeon sees lots of other patients at the outpatient department; For example, Korean breast surgeons were interested in cancer genetic testing in the clinic as a research tool but also a clinical tool to improve patient outcomes. The physicians had no time to give attention to patients' emotional distress during genetic testing or to guide them in communication about familial risk because they were busy in their clinical practices. The physician managed most of the follow up care long after completing the immediate post-operative care. Most patients trusted in a breast surgeon's direction and their recommendation. Consequently, they had a powerful position and had a huge impact on a patient's decisions about breast cancer course of treatment and follow up care.

Advantage of this model was economic in terms of cost effectiveness. The flow of the counselling process went forward automatically. As a consequence of the research laboratory, the cost of the genetic test was approximately 20% of the cost of the U.S.. Patient did not pay for the genetic counselling. Also In Korea, the physician's reputation about the breast cancer surgery attracted breast cancer patients and made them obey his order almost without questions. Autonomy of the counsellor was maximized due to physician's overloading clinical patient services. Overall decision-making for the patient and actual counselling processes were affected by the CNS. Disadvantage in this model was that the counselling team is lack in expertise about cancer genetics. They did not provide evidence based counselling guidelines and well established

educational materials. Neither physician nor counsellor had ever got any training related to genetic counselling.

Genetic information profoundly affected only a single patient but more importantly the whole family. This had great significance in Korean culture. Even though busy physicians (medical genetics, oncology specialists or surgeons) needed to collaborate with other health care professionals; such as nurses and genetic counsellors to deliver comprehensive cancer genetic services. The patient viewed the physician as omnipotent; thus they had a need to speak with the physician to hear expert medical genetics knowledge as applied to their situation and to confirm the legitimacy of any recommendations. This phenomenon was cross-culturally similar in that patients had greater respect and desire for physician's involvement in health care services. In the final analysis the ideal model must integrate the services of nurses/genetic counsellors in close collaboration with physicians who were able to provide the final discussion with patients and family. This model diluted the autonomy of the nurse's role however; in the best interest of patients and families this was the best model for Koreans.

## Discussion

Standardization of training for genetic counselors nationally and internationally is decidedly necessary to maintain quality assurance. The American Society of Clinical Oncology (ASCO) produced a resource document for curriculum development in cancer genetics education for ASCO members and other health care professionals designed for any professional who has a role in cancer predisposition analysis and testing in clinical oncology. The process and procedural knowledge labeled as the "standard protocol" (Kash & Lerman, 1998), or "structure of the genetic counseling session" (Butow & Lobb, 2004), or "algorithm for genetic risk counseling" (Washburn et al., 2005) have been well described and used nationally and internationally (Piniewski-Bond et al., 2003)

Policy makers, hospital administrators, and health care professionals in community and acute care settings need a better understanding of existing models as well as research evidence in order to predict which model(s) might be most cost effective, efficient and beneficial to people in any given setting, or country. Do we know for certain how new models of service should be structured? Do we need to create new

models of service? Can certain health professionals be just as efficient and cost effective without compromising patient outcomes? Should patients be stratified into one model versus another by their degree of risk: average, moderate, or high? Addressing these and other questions is not only economically prudent for administrators; it is socially and ethically responsible to patients and families who seek this service and to the limited numbers of health professionals who are available to provide these resources.

The purpose of delivering these services is to provide knowledge and support to those who might be at-risk which helps patients and at-risk relatives make informed choices about diagnosis, treatments, lifestyles, and to deal with social, legal and ethical dilemmas arising from genetic risk information. The question should be 'what is the best model for patients and families' not 'expert professionals maintaining strict adherence to standard protocol and procedures'. The best model of providing genetic services should be based on patient-centered outcomes not merely, professional practice and process. Analysis of these three exemplary models of delivering genetic services points to concepts that might be considered as part of a larger view of patient-centered outcomes.

Generally speaking, patients wanted to obtain information that would rule out their susceptibility to genetic disease, or identify a certain cause for why the genetic disease occurred. This expectation is impossible to answer. Their disappointment was unavoidable because interpretation of the medical genetics pedigree and the genetic testing is not yet 100% certain. Despite the hoped for certainty in the information provide by the experts they could understand a mutual sense of powerlessness on both sides of the genetic knowledge because it is by nature uncertain. Due to irresolvable uncertainty, their feelings of fear may not be resolvable immediately.

All three models result in the same residual feeling of uncertainty. The difference between the best quality services in terms on uncertainty is when the physician is able to explain why the information is uncertain then the synergy effect when both professionals explain about the uncertainty. But perhaps, an increase in uncertainty is a desirable patient outcome because it is a true component of genetic information and by it's very nature, patients are keenly aware that susceptibility to some degree remains, and should not be ignored.

In recognition of the benefits of a team approach for genetic



counseling services(MacDonald, Blazer, & Weitzel, 2010); clinically we can consider three major team models; multidisciplinary, interdisciplinary, and transdisciplinary. Few authors recognize the benefits of transdisciplinary teams especially in genetic counseling services over any other team approach (Anderson, Monsen, & Rorty, 2000; Greco & Anderson, 2002).

Providing genetic services is a kind of an artful balance between knowing the scientific knowledge but it needs to be interpreted in the context but also the artful skill and performance of knowing and interpreting the meaning information might have for patients and their families.

Sensitive communication and attending non verbal behavior are important element in the genetic counseling (MacDonald, Sarna, Uman, Grant, & Weitzel, 2006). This requires space or silence for looking and not speaking so that nonverbal cues can be seen and interpreted. When the technical expert focuses on the assessment tool and the counselor's attention is focused on the document then the focus is on the science. She did not sensibly interview the patient over the whole counseling procedure. The second model is best because the GC focuses on the documents but also interviews and listens to and looks at the patient from the beginning to end of the counseling. Also she can contextualize the patient.

In the second model(generalist expert model), the focus is on the patient and interviews the patient but the manner is not in depth and aimed at only specific types of data. The patient interview occurred over the telephone and that helped to create the pedigree. The content of the pedigree was superficial in comparison to the first model.

Based on the description of these three models the task is to find the best way to contextualize the data and the patient's story. This leads to the best interpretation of presenting what the patient faces in terms of the facts their situation and what decision making have to be made.

To reduce uncertainty it is impossible at this time in genetic science. So how do we show that we understand the patient situation, this is the dilemma to satisfy their needs and reduce anxiety due to uncertainty. This is impossible to do this by the facts alone, we have to understand how to do this using science and the patient's narrative understanding. This will guarantee quality assurance. We can satisfy the patient's needs scientifically but we must be able to demonstrate that we understand the patient's situation beyond merely their emotional

response.

In the medical geneticist expert model, if the egalitarian co-supportive relationship between the physician and the APN is established, this model can produce the best quality because it contextualize the patient situation and can create the best image of understanding the patient because communication is not information giving but the focus is on sharing and empathizing with the patients dilemma and desire to know. The structure of the egalitarian relationship between the physician and the APN can produce the best method because it is a transdisciplinary sharing of expertise and power is not monopolized but rather shifted back and forth to each team member as that expertise is needed.

A great deal of research about genetic services focuses on the process and content of risk communication and delivering genetic information with the intent to evidence patients' satisfaction with service, its cost-effectiveness (Holloway et al., 2004; Gustafson, et. al., 2011) or effects of risk communication by measuring relief of anxiety (Claes, et al., 2003; Wilson, et al., 2006; Phelps, Bennet, Iredale, Anstey, & Gray, 2006), level of emotional distress (Bowen, Burke, McTiernan, Yasui, & Anderson, 2004; Rimes, Salkovskis, Jones, & Lucassen, 2006) or depression (Wilson, et al., 2006).

The strongest criteria for quality assurance which are transdisciplinary teams, is the strong communication skills that focus on the patient and not on documents and also attentive to and reason nonverbal communication(Greco & Anderson, 2002). Also ability to retrace a systematic and comprehensive process of documentation of any many factors as relevant for a given situation and lack of subtle signs of power imbalance among all professionals and patients, patient centered process are important.

In Korea and other countries, the dominant method of delivering genetic counseling service is a physician-centered model. However, the authors recommend that the best model is a collaborative approach where physicians work closely with nurses who have expertise in cancer genetics. This suggestion may pose a conflict of interest for nurses' who desire increased autonomy based on expert knowledge and skills gained from advanced education and practice as nurse specialists or nurse practitioners. This conflict is well represented in all three models of delivering genetic services in the US. To maximize patient and family satisfaction and to create harmony between patients and the physician, it is

necessary to develop collaborative practice that emphasizes the complementary service of both physicians and expert nurses trained in cancer genetics (Feetham & Deatrck, 2002; Anderson, Jun, & Choi, 2007).

## Conclusion

The goal should be to design patient care services with a view to provide the best model of care for the patient and not merely focus on models that are ideal for clinicians or genetic researchers. Quality assurance means that services are evaluated for their effect on patients and families. This paper fills a gap in the literature because criteria for determining which model(s) might be most effective in certain patient populations, or clinical contexts has not been addressed in the literature. The authors identify new concepts that reflect a patient-centered perspective of quality assurance.

Researchers have measured patient outcomes before, during, and after genetic services to demonstrate cost effectiveness, patient satisfaction, knowledge gained, effect on treatment and genetic testing decisions, psychological effect of risk communication and other variables. However, without a clear understanding of the organizational structure of current service models, it is impossible to know which ones might be better than others, or whether certain models best serve specific populations. The authors invite discussion and debate to fill a gap in the literature about the relationship between models of service and patient outcomes.

Consequently, in Korea it is necessary to establish research evidence to demonstrate improved patient and family outcomes due to nurse's involvement in delivering cancer genetic risk information and testing. This is necessary to establish the educational program for nursing genetics and demonstrate the competence of nurses in providing these services and the most feature of transdisciplinary team model, counselor as a scientific expert as well as personhood expert who takes the time to be with, listen to and respond sensitively and compassionately as patients and family members try to understand and make the best possible decisions given the current state of science and medical practice. Also this model reflects careful cultural sensitivity. It must consider family oriented culture not individualism.

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## 유전상담 서비스 모델 분석: 이론적 탐색\*

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유전 위험 사정과 상담서비스가 임상실무에 널리 적용되어 감에 따라, 다양한 비용효율 면에서 다양한 상담서비스 모델을 사정하고, 대상자의 임상 요구와 건강문제를 해결하는데 어떤 모델이 유용한 지 확인할 필요가 있다. 본 연구의 목적은 114건의 현장 관찰과 문헌고찰을 통하여 3가지 유전상담 모형을 분석하였다. 유전의학 전문가 모델, 유전상담사 모델, 임상연구전문가 모델을 중심으로 각 모델의 구조, 전문가의 역할 및 기능, 목표, 물리적 세팅, 교육도구 등을 분석하였다. 각 모형 안에서 환자에게 기대되는 결과 면에서 질적 서비스가 보장되는지 확인하기 위하여 이론적 분석을 실시하였다. 본 연구를 통하여 각 모형의 상담 전, 중, 후 환자 만족, 지식 변화, 상담 효과 및 커뮤니케이션 효과 등을 분석하였지만, 결론적으로 상담서비스가 이루어지고 있는 기관의 구조를 충분히 고려하지 않은 상태에서 최상의 서비스 모델을 제시하기 어려울 것임을 논의하였다.

**주요어** : Genetics, Genetic counseling, Health care quality assurance

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