

The Genetic Self: The Human Genome Project, Genetic Counseling, and Family Therapy

JUNE A. PETERS, MS
LUBA DJURDJINOVIC, MS
DIANE BAKER, MS

In the ideal world, genetic counseling, family therapy, and primary healthcare should blend into a seamless network of psychosocial services for families with genetic conditions. The discussions presented here were inspired by two interdisciplinary workshops titled "The Genetic Self". This paper introduces family therapists and primary care practitioners to the Human Genome Project and current applications in genetic counseling. The practical goal is to foster interdisciplinary teams and referral networks for management of families with or at risk for genetic disorders. Families with genetic conditions may need access to genetic diagnosis, possible genetic testing, tailored medical management, crisis interventions, follow-up at appropriate developmental stages, family therapy, individual psychotherapy, or pastoral counseling for dealing with spiritual issues elicited by

genetic conditions. We also hope to stimulate collaborative research on the impact of genetic conditions in families, to form advocacy partnerships on behalf of these families, and ultimately, to influence public policy.

Fam Syst & Health 17:5-25, 1999

Genetic information influences how we see ourselves, and how others label us, and it carries the potential to shift connections within families (Durfy & Peters, 1993). Our responses are in part molded by beliefs, attitudes, and experiences in our own lives as well as in those of our families and cultures (Kenen, 1980). It has been noted that "in the narrative of every human life and family, illness is a prominent character" (McDaniel, et al., 1997). We predict that the dimension of genetic risk will become integral to the illness narratives of the future (Kenen & Smith, 1995).

Traditionally, genetic counseling has been offered at the time of prenatal diagnosis of a fetus, or newborn diagnosis of a baby, with a genetic condition or birth defect. Questions such as "What happened to my baby?....How did this happen?..... Are my other children or future pregnancies at risk?" can create a "genetic moment" in which the provider makes a genetic

June A. Peters, MS, Assistant Professor of Human Genetics, University of Pittsburgh Graduate School of Public Health, and Genetic Counselor, University of Pittsburgh Medical Center and Magee Women's Hospital, Cancer Genetics Program, Pittsburgh, PA

Luba Djurdjinovic, MS, Director, Genetic Counseling Program, Binghamton, NY; Director, Ferre Institute, Utica, NY; Lecturer, Syracuse University, School of Social Work

Diane Baker, MS, Director, Graduate Program in Genetic Counseling; Lecturer, Department of Human Genetics, University of Michigan Department of Human Genetics, Ann Arbor, MI

counseling referral for discussions of diagnosis, prognosis, and risk of recurrence in ways that facilitate informed decision-making and bolster family coping. Further, the provider could also recognize it as a "psychological moment" and refer to a family therapist to address the anxieties and fears of the family in more psychotherapeutic ways. He or she might also recognize this as an "existential moment" and suggest pastoral counseling. This paper addresses the challenges posed by genetic information and promotes an interdisciplinary approach to meet these challenges.

THE HUMAN GENOME PROJECT

It is important to understand the origins of the Genetic Self in the context of the Human Genome Project (HGP). The human genome refers to all the genetic information that resides in human cells. Begun in 1990 by the U.S. Congress, the HGP is jointly supported by the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH) and the Department of Energy (DOE). The 15 year goal of the U.S. Human Genome Project and its worldwide counterpart, The Human Genome Organization (HUGO), is to develop methods, train scientists, and establish infrastructure to identify and determine the DNA sequence of all 80,000-100,000 genes contained in human cells. (Peters & Hadley, 1997). The HGP has coalesced and focused molecular genetic research, which was already in progress prior to this coordinated effort on specific scientific international goals.

For those interested in additional information about scientific aspects of human genetics, refer to a recent genetics primer by Middelton et al., (1997) or to resources listed in Table 1.

Because it has long been recognized that acquiring and using genetic knowledge has serious implications for individuals and

society, a portion of the annual HGP budget has always been devoted to consideration of the ethical, legal, and social implications (ELSI) of these scientific discoveries. Funded ELSI projects include examining the impact of integrating genetic technologies into healthcare practice, establishing an understanding of health professionals' knowledge of human genetics, educating professionals and the public, and developing recommendations about how best to incorporate new technologies and discoveries into practice. Efforts have already begun to form national consortia of hundreds of professional organizations to improve genetic literacy of their members (Collins, 1997).

ORIGINS OF THE GENETIC SELF

In 1995 a group of genetic and mental health professionals met with officials from the National Human Genome Center, predecessor of the National Human Genome Research Institute (NHGRI). Their challenge was to create an opportunity for mental health providers to consider the emerging issues in applications of genetic information and to integrate this knowledge base into clinical practice, research, scholarship, and training of colleagues. The title "Genetic Self" was proposed to bridge the separate bodies of concepts and literature that exist in the genetic, psychological, and pastoral communities. Two meetings with a total of 50 participants were held in 1996 and 1997. Evaluations of projects emerging from these meetings, such as this issue of *Families, Systems, & Health*, are in progress.

TABLE 1. Genetic Resources and Internet Websites

ORGANIZATION	ABBREVIATION	WEBSITE http://	ADDRESS
Alliance of Genetic Support Groups	The Alliance	www.geneticalliance.org	Washington, DC
American Society of Human Genetics	ASHG	www.faseb.org/genetics/ashg/ashgmenu.htm	Rockville, MD
American College of Medical Genetics	ACMG	www.faseb.org/genetics/acmg/acmgmenu.htm	Rockville, MD
American Board of Genetic Counseling	ABGC	www.faseb.org/genetics/abgc/abgcmenu.htm	Rockville, MD
American Board of Medical Genetics	ABMG	www.faseb.org/genetics/abmg/abmgmenu.htm	Rockville, MD
Association of Professors of Human or Medical Genetics	APHG	www.faseb.org/genetics/aphmg/aphmg1.htm	Rockville, MD
Centers for Disease Control, Office of Genetics and Disease Prevention	CDC Genetics	www.cdc.gov/genetics	Atlanta, GA
Department of Energy, Ethics, Legal, Social Issues	DOE-ELSI	www.ornl.gov/techresources/human-genome/resource/elsi.html	Washington, DC
National Action Plan on Breast Cancer Genetics Curriculum	NAPBC	www.napbc.org/napbc/hsedcurr.htm	Washington, DC
National Institutes of Health Ethical, Legal, Social Issues	NIH-ELSI	www.nhgri.nih.gov/ELSI	Bethesda, MD
National Cancer Institute CancerNet	NCI	cancernet.nci.nih.gov/	Bethesda, MD
NCI Cancer Genetics Anatomy Project	CGAP	www.ncbi.nlm.nih.gov/cgap/	Bethesda, MD
National Human Genome Research Institute	NHGRI	www.nhgri.nih.gov	Bethesda, MD
National Organization of Rare Disorders	NORD	www.rarediseases.org	Bethesda, MD
National Society of Genetic Counselors	NSGC	www.nsge.org	Wallingford, PA
OncoLink information Univ.Penn.	ONCOLINK	//oncolink.upenn.edu/causeprevent/genetics	Philadelphia, PA
Online Mendelian Inheritance in Man, catalog of genetic disorders	OMIM	www3.ncbi.nlm.nih.gov/omim/searchomim.html	Bethesda, MD
University of Kansas Genetic Education Center	KUMC	www.kumc.edu/gec/	Kansas City, KA

THE BIOPSYCHOSOCIAL CONCEPT OF THE GENETIC SELF

Biological Concept of Genetic Self

Genetic information can be an important factor in defining self, family, and community. Some argue that testing for genetic susceptibility is unlike other medical tests (Durfy & Peters, 1993). DNA is most basic to the concept of who we are since our genetic makeup is unlike that of any other person except an identical twin (see Green & Thomas, 1997 for a discussion of the ethical issues raised by identical twins who disagree about genetic testing).

Unique to genetic tests is their ability to generate information about relatives as well as about the individual being tested. This affects ideas about kinship (Richards, 1997). For example, the parents of a child diagnosed with cystic fibrosis may learn that they are mutation carriers. A woman whose father died of colon cancer may learn that she is predisposed, not only to colon, but also to endometrial and ovarian cancers. Healthy carriers of genetic alterations which predispose them to develop certain diseases in the future have created a new class of "at risk" individuals (Wexler, 1979; Kenen, 1996). The "at risk" individual is certainly not ill at present, but may not remain well as long as the "average" person.

The Psychological Concept of the Genetic Self

Psychodynamic theories of human development argue that "the self" emerges through complex biological and/or psychological processes involving cognitive maturation, inborn temperament, and experiential events in the context of meaningful relationships. In the relational psychologies, the quality of relationships

with primary caretakers lays the framework for the capacity to be in relation to others as well as the molding of the self (Kohut, 1972; Miller & Stiver, 1997). If early relationships are satisfying, the self becomes strong, cohesive, and interactive. If not, the self is immature, fragile, or damaged and cannot easily relate to others. These views overlap with family therapy in agreement about the importance of family and other close relationships.

Human responses to certain genetic conditions can be either disabling or empowering. When the focus is placed on the disfiguring or disabling aspects of the condition, it can potentially threaten the self-concept of the parent and/or child. On the other hand, successful coping with adversity can provide a source of strength and growth, when the parents help the child incorporate the sequelae of having a genetic condition into a cohesive self-concept (Royal, et al., 1995).

Family Therapy Concept of the Genetic Self

Family therapy tends to downplay the individual self to emphasize interaction and context. One might say that there are "genetic systems" in addition to "a genetic self". Patterns of interaction within a family and between a family and other systems determine individual behavior. In the case of a genetic condition, the primary focus is the system created by the interaction of that genetic condition with an individual, marital, family, healthcare, and other systems (Rolland, 1994). Eunpu demonstrates how assessment and treatment planning in families with genetic disorders ideally can include the individual's psychological framework, the couple's interactions, and the intergenerational influences (Eunpu, 1997) as well as all of the customary medical and genetic issues.

The Sociological/Cultural Concept of a Genetic Self

Genetic advances hold the promise of more accurate genetic diagnoses and new therapies. However, these same advances also raise a number of ethical, social, and legal concerns regarding eugenic misuses, possible discrimination, stigmatization, and/or disruption of the social fabric of families and societies (Durant, Hansen, Bauer, 1997). Genetic news may stigmatize if it affects how we see ourselves and how others label us, an undesirable outcome exacerbated by the human tendencies to reduce complex genetic information into simple binary categories and to label one as better than another.

Medical sociologists and anthropologists monitor the intersections of science, medicine, technology, and popular culture in shaping the cultural meaning of the gene (Nelkin & Lindee, 1995). They argue that the precise scientific legitimacy of any image, such as that of the gene, is less important than the cultural use that is made of it in serving social ideologies and institutional agendas. Our view is that practitioners assessing and treating families with genetic conditions need basic genetic knowledge while also being cognizant of the cultural meanings and individual subjective meanings of having, or being at risk for, genetic conditions.

The Practitioner's View of the Genetic Self

We know that as practitioners our attitudes and experiences can have an impact on practice (McDaniel, et al., 1997). Have we as practitioners had experiences with birth defects, chronic illnesses, genetic conditions? Do we assume that the genetic self is a defective self? Do we see the ideal genetic self as engineered to enhance certain desirable characteristics and

eliminate undesirable ones? Desirable and undesirable to whom? Do we advocate for fair access and other rights of individuals with disabilities? How are our attitudes made manifest in our own practice settings through the physical layout of the office, accessible TTY machine, knowledgeable staff who are familiar with the disabled, and intake procedures adapted to meet the needs of the individual with a disability?

APPLICATIONS OF GENETIC INFORMATION

Evolution of Genetic Counseling and Medical Genetics

The new post-eugenic era started 50 years ago when geneticist Sheldon Reed coined the term "genetic counseling" (Reed, 1955; Resta, 1997). He stressed the ethos of respect and caring for families that typifies modern genetic counseling practice. Reed appreciated that genetic information often results in changes in perception of risk for one's children and oneself. During the 1940s and 1950s genetics clinics and professional societies were established throughout the world.

Genetic counseling is "a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family..." (Fraser, 1974, p637). In 1975, an Ad Hoc Committee of the American Society of Human Genetics accepted this definition and further defined the basic components of the genetic counseling process as helping families to:

- comprehend the medical facts of the condition, including the diagnosis, probable course of the disorder, and available management;
- appreciate the hereditary contribution

and recurrence risk for the disorder in specific relatives;

- understand their options for dealing with the risk of recurrence in terms of medical care, reproduction, testing, etc.
- choose which of the options, including doing nothing, is currently appropriate for them in view of their risk, disease burden, and family goals and values; and
- make the best possible adjustment to the condition, or to the risk of recurrence of the disorder, in oneself and/or one's loved ones.

More specific psychosocial goals of genetic counseling are to help the family to: feel competent in coping with the risk and impact of the genetic condition; diminish guilt or blame and restore self-esteem; make decisions about testing, treatment, and/or reproduction; and identify and utilize resources for psychological, social, and financial support.

The professional credentials and training of those who currently offer genetic counseling include M.D., Ph.D., or M.S. Degrees, with certification by the American Board of Genetic Counseling and/or the American Board of Medical Genetics. As a distinct profession, genetic counseling has its own code of ethics, accredited training programs, and clinical internships (Fiddler, et al., 1996; Fine, et al., 1996; Benkendorf, et al., 1992; NSGC Code of Ethics, 1992). Specific effort has been made to develop cross-cultural competencies, since genetic diseases know no bounds of race, class, nor ethnicity (Dixon, et al., 1992; Punaless-Morejon & Rapp, 1993; Weil & Mittman, 1993; Smith, Warren & Misra, 1993; Ota Wang, 1994).

GENETIC COUNSELING AND PSYCHOTHERAPY

Psychosocial Aspect of Genetic Counseling

While the educational activities of

genetic counseling may be obvious, the enterprise occurs within a psychosocial milieu (Kessler, 1997b; Marks, 1993). The psychosocial evaluation within genetic counseling can be brief or comprehensive depending on the setting, reason for referral, family needs, and the training and expertise of the genetics team. Assessment looks at the consultand's motivation for seeking genetic evaluation at a given time, the expectations of what would be gained from a genetics consultation, the personal and family experiences, beliefs and attitudes about the genetic condition as well as standardized psychosocial information. Because genetic conditions affect whole families, spouses or family members may be invited to the counseling session. The genetic counselor seeks to understand family health beliefs and attitudes, communication patterns about medical information, family constellation and dynamics, secrecy about disease diagnoses, and effectiveness of coping styles and support systems (Richards, 1996). The role of uncertainty and perceptions of risk in the genetic context have proven to be major counseling issues (Shiloh, 1996; Hallowell, 1997). An individual's subjective experience of genetic risk may be influenced by a variety of other factors including: the natural history and outcome of the disease, one's closeness of relationship to affected individuals, degree of physical caretaking required by affected individuals, psychological identification with affected individuals, and individual as well as family lifecycle developmental stage.

The practice of genetic counseling assists the counselee in appreciating the meaning and challenges of genetic information and its associated risks. Genetic counselors are trained to interact with individuals and families in "a kind of psychotherapeutic encounter" (Kessler, 1979). This approach is crucial since counselees may not be able to hear, understand, or assimilate information if

they are having an emotional response to what is being presented (Frazer, 1976). Therefore, the foundation of genetic counseling practice has been built on principles of "client-centered" psychotherapy as defined by Carl Rogers. The qualities of genuineness, empathic understanding and unconditional regard of the counselee undergird the stance of non-directiveness in medical genetic decision-making (Kessler, 1997a; Fine, 1993; Wilfond & Baker, 1995).

The genetic counselor can make brief psychosocial interventions with families and individuals in distress. Typical examples include crisis intervention and stabilization following genetic diagnosis; grief counseling following reproductive or other losses; facilitating decision-making about genetic testing; encouraging active, constructive coping with genetic information; and promoting effective social support. Additionally, some counselors help families communicate about being at increased risk for genetic disease, deal with barriers to positive healthcare practices, and make adjustments to personal and family changes precipitated by genetic diagnoses.

Similarities and Differences Between Genetic Counseling and Family Psychotherapy

Familiarity with similarities and differences in roles and practices of various health professionals can facilitate successful referrals and cohesive interdisciplinary team functioning among the genetic counselor, family therapist, psychologist, pastoral counselor, and social worker. Taking and making use of the family history can help to illustrate. The family history is the crux of a genetic assessment. This family history is converted to a family pedigree, which is a shorthand, graphic representation of the

family's medical history. With it the genetic counselor notes the number, ages, and genders of affected individuals; tracks patterns of health and illness over several generations on both sides of the family; documents genetic conditions, birth defects, mental retardation and miscarriages; and predicts the possibility of future disease occurrences. Recommendations for standardized human pedigree nomenclature in the U.S. have been published (Bennett, 1995). The processes of eliciting the family illness narratives, contacting relatives for additional health information, collecting medical information, and drawing up the family tree in a genetic clinic may have profound effects on mood, individual self-concepts, and family relationships, independent of whatever genetic interpretations may follow (Richards, 1997, p 258).

Likewise, the family psychotherapist may use the genogram to document family structure, alliances, and communications patterns (McGoldrick & Gerson, 1985). The genogram may include individuals who are socially connected to the family in addition to biological relatives. Rolland (1994, p 82) has adopted the family systems genogram into a family health genogram which resembles the genetic disease pedigree but also includes past experiences with illness, loss, crisis, and adversity. The genogram may also be organized around family planning, infertility, pregnancy loss, or losses specifically due to cancer or other chronic illnesses (Eunpu, 1997). This expanded genogram also adds the element of how each adult's family of origin organized itself as a system to respond to these health challenges and how these patterns changed or evolved over time.

Blending of Medical and Psychosocial Counseling

There are several family therapy models

which recognize the interplay of biological and psychosocial factors in illness (Turk & Kerns, 1985; Seligman & Darling, 1989; McDaniel, Hepworth, & Doherty, 1992; Rolland, 1995; Pollin, 1995). Many of these emphasize the importance of individual and family perceptions, beliefs, and values about health and disease, the variety of ways that families adapt to chronic disease, interaction of individual and family life-span developmental stages, the intersection of family systems and healthcare systems, and anticipate "expectable issues". These models show great promise for intersecting successfully with genetic counseling missions, techniques, and processes. Eunpu (1997) has made concrete suggestions for assessment and treatment in genetic counseling situations based on the Intersystem model of Weeks and Treat (1992).

Genetic Counseling & Psychotherapy Case Vignettes

The following cases illustrate a blending of genetic counseling with psychosocial and family systems models in cases which might come to the attention of the family therapist or health practitioner. These cases are representative in that spouses and family members attend genetic counseling sessions only when they perceive this as absolutely necessary, as in Case 1 where a spouse requested testing. Kenen has shown that when the decision to seek genetic counseling and prenatal diagnosis is viewed primarily as an informational decision, the male partners cede their role to the female partner, in contrast to circumstances where the men become more involved in decisions about taking an action based on genetic information (Kenen, et al., 1997).

Case 1 A couple in their late 20s refer themselves for genetic counseling with a request for cystic fibrosis (CF) carrier

status testing for the husband. Three years earlier, the wife's sister delivered her first child, a daughter, who was diagnosed shortly after birth with CF. The consultands have two young sons and are happy about a current pregnancy in light of their long-standing desire to have a family of four children; however, they are concerned about their risk of having a child with CF.

CF is a multi-system genetic condition that affects the lungs and digestive systems with secondary consequences affecting growth and difficulties with managing infections. Children are affected with this condition when they inherit a mutation in the CFTR gene from both of their parents, who are said to be "CF carriers". Twenty years ago, the life span was severely curtailed, however, medical advances have made survival into the 30s and beyond possible.

The CFTR gene was identified and cloned in 1989 and commercial CF carrier testing is now widely available. Testing is not 100% sensitive, but rather, detects the most common mutations identified to date. If a family has a unique or uncommon mutation, it may not be recognized.

In this case, the wife had already been tested and knew that she carried a CFTR mutation. After discovering her carrier status, she decided it would be helpful if she and her husband sought formal genetic counseling and obtained testing on the husband since their risk for having children with CF depended on whether the husband was also a CFTR mutation carrier.

In the initial session, the genetic counselor began to understand some of the psychosocial and family issues that will need to be addressed for successful genetic counseling. First, the wife is a nurse and works on a pediatric pulmonary ward where she has exposure to children with the most severe complications of CF. As a result she holds a skewed view of CF prognosis as extremely poor. It is also

evident that she is very resourceful in accessing the medical system and in assuming a role of "healthcare advisor" in her immediate and extended families.

The genetic counselor reassured the couple that she would address the issues that they have identified, but first asked them to tell more about their experience with their niece and as parents themselves. The wife revealed that she has always wanted a daughter and that her niece holds a special place in their family. She and her sister live nearby each other and are often together with all the children, an opportunity that they both appreciate. The consultant is often called by her sister for advice about general pediatric and acute respiratory concerns for her daughter. This three-year old niece has already had three major hospitalizations related to respiratory complications and the consultant knows the serious prognostic implications of such hospitalizations.

The wife talked about the patients she works with, including her experiences attending the funerals of some children with CF she has cared for. She described her strong commitment to doing the best that she can for her niece and identified many positive factors about her niece's situation including the early diagnosis of CF which afforded the opportunity for improved care through closer monitoring and more effective treatments.

The consultants appreciate the positive aspects of their strong family bonds. The couple expressed their strong mutual desire for a "large" family. Additionally, they expressed their delight with their two young sons and their hope for a daughter in the future.

The genetic counselor reflected to the couple that she identified two voices (roles) from the wife, one family voice as a mother, wife and aunt; and another professional voice as a nurse. Both are strong, compassionate, and clear voices. However, it seems that during times of confusion or

uncertainty, she preferentially adopts the professional role. They talked about these two perspectives and how the nurse and the mother each view risk, future children, and the possibility of having a child with CF. Although this woman is very nurturing in all of her roles, her family and professional perspectives are sometimes in conflict. The nurse part of herself values medical care, the healthcare delivery system, testing, and medical interventions and recognizes that children die. The mother aspect wants children and wants them to live long, healthy lives. She does not believe in abortion. With her new recognition and articulation of her unique combination of roles, the consultant recognized that she has the capacity to raise a child with CF and would treasure and love such a child.

The genetic counseling process has helped the couple identify and deal with the series of interlocking decisions that they face: carrier testing on the husband, clarification of risks for future pregnancies, considering prenatal diagnosis testing, and implications of pursuing testing, including facing possible pregnancy termination decisions.

During the course of these explorations, the couple came to understand that they feel united in their belief that they would not abort a future CF pregnancy and hopeful in their ability to parent a child with CF in the context of a rich family life. They found comfort in viewing their situation from their perspective as parents. The husband expressed confidence in his wife's judgement despite his own conflicts. Although he also did not believe in abortion, he was concerned about having a child with CF. The couple declined carrier testing of the father and prenatal diagnosis for the current pregnancy. These tests were left as a future option should they change their minds in subsequent pregnancies.

Case 2 This case demonstrates the occasional need for collaboration among

genetic counselor, obstetrician, and mental health professional in the provision of genetic screening and testing. This case was initially reported by Greene-Simonson & Peters, 1992; and Peters, 1994, with subsequent commentary by Djurdjinovic, 1998.

The client (VB) was a 36 year-old married Caucasian woman who had been referred by her obstetrician for genetic counseling regarding prenatal diagnosis due to advanced maternal age and an abnormal maternal serum alpha-fetoprotein screening (MS-AFP) result. The MS-AFP results could indicate a pregnancy at increased risk for having a child with a chromosomal aberration such as Down syndrome. Further evaluation with more definitive diagnostic testing was indicated. After genetic counseling, including a prenatal diagnosis informed consent process, VB chose to have ultrasound and amniocentesis at a nearby genetics center. Shortly thereafter, she became extremely anxious, preoccupied with bodily sensations of fluid leakage from the vagina and worried about possible miscarriage of the pregnancy, as well as about a possible abnormal outcome of the test. VB reported difficulty concentrating at work, and an inability to sleep, eat, or care for her children. Although she was visiting the obstetrician daily to assess her sensations of pressure from the amniotic fluid, there was no evidence of amniotic fluid leakage on repeated evaluations. She had extreme ambivalence about knowing the results of the prenatal diagnosis testing, which customarily take a week or more to return. At this point the genetic counselor referred the case to a local psychotherapist familiar with genetic issues.

The client attended the psychotherapy session alone, indicating that her husband did not consider himself concerned with the prenatal diagnosis nor with the psychotherapy. In this case, her depressed and anxious symptoms appeared not solely

attributable to a testing procedure, but were affected by the client's psychosocial makeup and past history. The threat of a possible genetic abnormality proved additionally stressful to an already fragile self, which became symptomatic in response. In an attempt to appreciate the psychological state of VB, the counselor's exploration of past and present relationships revealed that VB saw herself as isolated, unattractive, and a failure. She also reported that she was withdrawn in childhood, had poor family ties, an unhappy marriage, and that her present social support was minimal. On direct inquiry about suicidal ideation, VB revealed that she was considering killing herself and the fetus if the amniocentesis detected anything seriously wrong.

VB was seen for two sessions during the week between her amniocentesis and obtaining results; however, she refused to see a psychiatrist as recommended. To lessen the risk of suicide, the therapist attempted to relieve some of the pressures that VB was experiencing, increased frequency of professional contact as well as social support, and encouraged her to vent repressed anger and grief in therapy and at home within safe parameters of a non-suicide contract and reliable support system.

Grief counseling figured prominently in this case. An elective abortion which VB had undergone many years ago was never grieved appropriately because of the guilt which she harbored over choosing abortion. She had not shared this history with her husband, physician, or genetic counselor. By grieving the prior loss and developing some insight into parallels between her previous and current relational situations, VB was able to make use of the psychotherapy opportunity to gain better perspective and equilibrium in the current situation. Enhancement of her coping skills and making a plan for increased social support provided acceptable alternatives to suicide.

Several days after the second psychotherapy session, VB called to report

that the prenatal diagnosis results were normal, her mood was improved, and she was functioning adequately at home and work. She did not wish further therapy to address family or other relationship issues; however, the door was open for future contact.

Case 3 A 32 year-old woman requested that her primary care physician provide her with medication for anxiety. Since this patient frequently requested medical appointments about benign symptoms, he encouraged her to see a psychotherapist prior to providing a medication.

After experiencing increasing periods of anxiety, the woman contacted the therapist and revealed that she was planning to relocate for an employment opportunity. During intake, the therapist constructed a genogram which revealed that the client comes from a family that has lost many relatives to breast cancer, including her mother and oldest half-sister when the client was a teenager. This woman has never married but has had a number of relationships, which she ended when intimacy developed.

Counseling sessions revealed the client's increasing fears of developing breast cancer despite normal examinations and mammography. She was given a referral to a breast cancer center for risk assessment. There the client met with a genetic counselor and shared her cancer concerns, which were exacerbated by the following circumstances. Recently, her two sisters had prophylactic mastectomies due to their perception of high risk for developing breast cancer. The client was now approaching her mother's age at the time of cancer diagnosis. These factors created a conflict in which the client felt caught between her cancer fear and repulsion at the thought of undergoing prophylactic surgery.

The familial cancer risk counseling helped the client to re-evaluate her risk for

developing breast cancer in light of current scientific evidence. She also examined her options for cancer prevention and early detection in the light of her genetic risks. This process is detailed in a later section of this paper. The client eventually decided to enroll in a medical surveillance program and defer medical consultations regarding prophylactic mastectomy. Following this decision, the client returned to her therapist and has continued to explore the psychosocial implications of her family history.

GENETIC COUNSELING ACROSS THE LIFESPAN

As the above cases illustrate, the existence of genetic conditions produces a need for psychosocial and family interventions throughout the lifespan. Rolland (1994, p 101) has noted that "when a condition is long term or chronic, the dimension of time becomes a central reference point. The family and each of its members face the formidable challenge of focusing simultaneously on the present and the future, on mastering the practical and emotional tasks of the immediate situation while charting a course for dealing with the complexities and uncertainties of their problem in an unknown future."

Traditionally, genetic counseling services are divided into stages in the following categories: prenatal or perinatal, newborn or neonatal, childhood, adolescent, and adult. In many of these life stages, the counseling involves negotiating the intersection of individual and family life stages. The next sections will consider some issues during each of these stages.

Genetic Counseling During the Preconception and Prenatal Periods

Genetic counseling issues arising during the prenatal period generally involve infertility, family history that is positive

for genetic conditions or birth defects, or an abnormal prenatal diagnosis. The psychotherapist and the genetic counselor may find themselves working together with the infertile couple, the couple having experienced one or more miscarriages due to a genetic cause, or the couple struggling with a decision about prenatal diagnosis procedures. The psychotherapist also may be called upon to offer support during the prenatal diagnosis process (see Case 2 above). Many couples continue pregnancies with affected fetuses and may require psychosocial support and preparation for future coping (Palmer, et al., 1993b). An initial positive therapeutic encounter may leave the door partially open for further psychosocial work.

Genetic Counseling in the Neonatal Period and Infancy

Many genetic conditions are diagnosed during the newborn period, because many congenital defects are obvious at birth; or newborn screening reveals an unanticipated hereditary disorder, or they are discovered during the first year of life when development is delayed. Congenital simply means present at birth; the problem may or may not be inherited, e.g., congenital rubella is due to a viral infection. The family's major psychosocial tasks include bereavement, parental adjustment to a new diagnosis, complex medical decision-making under stress, and addressing existential questions such as "Why me?" Resolving grief and re-framing the experience of diagnosis often can be expedited with information and support resources (see Table 1).

Family members adjusting to the birth of a new child are further challenged when the child requires additional time and attention. The genetic counselor, family therapist, or social worker can help families access essential medical and social service resources.

Another intersection of genetic counseling and family therapy is in the evaluation of children being considered for foster care or adoption. Often there is a need for genetic evaluation of congenital anomalies or maternal exposures to potentially damaging substances before placements can be finalized. There have been a number of attempts to foster partnerships between genetics professionals and adoption workers (Rauch & Plumridge, 1992; Burners & Reiser, 1992; Delp & Kaepernick, 1992).

Genetic Counseling Issues During Childhood

Once the child with a genetic condition has weathered the newborn period, the family gradually moves out of the crisis mode and into the phase of long term coping with chronic illness and/or disability. The parents may now be considering having another child, but are fearful of the outcome and seek genetic counseling. The imagined or actual birth of a subsequent child can disturb the fragile homeostasis which some families have achieved. For example, some parents may become emotionally conflicted if they fear that seeking genetic testing for a subsequent pregnancy would imply that they do not fully accept and love their affected child. Other parents are relieved by the availability of prenatal diagnosis and selective abortion of affected fetuses, especially if they fear that they would become overwhelmed if they had to care for another child with the same disorder. Still other parents are coping well and do not consider the possibility of a genetic condition a significant issue.

A peer support group can provide a valuable adjunct to counseling. Often the genetic counselor can put parents in touch with other parents whom she personally knows who have faced and surmounted similar issues. More commonly, the family is referred to a genetic support group, of

which there are several hundred in existence. For a guide to genetic support groups, see Weiss and Mackta (1996) or contact the Alliance of Genetic Support Groups, or National Organization of Rare Disorders (NORD) listed in Table 1.

As the affected child grows older, caretaker burnout and depletion of family resources may occur. The parents, nuclear, and extended families may find their emotional, economic, and social resources stretched thin. Social work interventions as well as marital and family therapy may become necessary.

Behavioral interventions could be helpful for the affected child who may be developing dysfunctional behaviors. Often it is a difficult diagnostic issue to determine whether behavioral problems such as hyperactivity, attention deficit, poor impulse control, temper tantrums, compulsive or autistic-like mannerisms are due to mental retardation, underlying psychiatric disorder, social learning, adjustment reaction, or combinations of these. Proper assessment is needed to identify and treat underlying organic and psychiatric disorders so that parents can be relieved of unnecessary guilt or a sense of inadequacy.

While parents are occupied with caring for the affected child, siblings' needs may be overshadowed. Through her intensive investigations of the long-term and far-reaching impact on growing children of the chronic illness and death of a sibling before reaching adulthood, Fanos (1996) has made a compelling case that professionals involved in the care of children with chronic genetic disorders be aware of the many issues facing the siblings. Most of the existing sibling studies have focused almost exclusively on the negative aspects of coping which stress the siblings' experiences of sibling rivalry, anger, embarrassment, loneliness, guilt, and premature assumption of caretaker responsibilities. One study which reverses

this trend is that of Royal, et al. (1995) which examines resilience in siblings of children with sickle cell disease.

Genetic Counseling in Adolescents with Genetic Conditions

Psychosocial issues emergent in adolescence and adulthood often come to the attention of the social worker or psychologist rather than the genetic counselor. While many of the issues are common to chronic conditions in general, the presence of genetic risk adds a particular twist.

Adolescence is a time when the normal developmental drive for autonomy can come into conflict with the limitations imposed by having a genetic condition in the family. Developmental tasks of adolescence include adapting to physical changes of puberty, coping with blossoming sexual feelings, separating from family to form meaningful peer relationships, developing individual values and selecting future goals, transitioning from the educational system to the workforce, and moving toward living away from family and home. Any or all of these processes could be affected by the presence of a genetic condition, which may pose reproductive risks of recurrence, developmental delay, physical stigmata, or social isolation. For example, adolescents with paralysis due to the birth defect called spina bifida often benefit from education regarding how they can function sexually and socially with their peers, however, they may not ask for this directly. Girls with infertility caused by Turner syndrome due to a chromosomal anomaly may not be diagnosed until adolescence when menses and secondary sexual characteristics fail to develop. This unexpected news is often shocking. The girls and their families may need time and support to adjust to short stature and loss of fertility, investigate alternative means of parenting, arrange for hormone therapy

to promote growth and sexual maturation, and explore the inter-related meanings of sexuality, fertility, and parenthood. In teenagers with mental retardation, experimenting with sexual activity, which is assumed to be part of normal functioning of most adolescents, may be seen by others in society as threatening or inappropriate.

Development of the self depends on a stable, predictable, appropriate relational context. If the relational context is skewed due to focus on a disability or disfigurement from a genetic condition, the individual may have a poor context for psychosocial development. Individuals and families may benefit from help in negotiating these complex issues in a safe, supportive, family therapy environment. The disabilities communities can also provide information, support, and positive role models for the affected adolescent.

There is scant literature on offering genetic counseling to adolescent mothers. Since the pregnancies of adolescent mothers can be at risk for genetic problems, just as can other pregnancies of women in their 20s, 30s, and 40s, they too may enter the genetic counseling process for indications such as carrier screening for conditions like cystic fibrosis, Tay Sachs disease, or sickle cell anemia. Sometimes prenatal diagnosis by amniocentesis or other means is recommended when certain conditions are suspected. Often the adolescent consultant has trouble identifying consequences of her actions, understanding probability, predicting future outcomes, making reasoned decisions, and communicating effectively with the healthcare team. One genetic counseling model tailored for adolescent culture and based on trust, patience, and nonjudgemental behaviors has been presented (Peters-Brown & Fry-Mehlretter, 1996), however, additional research is clearly needed.

Genetic Counseling for Adults

These clients comprise several distinct categories, including adult survivors of childhood genetic conditions and adults who are affected with, or at risk for developing, adult-onset disorders.

Adult Survivors of Childhood Genetic Conditions

The families of adult survivors of childhood disorders face unique challenges exacerbated by inadequate social services and support for such individuals within society. Furthermore, social acceptance may decrease as a person with a genetic condition ages and is seen by the majority culture as less attractive than in childhood.

Secondary medical conditions can also arise in those with a genetic condition. For example, survivors of retinoblastoma form of eye cancer can later develop bone or other tumors. Also, premature Alzheimer's disease is common among adults with Down syndrome. Although survival rates have increased for childhood disorders such as cystic fibrosis and muscular dystrophy, life-span is still shortened significantly.

Adult Onset Genetic Conditions

Many common medical and mental conditions develop in adulthood, e.g., mental illnesses, cardiovascular disease, diabetes, cancer, and neuro-degenerative diseases of aging such as Parkinson and Alzheimer's diseases. Subsets of these conditions seem to have a strong hereditary component that causes the condition to run in families and occur at unusually young ages. Recent discoveries make it possible for medical geneticists and genetic counselors to provide genetic risk assessment and counseling to the subset of families with such characteristics.

Individuals affected in adulthood often have difficulty coping with the loss of their

previous level of functioning and find the limitations imposed by the condition to be frustrating or frightening. Other issues include dealing with guilt and responsibility for lifestyle choices (e.g., smoking) made earlier in life, which may have contributed to the current condition, and the need to make current and future lifestyle choices (e.g., medical surveillance or altering diet). Additionally, the new wave of genetic technologies has provided the possibility of genetic susceptibility testing of healthy persons who are concerned about future chances of developing a condition such as breast cancer. The following example of familial cancer risk counseling illustrates the recent emergence of a new interdisciplinary area of practice.

Familial Cancer Risk Counseling

Familial cancer risk counseling (FCRC) is a communication process between a healthcare professional and an individual concerning the occurrence, or risk of occurrence, of cancer in his or her family. Ideally, FCRC addresses genetic risk, medical surveillance and management, and individual and family psychosocial issues as needed (Schneider, 1994; Peters & Stopfer, 1995; Peters & Biesecker, 1997).

Optimally, cancer risk counseling is provided by a multidisciplinary team of professionals with some combination of oncology, genetics, and psychosocial orientations. The main activities include obtaining detailed family, medical, and lifestyle histories, documenting of cancer related diagnoses, constructing and analyzing pedigrees, assessing risk, offering susceptibility testing when appropriate, discussing options for medical management of cancer risk, and providing counseling, reassurance, and support as needed.

Risk assessment is becoming increasingly important in medical care.

Risk is a complex concept that means different things to different people (Hallowell, et al., 1997). The concept of risk incorporates both a statistical or probabilistic notion, and some measure of adversity or threat (Palmer & Sainfort, 1993). Risk encompasses the attributes of ambiguity and uncertainty that make genetic inheritance so difficult to deal with. Kenen has done an elegant review of how women learn to evaluate new risks and hazards (Kenen, 1994) and has expanded this work in a preliminary look at the role of male spouses in genetic decisionmaking (Kenen, et al., 1997).

Cancer risk assessment refers to the process of quantifying the statistical probability for an individual to develop cancer due to the presence of variables such as family history, environmental exposures, lifestyle, and chance. "High risk" families with known hereditary cancer susceptibility syndromes can be ascertained primarily by characteristic family histories.

There are several benefits of identifying such families and providing cancer risk assessment as described in Table 2.

Despite efforts to communicate statistical risks clearly, it has been demonstrated that "efforts to counsel women about their breast cancer risks are not likely to be effective unless their breast cancer anxieties are also addressed" (Lerman, 1994). Sometimes familial cancer risk counseling raises awareness in the family of a relative's battle with cancer and the impact that it may have had on that person (Matloff, 1997; Djurdjinovic, 1997; Eunpu, 1997b). Individuals with moderate statistical risk for cancer may be just as anxious about their perceived risk as people at very high risk. Therefore, sufficient time and attention to medical, psychological, and social needs should be dedicated to all risk counseling interactions, not just to the very high risk hereditary cases.

It is important to elicit the person's

TABLE 2. Benefits of Familial Cancer Risk Assessment, Education, and Counseling

- For members of a family with an identified inherited predisposition, cancer risks will be higher than for those in the general population group;
 - Early identification for those “at increased risk” may allow for the possibilities of prevention, early detection, or early treatment;
 - Identify cancer risks beyond the obvious ones, e.g. pancreatic as well as breast cancer;
 - Genetic susceptibility testing may be available to allow relatives to know whether or not they have inherited a cancer susceptibility gene mutation and plan accordingly;
 - Those who learn that they have not inherited a mutation which runs in a family may be able to avoid unnecessary medical tests and the accompanying anxiety which these might engender;
 - There is an opportunity to address individual and family concerns, which may not have been previously articulated in the medical setting;
 - In the future, knowledge of genetic status may influence methods and outcomes of diagnosis and treatment.
-

understanding of his or her risk and the beliefs underlying this understanding prior to beginning to convey risk information. The ability to process and retain new information can be influenced by cultural beliefs, level of intelligence, education, and psychological distress or wellbeing. Prior experiences with persons with the same disorder, whether or not part of the biological family, can form lasting impressions. It is helpful to provide the client with a cognitive framework of background information about cancer, principles of heredity, and laws of probability.

It is also important to address active coping options such as hormone replacement, diet, exercise, complementary medicine, or other ways of modifying or coping with their perceived risk (Kelly, 1992).

Finally, because some persons may not want specific risk information, there is a strong appreciation within genetic counseling of “the right not to know” about one’s genetic

risk for a specific genetic diagnosis (Schneider, et al., 1997). The optimal timing of referrals for cancer risk assessment, counseling, and testing has yet to be determined.

The emotional impact of cancer goes well beyond the person diagnosed with malignancy; family and friends are also deeply affected. Supportive or grief counseling can be important for unaffected relatives dealing with actual or potential losses of loved ones, practical matters of adjustment to changing family roles, or fears about their own health.

Psychosocial interventions already identified in the growing literature of cancer risk counseling include: confronting the meaning of one’s risk status; venting strong feelings of fear or guilt; helping the consultand to face fears of harm, disfigurement, pain, or death from cancer; managing anxiety, cancer-related worry and

intrusive thoughts; coaching in problem solving; facilitating decision-making strategies; and teaching positive, active coping behaviors (Peters & Biesecker, 1997).

Gene discoveries leading to cancer susceptibility testing have expanded familial cancer risk counseling and education from a few selected centers to becoming widespread in clinical practice (Lynch, et al., 1979; Mulvihill, et al., 1982). The first reports of use of genetic testing information used to inform a family making medical decisions about hereditary breast cancer testing did not occur until 1993 (Biesecker, et al., 1993). These tests provide a very specialized type of genetic information. As such, they require thorough informed consent, extensive pre-test counseling, knowledgeable interpretation of results and follow-up counseling regarding implications of test results for individuals and families (ASCO Statement, 1996; Baty, 1997; Schneider, 1994; Geller, et al., 1997; McKinnon, et al., 1997; Peters & Biesecker, 1997).

It remains difficult to predict who may have long-term struggles in adjusting to results, and who will adjust adequately over time. Those with negative results may be less motivated to return for long-term follow-up. However, clinical experience with other genetic conditions has shown comparable rates of psychological distress at one year among those with negative and inconclusive rates as with positive test results. Thus the provider should make every effort to maintain some long-term contact with all tested individuals.

There will be many individuals in the risk counseling program who do not meet criteria for genetic testing, or who choose to decline testing or defer a decision to a later date. It is important to encourage regular contact with these individuals to update family records, and assess any new genetic, psychosocial, or medical issues that may have emerged.

Hopefully, the lessons learned in cancer genetic counseling will be applied in the

future to cardiovascular diseases, mental illness, and other common adult-onset disorders whose genetic contribution will be clarified over time..

SUMMARY

The purpose of this paper is to promote inter-disciplinary collaborations. Toward this goal, we have attempted to describe some of the historical and current issues in the applications of genetic discoveries. In reviewing genetic conditions throughout the lifespan, the practitioner may develop a sense of how ubiquitous genetic issues are. The brief case vignettes about genetic counseling illustrate existing practice, the competencies and roles of various healthcare professionals, and the parallel and collaborative work that is possible in families facing genetic conditions. The example of familial cancer risk counseling and disease susceptibility genetic testing illustrate a prototype for new areas of practice marked by significant inter-disciplinary management of complex conditions within families.

Genetic conditions also offer numerous opportunities for behavioral, medical, and family research. Current successful strategies involve multidisciplinary research protocols where medical and psychosocial researchers team up to identify and address relevant questions in basic science and clinical care. See Table 3 for a list of currently researchable questions in genetic counseling and Table 4 for possible policy issues to be addressed.

TABLE 3. Researchable Issues in Genetic Counseling

- Understand the expectations and beliefs that patients and counselors bring to the counseling consultations;
- Standardize measures of the input, process, and outcomes of genetic counseling;
- Broaden the range of possible outcome measures for genetic counseling;
- Understand of providers' perceptions of patients' concerns;
- Identify, adapt, and standardize psychometric tools for use in various genetic counseling populations of affected, unaffected, "at risk" relatives, siblings, and spouses;
- Conduct longitudinal descriptive studies of the effects of a hereditary chronic illness on family structure, dynamics, communications, and quality of life;
- Explore psychoneuroimmunology and understanding mechanisms of mind-body interactions that may influence the development or outcome of conditions with genetic components;
- Identify predictors of stress, coping, decision-making, problem-solving, and social support in the context of adjustment to genetic conditions;
- Elucidate the affective, behavioral, and cognitive manifestations of genetic conditions; e.g., psychiatric symptoms in persons with chromosomal abnormalities such as Fragile X, Smith-Magenis, and Prader-Willi syndromes;
- Understand the genetic education and counseling needs associated with the introduction of cancer risk assessment and genetic susceptibility testing for common adult disorders.

TABLE 4. Selected Genetic Counseling Policy Issues

- Determine the best ways to educate the public about technical and complex information being generated by genetic research;
- Ensure fair access to genetic counseling, psychological, and social work services in those with genetic conditions who also belong to special populations such as racial, ethnic, sexual orientation, and disability minorities;
- Determine how best to incorporate large amounts of rapidly changing technical genetic information into training, continuing education, and ongoing competency evaluations of a variety of medical and mental health professionals;
- Establish public policy that addresses some of the ethical, legal, social, and psychological issues associated with the introduction of new genetic technologies;
- Investigate how/why for-profit healthcare systems incorporate genetic information;
- Study the impact of the lack of universal healthcare on genetic testing in terms of putting individuals and families at risk for denial of coverage by healthcare insurers;
- Determine the need for research, clinical care, and policy formulations that incorporate attention to the intersection of the individual, the family, and the healthcare systems.

ACKNOWLEDGEMENTS

We wish to thank the National Human Genome Research Institute for funding and support of development of the 1996 and 1997 Genetic Self conferences. Specifically, we would like to recognize the contributions of other Genetic Self founders Paula Gregory, Kate Berg, Lindsay Middleton, Theresa Hadley, Joan Weiss, and Reginald Burgess along with the authors. The true richness of the Genetic Self Conferences was in the participants' own contributions to the mutual human interactions which leave us all changed. Thanks also to John Mulvihill, Peg Johnston, and Pamela Bram for editing earlier versions of this manuscript.

REFERENCES

- Ad Hoc Committee of the American Society of Human Genetics (ASHG) (1975). Genetic counseling definition. *American Journal of Human Genetics* 27:240-242.
- ASCO (1996) Statement of the American Society of Clinical Oncology: Genetic testing for cancer susceptibility. *Journal of Clinical Oncology* 14(5):1730-1736.
- Baty, B.J., Benne, V.L., McDonald, J., Croyle, R.T., Halls, C., Nash, J.E., Botkin, J.R. (1997). BRCA1 testing: Genetic counseling protocol development and counseling issues. *Journal of Genetic Counseling* 6(2): 223-244.
- Benkendorf, J.L., Callanan, N.P., Grobstein, R., Schmerler, S., Fitzgerald, K.T. (1992). An explication of the National Society of Genetic Counselors Code of Ethics. *Journal of Genetic Counseling* 1(1):31-40.
- Bennett, R.L. Steinhaus, K.A., Uhrich, S.B., O'Sullivan, C.K., Resta, R.G., Lochner-Doyle, D, et al. (1995). Recommendations for standardized human pedigree nomenclature. *American Journal of Human Genetics* 56:745-752.
- Biesecker, B.B., Boehnke, M., Calzone, K. Genetic counseling for families with inherited susceptibility to breast and ovarian cancer. *Journal of the American Medical Association* 269:1970-4.
- Burns, J.K., Reiser, G.A. (1992). Continuing education in genetics for adoption workers in Wisconsin. *Journal of Genetic Counseling* 1(2):187-202.
- Collins, F.C. (1997). Preparing health professionals for the genetic revolution. *Journal of the American Medical Association* 278(15):1285-1286.
- Delp, K.J., Kaepernick, L.A. (1992). Genetics and adoption: Forming a partnership. *Journal of Genetic Counseling* 1(2):203-209
- Dixson, B., Dang, V., Cleveland, J.O., Peterson, R.M. (1992). An education program to overcome language and cultural barriers to genetic services. *Journal of Genetic Counseling* 1(3):267-274.
- Djurdjinovic, L. (1998). Psychosocial counseling. In Baker, D.L., Schuette, Uhlmann, W. (eds) *Text in Genetic Counseling*. New York, NY: John Wiley & Sons.
- Djurdjinovic, L. (1997) Generations lost: A psychological discussion of a cancer genetics case report. *Journal of Genetic Counseling* 6(2):177-180.
- Durant, J., Hansen, A., Bauer, M. (1997) Public understanding of the new genetics. In Marteau, T., Richards, M. (eds.) *The troubled helix: Social and psychological implications of the new human genetics*. Cambridge, UK: Cambridge University Press.
- Durfy, S., Peters, J.A. (1993). For the benefit of all: Case study and commentary on breast cancer susceptibility. *Hastings Center Report*, 25(5):28-30.
- Eunpu, D.L. (1997). Systemically-based psychotherapeutic techniques in genetic counseling. *Journal of Genetic Counseling* 6(1):1-20.
- Fanos, J. (1996). *Sibling Loss*. Mahwah, NJ: Lawrence Erlbaum Associates.
- Fiddler, M.B., Fine, B.A., Baker, D.L., and ABGC Consensus Development Consortium. (1996). A case-based approach to the development of practice-based competencies for accreditation of and training in graduate programs in genetic counseling. *Journal of Genetic Counseling* 5(3):105-112.
- Fine, B.A., Baker, D.L., Fiddler, M.B. (1996). Practice-based competencies for accreditation of and training in graduate programs in genetic counseling. *Journal of Genetic Counseling* 5(3): 113-122.
- Fine, B.A. (1993). The evolution of nondirectiveness in genetic counseling and implications of the human genome project. In Bartels, D.M., LeRoy, B.S., Caplan, A.L. *Prescribing our future: Ethical challenges in genetic counseling*. New York, NY: Aldine de Gruyter.
- Fraser, F.C. (1974) Genetic counseling. *American Journal of Human Genetics* 26:636-59.
- Geller, G., Botkin, J.R., Green, M.J., Press, N., Biesecker, B.B., Wilfond, B., Grana, G., Daly, M.B., Schneider, K., Kahn, M.J.E. (1997) Genetic testing for susceptibility to adult-onset cancer: The process and content of informed consent. *Journal of the American Medical Association* 277(18):1467-74.

- Green, J., Richards, M., Murton, F., Statham, H., Hallowell, N. (1997) Family communication and genetic counseling: The case of hereditary breast and ovarian cancer. *Journal of Genetic Counseling* 6(1): 45-60.
- Green, R.M., Thomas, A.M. (1997) Whose gene is it? A case discussion about familial conflict over genetic testing for breast cancer. *Journal of Genetic Counseling*. 6(2):245-254.
- Greene-Simonsen, D., Peters, J.A.. (1992). A successful blending of genetic counseling and psychotherapy. *Perspectives in Genetic Counseling*. 14(2):5, 1992.
- Hallowell, N., Statham, H., Murton, F., Green, J., Richards, M. (1997). "Talking about chance": The presentation of risk information during genetic counseling for breast and ovarian cancer. *Journal of Genetic Counseling* 6(3): 269-286.
- Kelly P.T. (1992). Breast cancer risk analysis: A genetic epidemiology service for families. *Journal of Genetic Counseling* 1(2):155-168.
- Kenen, R. (1980). Negotiations, superstitions, and the plight of individuals born with severe birth defects. *Social Science and Medicine*, 14A.
- Kenen, R. (1994). Women and risk. In Hutner, F.C. (ed.) *Our vision and values: Women shaping the 21st century*. Westport, CT: Praeger.
- Kenen, R. (1996) The at-risk health status and technology: A diagnostic invitation and the gift of knowing. *Social Science and Medicine* 42(11):1545-1553.
- Kenen, R., & Smith, A.C.M. (1995) Genetic counseling for the next 25 years: Models for the future. *Journal of Genetic Counseling* 4(2):115-124.
- Kenen, R., Smith, A.C.M., Watkins, C., Zuber-Pittore, C. (1997) To use or not to use?: Factors involved in decision-making regarding genetic counseling and prenatal diagnosis from the male partner's perspective. *Journal of Genetic Counseling* 6(4):443-4.
- Kessler, S. (1979) *Genetic Counseling: Psychological Dimensions*. New York: Academic Press.
- Kessler, S. (1997a). Psychological aspects of genetic counseling. VII. Thoughts on Directiveness. *Journal of Genetic Counseling* 1(1):9-18.
- Kessler, S. (1997b). Psychological aspects of genetic counseling. IX. Teaching and counseling. *Journal of Genetic Counseling* 6(3):287-296.
- Kohut, H. (1972) *The Restoration of the Self*. New Haven, CT: International University Press
- Lerman, C., Lustbader, E., Rimer, B., Daly, M., Miller, S., Sands, C. (1994) Effects of individualized breast cancer risk counseling: a randomized trial. *Journal of the National Cancer Institute* 87:286-92.
- Lynch, H.T., Lynch, P.M., Lynch, J.F. (1979). Genetic counseling and cancer. In S. Kessler. *Genetic Counseling: Psychological Dimensions*. New York: Academic Press, pp. 221-241.
- Marks, J.H. (1993). The training of genetic counselors: Origins of a psychosocial model. In Bartels, D.M., LeRoy, B.S., Caplan, A.L. (eds.) *Prescribing our future: Ethical challenges in genetic counseling*. New York, NY: Aldine de Gruyter.
- Matloff, E.T. (1997). Generations lost: A cancer genetics case report. *Journal of Genetic Counseling*. 6(2):169-172.
- McDaniel, S.H., Hepworth, J., Doherty, W.J. (1992) *Medical Family Therapy: A Biopsychosocial Approach to Families with Health Problems*. New York, NY: Basic Books, Harper Collins.
- McDaniel, S.H., Hepworth, J., Doherty, W.J. (1997). The shared emotional themes of illness. In McDaniel, S.H., Hepworth, J., Doherty, W.J. (eds.) *The shared experience of illness: Stories of patients, families and their therapists*. New York, NY: Basic Books, Harper Collins.
- McGoldrick, M., Gerson, R. (1985). *Genograms in Family Assessment*. New York, NY: Norton.
- McKinnon, W.C., Baty, B.J., Bennett, R.L., Magee, M., Neufeld-Kaiser, W.A., Peters, K.F., Sawyer, J.C., Schneider, K.A., and the Social Issues Committee of the National Society of Genetic Counselors (1997). Predisposition genetic testing for late-onset disorders in adults. *Journal of the American Medical Association* 278(15):1217-1220.
- Michie S., Marteau, T. (1997) Genetic counseling. In Marteau T., Richards M. (eds.) *The troubled helix: Social and psychological implications of the new human genetics*. Cambridge, UK: Cambridge University Press.
- Middelton, L.A., Peters, K.F., Helmbold, E.A. (1997). Genes and inheritance. *Cancer Nursing*. 20(2):129-151.
- Miller JB, Stiver IP (1997) *The Healing Connection*. Boston, MA: Beacon Press.
- Mulvihill, J.J., Safyer, A.W., Bening, J.K. (1982) Prevention in familial breast cancer: Counseling and prophylactic mastectomy. *Preventive Medicine* 11:500-511.
- National Society of Genetic Counselors Code of Ethics. *Journal of Genetic Counseling* 1(1):41-44.

- Nelkin, D., Lindee, M.S. (1995). *The DNA Mystique: The gene as cultural icon*. New York, NY: W.H. Freeman & Co.
- Offit, K. (1998). *Clinical Cancer Genetics*. New York, NY: Wiley-Liss.
- Ota Wang, V. (1994). Cultural competency in genetic counseling. *Journal of Genetic Counseling* 3(4):267-278.
- Palmer, C.G.S., Sainfort, F. (1993a). Toward a new conceptualization and operationalization of risk perception within the genetic counseling domain. *Journal of Genetic Counseling* 2(4): 275-294.
- Palmer, S., Spencer, J., Kushnick, T., Wiley, J., Bowyer, S. (1993b) Follow-up survey of pregnancies with diagnoses of chromosomal abnormality. *Journal of Genetic Counseling*. 2(3):139-152.
- Peters, J.A. (1994). Suicide prevention in the genetic counseling context. *Journal of Genetic Counseling*. 3(3):199-213.
- Peters, J.A., Stopfer, J.E. (1996). The genetic counselor's role in familial cancer. *Oncology* 10(2):159-166.
- Peters, J.A., Biesecker, B.B. (1997). Genetic counseling and hereditary cancer. *Cancer (Supplement)*, 80(3):576-586.
- Peters, K.F., Hadley, D.W. (1997). The human genome project. *Cancer Nursing* 20(1):62-75.
- Peters-Brown, T. & Fry-Mehlreter, L. (1996). Genetic counseling for pregnant adolescents. *Journal of Genetic Counseling* 5(4):155-168.
- Pollin, I., Kanaan, S.B. (1995). *Medical Crisis Counseling: Short-term therapy for long-term illness*. New York, NY: BasicBooks, HarperCollins Publishers.
- Punales-Morejon, D., Rapp, R. (1993). Ethnocultural diversity and genetic counseling training: The challenge for a twenty-first century. *Journal of Genetic Counseling* 2(3):155-8.
- Rauch, J.B., Plumridge, D. (1992) A project to strengthen linkages between genetic centers and child welfare services. *Journal of Genetic Counseling*. 1(2): 169-186.
- Resta, R.G. (1997). The historical perspective: Sheldon Reed and 50 years of genetic counseling. *Journal of Genetic Counseling* 6(4):375-377.
- Richards, M. (1997) Families, kinship and genetics. In Marteau, T., Richards, M. (eds.) *The troubled helix: Social and psychological implications of the new human genetics*. Cambridge, UK: Cambridge University Press.
- Rolland, J.S. (1994). *Families, Illness & Disability: An integrative treatment model*. New York, NY: Basic Books, Harper Collins Publishers.
- Royal, C.D., Headings, V.E., Molnar, E.T., Ampy, F.R. (1995) Resilience in siblings of children with sickle cell disease. *Journal of Genetic Counseling* 4(3):199-218.
- Schneider, K.A. (1994). *Counseling about cancer: Strategies for genetic counselors*. Wallingford, PA: National Society of Genetic Counselors.
- Schneider, K.A., Stopfer, J.E., Peters, J.A., Knell, E., Rosenthal, G. (1997) Complexities in cancer risk counseling: Presentation of three cases. *Journal of Genetic Counseling* 6(2):147-168.
- Seligman, M., Darling, R.B. (1989). *Ordinary families, special children: A systems approach to childhood disability*. New York, NY: Guilford Press.
- Shiloh, S. (1997) Decision-making in the context of genetic risk. In Marteau T., Richards M. (eds.) *The troubled helix: Social and psychological implications of the new human genetics*. Cambridge, UK: Cambridge University Press.
- Smith, S.C., Warren, N.S., Misra, L. (1993). Minority recruitment into the genetic counseling profession. *Journal of Genetic Counseling* 2(3):171-182.
- Turk D.C., Kerns R.D. (1985). *Health, Illness, and Families: A Life-span Perspective*. New York: Wiley & Sons.
- Weeks, G.R., Treat S. (1992) *Couples in Treatment*. New York: Brunner/Mazel.
- Weil, J., Mittman, I. (1993) A teaching framework for cross-cultural genetic counseling. *Journal of Genetic Counseling* 2(3): 159-170.
- Weiss, J.O., Mackta, J.S. (1996). *Starting and Sustaining Genetic Support Groups*. Baltimore, MD: Johns Hopkins University Press.
- Wexler, N.S. (1979). Genetic Russian roulette: The experience of being "at risk" for Huntington disease. In Kessler, S. (ed.) *Genetic counseling: Psychological dimensions*. New York, NY: Academic Press.
- Wilfond, B., Baker D. 1995. Genetic counseling, non-directiveness, and clients' values: Is what clients say what they mean? *Journal of Clinical Ethics*: summer: 180-182.