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Genetic counselling

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Summary

This chapter focuses primarily on genetic counselling for susceptibility to the common familial cancers such as breast and colon, with occasional reference to other hereditary cancer syndromes. Genetic issues commonly encountered by health professionals who are seeing families with cancer clusters are discussed. The chapter first addresses the general field of genetic counselling and education, then turns to familial cancer risk counselling, and finally to genetic, medical, psychological, ethical, and social concerns in genetic susceptibility testing.

Introduction: what is genetic counselling?

The worldwide effort to map the human genome is already having major effects on medical care. A wide variety of health professionals will soon be dealing routinely with applications of numerous technological advances in cancer genetics. These developments will increasingly be applied to clinical use in cancer diagnosis, prevention, treatment, monitoring for recurrence, and susceptibility, and the role of the genetic counsellor in the provision of information relating to the risk of adult-onset diseases, such as cancer, is likely to become more important.

Genetic counselling definition

Genetic counselling during the twentieth century has evolved from several streams of input: eugenics, public health, academic study of human genetics, health psychology, and medicine (Kenen, 1984; Fine, 1993; Sorenson, 1993). Reed coined the modern term 'genetic counselling' and defined it as 'encompassing knowledge of human genetics, respect

for the sensitivities, attitudes and reactions of the client, and the desire to teach the truth to the full extent that it is possible' (Reed, 1955).

In the 1970s an attempt was made by professional societies to make this definition more explicit. Since that time, genetic counselling has been defined as a communication process which deals with the human problems associated with the occurrence, or risk of occurrence of a genetic disorder in a family (Ad Hoc Committee of ASHG, 1975). Explicitly included in this definition are helping the individual or family to: (1) comprehend the medical facts; (2) appreciate the way that heredity contributes to the disorder, and to the risk of recurrence in specified relatives; (3) understand the alternatives for dealing with the risk of occurrence; (4) choose among alternative courses of action; and (5) make the best possible adjustment to the diagnosis of the disorder in an affected family member and/or to the risk of recurrence of that disorder.

Genetic counselling requires the following elements: (1) eliciting a complete individual and family social, reproductive, and health history; (2) assessing genetic risk; (3) consulting with the individual and family about available clinical evaluation and testing options including risks, benefits, limitations, interpretation and possible psychological and economic consequences of genetic testing and diagnosis; (4) assessing psychosocial needs and making psychosocial interventions; (5) facilitating medical and reproductive decision-making in a non-directive fashion; (6) anticipatory grief and crisis counselling; and (7) facilitating medical screening, testing, or management options as requested by the individual or family.

Genetic counselling is likely to evolve further with the pressures of providing counselling for new diagnostic genetic tests for an increasing and diversified client population (Kenen and Smith, 1995). Changes are probable in both genetic service delivery systems and the development of alternative models of the genetic counselling process itself.

Genetic issues in genetic counselling

Genetic counselling involves the collection and documentation of genetic information in the family history, the educational opportunity to disseminate genetic information, and the establishment of statistical risk for occurrence or recurrence of disease. These are covered below with respect to cancer.

Family history

A detailed family history is elicited from the consultand (the person coming for consultation) and recorded as a pedigree (Resta, 1994). In the US, a Pedigree Standardization Task Force (PSTF) established recommendations for universal standards in human pedigree nomenclature (Bennett et al., 1993, 1995). These data include information about births, deaths, miscarriages, and abortions in a minimum of three generations, plus mention of mental retardation, birth defects, genetic conditions, and other illnesses such as cancer and heart disease. The ratio and pattern of affected and unaffected individuals influences risk analysis.

Genetic education

An important part of the genetic counselling process is to explain in understandable terms the principles of medical genetics, patterns of inheritance, and probability. Specific mention is made of reproductive recurrence risks, availability of genetic diagnostic testing, and reproductive options including availability of prenatal diagnosis.

Risk assessment in genetic counselling

Transmission and interpretation of risk information is difficult due to: problems for the layperson in understanding the laws of probability; applying an average population risk to the individual whose personal risk may in fact be much higher or lower than the average; preconceived notions about whether or not one will be affected; and the tendency to simplify ambiguous risks into simple binary categories that resolve uncertainty about outcome (Lippmann-Hand and Fraser, 1979a,b,c; Kessler and Levine, 1987; Evans, 1993; Palmer and Saintfort, 1993; Richards et al., 1995). Risk assessment may be offered in a variety of ways, including proportions, percentages, qualitative estimates of low, moderate or high, and the chances of not getting the disease as well as chances of having it. Sometimes risk figures are compared to another more familiar point of reference such as having another disease or accident.

The content of the genetic counselling session is generally recorded in a lengthy letter both to the referring physician and to the family. In addition, appropriate written literature, such as brochures and fact sheets, may be offered. For expediency, the content of some of the education for now commonplace procedures (e.g., amniocentesis or CF carrier testing) may be standardized in a set of slides, video, or even by using an interactive computer programme.

Medical content of genetic counselling

The scope of genetic counselling includes discussion of disease diagnosis, including the criteria for and certainty of diagnosis, the natural history of the disease over the lifespan, and whether the disease is expected to alter the natural lifespan. Although the presenting problem may involve just one organ, genetic diseases often involve disruption of multiple biological systems. Medical testing may be ordered to confirm diagnoses or to find occult signs of the disease in affected individuals and their relatives. Prenatal diagnosis refers to the diagnosis of genetic disease before birth and is accomplished by ultrasound, chorionic villus sampling, amniocentesis, percutaneous umbilical blood sampling, or fetal biopsy. Effective prevention or curative treatments rarely have been available for genetic diseases, so most medical interventions for genetic conditions involve secondary or tertiary prevention and symptom treatment.

Psychosocial assessment in genetic counselling

General areas of assessment include current mental status, mood, and the person's behaviour and responses during the genetic counselling process. Psychosocial information should include cultural background, childhood traumas, losses, or abuse; relationships and sexual history, family cohesiveness and communication styles; coping strategies, competencies, and support resources; and general psychosocial history. In addition, it is important to discuss beliefs, attitudes, and experiences when the specific genetic condition is being considered.

Psychosocial interventions in genetic counselling

The more cognitive aspects of genetic counselling will not be successful unless the counsellor attends to the emotional aspects concurrently (Epstein, 1975; Kessler, 1980). The genetic counsellor may need to help the consultand adjust to the knowledge of a genetic condition in the family (Peters, 1995).

Counsellors should be familiar with indications for psychiatric referral when signs of mental illness or distress are noted (Schneider, 1994). Suicide has been reported in some persons with high risk for expressing genetic disorders and the genetic counsellor should be aware of this possibility (Peters, 1994c). Other indications for referral are: clinical levels of depression, guilt, or anxiety; unresolved grief; obsessive, intrusive thoughts; overzealous health vigilance; severe sleep or eating disturbance;

drug or alcohol addiction; somatization; or disruptions in sexual functioning or satisfaction.

Genetic counselling process

Genetic counselling is practised in a variety of settings – academic medical centres, private testing sites, inter-disciplinary specialty clinics, and in research settings. In the US and Canada, performance of the various tasks of genetic counselling are often shared by a team of individuals including PhD or MD geneticists, post-doctoral fellows, masters level genetic counsellors or genetic nurses, and sometimes a variety of other medical specialists are needed to care for persons with multiple manifestations of genetic disease. Frequently, entire families are seen conjointly.

Directive counselling and non-directive counselling are generally taken to mean the offering or withholding of direct advice, often about reproduction and abortion (Kessler, 1992). Non-directiveness generally implies an assumption that consultands can and should make their own decisions about certain aspects of their healthcare.

Although non-directiveness is endorsed in the UK, a number of investigators have observed that all genetic counselling involves some element of direction given to participants, be it overtly delivered as medical advice, or covertly conveyed by counsellor selection of which information is given (Kessler, 1992). Cross-cultural experiences show that receiving advice from an expert is preferable to individual autonomy for a number of consultands of certain ethnic and cultural groups (McGoldrick, 1982; Weil and Mittman, 1993; Geller et al., 1995).

Familial cancer risk counselling

This is a communication process between healthcare professionals and individuals concerning the occurrence, of cancer in their families (Peters, 1994a; Table 5.1). These families are often seeking more information about the hereditary basis of cancer in the family, genetic testing, and advice about medical surveillance. These needs can most often be met by multi-disciplinary teams consisting of individuals with expertise in oncology, surgery, genetics, and counselling psychology.

The scope of this counselling is wide (Figure 5.1; Kelly, 1991; Schneider, 1994; Peters and Stopfer, 1996, table 1) and its aim includes reducing mortality through prevention and early detection, enhancing quality of care, establishing a co-ordinated approach to ascertainment,

Table 5.1. *Suggested components of comprehensive familial cancer risk counselling*

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- Full, three-generation pedigree taken and extended as necessary
 - Primary diagnosis documentation on all affected relatives
 - Background information on genetics, oncology, and testing
 - Statistical genetic risk assessment and counselling
 - Identification of genetic susceptibility syndromes
 - Psychosocial assessment and interventions
 - Genetic susceptibility testing as appropriate
 - Pre-test informed consent
 - Susceptibility test result notification
 - Post-test counselling and follow-up
 - DNA banking on affected relatives as needed
 - Recommendations for medical surveillance
 - Referral to prevention trials
 - Participate in multidisciplinary management teams
 - Establish/liaison with hereditary disease registries
 - Referral for additional consultations as needed
 - Advocacy for patient rights to healthcare
 - Establish/sustain support groups
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Source: Adapted from Peters (1994a), with permission.

screening, diagnosis, treatment, and follow-up care for cancer, and implementation of future genetic technology such as gene-based treatments.

Family history screening and indications for referral

Ascertainment may occur at the time of diagnosis of a cancer, when considering reproductive options during medical screening, or when considering genetic susceptibility testing (Petersen, 1996b). Each consultand should have a family history taken, particularly noting relatives diagnosed with cancer, their age at diagnosis, current age, bilaterality, and occurrence of multiple different cancers.

Documentation of diagnoses is necessary by review of the pathology reports of biopsies and surgical specimens (reviews of diagnoses on death certificates may be inaccurate) or with reference to disease registries.

Genetic education

Families with cancer need varying degrees of background information about biology, genetics, oncology, epidemiology, and probability in order to comprehend fully the risk information they will later be given (Kelly,

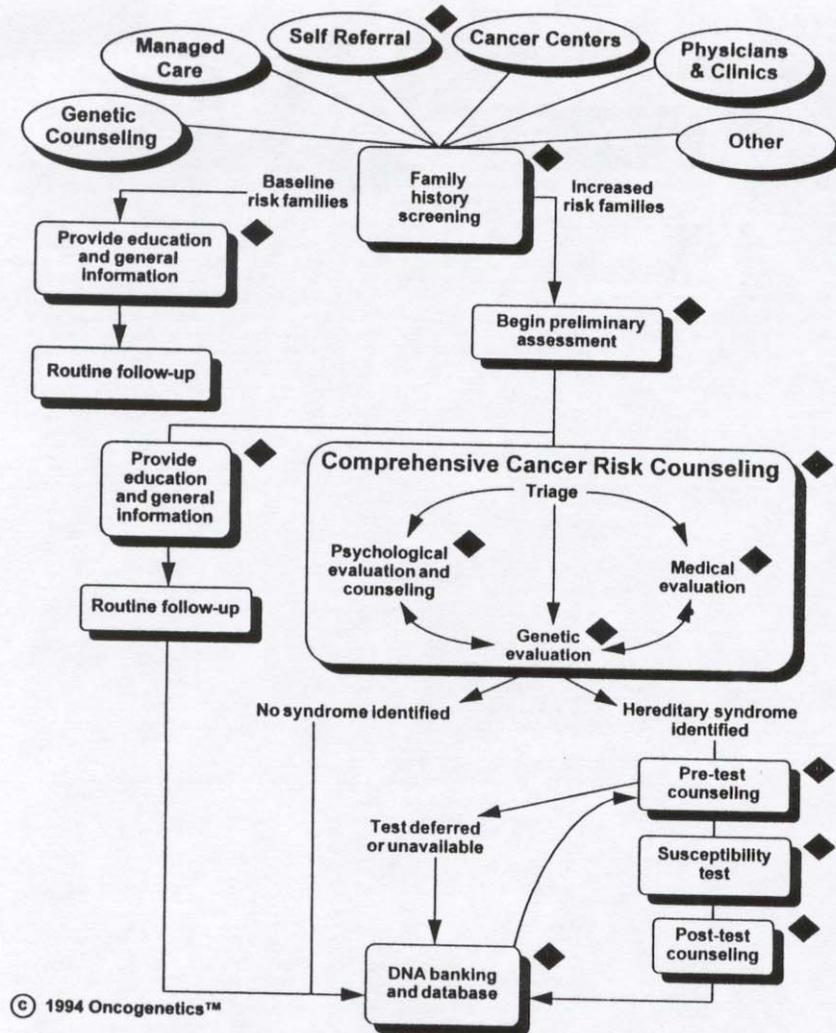


Figure 5.1 Cancer risk assessment and counselling protocol.

1991, 1992; Peters, 1994a,b, 1995; Schneider, 1994; Hoskins et al., 1995; Peters and Stopfer, 1996).

Popular sources of information that can be provided are books, brochures, newsletters, etc., written at the level of lay audiences (e.g., Kelly, 1991; Cooper, 1993), many of which are provided by specific hereditary cancer registries and support groups (e.g., in the US, The Von Hippel Lindau Family Alliance, and the *Hereditary Colon Cancer Newsletter*).

The National Cancer Institute of the US also provides written information, slide sets, telephone consultations, referral directories, and internet sites for supplemental cancer genetics information. In the US, these can be accessed by calling 1-800-4CANCER or through the NCI cancer net website at: www.cancernet.nci.nih.gov.

Familial cancer risk assessment

Cancer risk assessment refers to the process of quantifying the probability for an individual to develop cancer due to the presence of variables such as family history, environmental exposures, lifestyle, and chance, often in comparison to the general population 'baseline risk' of cancer.

Hoskins et al. (1995) present a guide for primary care clinicians for use in evaluating inherited breast cancer risk. Offit and Brown (1994) reviewed four different models for breast cancer risk assessment based on family history and other recognized risk factors. The most commonly used means of measuring risk are by estimating relative risk or cumulative risk. Individuals seem better to be able to understand and integrate cumulative risks over time in order to make concrete life decisions, for example, about career, family planning, and prophylactic surgery (Kelly, 1992). Methods for this analysis are detailed elsewhere (Anderson and Badzioch, 1985, 1989; Gail et al., 1989; Claus et al., 1994; Hoskins et al., 1995).

Diagnosing hereditary cancer syndromes

If the pedigree suggests a mendelian inheritance pattern of a cancer-predisposing condition and particularly when such a condition has been diagnosed in the family, risk assessment involves the discussion of two individual probabilistic events: (1) the chance that they will inherit the cancer susceptibility gene mutation; and (2) the manifestation of the disorder and the chance that people with this specific mutation will eventually develop cancer (penetrance). Reaching a correct diagnosis is crucial for genetic counselling. Hodgson and Maher (1993) have compiled a very useful text of known hereditary cancer syndromes, organized by site of origin and by syndrome.

When no specific syndrome can be identified, DNA banking from affected relatives within these families may be appropriate. The purpose would be for relatives at risk to have DNA available for testing when new

gene discoveries are made. However, tissue storage itself has many ethical, legal, and social ramifications and should be undertaken only with fully informed consent.

Medical recommendations

Richards et al. (1995) have noted that for many consultands the central issue of genetic risk counselling is what to do about a risk they already perceive as high. Surveillance and prophylactic measures should therefore be discussed. Counsellors may also help the family to implement any recommendations made, and to address anxieties and explore obstacles to surveillance (Peters and Stopfer, 1996).

Medical treatment

Knowledge of genetic risk status may alter the appropriate treatment and the decision-making process for some individuals who learn that they are at high risk for development of subsequent tumours.

Psychosocial issues

Cancer risk counselling is conducted simultaneously on two different levels – the medical and the emotional (Royak-Schaler and Benderley, 1992). Lerman and colleagues (1995) have noted that efforts to counsel women about their breast cancer risks are not likely to be effective unless their breast cancer anxieties are also addressed.

Psychosocial assessment

The counsellor should explore the meaning of cancer to the individual and to the family and their theories of causation (Kenen, 1980; Kelly, 1992; Green et al., 1993; Richards et al., 1995; Grosfeld et al., 1996). Cancer may be perceived as a punishment, or risk status may be assumed to be based on physical or personality resemblance to an affected individual. Information offered by geneticists that is not in accordance with family explanations and expectations may be disregarded (Kenen, 1980; Grosfeld et al., 1996), so it is wise for counsellors to elicit underlying beliefs before undertaking genetic education.

Assessment should also include the emotional reactions to cancer and risk. Investigators in the Utah (USA) study of a large *BRCA1* family have observed that consultands with close relatives with cancer often expressed strong, long-term emotional effects of this legacy (Baty et al.,

1995) including, anxiety, anger, fear of developing cancer, fear of disfigurement and dying, grief, guilt, lack of control, negative body image, comparisons to affected relatives, altered sexual functioning and sense of isolation (Baker, 1991; Kelly, 1992; Wellisch et al., 1992; Lynch and Watson, 1992; Lynch and Lynch, 1993; Peters, 1995). Such anxiety and fear can have a major impact on daily activities, life decisions, and healthcare behaviour (Kash et al., 1992; Lerman et al., 1993), and may fluctuate with time. Genetic disorders also affect relationships, and adequate marital and family assessment, therefore, may be necessary.

There may also be differences in psychosocial issues depending on whether the family is newly ascertained or whether it has been known for years to have a hereditary cancer predisposition (Berk, 1996) and, on which cancers the relatives develop (Baker, 1991; Williams, 1991; Peters, ASHG lecture, 1994). In western culture, for example, breasts are symbolic of women's identity, nurturing, and sexuality, whereas the functions of the colon are often seen as dirty, shameful, and secret.

Psychosocial interventions

Grief counselling is often necessary and discussing the family history is an opportunity for relatives to remember and grieve their personal losses.

Many consultants enter the counselling process feeling fatalistic about getting and dying from cancer. Demonstrating a genetic model that gives a basis for believing that he or she has an equal chance of *not* getting cancer can be very therapeutic. Dispelling misconceptions can also serve to relieve emotional burdens. Individuals having trouble making decisions about genetic testing or about their healthcare may benefit from learning problem-solving or other decision-making skills (Lerman et al., 1996).

Kash et al. (1992), studying women at high risk who were attending a high risk surveillance programme, has shown that women at risk of breast cancer have psychological distress levels as great as women with actual cancer. A substantial subset of women who have an interest in breast cancer susceptibility testing have intrusive thoughts and other signs of anxiety and distress (Lerman et al., 1994b). Psychoeducational support group experience has been shown to improve surveillance adherence in this at-risk group, as has been demonstrated for women with cancer (Fawzy et al., 1990a,b; Spiegel, 1992).

Process of counselling

A key to advising consultands lies in knowing which proposed interventions have proven risks, benefits, and limitations, and which do not. A non-directive approach is particularly appropriate for reproductive decisions and in situations in which options have relatively equal risk:benefit ratios (e.g., choosing lumpectomy plus radiation versus mastectomy for certain breast cancers).

Schneider and Marnane (1995) conducted a survey of a subset of 50 genetic counsellors within the US who are currently conducting cancer risk counselling sessions. Whereas the majority believed that it was appropriate for counsellors to advise clients to follow widely accepted screening and lifestyle guidelines, many felt that it was inappropriate to advise regarding DNA testing options, prophylactic surgery, or disclose information to other family members at risk.

Organization of service

Peters (1994a, b, 1995) has adapted a comprehensive breast centre model (Lee et al., 1992a,b) to the development of genetic cancer risk counselling programmes (Figure 5.1). It is important for clinicians to consider how this might fit into various parts of their medical practice from referral through screening, triage, assessment, treatment and follow-up to optimize use of limited genetic counselling resources (Peshkin et al., 1995). Even some private laboratories have made testing available primarily within a genetic counselling infrastructure (e.g. OncorMed, Genetic Education and Testing Packets, 1996) or at the very least, profess a responsibility to prepare educational materials for physicians and their patients (Skolnick, 1996).

In contrast to this heterogeneity of approaches, co-ordinated national approaches are possible in smaller European countries. For example, in the Netherlands, genetic counselling is provided by clinical geneticists (with a genetic associate) at eight clinical genetics centres all linked to a university hospital. There is co-operation among the three main medical centres in Amsterdam in dealing with families in accordance with a single protocol known as the Amsterdam Protocol for Familial and Hereditary Tumours. This protocol specifies medical diagnosis and management recommendations as well as providing information for patients and family members (Fred Menko, 1995, pers. comm.).

Susceptibility testing – general concerns

Nature of genetic susceptibility tests

Cancer genetic susceptibility testing poses challenges due to genetic heterogeneity, profound personal and familial implications of results, and the possibilities of stigmatization, discrimination, misunderstanding and misuse of the information (Kenen, 1980; Durfy and Peters, 1993; Nelkin and Lindee, 1995; Biesecker, 1997; Peters and Biesecker, 1997). Genetic testing historically has been voluntary, available only for rare genetic conditions with a known monogenic basis, offered within the context of genetic counselling that supports autonomous decision-making, and with thorough informed consent about possible implications prior to test-taking. It is now being introduced for common diseases, in larger populations, and within mainstream medicine.

Laboratory selection

Laboratories performing research studies are often not the same as those providing clinical diagnoses for purposes of counselling. Not all laboratories perform all tests, and different laboratories may use non-overlapping methodologies, making comparisons difficult. The technical aspects of susceptibility testing are discussed in Chapter 8.

Selection of a laboratory that also offers tissue storage capabilities leaves open the possibility for testing at a later date.

Regulation of laboratory quality control

Assuring quality control for molecular testing is essential, and laboratories should adhere closely to general quality assurance programmes, be licensed, and participate in all applicable external proficiency testing programmes. In the US, clinical laboratories performing an examination of materials derived from the human body for the purpose of providing information for diagnosis, prevention, prognosis, or treatment of humans must obtain certification from the Health Care Finance Administration (HCFA) under the Clinical Laboratory Improvement Amendment (CLIA) of 1988 (Andrews et al., 1994).

Test interpretation, sensitivity, specificity, and genotype–phenotype correlations

It is appropriate to expect the laboratory to provide up-to-date information regarding test sensitivity and specificity (Boland, 1966; Menko et al., 1996). For example, interpretation of a particular genetic alteration may be influenced by whether it has ever been observed before in high risk

families; whether the alteration causes a change in protein length, configuration, or function; how closely the family medical history of the current patient resembles those of the research families on which test interpretation is being based; family ethnic background; and the state of technology development being used by the laboratory selected. The laboratory report should take into account relevant genetic risk factors and be sufficiently clear and specific about limitations (Petersen and Brensinger, 1996).

Indications for cancer susceptibility testing

Genetic testing up to this point has been undertaken largely in the context of research studies where indications were selected based on research needs. There is a number of policy statements urging caution in this until more information is known about test characteristics and implications (Li et al., 1992; Evans et al., 1992, 1994; Lynch and Watson, 1992; Biesecker et al., 1993; King et al., 1993; ASHG, 1994; National Advisory Council for Human Genome Research, 1994; ASCO Sub-committee on Genetic Testing for Cancer Susceptibility, 1996).

Professional organizations are beginning to establish standardized indications for genetic testing for cancer susceptibility in an individual affected with cancer. For example, the American Society of Clinical Oncology (ASCO) has issued a statement that includes recommended indications for referral (ASCO Sub-committee on Genetic Testing for Cancer Susceptibility, 1996). These are: (1) the person has a strong family history of cancer or very early onset disease; (2) the test can be adequately interpreted; and (3) the results will influence the medical management of the patient or family member. Combinations of these criteria are then used to create three categories of indications for cancer predisposition testing.

Group 1 consists of tests for families with well-defined hereditary syndromes for which either a positive or negative result will change medical care, and for which genetic testing may be considered part of the standard management of affected families (see Table 5.2). Most practitioners now agree that genetic diagnostic and susceptibility testing under controlled circumstances is indicated for these diseases.

Group 2 includes tests for known cancer susceptibility genes, for which the medical benefit of the identification of a heterozygote carrier is presumed but not established. The potential clinical value and reliability of the test is based on research studies. Included in group 2 are hereditary

Table 5.2. Selected hereditary cancer predisposition syndromes

| Syndrome | Gene |
|--|-------------------------------------|
| Hereditary breast and breast-ovarian | <i>BRCA1, BRCA2</i> |
| Li-Fraumeni syndrome (LFS) | <i>TP53</i> |
| Cowden syndrome | <i>PTEN</i> |
| Familial adenomatous polyposis (FAP) | <i>APC</i> |
| Hereditary non-polyposis colorectal cancer (HNPCC) | <i>MSH2, MLH1, PMS1, PMS2, MSH6</i> |
| Multiple endocrine neoplasia type 1 (MEN1) | <i>MEN1</i> |
| Multiple endocrine neoplasia (MEN2A,B) | <i>RET</i> |
| Neurofibromatosis 1 (NF1) | <i>NF1</i> |
| Neurofibromatosis 2 (NF2) | <i>NF2</i> |
| von Hippel-Lindau syndrome (VHL) | <i>VHL</i> |
| Basal cell nevus syndrome (Gorlin syndrome) | <i>PTC</i> |
| Retinoblastoma | <i>RBI</i> |
| Wilms' tumour | <i>WT1, FWT1</i> (not identified) |
| Ataxia-telangiectasia (AT) | <i>ATM</i> |
| Peutz-Jeghers syndrome | Not identified |

non-polyposis colorectal cancer (HNPCC), hereditary breast-ovarian cancer syndrome, and Li-Fraumeni syndrome. There is some controversy about whether, when, how and for whom testing should be offered in this group. A subset of members of the ASCO sub-committee differed from the majority report. In their view, genetic testing for breast cancer susceptibility should not be offered outside the context of hypothesis-driven research approved by institutional review boards.

Finally, group 3 is the most controversial. Included in this group are tests for individuals without a family history of cancer, conditions in which the significance of the detection of a germline mutation is not clear, and tests for hereditary syndromes for which the germline mutations have been identified in only a small subset of families.

Several commentaries to the ASCO Statement were published concurrently by representatives of the National Breast Cancer Coalition (Visco and the National Breast Cancer Coalition, 1996), Myriad Genetics Inc. (Skolnick, 1996), and the National Action Plan on Breast Cancer (Collins and NAPBC, 1996). The two statements from the consumer coalitions call for partnerships between patients, physicians, and researchers in attempts to recognize and address the many unanswered questions regarding the ramifications of susceptibility testing, and urge consumers to be wary of testing outside of research protocols.

Technical advances in the ability to detect mutations with improved levels of sensitivity, specificity and speed are evolving. These developments will undoubtedly alter indications for testing; however, the technical ability to perform tests for mutations should not be confused with a mandate to offer them (Collins and NAPBC, 1996).

Susceptibility test context

Genetic susceptibility testing should always occur within the context of a supportive professional relationship between counsellor/physician/nurse and the consultand (ASHG, 1994). This can be accomplished through a co-ordinated research investigation or a clinical programme.

With informed consent, individuals in the family who are affected with cancer are tested first to search for mutations in a particular gene known to cause a hereditary cancer syndrome. If a pathogenic mutation is found in one or two family members with cancer, the testing of their relatives will be informative.

Genetic counselling for susceptibility testing

Biesecker et al. (1993) set out the genetic counselling necessary for susceptibility testing for inherited breast cancer. This approach had a strong emphasis on the importance of pre-test counselling, the multi-disciplinary team approach, and the necessity of follow-up for family members tested. Grosfeld et al. (1996) has described genetic counselling for multiple endocrine neoplasia type 2 (MEN2), Lynch et al. (1996) and Menko et al. (1996) for HNPCC, and Petersen (1996b) for familial adenomatous polyposis (FAP).

Currently, testing is being offered mainly to self-referred persons within high risk families or from registries. As testing becomes more commonplace, the possibility of testing may be introduced to people who have never heard of susceptibility testing or who had never previously thought much about it. Presumably this group will need more basic pre-test education.

Schneider (1994) discusses genetic principles for genetic susceptibility testing, and observes that there are at least three distinct aspects of the testing process: (1) informed consent during pre-test counselling; (2) disclosure of results; and (3) follow-up counselling.

Informed consent

Informed consent refers to a communication process (usually both oral and written) between health care providers and consultands in which participants are provided with sufficient information to decide whether or not to be tested, and if so, when and how. The legal aspects of informed consent are discussed in Chapter 3, but here the focus is on the role of the genetic counsellor in ensuring that the rights and welfare of the individual are protected. Two worrisome sources of possible undue influence are professionals (including the counsellors themselves) and family members.

Informed consent and professional biases

Difficulties in achieving non-directiveness, and counsellor biases colouring the transmission of information from counsellor to consultand, may occur. Firstly, there is a pro-testing bias typical of medicine in which testing is generally viewed in a positive light. Aware of this bias, the genetic counsellor is challenged to present a balanced view of the risks, benefits, and limitations of genetic susceptibility testing in order to refrain from overtly or covertly swaying the consultand's decision regarding testing. For example, if a counsellor tells an individual, 'If you were my spouse, I would advise you to take the test', the consultand might agree to undergo a test just to please the counsellor, who is in a powerful role of trustee. In a more subtle scenario, the counsellor who believes that knowing genetic status is better than not knowing might bias the consultand towards testing in more subtle ways, such as overstating the potential for cancer prevention or therapy as one of the benefits of genetic testing, gently persuading the consultand to be tested. When clear-cut benefits of testing have yet to be demonstrated for some syndromes such as breast-ovarian cancer, it may be prudent to let individuals who are undecided wait to be tested until more data are available.

Informed consent and family biases

Another layer of potential interference with autonomous decision-making about genetic susceptibility testing could come from family and friends. Families have cultures, beliefs about health and illness, testing biases, spiritual and religious beliefs and dynamics of their own. The influence of loved ones can be a source of strength and camaraderie, but they may have an interest for themselves in persuading that relative to be tested.

When the family history is positive, members have already experienced multiple relatives developing cancer and thus, have had previous encounters with the medical system. These prior experiences may make relatives either wary of the medical profession, more motivated to use medical care, or in conflict about wanting the best of care. The counsellor needs to help the consultand sort through and become more aware of the effects of these family biases while also striving to protect the privacy of individuals. This can be delicate when the individual is being pushed toward testing by zealous relatives determined to stamp out cancer through genetic testing, or conversely, impeded from testing by relatives who do not wish to co-operate with family testing efforts. Some individuals do not consider a test decision to be solely for their own benefit (considering this selfish), and prefer to construe their decision to be tested altruistically, for example, for the sake of a spouse or the children.

Informed consent and culture

Another issue to be considered in the informed consent process regarding testing is the class and cultural characteristics of the person. Cross-cultural issues are increasingly important. For example, it was estimated in 1990 that one in four Americans was foreign-born or a member of a racial minority group (US Bureau of Census, 1990) and by the year 2010, one in three Americans will be non-white or Hispanic (US Bureau of Census, 1992). Racial and ethnic minority groups have already achieved majority status in certain cities such as Los Angeles and San Francisco (US Bureau of Census, 1990). In addition to facing many barriers to general health care, members of these minority cultural groups may have very different needs and perceptions of the use of genetic testing (Weil and Mittman, 1993).

Geller et al. (1995) and colleagues held a series of consumer focus groups to explore ethnically and socially diverse participants of beliefs about the causes of cancer, what they would want to know about a genetic test for breast cancer, their attitudes about the advantages and disadvantages of having such a test, what they would do with the test results, and their expectations of their role in decision-making. In comparisons of women in different socio-economic strata (SES), marked differences were found in preferences for the content of the informed consent. Women in high SES groups wanted information on the validity and accuracy of the test, cost of testing, follow-up recommendations and implications of test results for other family members. Women in low SES groups wanted answers to practical questions regarding testing – for

example, does testing involve drawing blood?; who will do the test?; when will the results be available?; does the test detect cancers? Overall, higher SES women preferred more autonomy than lower SES women. However, participants across SES groups believed that written materials would be a welcome addition to the informed consent process.

Working with a Navajo family with HNPCC that they have been following for about 12 years, Lynch et al. (1996) found other cross-cultural issues interacted with and altered the genetic counselling process. The use of some culturally familiar analogies such as those of agricultural and animal husbandry met with some success in explaining biological and genetic principles in ways that would not be upsetting or degrading to the traditional tribal beliefs. Investigators observed that the process would be enhanced further by incorporating a member of the native group into the genetic counselling process as an active participant.

Informed consent and declining testing

The 'right not to know' is one that should not be overlooked. From their work on the effects of carrier testing in childhood on adult siblings of cystic fibrosis patients, Fanos and Johnson (1995) concluded that remaining unaware of carrier status may serve a significant psychological function for some individuals at risk for genetic disorders. Research to determine whether the same may apply to hereditary cancers is not yet available.

The health psychology literature suggest that the benefits of prior awareness of risk may depend on individual coping styles. Among women seeking amniocentesis, those who coped by seeking information or 'monitoring' were significantly more likely to experience anxiety and depression than those who coped by 'blunting' information-avoidance (Miller, 1995). Lerman and colleagues (Lerman and Croyle, 1994; Lerman et al., 1994a,b) studying first degree relatives of women with cancer found results consistent with this. Those with more cancer worries and monitoring coping styles were significantly more likely to anticipate negative psychological consequences of *BRCAl* testing.

Denial is commonly addressed in the genetic literature (Lubinsky, 1994). Some people may ignore or decline counselling because they do not think of themselves as having a risk for inherited cancer. Grosfeld et al. (1996) have pointed out that the decision to have a genetic test can be seen as a sign that the individual is facing the threat of genetic disease consciously. Consultands may engage in advance-and-retreat strategies of making and breaking appointments in repeated cycles that parallel

their internal conflicts over accepting and rejecting new cancer risk information about themselves (author's pers. obs.). In light of this, it is important to consider the timing of offering genetic testing. Situations in which testing might be counterproductive are when a person is in denial about risk, is currently caring for an ill relative, or is immobilized by anxiety about cancer. All of these individuals need supportive counselling, with deferral of testing until a more receptive time.

Informed consent in cancer patients versus in relatives at risk for cancer

There are important differences between counselling relatives considering predictive testing and counselling persons with cancer who are considering testing to help establish the presence of a disease-conferring mutation in the family. Affected individuals will be getting information that elucidates the genetic basis for their cancer as well as information about risk for possible additional cancers. As Peshkin (1996) points out, the counsellor should be sensitive to the timing of an invitation for the affected individuals to participate. They may be currently undergoing treatment and concerned primarily about their recovery. Others may have had cancer years ago and want to keep all thought of cancer behind them. The prevention and early detection options for them may not be as much of a benefit as for the younger susceptible consultand.

Pre-test counselling for cancer susceptibility testing

The psycho-oncology literature suggests that genetic testing produces greater distress among persons who previously were unaware of the risk status (Croyle and Lerman, 1994). Therefore, it is essential that the foundational work for the risk notification session is laid during the pre-test counselling and informed consent process.

There is general agreement that samples should not be obtained without thorough pre-test counselling (Table 5.3; Schneider, 1994).

Testing motivations and expectations

The reasons for seeking cancer predictive testing are varied. Some people may value knowledge both for the sake of knowing and for the control over one's life that this knowledge implies. Others may hope to put uncertainty to rest. Some may wish to avoid expensive, risky, and uncomfortable medical surveillance programmes if they were to test negative. Many have altruistic reasons such as helping research or for the sake of

Table 5.3. *Issues in pre-test counselling*

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- Motivation and expectations
 - Review medical and genetic facts
 - Possible results and implications
 - Accuracy and limitations
 - Risks and benefits
 - Assess and support coping
 - Privacy and confidentiality
 - Programme specifics
-
-

protecting their children. Others may want to know their risk status when starting or continuing a family (Petersen and Boyd, 1995; Burt and Petersen, 1996). Sometimes patients and their families want more information to understand the biological basis of their cancer.

Surveys of the general population have demonstrated that the public is very interested in genetic susceptibility testing for cancer (Chaliki et al., 1995; Smith and Croyle, 1995). Over 90% of relatives of breast cancer patients and of ovarian cancer patients reported that they would want to be tested, once a test is available (Lerman et al., 1994b; Struewing et al., 1995). Women's interest in testing for any cancer susceptibility is usually greater than men's and there is greater interest among those with high self-perceived (rather than calculated) risk of cancer (Struewing et al., 1995; Lerman et al., 1994a).

Motivation for testing may be based on misconceptions about the nature of testing, for example, 'the test will give me a definitive answer about cancer'. It may also be based on assumptions about possible medical interventions. There is widespread belief in the general population that the medical community would not be offering cancer susceptibility testing if there were nothing that one could do about one's risk. These test expectations can be a source of later disappointment if the testing fails to meet the person's underlying needs and wishes.

For some genetic conditions, studies, however, find few people are presenting themselves for predictive testing. Schneider and her colleagues have shown that only 12 of 48 members of the first six extended kindred with Li-Fraumeni syndrome invited to be tested actually enrolled and completed an initial visit (Schneider and Marnane, 1995). The most common reasons for declining participating in the testing process were recent cancer diagnosis or death in a relative, fear of insurance problems, lack of options for prevention, inconvenience, 'not a good time in life', and not

interested. The investigators conclude that the demand for testing will be difficult to predict given the complex issues faced.

Most participants in one research study on susceptibility testing felt strongly that they knew what the outcome would be and were seeking testing as confirmation of that intuition (Grosfeld et al., 1996). There is some evidence from Huntington disease testing that those persons who receive results congruent with prior expectations show better adjustment, whereas those who receive results that differ significantly from what they expected will have the most long-term adjustments to make (Huggins et al., 1992). The genetic counsellor, therefore, should ascertain at the outset whether the participant believes that the results will turn out positive or negative.

Medical and genetic facts

Many consultands have little prior knowledge of genes, chromosomes and the relation of these to cancer. Richards and colleagues observe 'the fact that lay people use technical terms such as gene or chromosome does not necessarily mean that they understand these terms in the same way as geneticists' (Richards et al., 1995, p.229). Grosfeld et al. (1996) found that much of what passes for genetic knowledge is often derived from personal or family experience with a particular disease. Patterns unique to a given family – for example, only first born offspring are affected – may be erroneously generalized to a principle. Others may correctly understand a principle, for example, 50% risk, but inappropriately apply it to count up the number of affected persons in their own sibship and decide about one's own risk based on whether that 50% ratio has been met or not.

The primary genetic facts about the common inherited cancers that are essential are: basic laws of inheritance; gene penetrance; variable phenotypic expression; and occurrence of non-genetic cases even within hereditary families (Richards et al., 1995).

Prior to testing, the persons considering testing should also understand the medical options available for prevention, early detection, and treatment. Being at increased risk for cancer, they should be following surveillance recommendations that differ from the general public. These recommendations may change as the result of genetic testing.

In the first year of a study of *BRCA1* counselling and presymptomatic testing in Utah, qualitative observations of subject responses to genetic counselling showed that subjects often lacked certain knowledge prior to

counselling, for example, they did not know of the increased risk of gene positive males for cancer and the relative lack of increased risk for male breast cancer. Female subjects often did not know that they were at risk for both breast and ovarian cancer because their own nuclear family may have expressed only one or the other. Many were surprised to learn of the possibility of prophylactic mastectomy and oophorectomy (Baty et al., 1995).

Prophylactic surgery is a prevention option that is offered in many of the hereditary cancers, but the efficacy varies considerably from one condition to the next. For example, it is very effectively used in FAP (Petersen, 1996b) and MEN2. These conditions are also characterized by a very early onset of symptoms, almost 100% penetrance, and the ability to remove the organ at risk for cancer. The situation is much less clear-cut for hereditary colon (except FAP), breast, and ovarian cancer, because of heterogeneity, variable age at onset of cancer, reduced penetrance and a number of organs at risk for malignancy; also prophylactic surgery leaves tissue which remains at risk for malignant transformation.

Prophylactic mastectomy and oophorectomy are emotionally charged areas of prevention, with strong proponents and critics within both professional and lay groups (Lynch and Watson, 1992; King et al, 1993; Stefanek, 1995). There is lack of research regarding the decision-making process about using these procedures and only sporadic case reports about the effectiveness of the procedures. Because genetic risk is rarely the only factor that goes into making a decision about surgery, there is a strong need for multidisciplinary input, and thorough psychosocial counselling regarding use of prophylactic surgery to prevent cancer (Peters, 1994b). For recent ELSI (ethical, legal, and social issues) recommendations, see Chapter 20.

Susceptibility test implications

Participants should be prepared for results of genetic susceptibility testing that are negative, positive, or inconclusive (see Tables 5.4, 5.5a and 5.5b).

The implications differ depending on several factors:

- (1) The test result.
- (2) Whether or not the person being tested has already had cancer.
- (3) Whether a cancer-causing mutation has already been identified in the family or this is the first person to be tested.

Table 5.4. *Possible implications of positive results of cancer susceptibility test*

| | |
|-----------------|---|
| Genetic | The mutation can be passed on to offspring Notify other relatives at risk |
| Medical | Explanation for cancers in self and/or relatives Increased lifetime risk of one or more cancers Specialised surveillance and prevention advised Risk to more than one organ system |
| Psychological | Psychological difficulties common during adjustment May alter self-perception |
| Social/economic | Family relationships can be strained or altered Insurance and employment discrimination Possible stigmatization |

Source: Adapted from Schneider (1994), with permission.

Participants can have much greater confidence that negative results are truly negative when a mutation is already known to occur in a given family than when one must be discovered and demonstrated to be causative of cancer. The clinical significance of a particular mutation found in a high risk research population cannot necessarily be generalized to different populations without such high a priori risks (Schatzkin et al., 1995).

Accuracy/limitation of testing

The counsellor will need to be very clear with the family at the outset as to what types of genetic alterations will and will not be found, and the implications of each. When results are negative, there is a possibility that a mutation was missed or the results unclear. A test may be inconclusive if a missense alteration cannot be distinguished from a benign polymorphism. If a mutation is found, it does not mean that the person will definitely get cancer, which type, or when.

Risks of genetic susceptibility testing

The main risks of testing are psychological, social, and economic. These are summarized in Table 5.6. The psychological reactions often start at the time of informed consent, with high levels of anxiety, depression, sleep and other somatic complaints documented one week following

Table 5.5a. *Implications of negative result of susceptibility test when a mutation is already known to be in the family*

| | |
|-----------------|---|
| Genetic | This person does not have the mutation seen in affected relatives; does not exclude other genetic alterations Cancers in this person have other causes, which may be genetic, environmental, multifactorial, or chance This person cannot pass on this mutation |
| Medical | Cancer risks return to baseline unless other risk factors exist Medical surveillance resumes population recommendations |
| Psychological | May be relief or guilt at being spared May be disbelief in result after a lifetime at risk |
| Social/economic | May create gap between relatives with and without mutation Insurance risk status may improve |

Source: Adapted from Schneider (1994), with permission.

Table 5.5b. *Implications of negative result of susceptibility test when the cause of familial cancers is unknown in the family*

| | |
|-----------------|---|
| Genetic | Cannot distinguish false negative from true negative The person could still be at risk for a different mutation Risk to offspring remains increased |
| Medical | Cancer remains elevated Continue high risk medical surveillance |
| Psychological | Uncertainty about genetic risk continues Unable to obtain closure May become disillusioned with testing after repeated negative results in the face of obvious increased risk |
| Social/economic | Family relationships may be affected Insurance and employment status remain vulnerable |

Source: Adapted from Schneider (1994), with permission.

Table 5.6. *Potential risks associated with positive results of cancer susceptibility testing*

| | |
|-----------------|---|
| Genetic | <p>May offer inaccurate risk information due to:</p> <ul style="list-style-type: none"> • inappropriately applying cancer risks derived from one population to another; • failing to account for gene-gene and gene-environmental interactions; • unrecognized variations in penetrance and expressivity |
| Medical | <p>Increased lifetime cancer risk estimates uncertain regarding: individual age of onset, type and number of tumours Problems obtaining adequate medical surveillance</p> |
| Psychological | <p>Altered sense of personal identity, self-esteem, mood, function</p> |
| Social/economic | <p>Couple and family relationships altered Confidentiality and privacy threatened Insurance and employment vulnerable Financial burden of extra surveillance</p> |

Source: Adapted from Schneider (1994), with permission.

pre-test counselling and remaining high for the interim months until test results are available (Grosfeld et al., 1996).

The risk of adverse social implications within the family are common (Biesecker et al., 1993). There is the risk of communication barriers going up between those who chose testing and those who declined testing, as well as between those who tested positive and those who tested negative. As a result, family relationships and dynamics could be altered. Grosfeld et al. (1996) found that asymptomatic carriers were often stigmatized within the family as already sick. As a result, the family became preoccupied with looking for symptoms or for re-evaluation of past events as symptomatic. Some individuals felt almost no relief when they learned that they were non-carriers, but rather, felt empty and isolated from family members who were carriers. Lynch and Watson (1992) noted that some of those who tested negative for *BRCAl* expressed disbelief and wished to continue with intensive surveillance and still consider prophylactic surgery.

Benefits of cancer susceptibility testing

Reduction of uncertainty for at risk individuals and increased compliance with screening recommendations are important benefits of testing

programmes (Petersen, 1996a,b). Some families have reported increased supportiveness and better communications (Giambarresi and Kase, 1995, pers. comm.; Grosfeld et al., 1996). Risks for offspring may be clarified. Testing may provide an increased ability to plan for the future, such as choice of career, geographic location near a medical centre, or planning a family. For those whose test results are negative in a family with a known mutation, results can be reassuring and release them from surveillance programmes.

The benefits are summarized in Tables 5.7a and 5.7b.

Table 5.7a. *Potential benefits associated with cancer susceptibility testing when results are positive for a mutation*

| | |
|------------------|--|
| Genetic | Have a better understanding of the biological basis of the cancer Provide accurate risk analysis for relatives |
| Medical | Alter cancer prevention and detection |
| Psychological | Resolution of uncertainty about risk status Explanation for cancer Improve motivation for healthcare Opportunity for active coping strategies |
| Social/economics | Increased supportiveness and communications Children can be started on healthy habits Motivation to plan for future |

Source: Adapted from Schneider (1994), with permission.

Table 5.7b. *Potential benefits associated with cancer susceptibility testing when no mutation is found in person from family with known mutation*

| | |
|-----------------|---|
| Genetic | Person does not have the mutation causing cancer in a branch of the family Children cannot inherit mutation from parent who does not bear it |
| Medical | Cancer risk decreases to general population risk in absence of other risk factors Reduce unnecessary medical surveillance |
| Psychological | Feelings of relief and elation Enables chance to get on with life |
| Social/economic | Increased communication and support Financial savings from unnecessary medical visits and procedures |

Source: Adapted from Schneider (1994), with permission.

Coping resources and strategies

It is important to discuss the anticipated impact of results upon the client and his or her family and to identify maladaptive coping styles before the decision to be tested is made. Adaptive coping approaches can then be encouraged.

Privacy and confidentiality

Because the dangers of discrimination are very real to families with hereditary cancer susceptibility, issues of privacy and confidentiality should be discussed. Confidentiality pertains to the treatment of information that an individual has disclosed in a relationship of trust and with the expectation that it will not be divulged without permission to others in ways that are inconsistent with the understanding of the original disclosure (see Chapter 3).

Test specifics

The genetic counsellor (or nurse) is often the one to discuss other more mundane matters regarding the testing programme, including fees, that testing requires a tissue specimen (usually blood), how the test will be done, when results will be available, at what points clients can change their minds or withdraw from the testing programme, and who to contact for questions and support.

Cancer susceptibility test notification

See Table 5.8. The way in which results are to be given should be negotiated between participant and counsellor prior to the risk notification visit. Results are usually given in person, preferably with a support person in attendance. Most counsellors agree that results should be given near the beginning of the session once it is established that the person still wishes to receive the results. The content of the session should again cover the genetic, medical, and psychosocial issues raised in the pre-test counselling (Biesecker et al., 1993). Discussions of children's risk for inheriting a mutation are appropriate at this time. The issue of testing children will be dependent largely on whether the disease is manifested in childhood and what medical benefits can be derived from testing.

Medical issues at this point switch from theoretical generalizations to being applied to the individual who has just learned that the cancer risk

Table 5.8. *Susceptibility test result notification*

-
-
- Consider family preferences regarding setting and format
 - Disclose in person when possible
 - Invite a supportive relative/friend
 - Confirm that person still wants results
 - Offer results in simple, direct way
 - Allow for emotional reactions
 - Encourage active coping, increased social support, stress management
 - Personalize genetic and medical information
 - Be prepared for issues arising during pre-test counselling to re-emerge
 - Re-visit privacy, confidentiality, insurance issues
 - Discuss short-term and long-term plans
-
-

has substantially increased or decreased based on the test results. It is important for those who have learned that they are likely to develop cancer to feel that they can be active in trying to lower their risks.

Dealing with the potentially strong psychological reactions to hearing the results of testing is one of the primary functions of this meeting (Biesecker et al., 1993; Lynch and Watson, 1992; Grosfeld et al., 1996). Some people might subsequently feel differently about themselves or lower their goals, or be too frightened to seek medical care.

It is often wise to provide written or tape-recorded information about the genetic and medical consequences of the test result, since participants may be too stunned at the time of the visit to absorb much information. Additional telephone contacts or referral to an appropriate support group are often helpful.

Follow-up

Because the emotional impact of receiving test results may vary widely from person to person and over time, a significant duration may be necessary to work through the implications of test results emotionally (Table 5.9).

No variables have yet been found to predict infallibly who will have long-term trouble in adjusting to results, which may be important whether the results are favourable or unfavourable. Studies are in process through the NIH-ELSI hereditary cancer consortium to assess this. One key predictor may be the extent to which the consultand equates a positive test result with cancer and death. Results of a prospective cohort

Table 5.9. *Susceptibility test counselling follow-up*

-
-
- Coming to terms with test result implications
 - Positive, negative, and inconclusive results can all generate emotional reactions
 - Short-term contact for acute reactions
 - Offer at least one year long-term contact post-test
 - Update personal and family histories
 - Check understanding of test implications
 - Review medical surveillance plans
 - Enroll in prevention trials when available
 - Consider reproductive implications
 - Notify appropriate relatives of testing options
 - Explore family relationships and communications
-
-

study of 279 adult men and women of families with *BRCAl*-linked hereditary breast–ovarian cancer showed that of the almost 200 persons who completed a baseline interview, genetic education, and counselling, 60% requested test results (Lerman et al., 1996). At one-month follow-up, non-carriers of *BRCAl* mutations showed statistically significant reductions in depressive symptoms and functional impairment compared with carriers and non-tested individuals. However, individuals identified as mutation carriers did not exhibit increases in depression and functional impairment. Among unaffected women with no prior prophylactic surgery, 17% of carriers intended to have mastectomies and 33% to have oophorectomies.

Grosfeld and colleagues (1996) counselling families undergoing MEN-2 testing found that 43% of participants expressed anxiety complaints, 34% depression, 37% had somatic complaints, and 49% had sleep disturbances two weeks after disclosure. Psychological distress remained for up to a year following disclosure, although the levels dropped significantly.

Petersen and colleagues have reported on testing of 47 adults and 36 minors at risk for FAP (Petersen and Boyd, 1995). There was some evidence that family relationships and identity was linked to gene status. The value of counselling included reduction of uncertainty and adjustment of misperceptions. Testing of minors presented additional counselling challenges; predictive genetic testing of 41 children at risk for FAP showed that at three-month follow-up, there were no significant changes in the levels of clinical depression or anxiety in the children or in their parents. However, mutation-positive children with affected mothers had

significantly higher depression scores at follow-up, and regardless of test results, children with affected mothers had significantly increased anxiety scores after testing. In families with both mutation-positive and mutation-negative children, FAP-unaffected parents experienced significantly increased depressive symptoms at follow-up.

In grief counselling, counsellors generally caution consultands to avoid making any irrevocable life decisions within the first few months following a loss or crisis because the strong emotional response to the test result itself may be colouring decisions at this time. The same principle may be applied to susceptibility testing.

Individuals with negative results are often reluctant to return for follow-up, but experience with Huntington disease has shown that they had the comparable rates of psychological distress at one year post-testing as did those with positive results. These observations have been confirmed in MEN-2 (Grosfeld et al., 1996). Thus, the provider should make every effort to maintain contact with all tested individuals.

Conclusion

There are many technical, ethical, and counselling issues yet to be resolved about hereditary cancer susceptibility. Some of the ethical dilemmas include establishing policy about testing of fetuses and minors for adult-onset risks; how to maintain the confidentiality of results without impinging on family or primary care patient relationships; how to handle desires for different levels and types of information with families; how to provide equitable access to information with geographically dispersed families and with different socio-economic groups (Biesecker, 1997).

Richards and colleagues (1995) have suggested areas of research that are needed: consultands' accuracy of perception of their own and their close relatives genetic risk; knowledge and understanding of inheritance of susceptibility genes; communication within families about genetic risk; family patterns in use of appropriate screening and risk reduction methods; effects of genetic counselling and of genetic testing on levels of worry and anxiety; establishing the ideal levels of worry and anxiety to energize positive health behaviours; selective use or avoidance of genetic testing by various sub-sets of the population; and effective use of resources in providing genetic services including counselling, education, and testing.

As knowledge about inherited susceptibility to cancer increases and additional susceptibility genes are identified, there will be an increased need for multidisciplinary teams to form in order to care for the medical,

genetic, and psychosocial needs of families with hereditary cancer syndromes. It is hoped that forming familial cancer risk counselling programmes now will offer not only a suitable context for susceptibility testing, but could also serve as a framework for many of the technological advances to follow in every aspect of cancer care, from surveillance to diagnosis, treatment, and follow-up.

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Edited by

WILLIAM D. FOULKES
AND SHIRLEY V. HODGSON

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