The field of genetics is in the news daily, with researchers mapping the human genome, cloning animals, and identifying new disease genes. Among those members of the population who have a family history of cancer, researchers have identified inherited alterations in the genetic code (gene mutations) that increase the likelihood of developing cancer. Some of these mutations increase risk only slightly, and others make an eventual diagnosis of cancer nearly inevitable. These latter types of mutations are said to cause hereditary cancer syndromes, the most common of which are Lynch syndrome, also called hereditary nonpolyposis colorectal cancer (HNPCC); familial adenomatous polyposis (FAP); and hereditary breast and ovarian cancer syndrome (HBOC).

The main objective of genetic risk assessment and counseling is to prevent cancers from occurring (or at least to discover them at their earliest stages, when they are most curable) by identifying individuals and families at increased risk. This is a multifaceted process by which physicians, genetic counselors, and nurses determine the likelihood of inherited disease in a family, provide education about cancer risks for patients and other members of the family, facilitate genetic testing when appropriate, provide recommendations for cancer screening and prevention, and offer psychological support.

Overview of Genetics and Inheritance
It is worthwhile to review some basic genetic definitions and principles before discussing the relationship of inherited gene mutations to cancer. The human genetic code is contained in a long molecule called DNA, and virtually every cell in the human body has this molecule of DNA. The DNA is arranged in forty-six chromosomes and holds approximately 20,000–25,000 genes. Prior to conception, the sperm and the egg cells, also called germ cells, each carry twenty-three chromosomes. Upon fertilization, the sperm and egg cell fuse, resulting in an embryo with a total of forty-six chromosomes. Of these, forty-four are identical in men and women—these are called autosomes. The remaining two chromosomes are called sex chromosomes, which are designated X and Y. Women inherit two X chromosomes, one from their mother and one from their father; whereas men inherit one X chromosome from their mother and one Y chromosome from their father. Because of the way chromosomes are inherited, every person has two copies of each gene: one copy from your father and one copy from your mother. When the genes from your parents are combined to make a child, the DNA is “shuffled.” This shuffling produces a brand-new, unique individual. The human genetic code is so complex that each person’s combination...
of their father’s and mother’s genes is unique. That is why, except for identical twins, none of the 6.5 billion residents of planet Earth is the same as any other.

Genes determine all physical characteristics, such as skin and eye color, and all bodily functions, such as metabolism and growth. In scientific terms, a gene is a segment of DNA that codes for the production of a specific protein. The DNA code is much like a recipe made up of only four letters: A, C, G, and T. These letters, called bases, are the individual chemical units that “spell” out the protein recipe. Any change in the sequence of bases is called a mutation. Just like changing a letter in a word can change the word’s meaning, a mutation can change the instruction contained in the gene. Proteins are responsible for carrying out all of the functions that keep cells alive and healthy, thus the accurate production of proteins depends on having normal genes.

When discussing inheritance of a disease, there are two genetic scenarios: monogenic inheritance, where there is only one gene responsible for the disease, and polygenic inheritance, where many genes contribute. Hereditary cancer predisposition syndromes are all examples of monogenic inheritance. There are two main types of monogenic inheritance: dominant and recessive. Dominant inheritance defines a situation where only one copy of a gene mutation needs to be inherited for the person to develop the disease—that is, either the mother’s copy or the father’s copy, but not both. Recessive inheritance, on the other hand, means that two copies of a mutant gene must be inherited for the disease to develop—one from each parent.

When a disease is dominantly inherited, each child of an affected parent has a 50 percent chance of inheriting the disease, because the affected parent has two copies of the gene, one normal and the other with a mutation. It is a matter of equal chance which copy from the affected parent goes into the sperm or the egg cell that produces a child. Familial adenomatous polyposis (FAP) is an example of a dominantly inherited cancer predisposition syndrome.

When a disease is recessively inherited, each parent must have at least one copy of the predisposing gene mutation and the child must inherit the gene mutation from both parents for the disease to occur. An individual with just one gene mutation is called a carrier and does not show the disease him- or herself. Each child of parents who are both carriers for a recessive gene mutation has a 25 percent chance of inheriting the disease. An example of recessively inherited cancer predisposition is MYH-associated polyposis (MAP), in which the inheritance of two copies of a gene mutation increases the risk for multiple colorectal polyps and colorectal cancer.

Gene Mutations and Cancer

Everyone acquires some mutations in their DNA during the course of their lives. These mutations occur in a number of ways. Sometimes there are simple copying errors that are introduced when DNA replicates itself. As our cells age and die, new cells must be made to replace them. For this to happen, each of the 3 billion letters of DNA that are found within every cell has to be copied, one letter at a time. Not surprisingly, mistakes sometimes occur. Other mutations are introduced as a result of DNA damage through environmental agents such as sunlight, cigarette smoke, and radiation. Cells have built-in mechanisms that catch and repair most of the changes that occur during DNA replication or from environmental damage. However, as the human body ages, DNA repair does not work as effectively, resulting in the accumulation of gene mutations.

The most important thing for the cell to do is to stop damaged DNA from
Genetic risk assessment and counseling

being passed on to the next generation of cells. If genetic damage is not too bad, it is repaired. If the damage cannot be repaired, typically the cell dies. However, sometimes genetic damage results in the inability of a cell to control its own growth and death. After accumulated damage to both copies of multiple genes in charge of cell growth and death, a cell can no longer be controlled and the result is cancer.

Hereditary Cancer Syndromes

In hereditary cancer predisposition syndromes, a gene mutation exists in one of the germ cells (egg or sperm) at the time of conception. This is called a germ line mutation.

Most of the time, the mutation is in the germ cell because the parent received it from one of his or her parents. Sometimes it is a new mutation, meaning that the change in the gene occurred during the production of that germ cell and neither parent has the mutation in their other body cells. In both cases, the mutation will be copied during the development of the new individual, so that it will be present in every cell of the body. A germ cell mutation in a gene controlling cell growth or DNA repair is the first in the sequence of mutations leading to the development of cancer.

Since multiple mutations within successive generations of a cell are required for cancer to appear, most of the hereditary cancers occur in the adult years. On average, hereditary forms of cancer occur about ten to twenty years earlier than nonhereditary cancers because of the first critical mutation in each cell.

Hereditary cancer syndromes have specific patterns of cancer occurrences...
within the family. In Lynch syndrome, one or more members of the family may have multiple primary cancers such as colon and endometrial cancers. In HBOC, there is an excess of bilateral cancer in paired organs such as the breasts and ovaries. Within the same family, the expression of the cancer gene can vary in terms of the age of onset and the organs affected. The risk of cancer for a person with a germ line mutation is high but not 100 percent. This phenomenon of less than 100 percent risk of cancer in those with hereditary predisposition is referred to as the penetrance of the gene.

In the majority of hereditary cancer syndromes, the pattern of inheritance is dominant. The one known exception to this rule is MAP, in which the inheritance of two copies of a gene mutation increases the risk for multiple colorectal polyps and colorectal cancer. Some syndromes are heterogeneic, meaning that they can be the result of a mutation in more than one gene. Lynch syndrome can be caused by a germ line mutation in the MLH1, MSH2, MSH6, or PMS2 gene. Likewise, hereditary breast and ovarian cancer is caused by a germ line mutation in either the BRCA1 or BRCA2 gene.

The table on the following page lists some of the more common hereditary colon and breast cancer syndromes, the genes associated with them, and the clinical features of each.

**Cancer Risk Assessment**

Cancer is very common throughout the world. One out of three Americans will develop the disease at some point, so it should not be surprising that most people have a history of one or more cancer occurrences within a three-generation history of their families.

Most of these cancers will be sporadic or chance occurrences posing only a slight increased risk for cancer to those individuals’ closest relatives. In some families, the cancer may be familial; multiple genes (polygenic) and/or common environmental exposures are believed to be the cause of these cancers. Family members who are closely related to an affected individual have about two to three times the risk for the same cancer that the general population has. Only 5 to 10 percent of all cancers are truly hereditary.

The case of Mrs. B (page 406) will be used as an example to illustrate the clinical services provided during cancer risk evaluation, genetic counseling, and predictive testing.

**Family History** Mrs. B was a twenty-eight-year-old female who sought cancer risk assessment and genetic counseling services because of her concern about multiple occurrences of colon cancer in her family. Mrs. B’s mother (Mrs. H) had a history of colon cancer and her aunt had just died with colon cancer after surviving uterine cancer for ten years. Mrs. B’s maternal grandmother and a maternal uncle had also both died with cancer, but she didn’t know their ages or the sites of their cancers.

**Gathering Information** To determine the significance of the cancer occurrences in Mrs. B’s family, it was essential to gather the history on her parents, brothers, sisters, children, nieces, nephews, aunts, uncles, cousins, grandparents, and any other relatives for whom medical information was available. The information required for a genetic evaluation includes any significant health problems, the primary site of all cancer occurrences, the age of diagnosis, current age or age at death, and cause of death, and exposures to carcinogens such as high-fat diets or tobacco.

Since Mrs. B did not know the medical history for all of these relatives, she was encouraged to ask her mother and her maternal uncle’s widow to fill in the gaps. In many families, there is a “family historian” who can provide information
about previous generations. In others, information may have been recorded in the family Bible or a genealogy.

Families vary in how open they are to discussing health issues. Some individuals in Mrs. B's family were very reluctant to share information with her, possibly because of an inclination for privacy, a distrust of medical professionals, an avoidance of the emotional context of the family's history, or a fear of what might be learned from the evaluation.

**Documenting the History** Medical records, especially pathology reports for all reported cancers, are essential for identifying or verifying the primary site of the cancer. In addition, there are specific pathologic features of some tumors that are more frequently found in hereditary cancers.

Mrs. B, therefore, was asked to request a release of medical records from her mother, who had a history of cancer. Mrs. B asked her mother, cousin, and aunt (her maternal uncle's widow), as the nearest surviving relatives, to sign the release forms for her grandmother's, aunt's, and uncle's records. Unfortunately, the hospital where her grandmother was treated had destroyed the records and a death certificate was requested in hopes of

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### Common Hereditary Colon and Breast Cancer Syndromes

<table>
<thead>
<tr>
<th>SYNDROME</th>
<th>GENE(S)</th>
<th>CLINICAL FEATURES</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lynch syndrome (HNPCC)</td>
<td>MLH1, MSH2, MSH6, or PMS2</td>
<td>Up to 80% lifetime risk for colorectal cancer in males, somewhat less in females, and 20–60% lifetime risk for endometrial (uterine) cancer. In some families, the risks for cancers of the stomach, ovary, urinary tract, kidney, hepatobiliary tract, skin, and brain are increased.</td>
</tr>
<tr>
<td>HBOC</td>
<td>BRCA1</td>
<td>50–85% lifetime risk for breast cancer in women, and 20–50% risk for ovarian cancer. An increased risk for male breast cancer and prostate cancer also exists.</td>
</tr>
<tr>
<td></td>
<td>BRCA2</td>
<td>50–85% lifetime risk for breast cancer in women, 6% risk for male breast cancer, and 10–30% risk for ovarian cancer. The risks for melanoma and prostate and pancreatic cancers are also increased in some families.</td>
</tr>
<tr>
<td>FAP</td>
<td>APC</td>
<td>Hundreds to thousands of colonic adenomas develop at a young age. Without treatment, the lifetime risk for colorectal cancer is close to 100%. Risks for cancers of the stomach, small bowel, and thyroid are also increased. Some families have an increased risk for benign tumors in the abdomen (desmoid), bony growths (osteomas), epidermoid cysts, extra teeth, and extra pigment in the retina of the eye (CHRPE).</td>
</tr>
</tbody>
</table>
identifying the primary site of her cancer. Mrs. B’s aunt declined the release of her husband’s records.

**Clinical Evaluation** The clinical evaluation of the significance of the family history usually consists of a review of the individual’s medical history and the family history and a physical examination. It’s often helpful for the individual seeking the evaluation to bring someone along to provide emotional support or help clarify the family history. Several family members may choose to come together so they can provide and receive information at the same time. Mrs. B brought her husband and her mother with her for the evaluation.
While reviewing the history with the family, the genetic counselor also assessed their level of knowledge about cancer, the impact of early cancers and deaths in the family, and their perception of Mrs. B’s risk for cancer. The information they had gathered about the family history and the information obtained in the medical records were recorded in a pedigree, which made it possible to visualize the history and determine if there was a pattern to the cancer occurrences consistent with a hereditary syndrome.

**Diagnosis** A diagnosis of Lynch syndrome was suspected based on the pedigree analysis, medical history, and physical examination.

While the records on all family members were important because they provided information about the primary sites of cancers, the records of Mrs. B’s aunt’s first cancer occurrence were especially helpful because they provided documentation about the primary site of her uterine cancer (endometrium), and endometrial cancer is a common site in Lynch Syndrome.

**Genetic Counseling**

Genetic counseling has been defined as a communication process related to either a medical condition or the risk of its recurrence in which information about the medical and genetic facts is provided and assistance is offered with decision making and adapting to the situation.

Mrs. B was told that the colon and endometrial cancers in her family were most likely due to a mutation in one of the genes known to cause Lynch syndrome. The family was informed that genetic testing might provide the possibility of identifying gene carriers before they were affected with cancer.

**Predictive Testing** Because of the potential repercussions, DNA testing for a hereditary cancer syndrome is not like most other blood tests. It requires careful consideration by the individual to be tested. Testing is not offered for minors unless there is some definite benefit to be derived, such as in FAP, where there is an increased risk for cancer in childhood.

Another unique aspect about DNA testing is that it is best initiated by a family member who is affected with cancer. While genetic testing for hereditary cancer predisposition is quite accurate, it will not always be informative. This is because of limitations in genetic testing technology, or because a mutation exists in a cancer predisposing gene for which a genetic test is not yet available. This type of testing can help to pinpoint the gene mutation that is responsible for the cancer in the person who has a diagnosis of cancer. Without knowledge of the mutation in the family, a negative result for an unaffected member could mean that the individual did not inherit the mutation, but it could also mean that the mutated gene in the family was beyond the scope of the testing and that the individual is still at high risk of cancer. Figure 3 shows the process of predictive testing.

**Informed Consent** To ensure the informed consent of the person to be tested, the counselor discusses all aspects of the testing process and the potential impact of the results.

The individual is informed about the test and the limitations of the knowledge that may be derived from the result. For example, a negative result does not equate with no risk because the individual still has the same risk for cancer as the general population. A positive result does not provide any indication of when cancer will occur, if it does. And, although there are laboratory safeguards, there is a chance of technical or human error during the testing. There also is the chance that the result will be inconclusive if the
Figure 3. Flow Diagram of Predictive Testing

Gather Information
1. Oral history of cancer in family  
2. Have patient investigate history where not well known  
3. Request medical records where possible

Counseling about
1. Risks and benefits  
2. Limitations of predictive testing  
3. Meaning of potential test results

Personal and family history do reflect potential hereditary risk

Patient decides to test at this time
Informed consent and blood draw
Counseling session to interpret results and discuss implications for patient and specific family members

True negative
Mutation known to exist in the family; patient does not have the mutation
Implications for at-risk relatives
Discuss implications of family history for other family members. Recommend genetic counseling (with or without) testing for at-risk family members
Review suggested ACS guidelines

Uninformative negative
No mutation known to exist in the family; patient does not have a mutation

Issues
1. future testing options
2. medical management options
3. implications for relatives
4. potential causes for cancers in family
5. long-term follow-up
Develop surveillance program based on family history of cancer

Positive
Patient has a mutation

Issues
1. prophylactic options
2. medical management options
3. implications for relatives
4. potential causes for cancers in family (not deleterious mutation)
5. long-term follow-up
Discuss NCCI guidelines for mutation carriers

ACS = American Cancer Society Guidelines
family has a mutation that cannot be identified with the test performed.

The counselor will support the individual’s right to make the best decision for him- or herself, although there may be pressure from one or more family members to do the opposite of what that individual wishes. A difficult dilemma may arise if a parent without a history of cancer does not want to know his or her status, but an offspring wants to know his or hers. A positive result for the offspring would reveal the parent’s status as a gene carrier. In such a situation, family members are encouraged to discuss their positions and come to a mutually satisfying decision.

**Costs** DNA testing is expensive, often costing $3,000 or more for testing to identify the mutation in the family. Once the mutation is identified, other people in the family can usually be tested for $500 or less. Most insurance companies cover the expense of DNA testing.

Some individuals have questions about health insurance discrimination. To date, there have been no well-documented cases of health insurance discrimination based on results from hereditary cancer genetic testing. The Health Insurance Portability and Accountability Act (HIPAA) protects individuals in group health insurance plans by prohibiting the use of genetic test results to determine eligibility for or rates/cost of health insurance. Most states have additional laws that protect patients from genetic discrimination. Concerns about the potential for genetic discrimination and the use of the results should be discussed as part of the informed consent process.

The potential risks and benefits of both positive and negative results are also part of the informed consent discussion.

**Making the Decision** Mrs. B and her mother were encouraged to anticipate the impact of a positive result, a negative result, and a decision not to be tested and, in the process, to consider a number of questions:

- Do I cope better with a degree of uncertainty or with all the knowledge available to me?
- If my result is negative, will I feel guilty that others in the family have not been as fortunate?
- If my result is negative, will I feel left out because of the family focus on the risk or occurrence of cancer?
- If my result is positive, will I be able to deal with any sadness or depression?
- If my result is positive, will the fear of cancer interfere with my ability to follow through with the recommended surveillance plan?
- What impact will my results have on the important relationships in my life?
- Will the information gained allow me to consider medical options that are not available or relevant now?

Mrs. H wanted to proceed with the testing for the benefit of her children and her unaffected sister, nieces, and nephews.

**After the Results** For someone with a history of cancer, receiving a positive result may have different implications than for a family member without such a history. When informed that her test result identified a gene mutation, Mrs. H said she was relieved there would be a better chance for early identification of cancers in the next generation and also talked of her fears for her children who may have received the mutation from her. She then made plans for informing her family.

Genetic counseling includes working toward the best possible adjustment to each situation. For the genetic counselor, this aspect of the counseling process may include providing a supportive presence when times are difficult, helping to negotiate and resolve diverse opinions within a family, and providing referrals to social
workers, psychologists, or support groups. For the patient, it may mean an explanation for the personal and family history, and assistance with understanding and coming to grips with the implications of the test result.

For others, there may not be an answer, but by working together, there may be additional testing or new information that will provide answers for these other high-risk families in the future.

**Recommendations for High-Risk Individuals**

The ultimate goal of risk assessment and predictive testing is to reduce the morbidity and mortality of cancer in high-risk families. Most of the interventions recommended for this purpose are known to reduce cancer morbidity and mortality. However, a few are unproven at this time, and studies of their effectiveness are under way.

**HBOC** The current breast cancer screening recommendations for females at risk or known to have a BRCA1 or BRCA2 mutation include breast self-examination every month starting in the late teens and clinical breast examination by a health care provider twice a year. Annual mammography is recommended starting at age twenty-five and alternating with breast MRI, so that one type of scan is completed every six months.

Risk-reducing mastectomy is an option for the woman who considers the breast cancer risk to be too high to consider surveillance alone. Other women consider risk-reducing mastectomies if cancer risk is interfering with quality of life or if they have breasts that are difficult to evaluate by exam or mammography. Studies indicate there is a 90 percent reduction in breast cancer risk among women with a BRCA1 or BRCA2 mutation who have risk-reducing mastectomies. There are

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**Factors in the Informed Consent Decision**

<table>
<thead>
<tr>
<th>BENEFITS</th>
<th>POTENTIAL RISKS</th>
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<tr>
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<td>Regret about decisions made based on the assumption of increased risk</td>
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<td>Avoidance of cost and discomfort of unnecessary surveillance</td>
<td>Survivor guilt</td>
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<td>Knowledge for making plans for the future</td>
<td>Changes in family relationships</td>
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<td>Feelings of alienation from the family</td>
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<td>Positive Results</td>
<td></td>
</tr>
<tr>
<td>Targeted surveillance recommendations leading to early detection and treatment</td>
<td>Negative psychological impact (depression, anxiety, fear)</td>
</tr>
<tr>
<td>Information for making decisions about chemoprevention or preventive surgery options</td>
<td>Changes in personal relationships</td>
</tr>
<tr>
<td>Improved opportunity to plan for the future</td>
<td>Worry and guilt about potentially passing the mutation to one's children</td>
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<tr>
<td>Increased compliance with surveillance</td>
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<tr>
<td>Relief from uncertainty</td>
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**Table:**

- **Negative Results:**
  - Relief from anxiety and fear of increased risk of cancer
  - Avoidance of cost and discomfort of unnecessary surveillance
  - Knowledge for making plans for the future

- **Positive Results:**
  - Targeted surveillance recommendations leading to early detection and treatment
  - Information for making decisions about chemoprevention or preventive surgery options
  - Improved opportunity to plan for the future
  - Increased compliance with surveillance
  - Relief from uncertainty

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also chemoprevention trials using various agents such as tamoxifen (Nolvadex) and raloxifene (Evista) for women at high risk for breast cancer.

Ovarian cancer screening includes twice-yearly transvaginal ultrasound and cancer antigen 125 (CA-125) screening starting at age twenty-five. This screening is not as effective as breast cancer screening in detecting early-stage cancer, so mutation carriers are counseled to consider risk-reducing removal of the ovaries (oophorectomy) and fallopian tubes at age thirty-five or after they have had their children. It is recommended that the risk-reducing salpingo-oophorectomy surgery include a protocol of washings and fine sectioning of all of the tissue in the ovaries and fallopian tubes to rule out the possibility of a microscopic cancer.

**Lynch Syndrome** It is currently recommended that men and women at risk for Lynch syndrome have their first colonoscopy at age twenty (or ten years younger than the earliest colon cancer in the family, whichever is sooner), then repeat it every one to two years. Over the age of forty, annual colonoscopy is advised. Numerous studies have shown that regular colonoscopy can prevent colorectal cancer. Individuals with Lynch syndrome who are diagnosed with colorectal cancer are encouraged to consider colectomy (removal of the entire colon) when they undergo colon cancer surgery. For some individuals in whom colonoscopy is burdensome, removal of the colon prior to a diagnosis of cancer (called prophylactic colectomy) may be considered.

Women with Lynch syndrome also require annual screening for gynecologic cancers. While these gynecologic screening tests are not proven to prevent cancer, it is hoped that they will help identify cancers when they are at their earliest stages and therefore most curable. Gynecologic cancer screening includes annual biopsy of the endometrium, transvaginal ultrasound, and CA-125 blood tests beginning at twenty-five to thirty years of age. Women who are finished with childbearing are encouraged to consider surgical removal of the uterus and ovaries to prevent gynecologic cancers. Women with Lynch syndrome who are diagnosed with colorectal cancer may also consider hysterectomy and oophorectomy at the time of their colon cancer surgery.

Surveillance for other cancers that occur in Lynch syndrome are not routinely recommended unless there is a known family history of that type of cancer. For example, upper endoscopy is recommended if there is a family member with stomach or small intestine cancer. Chemoprevention trials utilizing agents such as aspirin and celecoxib (Celebrex) have shown mixed results in patients with Lynch syndrome. There is an active effort to identify additional chemopreventive agents for both colon and gynecologic cancers.

**Familial Adenomatous Polyposis (FAP)** Screening for FAP is initiated with annual colonoscopy by ten to twelve years of age. Upper endoscopy for polyps in the stomach and small intestine is performed every one to three years once colon polyps begin to develop. Prophylactic total colectomy (removal of the colon prior to a diagnosis of cancer) should be performed in all patients when the number and pathology of the polyps becomes unmanageable by colonoscopy. Most patients undergoing total colectomy are able to maintain normal bowel function through either preservation of the rectum or creation of a J-pouch (ileal pouch reconstruction), where the small intestine is used to create an internal reservoir for collecting stool. Following surgery, individuals with FAP still require surveillance of the rectum or pouch, stomach, and duodenum.

Individuals with FAP should also have annual thyroid exams. Chemoprevention
trials with sulindac (Clinoril) have shown some promise of preventing the development of polyps, and additional trials with celecoxib are ongoing.

**Patient and Professional Responsibilities**

Throughout cancer risk assessment, counseling, and predictive testing, the patient and the professional share responsibility for both the process and the outcome.

**Patient Responsibilities**

- Gather the family-history information.
- Assist in the collection of medical records.
- Ask for clarification or for information to be repeated as necessary.
- Weigh the options and make decisions about genetic testing, use of the genetic knowledge, prophylactic surgeries, etc.
- Share experiences and responses, and allow others to provide support.
- Communicate with family members about the outcome of genetic risk assessment.

**Professional Responsibilities**

- Listen and hear what the patient is saying and asking.
- Take the time required to give both information and anticipatory guidance about risk reduction and surveillance.
- Present information in understandable terms.
- Provide a balanced view of options, pros, and cons, when the patient has a choice to make.
- Support the decision of the patient.
- Preserve and protect privacy and confidentiality.
- Discuss the implications for other family members and facilitate family communication.
- Provide referrals when the patient’s needs are beyond the professional’s realm of practice.

**Future Directions**

The genetic advances of the last twenty years have provided the ability to predict accurately the risk of cancer for members of families with hereditary cancer. Genetic counseling is an integral part of the testing process to reduce the risk of harm and enhance the potential for benefit. It is also instrumental in facilitating the assessment of cancer risk and developing recommendations for other family members.

Further research is needed to improve the genetic risk assessment and counseling process, identify cancer risks associated with specific mutations, and identify the effectiveness of some surveillance and prevention measures as well as develop new ones. The results of a few studies indicate that some high-risk individuals are not following through with the recommendations for cancer screening. Research is needed to identify the most effective ways of overcoming the barriers to screening for these individuals.

More must also be done to assure affordable access to counseling, DNA testing, risk reduction, surveillance, and treatment measures for high-risk individuals. Without access for all, there will be no hope of reaching the ultimate goal of cancer prevention through the identification of all families at increased risk for cancer.