

# GENETIC COUNSELING FOR CANCER

## *A Colorado Facility Brings Genetic Testing and Assessment to Its Community*

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**I**t is an unfortunate fact of life that many women live in fear of contracting cancer, particularly breast or ovarian cancer. Genomic medicine is beginning to change this, however. Women who undergo genetic counseling and subsequent gene testing receive answers that dispel these fears and allow them to make decisions directly related to their personal situations. The Hereditary Cancer Service (HCS), at Penrose Cancer Center, Colorado Springs, CO, offers such counseling and testing.

### CANCERS AND THEIR THERAPIES

Two pairs of genes, BRCA1 and BRCA2, are involved in the normal control of breast cancer growth. Indeed, an abnormality in the BRCA1 or BRCA2 tumor-suppressor genes indicates a high lifetime risk of developing not only breast cancer but ovarian cancer as well. These cancers have a much greater chance of occurring early in life than do others.<sup>1</sup> Women who have positive results in genetic testing can choose to have their ovaries removed, thereby drastically reducing their risk of developing ovarian cancer.<sup>2</sup> They may also choose to undergo prophylactic mastectomy, which significantly reduces the risk of breast cancer, or to increase their vigilance through regular mammography or MRI screens of the breast.<sup>3</sup> Women who test negative, on the other hand, will learn that their risk levels are the same as that of the general population. Such knowledge should reduce anxiety, especially in women who have a family history of the disease.

The same is true of testing for colon cancer. People who carry a mutation in the APC gene, which causes familial adenomatous polyposis (FAP) should undergo flexible sigmoidoscopy or colonoscopy every year. Children with a parent who has FAP should begin these procedures at

age 10. The authors know a man who has a faulty APC gene and is the father of four children. The oldest child, a girl aged 10, recently was tested and found not to carry the mutation. Since she did not inherit the faulty gene from her father, her risk for colon cancer is no higher than that of anyone else in the general population. Her parents were greatly relieved to learn that she will neither have to live in fear of developing FAP nor undergo annual screening procedures. The other three children will be tested soon. If they are shown to have inherited the faulty gene, they will have a nearly 100 percent chance of developing colon cancer, and a 93 percent chance of developing it before they turn 50.<sup>4</sup>

### HEREDITARY CANCER SERVICE

We began the HCS at Penrose Cancer Center in 1997.\* In the program's early stages, we focused on breast cancer genetics, counseling from 37 to 59 patients each year. In late 1999, we decided to expand the program, launching a concerted effort to reach more at-risk families. Last year, we counseled 151 new patients. The HCS has three tracks:

- **Education and Information** We provide cancer genetics information to physicians, patients, families, and the general public through community-wide seminars, physician conferences, health fairs, and other programs.

- **Risk Assessment/Pedigree Analysis** Following referral by a physician or self-referral, we collect and evaluate the patient's family history. On the basis of that history, we assess the risk to the patient of developing the disease and help him or



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\*Penrose Cancer Center is a member of Penrose-St. Francis Health Services, Colorado Springs, which is in turn a member of Centura Health, Englewood, CO, a member of Catholic Health Initiatives, Denver.



her develop a surveillance plan for it.

- **Testing** When indicated, we conduct genetic testing to confirm the presence of a hereditary cancer syndrome.

# The cost of gene testing is a primary concern for many prospective patients.

In 2000, the program received grants from the Colorado Springs affiliate of the Susan G. Komen Breast Cancer Foundation and the El Pomar Foundation. These grants allowed us to provide care for women who had concerns about their breast and ovarian family histories but lacked money for testing and counseling. These patients paid a nominal fee of \$20 for the first visit and \$15

for follow-up visits. The grant covered the remaining program expenses.

The annual cost of HCS operations is approximately \$70,000, which includes salary and benefits for a full-time genetic counselor. The program provides services primarily for the metropolitan Colorado Springs area. However, once a month the genetic counselor travels to Pueblo, CO, to offer services at St. Mary-Corwin Medical Center, a sister Centura Health facility.

## REFERRAL SOURCES

In 1997, when the HCS began, physicians referred 72 percent of patients. Today, 64 percent of referrals come from physicians, 18 percent are referrals from other health care providers, and 18 percent are self-referrals.

Sixty percent of HCS referrals from physicians come from the standard cancer specialties, surgery and medical and radiation oncology, which is evidence that the relatives of a person diagnosed with cancer have become concerned about their own chances of developing the disease. The relatives certainly benefit from turning to a genetics service. Still, society has a long way to go toward establishing a preventive approach to cancer.

This is especially true with regard to risk assessment for colon cancer. Until 2004, our program had only a few colon cancer referrals each year. Assuming that 5 to 10 percent of colorectal cancer is inherited and that 10 to 20 percent is familial, we would expect, in our service area, a minimum of 40 patients with an inherited colorectal cancer predisposition per year.<sup>5</sup> In addition, there

would be some 200 others with a significantly increased risk for colorectal cancer, based on family history. In our experience, unfortunately, few primary care physicians refer patients for colorectal genetic assessment and counseling. There is obviously a need for more physician education about preventive care.

Toward this end, the HCS has developed a one-page newsletter that we call *Genetic Fax*. Several times a year, we send it via automatic fax to both referring and nonreferring physicians. *Genetic Fax*, which is meant to provide physicians with a snapshot concerning a specific topic of genetics, is inexpensive to produce. Probably because the newsletter reminds physicians about the importance of assessing colon cancer risks, we typically see an increase in referrals for a time after the newsletter is sent out.

We also see a jump in referrals after Myriad Genetics representatives visit physicians in the Colorado Springs area. Myriad Genetics is a Salt Lake City-based biopharmaceutical firm that specializes in the development of diagnostic and therapeutic products. Both the *Genetic Fax* and the Myriad Genetics visits help build and maintain awareness of our program.

Marketing to the general public via educational presentations and advertising can lead to increased self-referrals. In 2001, we created a print ad that boosted breast and ovarian cancer referrals during that year. Today, our community is reasonably well educated about the benefits of genetic counseling for women who are at risk of developing breast and ovarian cancers. That is not the case with colon cancer. To close that gap, we plan to create an ad that stresses the importance of genetic counseling for colon cancer.

## PATIENT CONCERNS

A primary concern for many prospective patients is the cost of counseling and gene testing. In our community, grant funding has eliminated many of the financial barriers to breast and ovarian cancer counseling. In addition, most insurance programs now cover gene testing when counseling shows a high risk of cancer and indicates that testing would be beneficial. In both the general population and in certain minority populations, people express concerns about confidentiality, particularly about potential discrimination from employers or insurance companies if testing results should become known to them.

Because of such fears, few African-American women undergo counseling or gene testing. In 2002, the ANGEL (African-American women Nurturing and Giving Each other Life) network

was formed in Colorado Springs to provide African-American women clinical breast exams and inform them of the importance of having mammograms done. Thanks to ANGEL, referrals of African-American women to the HCS for genetic counseling have increased from five, for the years 1997-2001, to 30 in 2002 and 2003. Even with these referrals, however, only 11 families actually have pursued genetic counseling.

There is still a great deal of fear of discrimination due to genetic information—as well as a general distrust of the health care system—among African-American people. Additional work is needed to break down African-American distrust in particular, and distrust of genetic counseling in the general population.

#### A NEW ACCEPTANCE

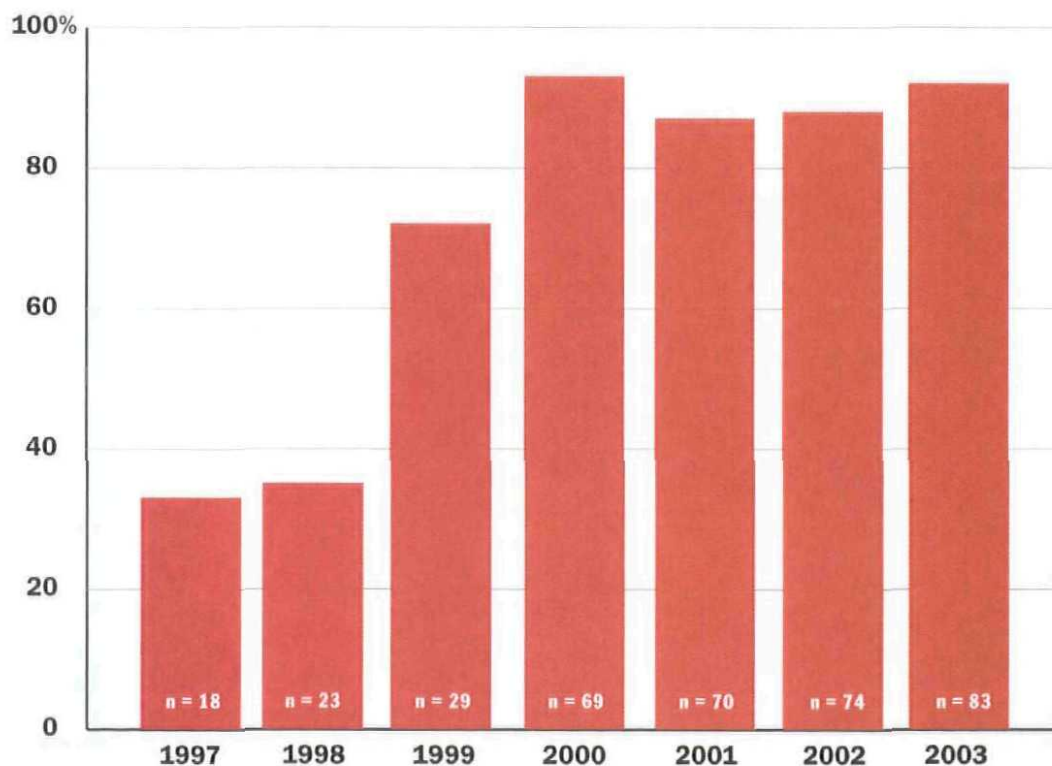
Of the patients our program sees, approximately 63 percent are eligible for genetic testing. The number of patients who actually decide to pursue such testing has increased significantly in

recent years (see **Graph**). In 1997, only 33 percent of those offered testing decided to pursue it after their initial genetic counseling appointment. Today, more than 90 percent of our patients choose testing. The exact reasons for this trend are unclear, but we suspect that it is due to increased awareness and acceptance of genetics on the part of the public, and to the fact that medical science now has data showing that screening and prophylactic surgical intervention can reduce breast and ovarian cancer mortality.

#### PATIENT SURVEYS

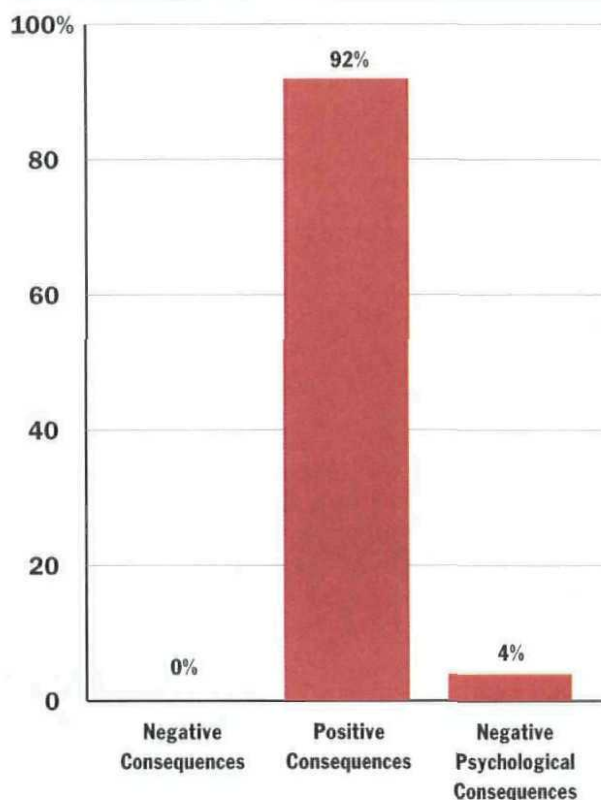
All of our patients who receive counseling are mailed a follow-up survey one year after their initial genetic counseling appointments. To date, we have had a 72 percent response rate. We preface each question with the phrase, "Since your initial appointment with the Hereditary Cancer Service . . ." The questions and responses are as follows:

### Percentage of Eligible Patients Who Pursue Gene Testing





## Perceived Consequences of Testing



*Have you made any changes in your breast cancer surveillance?*

Eighty percent of those respondents who had not already made such changes did so as a result of their breast cancer risk assessments.

*Has your anxiety level regarding your risk for cancer increased, decreased, or not changed?* One concern regarding genetic testing, counseling, and risk analysis is the possibility that a patient found to be at increased risk for cancer will experience increased anxiety. However, only five of our patients—a very small percentage—have reported increased anxiety regarding their risk for cancer. This is especially interesting considering that 59 percent of respondents said they believed that they had a moderate to significantly higher risk for breast cancer than other women their age.

*Have you made any lifestyle changes that you feel will help lower your risk for breast cancer?* Fifty-five percent said they had made healthy lifestyle changes since their genetic counseling. Thirty-seven percent said they

already had made such changes at the time of their counseling. Only 8 percent said they had made no changes.

*Have you experienced a negative psychological impact from your assessment?* One patient reported that she had experienced mild transitory depression. Two said they had had some difficulty with family members after informing them that gene testing had been performed and that a mutation was found. These results suggest that the negative psychological impact of gene testing is likely to be minimal.

The survey results stress the importance of making the genetic assessment team multidisciplinary. Such teams should include:

- An *oncology counselor* to provide individual and family counseling regarding the psychological impact of risk assessment and/or gene testing and the diagnosis and treatment of cancer
- An *oncology nurse educator* to provide any additional information the patient might need regarding the prevention, diagnosis, and treatment of cancer
- A *breast cancer coordinator* to provide patients with clarification of their cancer diagnoses, treatments, and reconstruction and prevention options, and to instruct them concerning optimal breast self-exam techniques

It is critical that the team communicate clearly with the patient's primary care physician, so that he or she can help counsel the patient.

The fact that few HCS patients report negative psychological consequences may be a result of our format, which involves risk analysis, in-depth informed consent counseling prior to testing, and post-test counseling sessions (see **Graph**).

In fact, 92 percent of our survey's respondents said they had positive consequences from their evaluation. Respondents said they felt relief and a certain measure of empowerment as a result of:

- Coming to a better understanding of the biology of cancer occurrence and development
- Having their risk of breast cancer clearly assessed and coming to understand the factors that contribute to an increased risk
- Understanding the screening and prevention options available and the risks, benefits, and limitations pertaining to these options
- Understanding how their particular family histories do or do not indicate an increase in the risk of breast cancer for other family members

None of the respondents reported negative consequences regarding discrimination by health or disability insurance carriers or by their employers.

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## "GOOD HELP" IN ST. PETERSBURG

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tance with necessary paperwork to enter training courses

- Placement in health-care related, entry-level training programs, such as CNA or LPN courses

- Resume preparation and interview coaching

- Assistance in employment placement in hospitals, nursing homes, or clinics

- Mentor programs to support refugees as they go through the training process and also after job placement

- Referrals to available community medical services

PREP's Corbin sees the RHP clientele has having changed dramatically since the program's founding. At that time, about 80 percent of the refugees served were Bosnians. (Clients also came from Colombia, Afghanistan, Cuba, Haiti, Liberia, the Ukraine, Guatemala, Moldavia, Vietnam, and Croatia.) By 2004, more than half of those served were Spanish-speaking Latin Americans, many of whom were asylees.

As noted, the RHP was initially funded through an \$80,000 grant from the Bon Secours Mission Fund. For its second and third years, the RHP received from the fund grants of \$99,698 (this included two RN scholarships) and \$75,585, respectively. The final Bon Secours Mission Fund grant expired in November 2004. However, the diocesan Catholic Charities agency has now received from the Department of Children and Families a \$450,000 "career-laddering" grant to help refugees advance in their new careers.

Thus the good work that was begun in 2001 with the Bon Secours Mission Fund grant has borne good fruit.

### LESSONS LEARNED

Those who have been involved in the RHP project say that several valuable

lessons have been learned from it.

- Refugees sometimes leave their new jobs because they feel overwhelmed by the work culture. Mentoring programs can help refugee employees feel welcome. And employers should consider assigning two refugees from the same country to the same work unit, so that they can give each other mutual support.

- Since September 11, 2001, it has taken immigration authorities longer to run background checks on refugees, thereby delaying vocational training for them. Florida has also had a shortage of teachers for RN and LPN classes, resulting in long waiting lists for those classes.

- Despite such problems, health care employers tend to find that refugees make excellent workers. Many who become CNAs and LPNs are better educated than native-born CNAs and LPNs (some were health care professionals in their own countries) and have a strong work ethic. □

### NOTES

1. 1998 Statistical Yearbook of the Immigration and Naturalization Service, which can be found at <http://uscis.gov/graphics/shared/aboutus/statistics/1998yb.pdf>, Table 36, p. 118. In 2003 the U.S. Immigration and Naturalization Service became the U.S. Citizenship and Immigration Services.
2. McKnight's Long-Term Care News, May 23, 2003; this publication can be found at [www.mcknightsonline.com/home/](http://www.mcknightsonline.com/home/).
3. American Health Care Association, *Results of the 2002 ACHA Survey of Nursing Staff Vacancies and Turnover in Nursing Homes*, Washington, DC, 2003, pp. ii, 5, available at [www.ahca.org/research/rpt\\_vts2002\\_final.pdf](http://www.ahca.org/research/rpt_vts2002_final.pdf).
4. American Health Care Association, p. 15.

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### THE ROAD AHEAD

It is increasingly obvious that genetics is the future of medicine. The leaders of Penrose Cancer Center and its parent organization, Penrose-St. Francis Health Services, are determined to bring high-quality genetic medicine to southern Colorado. Penrose representatives serve on the Genetics Advisory Committee of Catholic Health Initiatives (CHI), advising system leaders on how best to expand services to rural CHI facilities and offering guidance to other CHI cancer treatment facilities. Meanwhile, Penrose leaders are working to increase referrals for people at risk for colon cancer and to develop funding sources to support this and other program objectives.

The growth and acceptance of Penrose and the HCS indicate that genetic counseling and testing should be an integral part of a comprehensive oncology program in community hospitals. Bringing genomics to the community increases the quality of care and advances the hospital's mission. □

### NOTES

1. D. Ford, D. F. Easton, M. Stratton, et al., "Genetic Heterogeneity and Penetrance Analysis of the BRCA1 and BRCA2 Genes in Breast Cancer Families," *American Journal of Human Genetics*, vol. 62, no. 3, March 1998, pp. 676-689.
2. B. L. Weber, C. Punzalan, A. Eisen, et al., "Ovarian Cancer Risk Reduction after Bilateral Prophylactic Oophorectomy (BPO) in BRCA1 and BRCA2 Mutation Carriers," *American Journal of Human Genetics*, vol. 67, supplement 2, July 2000, pp. 59-66.
3. L. C. Hartmann, D. Schaid, T. Sellers, et al., "Efficacy of Bilateral Prophylactic Mastectomy in BRCA1 and BRCA2 Gene Mutation Carriers," *Journal of the National Cancer Institute*, vol. 93, no. 21, November 7, 2001, pp. 1,633-1,637.
4. See H. J. R. Bussey, *Familial Polyposis Coli: Family Studies, Histopathology, Differential Diagnosis, and Results of Treatment*, Johns Hopkins University Press, Baltimore, 1975.
5. Penrose Cancer Center registry, colorectal statistics, 1996-2001.

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