

Cancer Genetic Resources

WENDY H. VOGEL, MSN, FNP, AOCNP®, and DEBORAH F. PENCARINHA, MS, LGC

Human genetics is a complicated science. There are a multitude of terms to understand, such as cancer genetics, hereditary genetics, cellular genetics, genetic engineering, human genome, predictive genetic testing, Mendelian inheritance, and many more. If we, as advanced practitioners (APs) in oncology, are sometimes confused, imagine what our patients feel.

CANCER AND GENETICS

All cancer is genetic, but not all cancer is hereditary. In oncology, APs deal primarily with two types of genetic testing: tumor genetics and hereditary genetics. Tumor genetics is the study of acquired mutations of certain cells in the body. These mutations may cause abnormal cell growth and division, thus creating a tumor that continues to grow and invade other body tissues. These mutations cannot be passed to offspring, as they are only present in the cancerous cells. Some cancer treatments will target the cause of the harmful mutation. An example of this would be current treatments for chronic myelogenous leukemia, such as imatinib, which targets the *BCR-ABL* oncogene: the result of an acquired mutation.

Hereditary genetics is the study of mutations that are present in every cell of the body and thus may be passed on to offspring. Each of us has two copies of every gene: one from our mother and one from our father. We could inherit a defective gene

from our parent and in turn pass it on to a child. For example, the *BRCA1* gene functions as a cancer suppressor, protecting both men and women from breast cancer (and in women, from ovarian cancer as well). When a defective *BRCA1* gene is present, we have less protection. If the other *BRCA1* gene (inherited from our other parent) breaks (mutates), we no longer have protection against breast or ovarian cancer. Not everyone with a *BRCA1* mutation develops cancer, but the risk is significantly increased (up to 87% lifetime risk of breast cancer).

RESOURCES

As APs, we must have a basic knowledge of genetics. We help our patients understand the difference between the types of genetics and how that affects their disease and their treatment. We need to identify those patients and family members who would benefit from genetic counseling and testing. The partnership between the AP and the genetic counselor synergizes cancer genetics expertise with medical and clinical knowledge to further the best outcomes for the patient. In this article, you will find a collection of valuable Internet resources intended for us, as APs, as well as for our patients. The resources intended for patients appear grouped together on the last page of the article. Please feel free to reproduce the list for your patients or download a copy from the *JADPRO* website (www.advancedpractitioner.com).

Ms. Vogel is a nurse practitioner and Ms. Pencarinha is a genetic counselor, both at Wellmont Cancer Institute in Kingsport, Tennessee.

© 2013 Harborside Press®

Basic Human Genetics

Human Genome Project Information	web.ornl.gov/sci/techresources/Human_Genome/index.shtml US Department of Energy site describing the Human Genome Project and provides educational resources, fact sheets, and more
National Human Genome Research Institute	www.genome.gov/Health Information about genetics and genomics, rare diseases, patient care, resources, research, and genetics 101
The Tech Museum of Innovation: Understanding Genetics	genetics.thetech.org Stanford University website that contains basic genetic information, games, online exhibits, and a LISTSERV to ask a geneticist questions
University of Utah Genetic Science Learning Center	gslc.genetics.utah.edu Provides a learning and a teaching website on basic genetics

Books: Online and Print

Greco, K. E., Tinley, S., & Seibert, D. (2012). <i>Essential Genetic and Genomic Competencies for Nurses With Graduate Degrees</i> . Silver Spring, MD: American Nurses Association and International Society of Nurses in Genetics.	www.genome.gov/Pages/Health/HealthCareProvidersInfo/Grad_Gen_Comp.pdf Describes the 38 essential genetic and genomic competencies that inform the practice of graduate level nurses and summarizes key documents and processes used to identify these competencies
Consensus Panel on Genetic/Genomic Nursing Competencies. (2009). <i>Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators</i> , 2nd ed. Silver Spring, MD: American Nurses Association.	www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf Defines essential genetic and genomic competencies for all nurses; guide for nurse educators in the design and implementation of learning experiences that help students/learners/nurses achieve these genetic and genomic competencies

Peer-Reviewed Literature

<i>GeneReviews</i>	www.ncbi.nlm.nih.gov/books/NBK1116 Online publication with peer-reviewed articles on clinically relevant and medically actionable information on the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions
<i>Hereditary Cancer in Clinical Practice</i>	www.hccpjournals.com The official journal of the International Hereditary Cancer Centre; site provides open access to most articles; free registration is required

Professional Websites

American Academy of Physician Assistants	www.aapa.org Clinical policies on genetics, listing of resources, informational program called "Genetics in the Physician Assistant's Practice"
American Society of Clinical Oncology	www.asco.org E-courses on topics such as Genetics and Genomics for the Practicing Clinician and Genetic Testing in Oncology; policy statements
American Society of Human Genetics	www.ashg.org Video series on interpreting family health history, policy statements, current abstracts, and articles
International Society of Nurses in Genetics	www.isong.org Annual conferences, webinars, educational resources, ethical issues, position statements, public policy information, clinical protocols, and counseling checklists
National Society of Genetic Counselors	www.nsgc.org Genetic counselor locator, family history tools and resources, position statements, and parameters for referrals to genetic counseling
Oncology Nursing Society	www.ons.org Genetic information (including a cancer genetics online course), position statements, publications, and a special interest group

Cancer Genetics

Lynch Syndrome Screening Network

www.lynchscreening.net

Professional information on Lynch syndrome, genetic testing, resources, database, support groups, and research

National Cancer Institute (NCI): General Cancer Genetics Information

www.cancer.gov/cancertopics/genetics

Provides information on cancer genetics, testing, and syndromes; useful for both health-care providers and patients

NCCN Guidelines: Genetic/Familial High-Risk Assessment: Breast and Ovarian; Colorectal Cancer Screening

www.nccn.org/index.asp

Guidelines on screening, detection, and management of hereditary cancer syndromes, including hereditary breast and ovarian cancer syndrome, Li-Fraumeni syndrome, Cowden syndrome, Lynch syndrome, and familial adenomatous polyposis; free registration required

Cancer Risk Assessment Tools

Breast Cancer Genetics Referral Screening Tool

www.breastcancergenecscreen.org

Genetic hereditary breast and ovarian cancer risk assessment tool for health-care professionals to evaluate who should be referred for genetic counseling

BOADICEA (Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm)

ccge.medschl.cam.ac.uk/boadicea/modelWeb application for hereditary breast cancer risk assessment tool (*BRCA1* and *BRCA2*)

NCI: Breast Cancer Risk Assessment Tool

www.cancer.gov/bcrisktool

Tool using the Gail model to evaluate a woman's risk of developing breast cancer

NCI: Colorectal Cancer Risk Assessment Tool

www.cancer.gov/colorectalcancerrisk

Estimates risk for colorectal cancer in men and women between the ages of 50 and 85

NCI: Melanoma Risk Assessment Tool

www.cancer.gov/melanomariskscreen

Tool to estimate a person's absolute risk of developing invasive melanoma using the patient's self-reported history and a brief physical exam by a health-care professional

UT Southwestern Medical Center at Dallas: CancerGene

www4.utsouthwestern.edu/breasthealth/cageneDownloadable software tool that estimates *BRCA1*, *BRCA2*, *MLH1*, *MSH2*, and *MSH6* gene mutation probabilities; hypothetical pancreatic cancer gene mutation probability; and age-specific breast, ovarian, colorectal, endometrial, and pancreatic cancer probabilities**Searchable Databases**

Genetics Home Reference

ghr.nlm.nih.gov/

Consumer-friendly information about the effects of genetic variations on human health; searchable database on genetic disorders and genes; genetic information, glossary, and resources

National Institutes of Health: Genetic and Rare Diseases Information Center

rarediseases.info.nih.gov/gard

Searchable database for rare diseases with FDA-approved products for the disease, genetics resources, information on testing; has information in Spanish as well

NextGxDx

www.nextgxdx.com

Assists health-care professionals in finding the appropriate genetic test, comparing genetic tests, and finding a laboratory that performs the needed test; includes searchable database for disorders, genes, and genetic tests

OMIM (Online Mendelian Inheritance in Man)

www.omim.org

Online searchable comprehensive database of human genes and genetic disorders written and edited at Johns Hopkins University with input from scientists and physicians around the world

Patient Resources

BHD (Birt-Hogg-Dubé) Foundation	www.bhdsyndrome.org Information and support for an inherited genetic condition linked to benign skin growths, collapsed lung, and kidney cancers
Cancer.Net: Cancer Types	www.cancer.net/cancer-types Online information from the American Society of Clinical Oncology about cancer types, including most genetic related cancers
Centers for Disease Control and Prevention, Public Health Genomics	www.cdc.gov/genomics/famhistory/index.htm Activities, publications, and resources for patients and providers to collect and utilize family history information
Fertile Hope	www.fertilehope.org Reproductive information, support, and hope for cancer patients and survivors whose medical treatments may cause infertility
FORCE (Facing Our Risk of Cancer Empowered)	www.facingourrisk.org Nonprofit organization for individuals and families affected by hereditary breast and ovarian cancer
Genetic Alliance	www.geneticalliance.org Nonprofit organization website featuring information on genetic disorders, webinars, a searchable database, a family health history tool, and information about current legislation
Genetic Health	www.genetichealth.com Run by volunteer health-care professionals; provides basic genetic and genetic counseling information as well as information about various genetic conditions, including cancer, genetic discrimination, clinical trials, and genetic testing
The Gilda Radner Familial Ovarian Cancer Registry	www.ovariancancer.com National computer tracking system that stores data for women with two or more close relatives who have been diagnosed with ovarian cancer; education, information, and a helpline with peer support for women at high risk for ovarian cancer
My Family Health Portrait Tool	www.hhs.gov/familyhistory/portrait/index.html US Department of Health & Human Services' tool for collecting family medical history, available in English and Spanish; patients can print and bring to their health-care provider
MyGenerations (North Shore University Health System)	www.northshore.org/genetics/mygenerations Tool for documenting family cancer history and determining risk of developing cancer through building a family tree; patients can print and bring to their health-care provider
No Stomach for Cancer	www.nostomachforcancer.org Education about stomach cancer, including hereditary diffuse gastric cancer; support for affected families; promotes screening, detection, treatment, and prevention of stomach cancer
Ovarian Cancer National Alliance	www.ovariancancer.org Advocacy organization for women with ovarian cancer
Ovations for the Cure of Ovarian Cancer	www.ovationsforthecure.org Public awareness and patient programs
Right Action for Women, Christina Applegate Foundation	www.rightactionforwomen.org Aid for individuals who are at increased risk for breast cancer and do not have insurance or the financial flexibility to cover the high costs associated with breast screenings
Previvors and Survivors	previvorsandsurvivors.com/forum Discussion forum for (and by) women with a genetic predisposition for breast cancer; also provides links to relevant cancer information
von Hippel-Lindau (VHL) Family Alliance	www.vhl.org Information and support for VHL for the benefit of patients, caregivers, researchers, clinicians, and the general public
Young Survivor Coalition	www.youngsurvival.org Resources and support for young women facing breast cancer