Illinois Department of Public Health

BRCA1 and BRCA2: Information for Illinois Health Care Professionals

How common are BRCA1 and BRCA2 mutations?

- In the general population, between one in 300 and one in 800 individuals carry a BRCA1 or 2 or mutation.[1,3]
- For individuals of Ashkenazi Jewish ancestry, one in 40 individuals carry a BRCA1 or 2 mutation.

What percentage of breast and ovarian cancers are caused by BRCA1 or BRCA2 mutations?

- 5 percent-10 percent of all breast cancer.
- Up to 15 percent of ovarian cancers.[1-4]

What are the lifetime cancer risks associated with BRCA1 and BRCA2 mutations?

- Breast cancer: 44 percent-87 percent
- Ovarian cancer: 20 percent-50 percent
- Male breast cancer and pancreatic cancer: ~7 percent

Which patients should I refer to a genetic counselor for risk assessment and discussion of genetic testing for BRCA1 and BRCA2 mutations?

Patients who have a personal or family history (this includes maternal or paternal lineages) or those who have had:[1-3,5]

- Breast cancer at a young age (younger than age 50)
- Ovarian cancer at any age
- Bilateral breast cancer or 2 or more primary tumors of the breast
- Breast and ovarian cancer
- Breast cancer and are male
- Two or more biological relatives* on the same side of the family with breast and/or ovarian cancer (*parent, child, siblings, grandparent, grandchild, aunt/uncle, niece/nephew, first cousin)
- Breast or ovarian cancer and are of Ashkenazi Jewish ancestry
- Confirmed BRCA1 or BRCA2 mutation in their family

Specific indications for genetic counseling and testing vary among professional organizations. The indications above are to be used as a guide and are not a substitute for clinical judgment. Not all clinical scenarios can be anticipated such as when there is a limited family structure or family medical history.

What are the benefits of genetic counseling when offering BRCA1 and BRCA2 testing?

- Patient education and informed consent are critical aspects of the genetic testing process and will help individuals decide if genetic testing is right for them.
- Pre-test counseling addresses the implications of potential test results, medical management options, psychosocial challenges, laws protecting against genetic discrimination, other options for testing and testing costs. Examples include:
    - The possibility of false negative results because genes other thanBRCA1 and 2 may be responsible for cancers in the family
    - The possibility of test results with mutations noted to have "uncertain clinical significance"
    - The cost of BRCA gene testing differs depending on which level of testing is indicated ($440-$3,770)
    - The possibility of indirectly disclosing non-paternity, non-disclosed adoptions or use of egg or sperm donation

- If help is needed, a genetics health care professional also can provide assistance with dissemination of relevant information to at-risk family members.

Resources

Genetics health care professionals:

- Illinois Department of Public Health (http://www.idph.state.il.us/HealthWellness/genetics.htm)
- National Society of Genetic Counselors (www.nsgc.org)
- National Cancer Institute (www.cancer.gov/search/geneticsservices/)
- National Center for Biotechnology Information (www.ncbi.nlm.nih.gov/sites/genetests/clinic)

BRCA1 and BRCA2 referral, testing and risk management guidelines:

- American College of Obstetrics and Gynecology (ACOG)
  - ACOG Practice Bulletin. Obstet Gynecol. 113(4):957-966. 4/2009
- American Society of Clinical Oncology (ASCO)
  - JCO Vol 21(12):2397-2406. 6/2003.
- National Cancer Institute (NCI)
  - www.cancer.gov/cancertopics/pdq/genetics/breast-and-ovarian/healthprofessional
- National Comprehensive Cancer Network (NCCN)
  - www.nccn.org/professionals/physician_gls/PDF/genetics_screening.pdf
- U.S. Preventative Services Task Force (USPSTF)
  - www.ahrq.gov/clinic/uspstf/uspsbrgen.htm

Patient friendly facts about genetic testing for breast and ovarian cancer risk

- U.S. Centers for Disease Control and Prevention
  - www.cdc.gov/genomics/resources/diseases/breast_ovarian_cancer.htm
- Facing Our Risk of Cancer Empowered (FORCE)
  - www.facingourrisk.org
- National Cancer Institute
  - www.cancer.gov/cancertopics/genetic-testing-for-breast-and-ovarian-cancer-risk
- National Institute of Health – Genetics Through a Primary Care Lens: A Web-Based Resource for Faculty Development
  - www.genetests.org

References

1. Berliner, J.L. et al. (2007) Risk assessment and genetic counseling for hereditary breast and ovarian cancer: Recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling.* 16:241-260.
2. U.S. Centers for Disease Control and Prevention. Located September 4, 2009, at http://www.cdc.gov/genomics/training/perspectives/factshts/breastcancer.htm.
3. National Cancer Institute. *Genetic Testing for BRCA1 and BRCA2: It's Your Choice.* Located September 4, 2009, at http://www.cancer.gov/cancertopics/factsheet/Risk/BRCA/print?page=&keyword
4. Pinsky, L.E., et al. (2001) Why should primary care physicians know about breast cancer genetics? *Western Journal of Medicine.* 175:168-173.
5. Guidelines listed in Resources above.