

# City of Hope Division of Clinical Cancer Genetics Publications



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## 2014

1. Antoniou AC, Casadei S, Heikkinen T, Weitzel, JN, ...et al. **Inherited loss-of-function mutations in PALB2 and breast cancer risk.** New Engl J Med. 2014 [In Press].
2. Gronwald J, Robidoux A, Kim-Sing C, Weitzel, JN, ...et al. (2014) **Duration of tamoxifen use and the risk of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers.** Breast Cancer Res Treat. 146(2):421-427.
3. Hernandez JE, Llacuachaqui M, Palacio G, ...Weitzel, JN, Narod, S. (2014) **Prevalence of BRCA1 and BRCA2 mutations in unselected breast cancer patients from Medellin, Colombia.** Hered Cancer Clin Pract. 12(1):11.
4. Nelson DE, Faupel-Badger J, Phillips S, ...Weitzel, JN. (2014) **Future directions for postdoctoral training in cancer prevention: insights from a panel of experts.** Cancer Epidemiol Biomar. 23(4):679-683.
5. Osorio A, Milne RL, Vaclová T, Pita G, Weeman K, CIMBA. **DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers.** PLoS Genetics. 2014;10(4):e1004256.
6. Thompson BA, Spurdle AB, Plazzer JP, ...Espenschied, et al. **Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database.** Nat Genet. 2014;46(2):107-115.
7. Wain KE, ...Weitzel, JN, ...Lindor N. (2014 ePub ahead of print] **Appreciating the broad clinical features of SMAD4 mutation carriers: a multi-center chart review.** Genet Med. PMID: PMC Journal - In Process.
8. Wood M, Kadlubek P, Pham T, Weitzel, JN, ...et al. (2014) **Quality of cancer family history and referral for genetic counseling and testing among oncology practices: a pilot test of quality measures as part of the American Society of Clinical Oncology quality oncology practice initiative.** J Clin Oncol. 32(8):824-829.

## 2013

1. Ashing-Giwa K, Rosales M, Lai L, Weitzel JN. (2013) **Depressive Symptoms in Latina Breast Cancer Survivors.** Psycho-oncology, 22(4):845-853.

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3. Couch FJ, Wang X, McGuffog L, ...Weitzel, JN, ...et al. (2013) **Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk.** PLOS Genetics. 9(3):e1003212. PMID: PMC3609646.
4. Culver JO, Brinkerhoff CD, Clague J, K. Y, Singh K, Sand SR, Weitzel JN. (2013) **Variants of Uncertain Significance in BRCA Testing: Evaluation of Surgical Decisions, Risk Perception, and Cancer Distress.** Clin Genet. 84(5):464-472. PMID: PMC3751990.
5. Finch A, Valentini A, Greenblatt E, ...Weitzel, JN, et al. (2013) **Frequency of premature menopause in women who carry a BRCA1 or BRCA2 mutation.** Fertil Steril. 99(6):1724-1728.
6. Gaudet M, Kuchenbaecker K, Vijai J,...Weitzel JN, et al. (2013) **Identification of a BRCA2-specific Modifier Locus at 6p24 Related to Breast Cancer Risk.** PLoS Genet. 9(3):e1003173. PMID: PMC3609647.
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9. Rebbeck T, Mitra N, Wan F, ...Weitzel, JN, et al. (2013 In Press) **Mutation-specific Cancer Risks Estimated from 31,481 Female Carriers of BRCA1 or BRCA2 Mutations.** Cancer Res. PMID: PMC Journal - In Process.
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11. Segev Y, Iqbal J, Lubinski J, ...Weitzel, JN, et al. (2013) **The incidence of endometrial cancer in women with BRCA1 and BRCA2 mutations: An international prospective cohort study.** Gynecol Oncol. 130(1):127-131.
12. Silva TB, Macdonald DJ, et al. (2013) **Perception of cancer causes and risk, family history and preventive behaviors of users in oncogenetic counseling.** Rev Esc Enferm USP. 47: p. 377-384.
13. Sorrell AD, Espenschied C, Culver J, Weitzel J. (2013) **Tumor Protein p53 (TP53) Testing and Li-Fraumeni Syndrome: Current Status of Clinical Applications and Future Directions.** Mol Diagn Ther.17(1):31-47. PMID: PMC3627545.
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15. Valentini A, Lubinski J, Byrski T, ...Weitzel, JN, et al. (2013) **The impact of pregnancy on breast cancer survival in women who carry a BRCA1 or BRCA2 mutation.** Breast Cancer Res Treat 142 (1):177-185. PMID: PMC Journal - In Process.

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16. Weitzel JN, Clague J, Martir-Negron A, Ogaz R, Herzog J, Ricker C, Jungbluth C, Cina C, Duncan P, et al. (2013) **Prevalence and Type of BRCA Mutations in Hispanics Undergoing Genetic Cancer Risk Assessment in the Southwestern United States: A Report From the Clinical Cancer Genetics Community Research Network.** J Clin Oncol. 31(2):210-216. PMID: PMC3532393

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1. Beamer, L., Grant, M., Espenschied, C., Blazer, K., Hampel, H., Weitzel, JN, & MacDonald, DJ (2012) **Reflex Immunohistochemistry and Microsatellite Instability Testing of Colorectal Tumors for Lynch Syndrome among US Cancer Programs and Follow-up of Abnormal Results.** J Clin Oncol, 1;30(10):1058-63
2. Blazer, KR, Christie, C., Uman, G., & Weitzel, JN (2012). **Impact of Web-based Case Conferencing on Cancer Genetics Training Outcomes for Community-based Clinicians.** J Cancer Educ, 27(2):217-225. NIHMS ID: 431334
3. Clark CC, Weitzel JN, O'Connor TR (2012) **Enhancement of Synthetic Lethality via Combinations of ABT-888, a PARP Inhibitor, and Carboplatin In Vitro and In Vivo Using BRCA1 and BRCA2 Isogenic Models.** Molecular Cancer Therapeutics 11 (9):1948-1958. PMID: PMC3551628.
4. Ding YC, McGuffog L, Healey S, Friedman E, Laitman Y,...Weitzel, JN,...Neuhausen, SL (2012) **A non-synonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers.** Cancer Epidem Biomar, 21:1362-1370. PMID: PMC3415567.
5. Espenschied, CR, MacDonald, DJ, Culver, JO, Sand, S., Hurley, K., Banks, K. C., Weitzel, JN, & Blazer, KR (2012). **Closing the Loop: Action research in a multimodal hereditary cancer patient conference is an effective tool to assess and address patient needs.** Journal of Cancer Education, 27(3):467-477. PMID: PMC3540105.
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8. Liu, J., Cristea, M. C., Frankel, P., Neuhausen, SL, Steele, L., Engelstaedter, V., Matulonis, U., Sand, S., Tung N., Garber, JE, & Weitzel, JN (2012). **Clinical characteristics and outcomes of BRCA-associated ovarian cancer (OC): genotype and survival.** Cancer Genet, 205, 34-41
9. MacDonald DJ, Deri J, Ricker C, Perez MA, Ogaz R, Feldman N, Viveros LA, Paz B, Weitzel JN, Blazer, KR. (2012) **Closing the Loop: An interactive action-research conference format for delivering updated medical information while eliciting Latina patient/family experiences and psychosocial needs post-genetic cancer risk assessment.** Familial Cancer. 11(3):449-445 PMID: PMC3620038.

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13. Pilarski R, Patel DA, Weitzel JN, McVeigh T, Dorairaj JJ, Heneghan HM, Miller N, Weidhaas JB, Kerin MJ, et al. (2012) **The KRAS-Variant Is Associated with Risk of Developing Double Primary Breast and Ovarian Cancer.** *PLoS One*, 7(5):e37891. PMID: PMC3360659
14. Ratner, E. S., ...Weitzel, JN, Neuhausen, SL, Schwartz, P. E., Slack, FJ, Santin, A. D., Weidhaas, J. B. (2012). **A KRAS variant is a biomarker of poor outcome, platinum chemotherapy resistance and a potential target for therapy in ovarian cancer.** *Oncogene*, 31(42):4559-4566. PMID: PMC3342446.
15. Rodriguez, A. O., Llacuachaqui, M., Pardo, G. G., Royer, R., Larson, G., Weitzel, JN, & Narod, S. A. (2012). **BRCA1 and BRCA2 mutations among ovarian cancer patients from Colombia.** *Gynecol Oncol.*, 124, 236-243.
16. Sorrell A, Espenschied C, Wang W, Weitzel JN, Chu S, Parker P, Saldivar S, Bhatia R. (2012) **Hereditary leukemia due to rare RUNX1c splice variant (L472X) presents with eczematous phenotype.** *Int J Clin Med*, 3:607-613. PMID: PMC Journal - In Process

## 2011

1. Blazer, KR, MacDonald, DJ, Culver, JO, Huizenga, CR, Morgan, RJ, Uman, GC, & Weitzel, JN (2011). **Personalized cancer genetics training for personalized medicine: Improving community-based healthcare through a genetically literate workforce.** *Genet Med*. 13(9), 832-840.
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6. Kempers, M. J., Kuiper, R. P., Ockeloen, C. W., Chappuis, P. O., Hutter, P., Rahner, N.,...Culver, JO, et al. (2011). **Risk of colorectal and endometrial cancers in epcam deletion-positive Lynch Syndrome: A cohort study.** *Lancet Oncol*, 12 (1):49-55.
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11. Neuhausen, SL, Brummel, S., Ding, Y., Steele, L., Nathanson, K., Domchek, S. M., ...Weitzel, JN, et al. (2011). **Genetic Variation in IGF2 and HTRA1 and Breast Cancer Risk Among BRCA1 and BRCA2 Carriers.** *Cancer Epidem Biomar*, 2011; 8(20): 1690-1702.
12. Rebbeck, T., Mitra, N., Domchek, S. M., Wan, F., Friebel, T. M., Tran, T. V.,...Weitzel, JN, et al. (2011) **Modification of BRCA1-Associated Breast and Ovarian Cancer Risk by BRCA1-Interacting Genes.** *Cancer Res*. 71(17), 5792-5805.
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## 2010

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2. Antoniou, AC, Wang, X, Fredericksen, ZS, McGuffog, L, Tarrell, R, Sinilnikova, OM, ...Weitzel, JN, et al. (2010) **A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population.** *Nat Genet*, 42(10), 885-892.

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7. Domchek, SM, Friebel, TM, Garber, JE, Isaacs, C, Matloff, E, Eeles, R, ...Weitzel, JN, et al. (2010) **Occult ovarian cancers identified at risk-reducing salpingo-oophorectomy in a prospective cohort of BRCA1/2 mutation carriers.** *Breast Cancer Res Treat*, 124(1), 195-203.
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10. Huizenga, CR, Lowstuter, K. J., Banks, K. C., Lagos, V. I., Vandergon, V. O., & Weitzel, JN (2010). **Evolving perspectives on genetic discrimination in health insurance among health care providers.** *Fam Cancer*, 9, 253-260. PMID: PMC Journal - In Process
11. Jasperson, K., Vu, T. M., Schwab, A. L., Neklason, D. W., Rodriguez-Bigas, M. A., Burt, R. W., & Weitzel, JN (2010). **Evaluating Lynch Syndrome in very early onset colorectal cancer probands without apparent polyposis.** *Fam Cancer*, 9(2), 99-107. PMID: PMC3620042.
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13. MacDonald, DJ, Blazer, KR, & Weitzel, JN (2010). **Extending comprehensive cancer center expertise in clinical cancer genetics and genomics to diverse communities: The power of partnership.** *J Natl Compr Canc Netw*, 8(5), 615-624.
14. MacDonald, DJ, Sarna, L, Weitzel, JN, Ferrell, B (2010) **Women's perceptions of the personal and family impact of genetic cancer risk assessment: Focus group findings.** *J Genet Couns*, 19(2), 148-160.
15. Robson, M. E., Storm, C. D., Weitzel, JN, Wollins, D. S., & Offit, K. (2010). **American Society of Clinical Oncology policy statement update: Genetic and genomic testing for cancer susceptibility.** *J Clin Oncol*, 28(5), 893-901.
16. Stadler, Z. K., Thom, P., Robson, M. E., Weitzel, JN, Kauff, N. D., Hurley, K. E., et al. (2010). **Genome-wide association studies of cancer.** *J Clin Oncol*, 28(27), 4255-4267.

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17. Tutt, A., Robson, M., Garber, JE, Domchek, S. M., Audeh, MW, Weitzel, JN, et al. (2010). **Oral poly (adp-ribose) polymerase inhibitor olaparib in patients with BRCA1 or BRCA2 mutations and advanced breast cancer: A proof-of-concept trial.** Lancet, 376(9737), 211-213.

### 2009

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2. Gonzalez, K. D., Noltner, K. A., Buzin, C. H., Gu, D., Wen-Fong, C. Y., Nguyen, V. Q., ...Weitzel, JN (2009). **Beyond Li –Fraumeni Syndrome: Clinical characteristics of families with p53 germline mutations.** J Clin Oncol, 27( 8), 1250-1256.
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4. Neuhausen, SL, Brummel, S., Ding, Y. C., Singer, C. F., Pfeiler, G., Lynch, HT, ...Weitzel, JN, et al. (2009). **Genetic variation in insulin-like growth factor signaling genes and breast cancer risk among BRCA1 and BRCA2 carriers.** Breast Cancer Res, 11(5), R76.
5. Pal, S. K., Blazer, KR, Weitzel, JN, & Somlo, G. (2009). **An association between invasive breast cancer and familial idiopathic hyperparathyroidism: A case series and review of the literature.** Breast Cancer Res Treat, 115: 1-5.
6. Rebbeck, T. R., Mitra, N., Domchek, S. M., Wan, F., Chuai, S., Friebel, T. M., ...Weitzel, JN, et al. (2009). **Modification of ovarian cancer risk by BRCA1/2-interacting genes in a multicenter cohort of BRCA1/2 mutation carriers.** Cancer Res, 69(14), 5801-5810.
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### 2008

1. Antoniou, AC, Spurdle, A. B., Sinilnikova, O. M., Healey, S., Pooley, K. A., Schmutzler, R. K., ...Weitzel, JN, et al. (2008). **Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers.** Am J Hum Genet, 82(4), 937-948.
2. Jasperson, K. W., Blazer, KR, Lowstuter, K. J., & Weitzel, JN (2008). **Working through a diagnostic challenge: Colonic polyposis, Amsterdam criteria, and a mismatch repair mutation.** Fam Cancer, 7(4), 281-285.
3. Lagos, VI, Perez, MA, Ricker, CN, Blazer, K R, Santiago, NM, Feldman, N, ...Weitzel, JN. (2008) **Social cognitive aspects of underserved Latinas preparing to undergo genetic risk assessment for hereditary breast and ovarian cancer.** Psycho-oncol, 17(8), 774-782.
4. Lowstuter, K. J., Sand, S., Blazer, KR, MacDonald, DJ, Banks, K. C., Lee, C. A., et al. (2008). **Influence of genetic discrimination perceptions and knowledge on cancer genetics referral practice among clinicians.** Genet Med, 10(9), 691-698.

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5. MacDonald, DJ, Sarna, L., Giger, JN, Bastani, R., van Servellen, G., & Weitzel, JN (2008). **Comparison of Latina and non-Latina white women's beliefs about communicating genetic cancer risk to relatives.** J Health Commun, 13(5), 465-479.
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9. West, J. G., Weitzel, JN, Tao, M. L., Carpenter, M., West, JE, & Fanning, C. (2008). **BRCA1/2 mutations and the risk of angiosarcoma following breast cancer treatment.** Clin Breast Cancer, 8(6), 533-537.

### 2007

1. Antoniou, AC, Sinilnikova, OM, Simard, J, Leone, M, Dumont, M, Neuhausen, SL, ...Weitzel, JN, et al. (2007) **RAD51 135G→C modifies breast cancer risk among BRCA2 mutation carriers: Results from a combined analysis of 19 studies.** Am J Hum Genet, 81(6), 1186-1200.
2. Bevers TB, Armstrong DK, Arun B, Carlson RW, Cowan KH, Daly MB, Fleming I, Garber JE, Gemignani M, Gradishar WJ, Krontiras H, Kulkarni S, Laronga C, Lawton T, Loftus L, MacDonald DJ, Mahoney MC, Merajver SD, Seewaldt V, Sellin RV, Shapiro CL, Singletary E, Ward JH. (2007) **Breast cancer risk reduction.** J Natl Compr Canc Netw. 5(8): 676-701.
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5. Edwards, Q. T., Seibert, D., Maradiegue, A., MacDonald, DJ, Jasperson, K. W., Lowstuter, K. J., & Weitzel, JN (2007). **Breast cancer and the family tree: An issue for all practice settings.** Adv Nurse Pract, 15(5), 34-41.
6. Friebel, T. M., Domchek, S. M., Neuhausen, SL, Wagner, T., Evans, D. G., Isaacs, C., ...Weitzel, JN, et al. (2007). **Bilateral prophylactic oophorectomy and bilateral prophylactic mastectomy in a prospective cohort of unaffected BRCA1 and BRCA2 mutation carriers.** Clin Breast Cancer, 7(11), 875-882.
7. Kotsopoulos, J., Lubinski, J., Lynch, HT, Klijn, J., Ghadirian, P., Neuhausen, SL, ...Weitzel, JN, et al. (2007). **Age at first birth and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers.** Breast Cancer Res Treat, 105(2), 221-228.



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### Publications

8. MacDonald, DJ, Sarna, L, van Servellen, G, Bastani, R, Giger, JN, Weitzel, JN. (2007) **Selection of family members for communication of cancer risk and barriers to this communication before and after genetic cancer risk assessment.** Genet Med, 9(5), 275-282.
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10. Ricker, CN, Hiyama, S, Fuentes, S, Feldman, N, Kumar, V, ...Weitzel, JN (2007) **Beliefs and interest in cancer risk in an underserved Latino cohort.** Prev Med, 44(3), 241-245.
11. Weitzel, JN, Buys, SS, Sherman, WH, Daniels, AM, Ursin, G, ...MacDonald, DJ, Blazer, KR, et al. (2007) **Reduced mammographic density with use of a gonadotropin-releasing hormone agonist-based chemoprevention regimen in BRCA1 carriers.** Clin Cancer Res, 13(2), 654-658.
12. Weitzel, JN, Lagos, V. I., Cullinane, C. A., Gambol, P. J., Culver, JO, Blazer, KR, Palomares, MR, Lowstuter, K. J., & MacDonald, DJ (2007). **Limited family structure and BRCA gene mutation status in single cases of breast cancer.** JAMA, 297(23), 2587-2595.
13. Weitzel, JN, Lagos, VI, Herzog, JS, Judkins, T, Hendrickson, B, Ho, JS, ...Blazer, KR, et al. (2007) **Evidence for common ancestral origin of a recurring BRCA1 genomic rearrangement identified in high-risk Hispanic families.** Cancer Epidem Biomar, 16(8), 1615-1620.

### 2006

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3. Finch, A., Beiner, M., Lubinski, J., Lynch, HT, Moller, P., Rosen, B., ...Weitzel, JN, et al. (2006). **Salpingo-oophorectomy and the risk of ovarian, fallopian tube, and peritoneal cancers in women with a BRCA1 or BRCA2 mutation.** JAMA, 296(2), 185-192.
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5. Guillem, J. G., Wood, W. C., Moley, J. F., Berchuck, A., Karlan, B. Y., Mutch, D. G., ...Weitzel, JN, et al. (2006). **ASCO/SSO review of current role of risk-reducing surgery in common hereditary cancer syndromes.** J Clin Oncol, 24(28), 4642-4660.
6. Guillem, J. G., Wood, W. C., Moley, J. F., Berchuck, A., Karlan, B. Y., Mutch, D. G., ...Weitzel, JN, et al. (2006). **ASCO/SSO review of current role of risk-reducing surgery in common hereditary cancer syndromes.** Ann Surg Oncol, 13(10), 1296-1321.
7. Jasperson, K. W., Lowstuter, K. J., & Weitzel, JN (2006) **Assessing the predictive accuracy of HMLH1 and HMSH2 mutation probability models.** J Genet Couns, 15(5), 339-347.

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8. MacDonald, DJ, Sand, S., Kass, F., Blazer, KR, Congleton, J., Craig, J., & Weitzel, JN (2006). **The power of partnership: Extending comprehensive cancer center expertise in clinical cancer genetics to community breast care centers.** Semin Breast Dis, 9, 39-47.
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10. Ricker, C., Lagos, V. I., Feldman, N., Hiyama, S., Fuentes, S., Kumar, V., Gonzalez, K., Palomares, MR, Blazer, KR, Lowstuter, K. J., MacDonald, DJ, & Weitzel, JN (2006). **If we build it...will they come?--establishing a cancer genetics services clinic for an underserved predominantly Latina cohort.** J Genet Couns, 15(6), 505-514.

### 2005

1. Blazer, KR, MacDonald, DJ, Ricker, C, Sand, S, Uman, GC, Weitzel, JN (2005) **Outcomes from intensive training in genetic cancer risk counseling for clinicians.** Genet Med, 7(1), 40-47.
2. Cullinane, CA, Lubinski, J, Neuhausen, SL, Ghadirian, P, Lynch, HT, Isaacs, C, Weber, B, ...Weitzel, JN, et al. (2005). **The effect of pregnancy as a risk factor for breast cancer in BRCA1/BRCA2 mutation carriers.** Int J Cancer, 117(6), 988-991.
3. Culver, JO, Edwards, Q. T., MacDonald, DJ, & Weitzel, JN (2005). **USPSTF guidelines will miss BRCA families due to paternal inheritance or a truncated family tree.** Ann Intern Med, 143(5), 355. Retrieved from [http://www.annals.org/content/143/5/355.abstract/reply#nintmed\\_el\\_2247](http://www.annals.org/content/143/5/355.abstract/reply#nintmed_el_2247)
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6. Palomares, MR, Paz, IB, Weitzel, JN (2005) **Genetic cancer risk assessment in the newly diagnosed breast cancer patient is useful and possible in practice.** J Clin Oncol, 23(13), 3165-3166.
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8. Weitzel, JN, Robson, M, Pasini, B, Manoukian, S, ...D, Lynch, HT, et al. (2005) **A comparison of bilateral breast cancers in BRCA carriers.** Cancer Epidem Biomar, 14(6), 1534-1538.

### 2004

1. Blazer, KR, Grant, M., Sand, SR, MacDonald, DJ, Uman, GC, & Weitzel, JN (2004). **Effects of a cancer genetics education programme on clinicians knowledge and practice.** J Med Genet, 41(7), 518-522.

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### Publications

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3. Jardines, L., Haffty, B. G., Doroshow, J. H., Fisher, P., & Weitzel, JN (2004). **Breast cancer overview: Risk factors, screening, genetic testing, and prevention.** In R. Pazdur, L. R. Coia, W. J. Hoskins & L. D. Wagman (Eds.), *Cancer management: A multidisciplinary approach medical, surgical & radiation oncology* (8th ed., pp. 165-190). Manhasset, New York: CMP Healthcare Media, Oncology Publishing Group.
4. MacDonald, DJ, & Weitzel, JN (2004) **Keeping current with risk reduction strategies for patients at hereditary cancer risk.** *International Society of Nurses in Genetics Newsletter*, 12(2), Pages 1-15.
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8. Weitzel, JN (2004). **Evidence for advice: Reduction in risk of breast or ovarian cancer after salpingo-oophorectomy in carriers of BRCA1 or BRCA2 mutations.** *Breast Diseases: A Year Book Quarterly*, 14(4), 354-356.