HEREDITARY CANCER REFERENCE LIST

Note: This reference list is updated bi-annually and this version is current as of August 1, 2018.

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ALK	18724359	Mosse YP et al. Identification of ALK as a major familial neuroblastoma predisposition gene. Nature. 2008 Oct 16;455(7215):930-5.
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Gene	PMID	Reference Text
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	NCCN	NCCN Guidelines. Genetic/Familial High-Risk Assessment: Breast and Ovarian.
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BRCA2	10433620	The Breast Cancer Linkage Consortium. Cancer risks in BRCA2 mutation carriers. J Natl Cancer Inst. 1999 Aug;91(15):1310-6.
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	11170890	Thompson D, Easton D, Breast Cancer Linkage Consortium. Variation in cancer risks, by mutation position, in BRCA2 mutation carriers. Am J Hum Genet. 2001 Feb;68(2):410-9.
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	19858402	Graeser MK et al. Contralateral Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. J Clin Oncol. 2009 Dec 10;27(35): 5887-92.
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BRCA2	NCCN	NCCN Guidelines. Genetic/Familial High-Risk Assessment: Breast and Ovarian.
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BRIP1	17033622	Seal S et al. Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. Nat Genet. 2006 Nov;38(11):1239-41.
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CASR	11013439	Hendy GN et al. Mutations of the calcium-sensing receptor (CASR) in familial hypocalciuric hypercalcemia, neonatal severe hyperparathyroidism, and autosomal dominant hypocalcemia. Human Mutation. 2000 Oct 16(4):281-96.
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CASR	18751724	Obermannova B et al. Unusually severe phenotype of neonatal primary hyperparathyroidism due to a heterozygous inactivating mutation in the CASR gene. European Journal Of Pediatrics. 2009 168(5):569-73.
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CDC73	12434154	Carpten JD et al. HRPT2, encoding parafibromin, is mutated in hyperparathyroidism-jaw tumor syndrome. Nat Genet. 2002 Dec;32(4):676-80.
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CDH1	10072428	Richards FM et al. Germline E-cadherin gene (CDH1) mutations predispose to familial gastric cancer and colorectal cancer. Hum Mol Genet. 1999 Apr;8(4):607-10.
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Gene	PMID	Reference Text
CDH1	11968083	Oliveira C et al. Screening E-cadherin in gastric cancer families reveals germline mutations only in hereditary diffuse gastric cancer kindred. Hum Mutat. 2002 May;19(5):510-7.
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CDK4	17047042	Goldstein AM et al. High-risk Melanoma Susceptibility Genes and Pancreatic Cancer, Neural System Tumors, and Uveal Melanoma across GenoMEL. Cancer Res. 2006 Oct 15;66(20):9818-28.
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CDKN2A	10506626	Kefford RF et al. Counseling and DNA testing for individuals perceived to be genetically predisposed to melanoma: A consensus statement of the Melanoma Genetics Consortium. J Clin Oncol. 1999 Oct;17(10):3245-51.
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CDKN2A	18981015	de Snoo FA et al. Increased risk of cancer other than melanoma in CDKN2A founder mutation (p16-Leiden)-positive melanoma families. Clin Cancer Res. 2008 Nov 1;14(21):7151-7.
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CFTR	15379964	Cuppens H and Cassiman JJ. CFTR mutations and polymorphisms in male infertility. International Journal Of Andrology. 2004 Oct 27(5):251-6.
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CHEK2	14612911	Seppala EH et al. CHEK2 variants associate with hereditary prostate cancer. Br J Cancer. 2003 Nov 17;89(10):1966-70.
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CTRC	18059268	Rosendahl J et al. Chymotrypsin C (CTRC) variants that diminish activity or secretion are associated with chronic pancreatitis. Nature Genetics. 2008 Jan 40(1):78-82.
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MET	10417759	Olivero M et al. Novel mutation in the ATP-binding site of the MET oncogene tyrosine kinase in a HPRCC family. Int J Cancer. 1999 Aug 27;82(5):640-3
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MITF	22080950	Yokoyama S et al. A novel recurrent mutation in MITF predisposes to familial and sporadic melanoma. Nature. 2011 Dec 1 480(7375):99-103.
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MITF	26650189	Potrony M et al. Prevalence of MITF p.E318K in Patients With Melanoma Independent of the Presence of CDKN2A Causative Mutations. Jama Dermatology. 2016 Apr 1 152(4):405-12.
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MLH1	15937084	Quehenberger F et al. Risk of colorectal and endometrial cancer for carriers of mutations of the hMLH1 and hMSH2 gene: correction for ascertainment. J Med Genet.2005 Jun;42(6):491-6.
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Gene	PMID	Reference Text
MLH1	NCCN	NCCN Guidelines. Genetic/Familial High-Risk Assessment: Colorectal.
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MRE11A	10612394	Stewart GS et al. The DNA double-strand break repair gene hMRE11 is mutated in individuals with an ataxia-telangiectasia-like disorder. Cell. 1999 Dec 10;99(6):577-87.
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MSH2	15937084	Quehenberger F et al. Risk of colorectal and endometrial cancer for carriers of mutations of the hMLH1 and hMSH2 gene: correction for ascertainment. J Med Genet.2005 Jun;42(6):491-6.
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Gene	PMID	Reference Text
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	NCCN	NCCN Guidelines. Genetic/Familial High-Risk Assessment: Colorectal.
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MSH6	19125127	Palomaki GE et al. EGAPP supplementary evidence review: DNA testing strategies aimed at reducing morbidity and mortality from Lynch syndrome. Genet Med. 2009 Jan;11(1):42-65.
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Gene	PMID	Reference Text
MUTYH	12606733	Sieber OM. Multiple colorectal adenomas, classic adenomatous polyposis, and germ-line in MYH. N Engl J Med. 2003 Feb 27;348(9):791-9.
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	16492921	Jenkins MA et al. Risk of colorectal cancer in monoallelic and biallelic carriers of MYH mutations: a population-based case-family study. Cancer Epidemiol Biomarkers Prev. 2006;15:312-14.
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MUTYH homoz	28649662	Slavin TP et al. The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. NPJ Breast Cancer. 2017 Jun 9;3:22.
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MUTYH-single	28649662	Slavin TP et al. The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. NPJ Breast Cancer. 2017 Jun 9;3:22.
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Gene	PMID	Reference Text
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	16770759	Steffen J et al. Germline mutations 657del5 of the NBS1 gene contribute significantly to the incidence of breast cancer in Central Poland. Int J Cancer. 2006 Jul 15;119(2):472-5.
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