Bernard Peissel

List of Publications by Year
in descending order

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121 papers	7,432 citations	87843 38 h-index	60583 81 g-index
123 all docs	123 docs citations	123 times ranked	11714 citing authors

#	Article	IF	Citations
1	Pathology of Breast and Ovarian Cancers among BRCA1 and BRCA2 Mutation Carriers: Results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 134-147.	1.1	513
2	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
3	Perinatal lethality with kidney and pancreas defects in mice with a targetted Pkd1 mutation. Nature Genetics, 1997, 17, 179-181.	9.4	420
4	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	3.8	390
5	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
6	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
7	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
8	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
9	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
10	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
11	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	1.5	174
12	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.4	169
13	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	0.8	152
14	Incidental Carcinomas in Prophylactic Specimens in BRCA1 and BRCA2 Germ-line Mutation Carriers, With Emphasis on Fallopian Tube Lesions. American Journal of Surgical Pathology, 2006, 30, 1222-1230.	2.1	130
15	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125
16	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
17	LINE-1 Elements at the Sites of Molecular Rearrangements in Alport Syndromeâ€“Diffuse Leiomyomatosis. American Journal of Human Genetics, 1999, 64, 62-69.	2.6	106
18	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	1.4	99

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19	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
20	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. PLoS Biology, 2011, 9, e1001199.	2.6	91
21	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	1.4	91
22	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	2.2	88
23	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	1.1	82
24	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	1.4	80
25	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	2.2	78
26	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
27	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	2.2	71
28	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	1.4	68
29	Comparative In Vitro and In Silico Analyses of Variants in Splicing Regions of BRCA1 and BRCA2 Genes and Characterization of Novel Pathogenic Mutations. PLoS ONE, 2013, 8, e57173.	1.1	64
30	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. Genetics in Medicine, 2018, 20, 452-457.	1.1	59
31	miR-342 Regulates BRCA1 Expression through Modulation of ID4 in Breast Cancer. PLoS ONE, 2014, 9, e87039.	1.1	59
32	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	2.2	57
33	Cutaneous Melanoma in Childhood and Adolescence Shows Frequent Loss of INK4A and Gain of KIT. Journal of Investigative Dermatology, 2009, 129, 1759-1768.	0.3	54
34	Identification of fifteen novel germline variants in theBRCA13â€²UTR reveals a variant in a breast cancer case that introduces a functionalmiR-103target site. Human Mutation, 2012, 33, 1665-1675.	1.1	49
35	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	1.1	47
36	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	1.5	47

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37	Clinical genetic testing for familial melanoma in Italy: A cooperative study. Journal of the American Academy of Dermatology, 2009, 61, 775-782.	0.6	45
38	Germline mutations of TP53 and BRCA2 genes in breast cancer/sarcoma families. European Journal of Cancer, 2007, 43, 601-606.	1.3	44
39	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	3.0	40
40	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	1.4	40
41	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. Human Mutation, 2004, 24, 100-101.	1.1	39
42	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
43	MRI in the Early Detection of Breast Cancer in Women with High Genetic Risk. Tumori, 2006, 92, 517-523.	0.6	37
44	PALB2 germline mutations in familial breast cancer cases with personal and family history of pancreatic cancer. Breast Cancer Research and Treatment, 2011, 126, 825-828.	1.1	37
45	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	1.1	34
46	First description of an acinic cell carcinoma of the breast in a BRCA1 mutation carrier: a case report. BMC Cancer, 2013, 13, 46.	1.1	34
47	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	1.1	34
48	A large-scale assessment of two-way SNP interactions in breast cancer susceptibility using 46 450 cases and 42 461 controls from the breast cancer association consortium. Human Molecular Genetics, 2014, 23, 1934-1946.	1.4	32
49	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. Breast Cancer Research, 2016, 18, 64.	2.2	31
50	Wholeâ€exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. Cancer, 2017, 123, 210-218.	2.0	31
51	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. Journal of Human Genetics, 2006, 51, 209-216.	1.1	29
52	A cancer modifier role for parathyroid hormone-related protein. Oncogene, 2000, 19, 5324-5328.	2.6	28
53	The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians. Breast Cancer Research and Treatment, 2012, 133, 805-807.	1.1	28
54	The psychological impact of breast and ovarian cancer preventive options in BRCA1 and BRCA2 mutation carriers. Clinical Genetics, 2014, 85, 7-15.	1.0	28

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55	Adherence to Mediterranean Diet and Metabolic Syndrome in BRCA Mutation Carriers. Integrative Cancer Therapies, 2018, 17, 153-160.	0.8	28
56	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
57	Four new cases of double heterozygosity for BRCA1 and BRCA2 gene mutations: clinical, pathological, and family characteristics. Breast Cancer Research and Treatment, 2010, 124, 251-258.	1.1	27
58	Serum levels of IGF-I and BRCA penetrance: a case control study in breast cancer families. Familial Cancer, 2011, 10, 521-528.	0.9	27
59	Functional characterization of the MTC-associated germline RET-K666E mutation: evidence of oncogenic potential enhanced by the G691S polymorphism. Endocrine-Related Cancer, 2011, 18, 519-527.	1.6	27
60	Association of PHB 1630 C>T and MTHFR 677 C>T polymorphisms with breast and ovarian cancer risk in BRCA1/2 mutation carriers: results from a multicenter study. British Journal of Cancer, 2012, 106, 2016-2024.	2.9	27
61	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	2.2	26
62	PALB2 sequencing in Italian familial breast cancer cases reveals a high-risk mutation recurrent in the province of Bergamo. Genetics in Medicine, 2014, 16, 688-694.	1.1	25
63	Fine-Scale Mapping of the 4q24 Locus Identifies Two Independent Loci Associated with Breast Cancer Risk. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1680-1691.	1.1	24
64	Characterization of an Italian Founder Mutation in the RING-Finger Domain of BRCA1. PLoS ONE, 2014, 9, e86924.	1.1	24
65	SNPs in ultraconserved elements and familial breast cancer risk. Carcinogenesis, 2009, 30, 544-545.	1.3	23
66	Exploring the link between MORF4L1 and risk of breast cancer. Breast Cancer Research, 2011, 13, R40.	2.2	23
67	Reconstructing the Genealogy of a BRCA1 Founder Mutation by Phylogenetic Analysis. Annals of Human Genetics, 2008, 72, 310-318.	0.3	22
68	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	1.1	22
69	Constitutive BRCA1 Promoter Hypermethylation Can Be a Predisposing Event in Isolated Early-Onset Breast Cancer. Cancers, 2019, 11, 58.	1.7	22
70	Loss of tyrosinase activity confers increased skin tumor susceptibility in mice. Oncogene, 2004, 23, 4130-4135.	2.6	21
71	Mutation detection rates associated with specific selection criteria for BRCA1/2 testing in 1854 high-risk families: A monocentric Italian study. European Journal of Internal Medicine, 2016, 32, 65-71.	1.0	21
72	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19

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73	Single base pair deletions in exons 39 and 42 of the COL4A5 gene in Alport syndrome. Human Molecular Genetics, 1994, 3, 201-202.	1.4	18
74	A novel frameshift deletion in type IV collagen Î±5 gene in a juvenile-type Alport syndrome patient: An adenine deletion (2940/2943 del A) in exon 34 of COL4A5. Human Mutation, 1994, 3, 386-390.	1.1	18
75	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	1.8	18
76	A large de novo9p21.3 deletion in a girl affected by astrocytoma and multiple melanoma. BMC Medical Genetics, 2014, 15, 59.	2.1	18
77	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	0.6	18
78	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	1.1	18
79	A Dietary Intervention to Lower Serum Levels of IGF-I in BRCA Mutation Carriers. Cancers, 2018, 10, 309.	1.7	18
80	Cyclin D1 expression analysis in familial breast cancers may discriminate BRCAX from BRCA2-linked cases. Modern Pathology, 2008, 21, 1262-1270.	2.9	16
81	Linkage disequilibrium and haplotype mapping of a skin cancer susceptibility locus in outbred mice. Mammalian Genome, 2000, 11, 979-981.	1.0	15
82	An evaluation of the polymorphisms Ins16bp and Arg72Pro in p53 as breast cancer risk modifiers in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2008, 99, 974-977.	2.9	14
83	Mapping of Melanoma Modifier Loci inRETTransgenic Mice. Japanese Journal of Cancer Research, 2000, 91, 1142-1147.	1.7	13
84	Linkage analysis for the diagnosis of autosomal dominant polycystic kidney disease, and for the determination of genetic heterogeneity in Italian families. Clinical Genetics, 1991, 40, 287-297.	1.0	13
85	Two new CHEK2 germ-line variants detected in breast cancer/sarcoma families negative for BRCA1, BRCA2, and TP53 gene mutations. Breast Cancer Research and Treatment, 2011, 130, 207-215.	1.1	13
86	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. Journal of Medical Genetics, 2011, 48, 703-704.	1.5	13
87	Lifestyle Characteristics in Women Carriers of BRCA Mutations: Results From an Italian Trial Cohort. Clinical Breast Cancer, 2021, 21, e168-e176.	1.1	13
88	Analysis of BRCA1 and RAD51C Promoter Methylation in Italian Families at High-Risk of Breast and Ovarian Cancer. Cancers, 2020, 12, 910.	1.7	13
89	Evidence for a link between TNFRSF11A and risk of breast cancer. Breast Cancer Research and Treatment, 2011, 129, 947-954.	1.1	12
90	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. Human Molecular Genetics, 2014, 23, 6034-6046.	1.4	12

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91	Estimate of the penetrance of BRCA mutation and the COS software for the assessment of BRCA mutation probability. Familial Cancer, 2015, 14, 117-128.	0.9	12
92	The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers. Breast Cancer Research and Treatment, 2011, 125, 855-860.	1.1	11
93	X chromosome inactivation pattern in BRCA gene mutation carriers. European Journal of Cancer, 2013, 49, 1136-1141.	1.3	11
94	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	1.7	11
95	Sequencing Analysis of SLX4/FANCP Gene in Italian Familial Breast Cancer Cases. PLoS ONE, 2012, 7, e31038.	1.1	10
96	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. Scientific Reports, 2015, 5, 15454.	1.6	10
97	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS ONE, 2016, 11, e0158801.	1.1	10
98	Prenatal diagnosis of autosomal dominant polycystic kidney disease using flanking DNA markers and the polymerase chain reaction. Prenatal Diagnosis, 1992, 12, 513-524.	1.1	9
99	Two Novel Mutations Affecting Splicing in the IRF6 Gene Associated With van der Woude Syndrome. Journal of Craniofacial Surgery, 2010, 21, 1654-1656.	0.3	9
100	Riskâ€reducing surgery in BRCA1/BRCA2 mutation carriers: Are there factors associated with the choice?. Psycho-Oncology, 2019, 28, 1871-1878.	1.0	9
101	Use of intercross outbred mice and single nucleotide polymorphisms to map skin cancer modifier loci. Mammalian Genome, 2001, 12, 291-294.	1.0	8
102	Allele-specific patterns of the mouse parathyroid hormone-related protein: influences on cell adhesion and migration. Oncogene, 2003, 22, 7711-7715.	2.6	8
103	An unusual BRCA2 allele carrying two splice site mutations. Annals of Oncology, 2009, 20, 1143-1144.	0.6	8
104	Analysis of Italian BRCA1/2 Pathogenic Variants Identifies a Private Spectrum in the Population from the Bergamo Province in Northern Italy. Cancers, 2021, 13, 532.	1.7	8
105	Prenatal testing in a fetus at risk for autosomal dominant polycystic kidney disese and autosomal recessive junctional epidermolysis bullosa with pyloric atresia. American Journal of Medical Genetics Part A, 1993, 47, 1225-1230.	2.4	7
106	Evaluation of the XRCC1 gene as a phenotypic modifier in BRCA1/2 mutation carriers. Results from the consortium of investigators of modifiers of BRCA1/BRCA2. British Journal of Cancer, 2011, 104, 1356-1361.	2.9	7
107	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. Breast Cancer Research, 2021, 23, 86.	2.2	7
108	Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. PLoS ONE, 2017, 12, e0171663.	1.1	7

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109	Gene linkage analysis and DNA based detection of autosomal dominant polycystic kidney disease (ADPKD) in a newborn infant. Case report. Journal of Perinatal Medicine, 1995, 23, 205-212.	0.6	4
110	Inhibition of both skin and lung tumorigenesis by Car-R mouse-derived cancer modifier loci. International Journal of Cancer, 2002, 97, 580-583.	2.3	4
111	A Targeted Approach to Genetic Counseling in Breast Cancer Patients: The Experience of an Italian Local Project. Tumori, 2016, 102, 45-50.	0.6	4
112	Survey of gynecological carcinosarcomas in families with breast and ovarian cancer predisposition. Cancer Genetics, 2018, 221, 38-45.	0.2	4
113	Rapid DNA-Based Prenatal Diagnosis of Autosomal Dominant Polycystic Kidney Disease. JAMA Pediatrics, 1994, 148, 1101.	3.6	2
114	Re: Molecular Basis for Estrogen Receptor Â Deficiency in BRCA1-Linked Breast Cancer. Journal of the National Cancer Institute, 2008, 100, 752-753.	3.0	2
115	Is there a Specific Magnetic Resonance Phenotype Characteristic of Hereditary Breast Cancer?. Tumori, 2010, 96, 363-384.	0.6	2
116	Cardio-Oncology. Journal of the American College of Cardiology, 2016, 68, 1921-1923.	1.2	2
117	BRCA1/2 Variants and Metabolic Factors: Results From a Cohort of Italian Female Carriers. Cancers, 2020, 12, 3584.	1.7	2
118	Genetic Variants and Somatic Alterations Associated with MITF-E318K Germline Mutation in Melanoma Patients. Genes, 2021, 12, 1440.	1.0	2
119	The Impact of Mediterranean Dietary Intervention on Metabolic and Hormonal Parameters According to BRCA1/2 Variant Type. Frontiers in Genetics, 2022, 13, 820878.	1.1	2
120	Clinical Applications of Genetic Linkage Analysis for the Molecular Diagnostics of ADPKD, Using DNA Markers Linked to the PKD1 and PKD2 Genes. Contributions To Nephrology, 1995, 115, 88-92.	1.1	1
121	Management of BRCA Tumour Testing in an Integrated Molecular Tumour Board Multidisciplinary Model. Frontiers in Oncology, 2022, 12, 857515.	1.3	1