siranoush manoukian

List of Publications by Year in Descending Order

143 papers	13,562 citations	50 h-index	115 g-index
144 ext. papers	15,183 ext. citations	7.8 avg, IF	4.35 L-index

#	Paper	IF	Citations
143	Malignant salivary gland tumours in families with breast cancer susceptibility. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2021**, 479, 221-226	5.1	
142	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	4.4	15
141	Whole-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	6.4	22
140	Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. *PLoS ONE*, **2017**, 12, e0171663	3.7	6
139	Haplotype analyses of the c.1027C>T and c.2167_2168delAT recurrent truncating mutations in the breast cancer-predisposing gene PALB2. *Breast Cancer Research and Treatment*, **2016**, 160, 121-129	4.4	7
138	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. *Breast Cancer Research*, **2016**, 18, 112	8.3	25
137	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
136	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
135	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). *Scientific Reports*, **2016**, 6, 32512	4.9	16
134	Cardio-Oncology: The Carney Complex Type I. *Journal of the American College of Cardiology*, **2016**, 68, 1921-1923	15.1	2
133	Multiple primary melanomas (MPMs) and criteria for genetic assessment: MultiMEL, a multicenter study of the Italian Melanoma Intergroup. *Journal of the American Academy of Dermatology*, **2016**, 74, 325-32	4.5	23
132	Treatment of infertility does not increase the risk of ovarian cancer among women with a BRCA1 or BRCA2 mutation. *Fertility and Sterility*, **2016**, 105, 781-785	4.8	28
131	The incidence of leukaemia in women with BRCA1 and BRCA2 mutations: an International Prospective Cohort Study. *British Journal of Cancer*, **2016**, 114, 1160-4	8.7	19
130	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. *Journal of Medical Genetics*, **2016**, 53, 298-309	5.8	83
129	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
128	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65
127	Triple-Negative versus Non-Triple-Negative Breast Cancers in High-Risk Women: Phenotype Features and Survival from the HIBCRIT-1 MRI-Including Screening Study. *Clinical Cancer Research*, **2016**, 22, 895-904	12.9	25

126	A targeted approach to genetic counseling in breast cancer patients: the experience of an Italian local project. *Tumori*, **2016**, 102, 45-50	1.7	3
125	Fine-scale mapping of 8q24 locus identifies multiple independent risk variants for breast cancer. *International Journal of Cancer*, **2016**, 139, 1303-1317	7.5	26
124	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	8.3	25
123	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	3.9	18
122	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. *Cancer Causes and Control*, **2016**, 27, 679-93	2.8	15
121	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	8.3	58
120	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
119	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
118	Novel and known genetic variants for male breast cancer risk at 8q24.21, 9p21.3, 11q13.3 and 14q24.1: results from a multicenter study in Italy. *European Journal of Cancer*, **2015**, 51, 2289-95	7.5	20
117	FANCM c.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, 5345-55	5.6	68
116	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
115	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74
114	Fine-scale mapping of the 4q24 locus identifies two independent loci associated with breast cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 1680-91	4	17
113	Estimate of the penetrance of BRCA mutation and the COS software for the assessment of BRCA mutation probability. *Familial Cancer*, **2015**, 14, 117-28	3	12
112	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
111	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
110	Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. *Scientific Reports*, **2015**, 5, 15454	4.9	10
109	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	8.3	16

108	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
107	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
106	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
105	PALB2 sequencing in Italian familial breast cancer cases reveals a high-risk mutation recurrent in the province of Bergamo. *Genetics in Medicine*, **2014**, 16, 688-94	8.1	21
104	A randomized controlled trial of diet and physical activity in BRCA mutation carriers. *Familial Cancer* , **2014**, 13, 181-7	3	12
103	The psychological impact of breast and ovarian cancer preventive options in BRCA1 and BRCA2 mutation carriers. *Clinical Genetics*, **2014**, 85, 7-15	4	26
102	FGF receptor genes and breast cancer susceptibility: results from the Breast Cancer Association Consortium. *British Journal of Cancer*, **2014**, 110, 1088-100	8.7	20
101	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
100	Duration of tamoxifen use and the risk of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2014**, 146, 421-7	4.4	29
99	A large de novo 9p21.3 deletion in a girl affected by astrocytoma and multiple melanoma. *BMC Medical Genetics*, **2014**, 15, 59	2.1	8
98	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. *Human Molecular Genetics*, **2014**, 23, 6034-46	5.6	11
97	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
96	Association of SULT1A1 Arg□□His polymorphism with male breast cancer risk: results from a multicenter study in Italy. *Breast Cancer Research and Treatment*, **2014**, 148, 623-8	4.4	4
95	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
94	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	8.3	46
93	Characterization of an Italian founder mutation in the RING-finger domain of BRCA1. *PLoS ONE*, **2014**, 9, e86924	3.7	18
92	miR-342 regulates BRCA1 expression through modulation of ID4 in breast cancer. *PLoS ONE*, **2014**, 9, e87039	3.7	54
91	First description of an acinic cell carcinoma of the breast in a BRCA1 mutation carrier: a case report. *BMC Cancer*, **2013**, 13, 46	4.8	28

90	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
89	Association of low-penetrance alleles with male breast cancer risk and clinicopathological characteristics: results from a multicenter study in Italy. *Breast Cancer Research and Treatment*, **2013**, 138, 861-8	4.4	26
88	The impact of pregnancy on breast cancer survival in women who carry a BRCA1 or BRCA2 mutation. *Breast Cancer Research and Treatment*, **2013**, 142, 177-85	4.4	40
87	X chromosome inactivation pattern in BRCA gene mutation carriers. *European Journal of Cancer*, **2013**, 49, 1136-41	7.5	8
86	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
85	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
84	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
83	Whole exome sequencing suggests much of non-BRCA1/BRCA2 familial breast cancer is due to moderate and low penetrance susceptibility alleles. *PLoS ONE*, **2013**, 8, e55681	3.7	77
82	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	3.7	46
81	Breastfeeding and the risk of breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R42	8.3	68
80	What is specific in hereditary breast cancer? High T2 signal intensity as a new semeiotic pattern?. *European Journal of Radiology*, **2012**, 81 Suppl 1, S165-70	4.7	1
79	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
78	Rare variants in XRCC2 as breast cancer susceptibility alleles. *Journal of Medical Genetics*, **2012**, 49, 618-20	5.8	37
77	9q31.2-rs865686 as a susceptibility locus for estrogen receptor-positive breast cancer: evidence from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1783-91	4	17
76	Sequencing analysis of SLX4/FANCP gene in Italian familial breast cancer cases. *PLoS ONE*, **2012**, 7, e31038	3.7	9
75	11q13 is a susceptibility locus for hormone receptor positive breast cancer. *Human Mutation*, **2012**, 33, 1123-32	4.7	33
74	Identification of fifteen novel germline variants in the BRCA1 3′UTR reveals a variant in a breast cancer case that introduces a functional miR-103 target site. *Human Mutation*, **2012**, 33, 1665-75	4.7	42
73	Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter study in Italy. *Breast Cancer Research and Treatment*, **2012**, 134, 411-8	4.4	58

72	The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians. *Breast Cancer Research and Treatment*, **2012**, 133, 805-7	4.4	28
71	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. *Cancer Research*, **2012**, 72, 1795-803	13.1	93
70	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, 645-57	4	44
69	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, 134-47	4	411
68	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
67	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
66	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	8.3	62
65	Multicenter surveillance of women at high genetic breast cancer risk using mammography, ultrasonography, and contrast-enhanced magnetic resonance imaging (the high breast cancer risk italian 1 study): final results. *Investigative Radiology*, **2011**, 46, 94-105	10.1	243
64	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, 1356-61	8.7	6
63	Serum levels of IGF-I and BRCA penetrance: a case control study in breast cancer families. *Familial Cancer*, **2011**, 10, 521-8	3	25
62	The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers. *Breast Cancer Research and Treatment*, **2011**, 125, 855-60	4.4	10
61	No evidence for an association between the earwax-associated polymorphism in ABCC11 and breast cancer risk in Caucasian women. *Breast Cancer Research and Treatment*, **2011**, 126, 235-9	4.4	16
60	PALB2 germline mutations in familial breast cancer cases with personal and family history of pancreatic cancer. *Breast Cancer Research and Treatment*, **2011**, 126, 825-8	4.4	32
59	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11
58	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, 207-15	4.4	11
57	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
56	Associations of breast cancer risk factors with tumor subtypes: a pooled analysis from the Breast Cancer Association Consortium studies. *Journal of the National Cancer Institute*, **2011**, 103, 250-63	9.7	513
55	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, 3304-21	5.6	62

54	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
53	The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain. *Journal of Medical Genetics*, **2011**, 48, 703-4	5.8	13
52	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
51	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
50	Evaluation of variation in the phosphoinositide-3-kinase catalytic subunit alpha oncogene and breast cancer risk. *British Journal of Cancer*, **2011**, 105, 1934-9	8.7	4
49	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. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
48	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, 9742-54	10.1	147
47	A BRCA1 promoter variant (rs11655505) and breast cancer risk. *Journal of Medical Genetics*, **2010**, 47, 268-70	5.8	5
46	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
45	Single-nucleotide polymorphisms inside microRNA target sites influence tumor susceptibility. *Cancer Research*, **2010**, 70, 2789-98	10.1	314
44	HMGA1 protein expression in familial breast carcinoma patients. *European Journal of Cancer*, **2010**, 46, 332-9	7.5	16
43	Four new cases of double heterozygosity for BRCA1 and BRCA2 gene mutations: clinical, pathological, and family characteristics. *Breast Cancer Research and Treatment*, **2010**, 124, 251-8	4.4	19
42	Evaluation of SNPs in miR-146a, miR196a2 and miR-499 as low-penetrance alleles in German and Italian familial breast cancer cases. *Human Mutation*, **2010**, 31, E1052-7	4.7	138
41	An unusual BRCA2 allele carrying two splice site mutations. *Annals of Oncology*, **2009**, 20, 1143-4	10.3	7
40	SNPs in ultraconserved elements and familial breast cancer risk. *Carcinogenesis*, **2009**, 30, 544-5; author reply 546	4.6	22
39	Analysis of a set of missense, frameshift, and in-frame deletion variants of BRCA1. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2009**, 660, 1-11	3.3	31
38	Evidences for association of the CASP8 -652 6N del promoter polymorphism with age at diagnosis in familial breast cancer cases. *Breast Cancer Research and Treatment*, **2009**, 113, 607-8	4.4	18
37	Cutaneous melanoma in childhood and adolescence shows frequent loss of INK4A and gain of KIT. *Journal of Investigative Dermatology*, **2009**, 129, 1759-68	4.3	46

36	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2009**, 101, 1456-60	8.7	17
35	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). *British Journal of Cancer*, **2009**, 101, 2048-54	8.7	13
34	Clinical genetic testing for familial melanoma in Italy: a cooperative study. *Journal of the American Academy of Dermatology*, **2009**, 61, 775-82	4.5	39
33	Cyclin D1 expression analysis in familial breast cancers may discriminate BRCAX from BRCA2-linked cases. *Modern Pathology*, **2008**, 21, 1262-70	9.8	15
32	The BOADICEA model of genetic susceptibility to breast and ovarian cancers: updates and extensions. *British Journal of Cancer*, **2008**, 98, 1457-66	8.7	358
31	Rapid progression of prostate cancer in men with a BRCA2 mutation. *British Journal of Cancer*, **2008** , 99, 371-4	8.7	112
30	The p53 Arg72Pro and Ins16bp polymorphisms and their haplotypes are not associated with breast cancer risk in BRCA-mutation negative familial cases. *Cancer Detection and Prevention*, **2008**, 32, 140-3		16
29	BRCA1 p.Val1688del is a deleterious mutation that recurs in breast and ovarian cancer families from Northeast Italy. *Journal of Clinical Oncology*, **2008**, 26, 26-31	2.2	39
28	Predictors of contralateral prophylactic mastectomy in women with a BRCA1 or BRCA2 mutation: the Hereditary Breast Cancer Clinical Study Group. *Journal of Clinical Oncology*, **2008**, 26, 1093-7	2.2	141
27	Re: Molecular basis for estrogen receptor alpha deficiency in BRCA1-linked breast cancer. *Journal of the National Cancer Institute*, **2008**, 100, 752-3; author reply 753-4	9.7	2
26	Hormone therapy and the risk of breast cancer in BRCA1 mutation carriers. *Journal of the National Cancer Institute*, **2008**, 100, 1361-7	9.7	152
25	Genomic rearrangements of the CDKN2A locus are infrequent in Italian malignant melanoma families without evidence of CDKN2A/CDK4 point mutations. *Melanoma Research*, **2008**, 18, 431-7	3.3	7
24	Infertility, treatment of infertility, and the risk of breast cancer among women with BRCA1 and BRCA2 mutations: a case-control study. *Cancer Causes and Control*, **2008**, 19, 1111-9	2.8	63
23	Indications for breast magnetic resonance imaging. Consensus document "Attualit□n senologia", Florence 2007. *Radiologia Medica*, **2008**, 113, 1085-95	6.5	34
22	International variation in rates of uptake of preventive options in BRCA1 and BRCA2 mutation carriers. *International Journal of Cancer*, **2008**, 122, 2017-22	7.5	268
21	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *American Journal of Human Genetics*, **2008**, 82, 937-48	11	218
20	New Trends of MRI in Breast Cancer Diagnosis **2008**, 127-144		
19	Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian BRCA1/2 carriers. *Breast Cancer Research and Treatment*, **2007**, 103, 29-36	4.4	22

18	A CDKN2A mutation in familial melanoma that abrogates binding of p16INK4a to CDK4 but not CDK6. *Cancer Research*, **2007**, 67, 9134-41	10.1	38
17	Multicenter comparative multimodality surveillance of women at genetic-familial high risk for breast cancer (HIBCRIT study): interim results. *Radiology*, **2007**, 242, 698-715	20.5	287
16	Determination of cancer risk associated with germ line BRCA1 missense variants by functional analysis. *Cancer Research*, **2007**, 67, 1494-501	10.1	98
15	Age at menarche and menopause and breast cancer risk in the International BRCA1/2 Carrier Cohort Study. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 740-6	4	56
14	Germline mutations of TP53 and BRCA2 genes in breast cancer/sarcoma families. *European Journal of Cancer*, **2007**, 43, 601-6	7.5	38
13	Incidental carcinomas in prophylactic specimens in BRCA1 and BRCA2 germ-line mutation carriers, with emphasis on fallopian tube lesions: report of 6 cases and review of the literature. *American Journal of Surgical Pathology*, **2006**, 30, 1222-30	6.7	112
12	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. *Journal of Human Genetics*, **2006**, 51, 209-216	4.3	23
11	Bilateral preaxial polydactyly in a WAGR syndrome patient. *American Journal of Medical Genetics, Part A*, **2005**, 134, 426-9	2.5	4
10	Breast and ovarian cancer risks to carriers of the BRCA1 5382insC and 185delAG and BRCA2 6174delT mutations: a combined analysis of 22 population based studies. *Journal of Medical Genetics*, **2005**, 42, 602-3	5.8	107
9	A comparison of bilateral breast cancers in BRCA carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 1534-8	4	42
8	Classification of BRCA1 missense variants of unknown clinical significance. *Journal of Medical Genetics*, **2005**, 42, 138-46	5.8	73
7	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. *Human Mutation*, **2004**, 24, 100-1	4.7	36
6	Atypical epithelial proliferation in fallopian tubes in prophylactic salpingo-oophorectomy specimens from BRCA1 and BRCA2 germline mutation carriers. *International Journal of Gynecological Pathology*, **2004**, 23, 35-40	3.2	104
5	Different expressivity of BRCA1 and BRCA2: analysis of 179 Italian pedigrees with identified mutation. *Breast Cancer Research and Treatment*, **2003**, 81, 71-9	4.4	22
4	Average risks of breast and ovarian cancer associated with BRCA1 or BRCA2 mutations detected in case Series unselected for family history: a combined analysis of 22 studies. *American Journal of Human Genetics*, **2003**, 72, 1117-30	11	2643
3	Characterization of a cytogenetic 17q11.2 deletion in an NF1 patient with a contiguous gene syndrome. *Human Genetics*, **1996**, 98, 646-50	6.3	23
2	Kaufman oculocerebrofacial syndrome in a girl of 15 years. *American Journal of Medical Genetics Part A*, **1995**, 58, 21-3		6
1	Prenatal testing in a fetus at risk for autosomal dominant polycystic kidney disease and autosomal recessive junctional epidermolysis bullosa with pyloric atresia. *American Journal of Medical Genetics Part A*, **1993**, 47, 1225-30		5