

# Brigitte Bressac-de Paillerets

## List of Publications by Citations

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157  
ext. papers

13,455  
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L-index

#	Paper	IF	Citations
146	Selective G to T mutations of p53 gene in hepatocellular carcinoma from southern Africa. <i>Nature</i> , <b>1991</b> , 350, 429-31	50.4	1185
145	p53 functions as a cell cycle control protein in osteosarcomas. <i>Molecular and Cellular Biology</i> , <b>1990</b> , 10, 5772-81	4.8	719
144	Genetic testing in pheochromocytoma or functional paraganglioma. <i>Journal of Clinical Oncology</i> , <b>2005</b> , 23, 8812-8	2.2	529
143	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , <b>2013</b> , 45, 371-84, 384e1-2	36.3	422
142	A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. <i>Nature</i> , <b>2011</b> , 480, 94-8	50.4	365
141	Geographical variation in the penetrance of CDKN2A mutations for melanoma. <i>Journal of the National Cancer Institute</i> , <b>2002</b> , 94, 894-903	9.7	363
140	Revisiting Li-Fraumeni Syndrome From TP53 Mutation Carriers. <i>Journal of Clinical Oncology</i> , <b>2015</b> , 33, 2345-52	2.2	360
139	Genome-wide association study identifies three loci associated with melanoma risk. <i>Nature Genetics</i> , <b>2009</b> , 41, 920-5	36.3	360
138	Abnormal structure and expression of p53 gene in human hepatocellular carcinoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>1990</b> , 87, 1973-7	11.5	343
137	High-risk melanoma susceptibility genes and pancreatic cancer, neural system tumors, and uveal melanoma across GenoMEL. <i>Cancer Research</i> , <b>2006</b> , 66, 9818-28	10.1	313
136	Prevalence of p16 and CDK4 germline mutations in 48 melanoma-prone families in France. The French Familial Melanoma Study Group. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 209-16	5.6	284
135	2009 version of the Chompret criteria for Li Fraumeni syndrome. <i>Journal of Clinical Oncology</i> , <b>2009</b> , 27, e108-9; author reply e110	2.2	236
134	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. <i>Nature Genetics</i> , <b>2014</b> , 46, 482-6	36.3	220
133	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 937-48	11	218
132	Genome-wide association study identifies three new melanoma susceptibility loci. <i>Nature Genetics</i> , <b>2011</b> , 43, 1108-13	36.3	203
131	Germline BAP1 mutations predispose to renal cell carcinomas. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 974-80	11	188
130	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , <b>2017</b> , 49, 1767-1778	36.3	186

129	PDGFRA germline mutation in a family with multiple cases of gastrointestinal stromal tumor. <i>Gastroenterology</i> , <b>2004</b> , 126, 318-21	13.3	185
128	Guidelines for splicing analysis in molecular diagnosis derived from a set of 327 combined in silico/in vitro studies on BRCA1 and BRCA2 variants. <i>Human Mutation</i> , <b>2012</b> , 33, 1228-38	4.7	171
127	Senescent cells develop a PARP-1 and nuclear factor- $\kappa$ B-associated secretome (PNAS). <i>Genes and Development</i> , <b>2011</b> , 25, 1245-61	12.6	168
126	Molecular basis of the Li-Fraumeni syndrome: an update from the French LFS families. <i>Journal of Medical Genetics</i> , <b>2008</b> , 45, 535-8	5.8	155
125	Genome-wide association study identifies a new melanoma susceptibility locus at 1q21.3. <i>Nature Genetics</i> , <b>2011</b> , 43, 1114-8	36.3	126
124	Common variants near TARDBP and EGR2 are associated with susceptibility to Ewing sarcoma. <i>Nature Genetics</i> , <b>2012</b> , 44, 323-7	36.3	124
123	Selection criteria for genetic assessment of patients with familial melanoma. <i>Journal of the American Academy of Dermatology</i> , <b>2009</b> , 61, 677.e1-14	4.5	115
122	Overexpression of MDM2, due to enhanced translation, results in inactivation of wild-type p53 in BurkittB lymphoma cells. <i>Oncogene</i> , <b>1998</b> , 16, 1603-10	9.2	112
121	Cancer risk in heterozygotes for ataxia-telangiectasia. <i>International Journal of Cancer</i> , <b>2001</b> , 93, 288-93	7.5	109
120	The founder mutation MSH2*1906G-->C is an important cause of hereditary nonpolyposis colorectal cancer in the Ashkenazi Jewish population. <i>American Journal of Human Genetics</i> , <b>2002</b> , 71, 1395-412	11	109
119	Localization of a novel melanoma susceptibility locus to 1p22. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 301-13	11	103
118	Clinical impact of the NKp30/B7-H6 axis in high-risk neuroblastoma patients. <i>Science Translational Medicine</i> , <b>2015</b> , 7, 283ra55	17.5	97
117	Comprehensive Study of the Clinical Phenotype of Germline BAP1 Variant-Carrying Families Worldwide. <i>Journal of the National Cancer Institute</i> , <b>2018</b> , 110, 1328-1341	9.7	97
116	Novel FH mutations in families with hereditary leiomyomatosis and renal cell cancer (HLRCC) and patients with isolated type 2 papillary renal cell carcinoma. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 226-34	5.8	94
115	Accumulation of multiple mutations in tumour suppressor genes during colorectal tumorigenesis in HNPCC patients. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 2257-60	5.6	92
114	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 4442-56	5.6	91
113	High frequency of germline SUFU mutations in children with desmoplastic/nodular medulloblastoma younger than 3 years of age. <i>Journal of Clinical Oncology</i> , <b>2012</b> , 30, 2087-93	2.2	91
112	Breast cancer risk in ataxia telangiectasia (AT) heterozygotes: haplotype study in French AT families. <i>British Journal of Cancer</i> , <b>1999</b> , 80, 1042-5	8.7	90

111	Melanoma prone families with CDK4 germline mutation: phenotypic profile and associations with MC1R variants. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 264-70	5.8	89
110	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. <i>American Journal of Human Genetics</i> , <b>2000</b> , 67, 207-212	11	89
109	Significant contribution of germline BRCA2 rearrangements in male breast cancer families. <i>Cancer Research</i> , <b>2004</b> , 64, 8143-7	10.1	86
108	Influence of genes, nevi, and sun sensitivity on melanoma risk in a family sample unselected by family history and in melanoma-prone families. <i>Journal of the National Cancer Institute</i> , <b>2004</b> , 96, 785-95	9.7	85
107	Therapeutic activity of CPT-11, a DNA-topoisomerase I inhibitor, against peripheral primitive neuroectodermal tumour and neuroblastoma xenografts. <i>British Journal of Cancer</i> , <b>1996</b> , 74, 537-45	8.7	83
106	Association of MC1R variants and host phenotypes with melanoma risk in CDKN2A mutation carriers: a GenoMEL study. <i>Journal of the National Cancer Institute</i> , <b>2010</b> , 102, 1568-83	9.7	81
105	Primary proliferative T cell response to wild-type p53 protein in patients with breast cancer. <i>European Journal of Immunology</i> , <b>1995</b> , 25, 1765-9	6.1	81
104	Genetic testing for melanoma. <i>Lancet Oncology, The</i> , <b>2002</b> , 3, 653-4	21.7	80
103	A single genetic origin for the G101W CDKN2A mutation in 20 melanoma-prone families. <i>American Journal of Human Genetics</i> , <b>2000</b> , 67, 311-9	11	75
102	Resistance of MCF7 human breast carcinoma cells to TNF-induced cell death is associated with loss of p53 function. <i>Oncogene</i> , <b>1997</b> , 15, 2817-26	9.2	74
101	Germline BAP1 mutations predispose also to multiple basal cell carcinomas. <i>Clinical Genetics</i> , <b>2015</b> , 88, 273-7	4	71
100	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2012</b> , 14, R33	8.3	70
99	Extensive molecular screening for hereditary non-polyposis colorectal cancer. <i>British Journal of Cancer</i> , <b>2000</b> , 82, 871-80	8.7	65
98	Screening for TP53 rearrangements in families with the Li-Fraumeni syndrome reveals a complete deletion of the TP53 gene. <i>Oncogene</i> , <b>2003</b> , 22, 840-6	9.2	64
97	Reassessing the clinical spectrum associated with hereditary leiomyomatosis and renal cell carcinoma syndrome in French FH mutation carriers. <i>Clinical Genetics</i> , <b>2017</b> , 92, 606-615	4	63
96	Prevalence of the E318K MITF germline mutation in Italian melanoma patients: associations with histological subtypes and family cancer history. <i>Pigment Cell and Melanoma Research</i> , <b>2013</b> , 26, 259-62	4.5	62
95	Mutations in BHD and TP53 genes, but not in HNF1beta gene, in a large series of sporadic chromophobe renal cell carcinoma. <i>British Journal of Cancer</i> , <b>2007</b> , 96, 336-40	8.7	59
94	Melanocortin-1 receptor (MC1R) gene variants and dysplastic nevi modify penetrance of CDKN2A mutations in French melanoma-prone pedigrees. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2005</b> , 14, 2384-90	4	59

93	Familial non-VHL clear cell (conventional) renal cell carcinoma: clinical features, segregation analysis, and mutation analysis of FLCN. <i>Clinical Cancer Research</i> , <b>2008</b> , 14, 5925-30	12.9	54
92	BRCA Share: A Collection of Clinical BRCA Gene Variants. <i>Human Mutation</i> , <b>2016</b> , 37, 1318-1328	4.7	48
91	Sporadic multiple primary melanoma cases: CDKN2A germline mutations with a founder effect. <i>Genes Chromosomes and Cancer</i> , <b>2001</b> , 32, 195-202	5	48
90	Contribution of de novo and mosaic mutations to Li-Fraumeni syndrome. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 173-180	5.8	47
89	PARKIN Inactivation Links Parkinson Disease to Melanoma. <i>Journal of the National Cancer Institute</i> , <b>2016</b> , 108,	9.7	46
88	Testing for BRCA1 mutations: a cost-effectiveness analysis. <i>European Journal of Human Genetics</i> , <b>2002</b> , 10, 599-606	5.3	46
87	Incomplete penetrance of the predisposition to medulloblastoma associated with germ-line SUFU mutations. <i>Journal of Medical Genetics</i> , <b>2010</b> , 47, 142-4	5.8	44
86	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. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 645-57	4	44
85	BRAF as a melanoma susceptibility candidate gene?. <i>Cancer Research</i> , <b>2003</b> , 63, 3061-5	10.1	44
84	Comprehensive analysis of CDKN2A (p16INK4A/p14ARF) and CDKN2B genes in 53 melanoma index cases considered to be at heightened risk of melanoma. <i>Journal of Medical Genetics</i> , <b>2006</b> , 43, 39-47	5.8	43
83	Individuals with presumably hereditary uveal melanoma do not harbour germline mutations in the coding regions of either the P16INK4A, P14ARF or cdk4 genes. <i>British Journal of Cancer</i> , <b>2000</b> , 82, 818-22	8.7	42
82	Gene expression signature associated with BRAF mutations in human primary cutaneous melanomas. <i>Molecular Oncology</i> , <b>2008</b> , 1, 425-30	7.9	41
81	Real-time PCR-based gene dosage assay for detecting BRCA1 rearrangements in breast-ovarian cancer families. <i>Clinical Genetics</i> , <b>2004</b> , 65, 131-6	4	41
80	Distinct deregulation of the hypoxia inducible factor by PHD2 mutants identified in germline DNA of patients with polycythemia. <i>Haematologica</i> , <b>2012</b> , 97, 9-14	6.6	40
79	p53 STATUS DOES NOT PREDICT INITIAL CLINICAL RESPONSE TO BACILLUS CALMETTE-GUERIN INTRAVESICAL THERAPY IN T1 BLADDER TUMORS. <i>Journal of Urology</i> , <b>1998</b> , 159, 1079-1084	2.5	39
78	Cutaneous phenotype and MC1R variants as modifying factors for the development of melanoma in CDKN2A G101W mutation carriers from 4 countries. <i>International Journal of Cancer</i> , <b>2007</b> , 121, 825-31	7.5	39
77	Correlation between p53 gene expression and tumor-cell proliferation in oropharyngeal cancer. <i>International Journal of Cancer</i> , <b>1994</b> , 57, 458-62	7.5	39
76	Genetic and environmental factors in cutaneous malignant melanoma. <i>Biochimie</i> , <b>2002</b> , 84, 67-74	4.6	38

75	Novel diagnostic tool for prediction of variant spliceogenicity derived from a set of 395 combined in silico/in vitro studies: an international collaborative effort. <i>Nucleic Acids Research</i> , <b>2018</b> , 46, 7913-7923 <sup>20.1</sup>	37
74	Identification of a new exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. <i>Blood</i> , <b>2018</b> , 132, 469-483	2.2 37
73	Germline SUFU mutation carriers and medulloblastoma: clinical characteristics, cancer risk, and prognosis. <i>Neuro-Oncology</i> , <b>2018</b> , 20, 1122-1132	1 35
72	Mapping and ranking of potential cytotoxic T epitopes in the p53 protein: effect of mutations and polymorphism on peptide binding to purified and refolded HLA molecules. <i>European Journal of Immunology</i> , <b>1995</b> , 25, 1638-42	6.1 34
71	Functional, structural, and genetic evaluation of 20 CDKN2A germ line mutations identified in melanoma-prone families or patients. <i>Human Mutation</i> , <b>2009</b> , 30, 564-74	4.7 33
70	Pattern multiplicity and fumarate hydratase (FH)/S-(2-succino)-cysteine (2SC) staining but not eosinophilic nucleoli with perinucleolar halos differentiate hereditary leiomyomatosis and renal cell carcinoma-associated renal cell carcinomas from kidney tumors without FH gene alteration. <i>Modern Pathology</i> , <b>2018</b> , 31, 674-683	9.8 32
69	Lack of referral for genetic counseling and testing in BRCA1/2 and Lynch syndromes: a nationwide study based on 240,134 consultations and 134,652 genetic tests. <i>Breast Cancer Research and Treatment</i> , <b>2013</b> , 141, 135-44	4.4 31
68	BRCA1, BRCA2, TP53, and CDKN2A germline mutations in patients with breast cancer and cutaneous melanoma. <i>Familial Cancer</i> , <b>2007</b> , 6, 453-61	3 31
67	CDKN2A as a uveal and cutaneous melanoma susceptibility gene. <i>Genes Chromosomes and Cancer</i> , <b>2003</b> , 38, 265-8	5 28
66	Four novel RET germline variants in exons 8 and 11 display an oncogenic potential in vitro. <i>European Journal of Endocrinology</i> , <b>2010</b> , 162, 771-7	6.5 25
65	A germline mutation in PBRM1 predisposes to renal cell carcinoma. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 426-30	5.8 24
64	A single Mediterranean, possibly Jewish, origin for the Val59Gly CDKN2A mutation in four melanoma-prone families. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 288-96	5.3 24
63	Guidelines for reporting secondary findings of genome sequencing in cancer genes: the SFMPP recommendations. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 1732-1742	5.3 23
62	Tracking of second primary melanomas in vemurafenib-treated patients. <i>JAMA Dermatology</i> , <b>2013</b> , 149, 488-90	5.1 23
61	The contribution of large genomic deletions at the CDKN2A locus to the burden of familial melanoma. <i>British Journal of Cancer</i> , <b>2008</b> , 99, 364-70	8.7 23
60	Breast cancer risk in BRCA1 and BRCA2 mutation carriers and polyglutamine repeat length in the AIB1 gene. <i>International Journal of Cancer</i> , <b>2005</b> , 117, 230-3	7.5 23
59	A germline oncogenic MITF mutation and tumor susceptibility. <i>European Journal of Cell Biology</i> , <b>2014</b> , 93, 71-5	6.1 22
58	Impact of gene patents on the cost-effective delivery of care: the case of BRCA1 genetic testing. <i>International Journal of Technology Assessment in Health Care</i> , <b>2003</b> , 19, 287-300	1.8 22

57	Primary leptomeningeal melanoma is part of the BAP1-related cancer syndrome. <i>Acta Neuropathologica</i> , <b>2015</b> , 129, 921-3	14.3	21
56	Relationship between genome and epigenome--challenges and requirements for future research. <i>BMC Genomics</i> , <b>2014</b> , 15, 487	4.5	21
55	Familial melanoma: clinical factors associated with germline CDKN2A mutations according to the number of patients affected by melanoma in a family. <i>Journal of the American Academy of Dermatology</i> , <b>2012</b> , 67, 1257-64	4.5	21
54	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. <i>Breast Cancer Research and Treatment</i> , <b>2011</b> , 127, 671-9	4.4	21
53	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 4732-47	5.6	21
52	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2015</b> , 24, 308-16	4	20
51	Contribution of CDKN2A/P16 (INK4A), P14 (ARF), CDK4 and BRCA1/2 germline mutations in individuals with suspected genetic predisposition to uveal melanoma. <i>Familial Cancer</i> , <b>2010</b> , 9, 663-7	3	20
50	Structure-activity relationships in glucocorticoid-induced apoptosis in T lymphocytes. <i>Biochemical Pharmacology</i> , <b>1995</b> , 50, 103-10	6	20
49	Activating point mutations in cyclin-dependent kinase 4 are not seen in sporadic pituitary adenomas, insulinomas or Leydig cell tumours. <i>Journal of Endocrinology</i> , <b>2003</b> , 178, 301-10	4.7	19
48	Deciphering the Role of Oncogenic MITF E318K in Senescence Delay and Melanoma Progression. <i>Journal of the National Cancer Institute</i> , <b>2017</b> , 109,	9.7	18
47	Genetic evidence of a precisely tuned dysregulation in the hypoxia signaling pathway during oncogenesis. <i>Cancer Research</i> , <b>2014</b> , 74, 6554-64	10.1	18
46	Soft tissue sarcomas. Case 3. Gastrointestinal stromal tumor and Carney's triad. <i>Journal of Clinical Oncology</i> , <b>2004</b> , 22, 2029-31	2.2	16
45	Genetic transmission of susceptibility to cancer in families of children with soft tissue sarcomas. <i>Cancer</i> , <b>1996</b> , 78, 1483-91	6.4	16
44	Diversity of the clinical presentation of the MMR gene biallelic mutations. <i>Familial Cancer</i> , <b>2014</b> , 13, 131-5		15
43	Characteristics of the coexistence of melanoma and renal cell carcinoma. <i>Cancer</i> , <b>2010</b> , 116, 5716-24	6.4	15
42	Association of the POT1 Germline Missense Variant p.I78T With Familial Melanoma. <i>JAMA Dermatology</i> , <b>2019</b> , 155, 604-609	5.1	15
41	Clinical relevance of 8q23, 15q13 and 18q21 SNP genotyping to evaluate colorectal cancer risk. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 99-105	5.3	14
40	Germline /P16INK4A mutations contribute to genetic determinism of sarcoma. <i>Journal of Medical Genetics</i> , <b>2017</b> , 54, 607-612	5.8	14

39	Patterns of familial aggregation of three melanoma risk factors: great number of naevi, light phototype and high degree of sun exposure. <i>International Journal of Epidemiology</i> , <b>2000</b> , 29, 408-415	7.8	14
38	Combining Homologous Recombination and Phosphopeptide-binding Data to Predict the Impact of BRCT Variants on Cancer Risk. <i>Molecular Cancer Research</i> , <b>2019</b> , 17, 54-69	6.6	14
37	Genital and anorectal mucosal melanoma is associated with cutaneous melanoma in patients and in families. <i>British Journal of Dermatology</i> , <b>2013</b> , 169, 594-9	4	13
36	Factors associated with altered long-term well-being after prophylactic salpingo-oophorectomy among women at increased hereditary risk for breast and ovarian cancer. <i>Oncologist</i> , <b>2011</b> , 16, 1250-7	5.7	13
35	Characteristics, treatment, and outcome of breast cancers diagnosed in BRCA1 and BRCA2 gene mutation carriers in intensive screening programs including magnetic resonance imaging. <i>Clinical Breast Cancer</i> , <b>2010</b> , 10, 113-8	3	13
34	Occurrence of BAP1 germline mutations in cutaneous melanocytic tumors with loss of BAP1-expression: A pilot study. <i>Genes Chromosomes and Cancer</i> , <b>2017</b> , 56, 691-694	5	12
33	eMelanoBase: an online locus-specific variant database for familial melanoma. <i>Human Mutation</i> , <b>2003</b> , 21, 2-7	4.7	12
32	Search for germline alterations in CDKN2A/ARF and CDK4 of 42 Jewish melanoma families with or without neural system tumours. <i>British Journal of Cancer</i> , <b>2005</b> , 92, 2278-85	8.7	12
31	The 5' untranslated region of p16INK4a melanoma tumor suppressor acts as a cellular IRES, controlling mRNA translation under hypoxia through YBX1 binding. <i>Oncotarget</i> , <b>2015</b> , 6, 39980-94	3.3	12
30	The MITF, p.E318K Variant, as a Risk Factor for Pheochromocytoma and Paraganglioma. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2016</b> , 101, 4764-4768	5.6	12
29	Germline mutations of inhibins in early-onset ovarian epithelial tumors. <i>Human Mutation</i> , <b>2014</b> , 35, 294-7	4.7	11
28	New founder germline mutations of CDKN2A in melanoma-prone families and multiple primary melanoma development in a patient receiving levodopa treatment. <i>Genes Chromosomes and Cancer</i> , <b>2007</b> , 46, 751-60	5	11
27	Absence of microsatellite instability in thyroid carcinomas. <i>European Journal of Cancer</i> , <b>1995</b> , 31A, 128	7.5	11
26	Multiple skin hamartomata: a possible novel clinical presentation of SUFU neoplasia syndrome. <i>Familial Cancer</i> , <b>2015</b> , 14, 151-5	3	10
25	Germline Variation at CDKN2A and Associations with Nevus Phenotypes among Members of Melanoma Families. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 2606-2612	4.3	10
24	Hereditary breast cancer associated with a germline BRCA2 mutation in identical female twins with similar disease expression. <i>Cancer Genetics and Cytogenetics</i> , <b>2002</b> , 133, 24-8		10
23	GEMO, a National Resource to Study Genetic Modifiers of Breast and Ovarian Cancer Risk in and Pathogenic Variant Carriers. <i>Frontiers in Oncology</i> , <b>2018</b> , 8, 490	5.3	10
22	Phenotypic and Histopathological Tumor Characteristics According to CDKN2A Mutation Status among Affected Members of Melanoma Families. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 1066-1069	4.3	9



21	Estimating CDKN2A mutation carrier probability among global familial melanoma cases using GenoMELPREDICT. <i>Journal of the American Academy of Dermatology</i> , <b>2019</b> , 81, 386-394	4.5	9
20	Protective effect of copy number polymorphism of glutathione S-transferase T1 gene on melanoma risk in presence of CDKN2A mutations, MC1R variants and host-related phenotypes. <i>Familial Cancer</i> , <b>2009</b> , 8, 371-7	3	9
19	MC1R variants in childhood and adolescent melanoma: a retrospective pooled analysis of a multicentre cohort. <i>The Lancet Child and Adolescent Health</i> , <b>2019</b> , 3, 332-342	14.5	8
18	The CDKN2A/p16(INK) (4a) 5'RTR sequence and translational regulation: impact of novel variants predisposing to melanoma. <i>Pigment Cell and Melanoma Research</i> , <b>2016</b> , 29, 210-21	4.5	8
17	Non-HodgkinB lymphomas and myeloid disorders: deletions associated with t(2;5) and t(3;5) detected by FISH. <i>Leukemia</i> , <b>1998</b> , 12, 1159-62	10.7	8
16	Optimization of Next-Generation Sequencing Technologies for von Hippel Lindau (VHL) Mosaic Mutation Detection and Development of Confirmation Methods. <i>Journal of Molecular Diagnostics</i> , <b>2019</b> , 21, 462-470	5.1	7
15	Germline mutations in the new E1Pcryptic exon of the gene in patients with tumours of von Hippel-Lindau disease spectrum or with paraganglioma. <i>Journal of Medical Genetics</i> , <b>2020</b> , 57, 752-759	5.8	7
14	Integrative analysis of dysregulated microRNAs and mRNAs in multiple recurrent synchronized renal tumors from patients with von Hippel-Lindau disease. <i>International Journal of Oncology</i> , <b>2018</b> , 53, 1455-1468	4.4	7
13	Association between germ cell tumours, large numbers of naevi, atypical naevi and melanoma. <i>Melanoma Research</i> , <b>2001</b> , 11, 117-22	3.3	7
12	Novel FH mutation in a patient with cutaneous leiomyomatosis associated with cutis verticis gyrata, eruptive collagenoma and Charcot-Marie-Tooth disease. <i>British Journal of Dermatology</i> , <b>2010</b> , 163, 1337-9	4	6
11	Absence of somatic alterations of the EB1 gene adenomatous polyposis coli-associated protein in human sporadic colorectal cancers. <i>British Journal of Cancer</i> , <b>1998</b> , 78, 1356-60	8.7	6
10	Melanoma Risk and Melanocyte Biology. <i>Acta Dermato-Venereologica</i> , <b>2020</b> , 100, adv00139	2.2	6
9	Improvement of Genetic Testing for Cutaneous Melanoma in Countries With Low to Moderate Incidence: The Rule of 2 vs the Rule of 3. <i>JAMA Dermatology</i> , <b>2017</b> , 153, 1122-1129	5.1	5
8	FBXO32 links ubiquitination to epigenetic reprogramming of melanoma cells. <i>Cell Death and Differentiation</i> , <b>2021</b> , 28, 1837-1848	12.7	3
7	Genetic Testing for Melanoma-Where Are We With Moderate-Penetrance Genes?. <i>JAMA Dermatology</i> , <b>2016</b> , 152, 375-6	5.1	2
6	The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1726-1737	8.1	2
5	Response to systemic therapy in fumarate hydratase-deficient renal cell carcinoma. <i>European Journal of Cancer</i> , <b>2021</b> , 151, 106-114	7.5	2
4	Is systemic disease in the coelomic epithelium associated with BRCA1 germline mutations?. <i>Journal of the National Cancer Institute</i> , <b>2004</b> , 96, 488-9	9.7	1

3	The PI3K/mTOR Pathway Is Targeted by Rare Germline Variants in Patients with Both Melanoma and Renal Cell Carcinoma. <i>Cancers</i> , <b>2021</b> , 13,	6.6	1
2	Germline mutation in the NBR1 gene involved in autophagy detected in a family with renal tumors. <i>Cancer Genetics</i> , <b>2021</b> , 258-259, 51-56	2.3	1
1	Prise en charge des formes familiales de tumeurs rares : mélanomes familiaux et primitifs multiples. <i>Oncologie</i> , <b>2008</b> , 10, 411-414	1	