

Sylviane Olschwang

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

116
papers

12,199
citations

44
h-index

110
g-index

134
ext. papers

13,393
ext. citations

8.7
avg, IF

4.86
L-index

#	Paper	IF	Citations
116	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. <i>Lancet Oncology, The</i> , 2021 , 22, 1014-1022	21.7	5
115	Characterisation of heterozygous variants in French patients with Lynch syndrome. <i>Journal of Medical Genetics</i> , 2020 , 57, 487-499	5.8	3
114	The Balance Between Cytotoxic T-cell Lymphocytes and Immune Checkpoint Expression in the Prognosis of Colon Tumors. <i>Journal of the National Cancer Institute</i> , 2018 , 110,	9.7	64
113	Improving Mutation Screening in Patients with Colorectal Cancer Predisposition Using Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2017 , 19, 589-601	5.1	5
112	Molecular combing reveals complex 4q35 rearrangements in Facioscapulohumeral dystrophy. <i>Human Mutation</i> , 2017 , 38, 1432-1441	4.7	22
111	Actionable Genes, Core Databases, and Locus-Specific Databases. <i>Human Mutation</i> , 2016 , 37, 1299-1307	4.7	5
110	Point Mutations in Exon 1B of APC Reveal Gastric Adenocarcinoma and Proximal Polyposis of the Stomach as a Familial Adenomatous Polyposis Variant. <i>American Journal of Human Genetics</i> , 2016 , 98, 830-842	11	153
109	Frequent intragenic rearrangements of DPYD in colorectal tumours. <i>Pharmacogenomics Journal</i> , 2015 , 15, 211-8	3.5	3
108	Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. <i>Journal of Clinical Oncology</i> , 2015 , 33, 3591-7	2.2	71
107	Genomic variations integrated database for MUTYH-associated adenomatous polyposis. <i>Journal of Medical Genetics</i> , 2015 , 52, 25-7	5.8	2
106	Clinical Utility Gene Card for: Familial adenomatous polyposis (FAP) and attenuated FAP (AFAP)--update 2014. <i>European Journal of Human Genetics</i> , 2015 , 23,	5.3	6
105	PIK3CA mutations predict recurrence in localized microsatellite stable colon cancer. <i>Cancer Medicine</i> , 2015 , 4, 371-82	4.8	20
104	Vascular Endothelial Growth Factor A c.*237C>T polymorphism is associated with bevacizumab efficacy and related hypertension in metastatic colorectal cancer. <i>Digestive and Liver Disease</i> , 2015 , 47, 331-7	3.3	20
103	The UMD-APC database, a model of nation-wide knowledge base: update with data from 3,581 variations. <i>Human Mutation</i> , 2014 , 35, 532-6	4.7	12
102	Molecular patterns in deficient mismatch repair colorectal tumours: results from a French prospective multicentric biological and genetic study. <i>British Journal of Cancer</i> , 2014 , 110, 2728-37	8.7	18
101	High-resolution analysis of DNA copy number alterations in rectal cancer: correlation with metastasis, survival, and mRNA expression. <i>Strahlentherapie Und Onkologie</i> , 2014 , 190, 1028-36	4.3	3
100	A multicenter blinded study evaluating EGFR and KRAS mutation testing methods in the clinical non-small cell lung cancer setting--IFCT/ERMETIC2 Project Part 1: Comparison of testing methods in 20 French molecular genetic National Cancer Institute platforms. <i>Journal of Molecular Diagnostics</i> , 2014 , 16, 45-55	5.1	27

99	Gastric adenocarcinoma in familial adenomatous polyposis can occur without previous lesions. <i>Journal of Gastrointestinal Cancer</i> , 2014 , 45, 377-9	1.6	3
98	Quality Assurance in Molecular Testing of Cancer 2014 , 423-431		
97	Semiparametric inference on the penetrances of rare genetic mutations based on a case-family design. <i>Journal of Statistical Planning and Inference</i> , 2013 , 143, 368-377	0.8	
96	Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM) - update 2012. <i>European Journal of Human Genetics</i> , 2013 , 21,	5.3	12
95	Gene expression classification of colon cancer into molecular subtypes: characterization, validation, and prognostic value. <i>PLoS Medicine</i> , 2013 , 10, e1001453	11.6	794
94	UMD-MLH1/MSH2/MSH6 databases: description and analysis of genetic variations in French Lynch syndrome families. <i>Database: the Journal of Biological Databases and Curation</i> , 2013 , 2013, bat036	5	11
93	Design of a core classification process for DNA mismatch repair variations of a priori unknown functional significance. <i>Human Mutation</i> , 2013 , 34, 920-2	4.7	3
92	Long-term effect of resistant starch on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. <i>Lancet Oncology</i> , 2012 , 13, 1242-9	21.7	70
91	Expression Profiles in Stage II Colon Cancer According to APC Gene Status. <i>Translational Oncology</i> , 2012 , 5, 72-6	4.9	15
90	Is the controversy on breast cancer as part of the Lynch-related tumor spectrum still open?. <i>Familial Cancer</i> , 2012 , 11, 681-3	3	10
89	8q24 Cancer risk allele associated with major metastatic risk in inflammatory breast cancer. <i>PLoS ONE</i> , 2012 , 7, e37943	3.7	27
88	A seven-gene signature aggregates a subgroup of stage II colon cancers with stage III. <i>OMICS A Journal of Integrative Biology</i> , 2012 , 16, 560-5	3.8	41
87	KRAS mutation spectrum notably diverges between non-small cell lung and colorectal carcinomas. <i>Journal of Thoracic Oncology</i> , 2012 , 7, 773-4; author reply 774	8.9	3
86	Cancer risks associated with germline mutations in MLH1, MSH2, and MSH6 genes in Lynch syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2011 , 305, 2304-10	27.4	692
85	Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. <i>Lancet</i> , 2011 , 378, 2081-7	40	715
84	Cross-validation study for epidermal growth factor receptor and KRAS mutation detection in 74 blinded non-small cell lung carcinoma samples: a total of 5550 exons sequenced by 15 molecular French laboratories (evaluation of the EGFR mutation status for the administration of EGFR-TKIs in non-small cell lung carcinoma [ERMETIC] project—part 1). <i>Journal of Thoracic Oncology</i> , 2011 , 6, 1006-15	8.9	43
83	Clinical utility gene card for: familial adenomatous polyposis (FAP) and attenuated FAP (AFAP). <i>European Journal of Human Genetics</i> , 2011 , 19,	5.3	9
82	The LKB1 complex-AMPK pathway: the tree that hides the forest. <i>Familial Cancer</i> , 2011 , 10, 415-24	3	25

81	SMAD4 protein expression and cell proliferation in colorectal adenocarcinomas. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2011 , 459, 511-9	5.1	7
80	SMARCB1/INI1 germline mutations contribute to 10% of sporadic schwannomatosis. <i>BMC Neurology</i> , 2011 , 11, 9	3.1	61
79	The educational role of external quality assessment in genetic testing: a 7-year experience of the European Molecular Genetics Quality Network (EMQN) in Lynch syndrome. <i>Human Mutation</i> , 2011 , 32, 696-7	4.7	6
78	Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2). <i>European Journal of Human Genetics</i> , 2010 , 18,	5.3	8
77	Statistical inference on the penetrances of rare genetic mutations based on a case-family design. <i>Biostatistics</i> , 2010 , 11, 519-32	3.7	5
76	Germline APC mutation spectrum derived from 863 genomic variations identified through a 15-year medical genetics service to French patients with FAP. <i>Journal of Medical Genetics</i> , 2010 , 47, 721-2	5.8	30
75	Contribution of ultrasonography to endometrial cancer screening in patients with hereditary nonpolyposis colorectal cancer/Lynch syndrome. <i>International Journal of Gynecological Cancer</i> , 2010 , 20, 583-7	3.5	12
74	Combined mutations of ASXL1, CBL, FLT3, IDH1, IDH2, JAK2, KRAS, NPM1, NRAS, RUNX1, TET2 and WT1 genes in myelodysplastic syndromes and acute myeloid leukemias. <i>BMC Cancer</i> , 2010 , 10, 401	4.8	125
73	Leiden Open Variation Database of the MUTYH gene. <i>Human Mutation</i> , 2010 , 31, 1205-15	4.7	61
72	Cancer occurrence during follow-up of the CAPP2 study -aspirin use for up to four years significantly reduces Lynch syndrome cancers for up to several years after completion of therapy. <i>Hereditary Cancer in Clinical Practice</i> , 2010 , 8, O5	2.3	1
71	Chronic iron-deficiency anemia caused by a jejunojejunal intussusception on a solitary hamartomatous polyp. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2010 , 50, 450-2	2.8	1
70	iCOMET, projet national de recherche de gènes de prédisposition au développement des métastases chez les patients traités pour un cancer colique sporadique de stade II 2009 , 7		
69	Analysis of candidate genes in occurrence and growth of colorectal adenomas. <i>Journal of Oncology</i> , 2009 , 2009, 306786	4.5	2
68	Age-Dependent Cancer Risk Is Not Different in between MSH2 and MLH1 Mutation Carriers. <i>Journal of Cancer Epidemiology</i> , 2009 , 2009, 791754	2.8	4
67	Quantitative PCR high-resolution melting (qPCR-HRM) curve analysis, a new approach to simultaneously screen point mutations and large rearrangements: application to MLH1 germline mutations in Lynch syndrome. <i>Human Mutation</i> , 2009 , 30, 867-75	4.7	55
66	Mutations of polycomb-associated gene ASXL1 in myelodysplastic syndromes and chronic myelomonocytic leukaemia. <i>British Journal of Haematology</i> , 2009 , 145, 788-800	4.5	460
65	A polymorphism of EGFR extracellular domain is associated with progression free-survival in metastatic colorectal cancer patients receiving cetuximab-based treatment. <i>BMC Cancer</i> , 2008 , 8, 169	4.8	60
64	Genome profiling of chronic myelomonocytic leukemia: frequent alterations of RAS and RUNX1 genes. <i>BMC Cancer</i> , 2008 , 8, 299	4.8	102

63	Identification in daily practice of patients with Lynch syndrome (hereditary nonpolyposis colorectal cancer): revised Bethesda guidelines-based approach versus molecular screening. <i>American Journal of Gastroenterology</i> , 2008 , 103, 2825-35; quiz 2836	0.7	101
62	Usefulness of prophylactic gastrectomy in a novel large hereditary diffuse gastric cancer (HDGC) family. <i>American Journal of Gastroenterology</i> , 2008 , 103, 2160-1	0.7	22
61	Effect of aspirin or resistant starch on colorectal neoplasia in the Lynch syndrome. <i>New England Journal of Medicine</i> , 2008 , 359, 2567-78	59.2	228
60	Multicenter study of ZAP-70 expression in patients with B-cell chronic lymphocytic leukemia using an optimized flow cytometry method. <i>Haematologica</i> , 2008 , 93, 215-23	6.6	30
59	High-resolution genotyping of chromosome 8 in colon adenocarcinomas reveals recurrent break point but no gene mutation in the 8p21 region. <i>Diagnostic Molecular Pathology</i> , 2008 , 17, 90-3		6
58	Duodenal adenocarcinoma and Mut Y human homologue-associated polyposis. <i>European Journal of Gastroenterology and Hepatology</i> , 2008 , 20, 1024-7	2.2	8
57	A large fraction of unclassified variants of the mismatch repair genes MLH1 and MSH2 is associated with splicing defects. <i>Human Mutation</i> , 2008 , 29, 1412-24	4.7	123
56	Mitochondrial D310 mutations in colorectal adenomas: an early but not causative genetic event during colorectal carcinogenesis. <i>International Journal of Cancer</i> , 2008 , 122, 2242-8	7.5	16
55	Y253H mutation appearing in a micro-BCR-ABL (e19a2) CML. <i>Leukemia Research</i> , 2008 , 32, 361-2	2.7	12
54	Colonoscopic screening of first-degree relatives of patients with large adenomas: increased risk of colorectal tumors. <i>Gastroenterology</i> , 2007 , 133, 1086-92	13.3	70
53	Genome-wide association scan identifies a colorectal cancer susceptibility locus on chromosome 8q24. <i>Nature Genetics</i> , 2007 , 39, 989-94	36.3	609
52	Partial duplications of the MSH2 and MLH1 genes in hereditary nonpolyposis colorectal cancer. <i>European Journal of Human Genetics</i> , 2007 , 15, 383-6	5.3	13
51	Rearrangements involving 12q in myeloproliferative disorders: possible role of HMGA2 and SOCS2 genes. <i>Cancer Genetics and Cytogenetics</i> , 2007 , 176, 80-8		25
50	Hysteroscopic findings in women at risk of HNPCC. Results of a prospective observational study. <i>Familial Cancer</i> , 2007 , 6, 295-9	3	21
49	High frequency of chromosome 14 deletion in early-onset colon cancer. <i>Diseases of the Colon and Rectum</i> , 2007 , 50, 1881-6	3.1	18
48	The thorough screening of the MUTYH gene in a large French cohort of sporadic colorectal cancers. <i>Genetic Testing and Molecular Biomarkers</i> , 2007 , 11, 373-9		15
47	Founder and recurrent CDH1 mutations in families with hereditary diffuse gastric cancer. <i>JAMA - Journal of the American Medical Association</i> , 2007 , 297, 2360-72	27.4	324
46	New types of MYST3-CBP and CBP-MYST3 fusion transcripts in t(8;16)(p11;p13) acute myeloid leukemias. <i>Haematologica</i> , 2007 , 92, 262-3	6.6	10

45	Similar colorectal cancer risk in patients with monoallelic and biallelic mutations in the MYH gene identified in a population with adenomatous polyposis. <i>Genetic Testing and Molecular Biomarkers</i> , 2007 , 11, 315-20		31
44	Genetic polymorphisms of MMP1, MMP3 and MMP7 gene promoter and risk of colorectal adenoma. <i>BMC Cancer</i> , 2006 , 6, 270	4.8	45
43	Retention of chromosome arm 5q in stage II colon cancers identifies 83% of liver metastasis occurrences. <i>Genes Chromosomes and Cancer</i> , 2006 , 45, 94-102	5	7
42	Frequency and spectrum of cancers in the Peutz-Jeghers syndrome. <i>Clinical Cancer Research</i> , 2006 , 12, 3209-15	12.9	627
41	SMAD4 germinal mosaicism in a family with juvenile polyposis and hypertrophic osteoarthropathy. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2005 , 41, 117-20	2.8	4
40	Value of microsatellite instability typing in detecting hereditary non-polyposis colorectal cancer. A prospective multicentric study by the Association Aquitaine Gastro. <i>Gastroenterologie Clinique Et Biologique</i> , 2005 , 29, 667-75		8
39	The 5' region of the MSH2 gene involved in hereditary non-polyposis colorectal cancer contains a high density of recombinogenic sequences. <i>Human Mutation</i> , 2005 , 26, 255-61	4.7	31
38	Vessel morphology in SMAD4 and BMPR1A-related juvenile polyposis. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 138A, 113-7	2.5	32
37	Functional analysis of Peutz-Jeghers mutations reveals that the LKB1 C-terminal region exerts a crucial role in regulating both the AMPK pathway and the cell polarity. <i>Human Molecular Genetics</i> , 2005 , 14, 1283-92	5.6	119
36	Analysis of the allele-specific expression of the mismatch repair gene MLH1 using a simple DHPLC-Based Method. <i>Human Mutation</i> , 2004 , 23, 379-84	4.7	20
35	Genotype-phenotype correlation in von Hippel-Lindau families with renal lesions. <i>Human Mutation</i> , 2004 , 24, 215-24	4.7	69
34	Relative frequency and morphology of cancers in STK11 mutation carriers. <i>Gastroenterology</i> , 2004 , 126, 1788-94	13.3	193
33	Mucinous colon carcinomas with microsatellite instability have a lower microvessel density and lower vascular endothelial growth factor expression. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2003 , 442, 111-7	5.1	16
32	Tissue microarray technology: validation in colorectal carcinoma and analysis of p53, hMLH1, and hMSH2 immunohistochemical expression. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2003 , 443, 115-21	5.1	98
31	Association between mutations in the CARD15 (NOD2) gene and Crohn's disease in Israeli Jewish patients. <i>American Journal of Medical Genetics Part A</i> , 2003 , 121A, 240-4		25
30	Microsatellite instability in colorectal carcinoma. The comparison of immunohistochemistry and molecular biology suggests a role for hMSH6 [correction of hMLH6] immunostaining. <i>Archives of Pathology and Laboratory Medicine</i> , 2003 , 127, 694-700	5	73
29	MSH2 in contrast to MLH1 and MSH6 is frequently inactivated by exonic and promoter rearrangements in hereditary nonpolyposis colorectal cancer. <i>Cancer Research</i> , 2002 , 62, 848-53	10.1	78
28	Peutz-Jeghers families unlinked to STK11/LKB1 gene mutations are highly predisposed to primitive biliary adenocarcinoma. <i>Journal of Medical Genetics</i> , 2001 , 38, 356-60	5.8	75

27	Kirsten ras mutations in patients with colorectal cancer: the QASCAL II study. <i>British Journal of Cancer</i> , 2001 , 85, 692-6	8.7	701
26	Familial adenomatous polyposis: prevalence of adenomas in the ileal pouch after restorative proctocolectomy. <i>Annals of Surgery</i> , 2001 , 233, 360-4	7.8	117
25	Deletion mapping of the tumor suppressor locus involved in colorectal cancer on chromosome band 8p21. <i>Genes Chromosomes and Cancer</i> , 1999 , 25, 147-53	5	23
24	Fine deletion mapping of chromosome 8p in non-small-cell lung carcinoma. <i>International Journal of Cancer</i> , 1999 , 81, 854-8	7.5	38
23	A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. <i>Nature</i> , 1998 , 391, 184-7	50.4	1284
22	PTEN germ-line mutations in juvenile polyposis coli. <i>Nature Genetics</i> , 1998 , 18, 12-4	36.3	132
21	Somatically acquired genetic alterations in flat colorectal neoplasias. <i>International Journal of Cancer</i> , 1998 , 77, 366-9	7.5	30
20	Germline mutation profile of the VHL gene in von Hippel-Lindau disease and in sporadic hemangioblastoma. <i>Human Mutation</i> , 1998 , 12, 424-30	4.7	71
19	Genetic alterations in thyroid carcinoma associated with familial adenomatous polyposis: clinical implications and suggestions for early detection. <i>World Journal of Surgery</i> , 1998 , 22, 1231-6	3.3	67
18	Alternative genetic pathways in colorectal carcinogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997 , 94, 12122-7	11.5	194
17	Germline mutations in the Von Hippel-Lindau disease (VHL) gene in families from North America, Europe, and Japan. <i>Human Mutation</i> , 1996 , 8, 348-57	4.7	358
16	Germline mutations in the Von Hippel-Lindau disease (VHL) gene in families from North America, Europe, and Japan 1996 , 8, 348		17
15	High resolution genetic map of the adenomatous polyposis coli gene (APC) region. <i>American Journal of Medical Genetics Part A</i> , 1995 , 56, 413-9		14
14	Predominance of normal karyotype in colorectal tumors from hereditary non-polyposis colorectal cancer patients. <i>Genes Chromosomes and Cancer</i> , 1995 , 14, 223-6	5	23
13	Association of p53 mutations with short survival in colorectal cancer. <i>Gastroenterology</i> , 1994 , 106, 42-8	13.3	323
12	Alleles of the APC gene: an attenuated form of familial polyposis. <i>Cell</i> , 1993 , 75, 951-7	56.2	545
11	Restriction of ocular fundus lesions to a specific subgroup of APC mutations in adenomatous polyposis coli patients. <i>Cell</i> , 1993 , 75, 959-68	56.2	269
10	Is the multiple endocrine neoplasia type 1 gene a suppressor for fundic argyrophil tumors in the Zollinger-Ellison syndrome?. <i>Gastroenterology</i> , 1993 , 105, 579-82	13.3	69

9	Survival and acquired genetic alterations in colorectal cancer. <i>Gastroenterology</i> , 1992 , 102, 1136-1141	13.3	161
8	Detection by DGGE of a new polymorphism closely linked to the adenomatous polyposis coli region. <i>Human Genetics</i> , 1992 , 88, 658-60	6.3	6
7	Genetic characterization of the APC locus involved in familial adenomatous polyposis. <i>Gastroenterology</i> , 1991 , 101, 154-60	13.3	14
6	Association of Ki-ras mutation with differentiation and tumor-formation pathways in colorectal carcinoma. <i>International Journal of Cancer</i> , 1991 , 49, 220-3	7.5	57
5	Reliability of presymptomatic test for adenomatous polyposis coli. <i>Lancet, The</i> , 1991 , 337, 1171-1172	4.0	4
4	Cytogenetic and molecular approaches of polyploidization in colorectal adenocarcinomas. <i>Cancer Genetics and Cytogenetics</i> , 1990 , 44, 107-18		38
3	Preservation of chromosome and DNA characteristics of human colorectal adenocarcinomas after passage in nude mice. <i>International Journal of Cancer</i> , 1989 , 44, 871-8	7.5	21
2	Chromosomes in Ewing@ sarcoma. I. An evaluation of 85 cases of remarkable consistency of t(11;22)(q24;q12). <i>Cancer Genetics and Cytogenetics</i> , 1988 , 32, 229-38		447
1	RFLP identified by the anonymous DNA segment OL VII E10 at 18q21.3 (HGM no. D18S8). <i>Nucleic Acids Research</i> , 1987 , 15, 1348	20.1	19