

# Sylviane Olschwang

## List of Publications by Year in descending order

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118  
papers

14,426  
citations

50276

46  
h-index

19190

118  
g-index

134  
all docs

134  
docs citations

134  
times ranked

15985  
citing authors

#	ARTICLE	IF	CITATIONS
1	A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. <i>Nature</i> , 1998, 391, 184-187.	27.8	1,451
2	Gene Expression Classification of Colon Cancer into Molecular Subtypes: Characterization, Validation, and Prognostic Value. <i>PLoS Medicine</i> , 2013, 10, e1001453.	8.4	1,064
3	Cancer Risks Associated With Germline Mutations in <i>MLH1</i> , <i>MSH2</i> , and <i>MSH6</i> Genes in Lynch Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2011, 305, 2304.	7.4	878
4	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> , The, 2011, 378, 2081-2087.	13.7	849
5	Kirsten ras mutations in patients with colorectal cancer: the RASCAL II study. <i>British Journal of Cancer</i> , 2001, 85, 692-696.	6.4	790
6	Frequency and Spectrum of Cancers in the Peutz-Jeghers Syndrome. <i>Clinical Cancer Research</i> , 2006, 12, 3209-3215.	7.0	746
7	Genome-wide association scan identifies a colorectal cancer susceptibility locus on chromosome 8q24. <i>Nature Genetics</i> , 2007, 39, 989-994.	21.4	676
8	Alleles of the APC gene: An attenuated form of familial polyposis. <i>Cell</i> , 1993, 75, 951-957.	28.9	611
9	Mutations of polycomb-associated gene <i>ASXL1</i> in myelodysplastic syndromes and chronic myelomonocytic leukaemia. <i>British Journal of Haematology</i> , 2009, 145, 788-800.	2.5	537
10	Chromosomes in Ewing's sarcoma. I. An evaluation of 85 cases and remarkable consistency of t(11;22)(q24;q12). <i>Cancer Genetics and Cytogenetics</i> , 1988, 32, 229-238.	1.0	488
11	Germline mutations in the Von Hippel-Lindau disease (VHL) gene in families from North America, Europe, and Japan. <i>Human Mutation</i> , 1996, 8, 348-357.	2.5	436
12	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2007, 297, 2360.	7.4	394
13	Association of p53 mutations with short survival in colorectal cancer. <i>Gastroenterology</i> , 1994, 106, 42-48.	1.3	348
14	Restriction of ocular fundus lesions to a specific subgroup of APC mutations in adenomatous polyposis coli patients. <i>Cell</i> , 1993, 75, 959-968.	28.9	307
15	Effect of Aspirin or Resistant Starch on Colorectal Neoplasia in the Lynch Syndrome. <i>New England Journal of Medicine</i> , 2008, 359, 2567-2578.	27.0	273
16	Relative frequency and morphology of cancers in STK11 mutation carriers. <i>Gastroenterology</i> , 2004, 126, 1788-1794.	1.3	228
17	Alternative genetic pathways in colorectal carcinogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997, 94, 12122-12127.	7.1	209
18	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.	6.2	201

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19	Survival and acquired genetic alterations in colorectal cancer. <i>Gastroenterology</i> , 1992, 102, 1136-1141.	1.3	182
20	PTEN germ-line mutations in juvenile polyposis coli. <i>Nature Genetics</i> , 1998, 18, 12-14.	21.4	153
21	A large fraction of unclassified variants of the mismatch repair genes <i>MLH1</i> and <i>MSH2</i> is associated with splicing defects. <i>Human Mutation</i> , 2008, 29, 1412-1424.	2.5	150
22	Combined mutations of <i>ASXL1</i> , <i>CBL</i> , <i>FLT3</i> , <i>IDH1</i> , <i>IDH2</i> , <i>JAK2</i> , <i>KRAS</i> , <i>NPM1</i> , <i>NRAS</i> , <i>RUNX1</i> , <i>TET2</i> and <i>WT1</i> genes in myelodysplastic syndromes and acute myeloid leukemias. <i>BMC Cancer</i> , 2010, 10, 401.	2.6	140
23	Familial Adenomatous Polyposis: Prevalence of Adenomas in the Ileal Pouch After Restorative Proctocolectomy. <i>Annals of Surgery</i> , 2001, 233, 360-364.	4.2	139
24	Functional analysis of Peutz-Jeghers mutations reveals that the <i>LKB1</i> C-terminal region exerts a crucial role in regulating both the AMPK pathway and the cell polarity. <i>Human Molecular Genetics</i> , 2005, 14, 1283-1292.	2.9	131
25	Identification in Daily Practice of Patients With Lynch Syndrome (Hereditary Nonpolyposis Colorectal) Tj ETQq1 1 0.784314 rgBT /Over <i>Journal of Gastroenterology</i> , 2008, 103, 2825-2835.	0.4	118
26	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, 115-121.	2.8	109
27	Genome profiling of chronic myelomonocytic leukemia: frequent alterations of <i>RAS</i> and <i>RUNX1</i> genes. <i>BMC Cancer</i> , 2008, 8, 299.	2.6	109
28	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> , The, 2012, 13, 1242-1249.	10.7	95
29	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, 3591-3597.	1.6	91
30	Germline mutation profile of the <i>VHL</i> gene in von Hippel-Lindau disease and in sporadic hemangioblastoma. <i>Human Mutation</i> , 1998, 12, 424-430.	2.5	89
31	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, 68-77.	6.3	89
32	Colonoscopic Screening of First-Degree Relatives of Patients With Large Adenomas: Increased Risk of Colorectal Tumors. <i>Gastroenterology</i> , 2007, 133, 1086-1092.	1.3	87
33	<i>MSH2</i> in contrast to <i>MLH1</i> and <i>MSH6</i> is frequently inactivated by exonic and promoter rearrangements in hereditary nonpolyposis colorectal cancer. <i>Cancer Research</i> , 2002, 62, 848-53.	0.9	86
34	Peutz-Jeghers families unlinked to <i>STK11/LKB1</i> gene mutations are highly predisposed to primitive biliary adenocarcinoma. <i>Journal of Medical Genetics</i> , 2001, 38, 356-360.	3.2	81
35	Genotype-phenotype correlation in von Hippel-Lindau families with renal lesions. <i>Human Mutation</i> , 2004, 24, 215-224.	2.5	81
36	Microsatellite Instability in Colorectal Carcinoma. <i>Archives of Pathology and Laboratory Medicine</i> , 2003, 127, 694-700.	2.5	81

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37	Is the multiple endocrine neoplasia type 1 gene a suppressor for fundic argyrophil tumors in the Zollinger-Ellison syndrome?. <i>Gastroenterology</i> , 1993, 105, 579-582.	1.3	79
38	SMARCB1/INI1 germline mutations contribute to 10% of sporadic schwannomatosis. <i>BMC Neurology</i> , 2011, 11, 9.	1.8	76
39	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, 1231-1236.	1.6	75
40	Leiden open variation database of the MUTYH gene. <i>Human Mutation</i> , 2010, 31, 1205-1215.	2.5	72
41	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, 560-565.	2.0	69
42	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.	2.6	65
43	Association of ki-ras mutation with differentiation and tumor-formation pathways in colorectal carcinoma. <i>International Journal of Cancer</i> , 1991, 49, 220-223.	5.1	61
44	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, 867-875.	2.5	58
45	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. <i>Lancet Oncology</i> , The, 2021, 22, 1014-1022.	10.7	58
46	Genetic polymorphisms of MMP1, MMP3 and MMP7 gene promoter and risk of colorectal adenoma. <i>BMC Cancer</i> , 2006, 6, 270.	2.6	56
47	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 Tj ETQq1 1 0.784314 rg5E /Overbo 1006-1015.	1.0	51
48	Vessels' morphology in SMAD4 and BMPR1A-related juvenile polyposis. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 113-117.	1.2	45
49	Cytogenetic and molecular approaches of polyploidization in colorectal adenocarcinomas. <i>Cancer Genetics and Cytogenetics</i> , 1990, 44, 107-118.	1.0	41
50	Fine deletion mapping of chromosome 8p in non-small-cell lung carcinoma. , 1999, 81, 854-858.		40
51	Molecular combing reveals complex 4q35 rearrangements in Facioscapulohumeral dystrophy. <i>Human Mutation</i> , 2017, 38, 1432-1441.	2.5	39
52	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, 315-320.	1.7	37
53	Somatically acquired genetic alterations in flat colorectal neoplasias. , 1998, 77, 366-369.		34
54	8q24 Cancer Risk Allele Associated with Major Metastatic Risk in Inflammatory Breast Cancer. <i>PLoS ONE</i> , 2012, 7, e37943.	2.5	34

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55	Germline mutations in the Von Hippel-Lindau disease (VHL) gene in families from North America, Europe, and Japan. <i>Human Mutation</i> , 1996, 8, 348-357.	2.5	33
56	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, 215-223.	3.5	32
57	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, 721-722.	3.2	32
58	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, 255-261.	2.5	31
59	A Multicenter Blinded Study Evaluating EGFR and KRAS Mutation Testing Methods in the Clinical Non-Small Cell Lung Cancer Setting: ICFCT/ERMETIC2 Project Part 1. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 45-55.	2.8	31
60	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, 240-244.	2.4	29
61	Usefulness of Prophylactic Gastrectomy in a Novel Large Hereditary Diffuse Gastric Cancer (HDGC) Family. <i>American Journal of Gastroenterology</i> , 2008, 103, 2160-2161.	0.4	28
62	The LKB1 complex-AMPK pathway: the tree that hides the forest. <i>Familial Cancer</i> , 2011, 10, 415-424.	1.9	27
63	Deletion mapping of the tumor suppressor locus involved in colorectal cancer on chromosome band 8p21. <i>Cancer</i> , 1999, 25, 147-153.		26
64	Rearrangements involving 12q in myeloproliferative disorders: possible role of HMGA2 and SOCS2 genes. <i>Cancer Genetics and Cytogenetics</i> , 2007, 176, 80-88.	1.0	26
65	PIK3CA mutations predict recurrence in localized microsatellite stable colon cancer. <i>Cancer Medicine</i> , 2015, 4, 371-382.	2.8	25
66	Predominance of normal karyotype in colorectal tumors from hereditary non-polyposis colorectal cancer patients. <i>Genes Chromosomes and Cancer</i> , 1995, 14, 223-226.	2.8	24
67	Analysis of the allele-specific expression of the mismatch repair gene MLH1 using a simple DHPLC-Based Method. <i>Human Mutation</i> , 2004, 23, 379-384.	2.5	24
68	Hysteroscopic findings in women at risk of HNPCC. Results of a prospective observational study. <i>Familial Cancer</i> , 2007, 6, 295-299.	1.9	24
69	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, 331-337.	0.9	23
70	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, 111-117.	2.8	22
71	High Frequency of Chromosome 14 Deletion in Early-Onset Colon Cancer. <i>Diseases of the Colon and Rectum</i> , 2007, 50, 1881-1886.	1.3	22
72	Preservation of chromosome and DNA characteristics of human colorectal adenocarcinomas after passage in nude mice. <i>International Journal of Cancer</i> , 1989, 44, 871-878.	5.1	21

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73	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, 2728-2737.	6.4	20
74	RFLP identified by the anonymous DNA segment OL VII E10 at 18q21.3 (HGM do. D18S8). <i>Nucleic Acids Research</i> , 1987, 15, 1348-1348.	14.5	19
75	Mitochondrial D310 mutations in colorectal adenomas: An early but not causative genetic event during colorectal carcinogenesis. <i>International Journal of Cancer</i> , 2008, 122, 2242-2248.	5.1	19
76	Expression Profiles in Stage II Colon Cancer According to APC Gene Status. <i>Translational Oncology</i> , 2012, 5, 72-76.	3.7	18
77	Genetic characterization of the APC locus involved in familial adenomatous polyposis. <i>Gastroenterology</i> , 1991, 101, 154-160.	1.3	17
78	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, 583-587.	2.5	17
79	Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM) - update 2012. <i>European Journal of Human Genetics</i> , 2013, 21, 118-118.	2.8	16
80	High resolution genetic map of the adenomatous polyposis coli gene (APC) region. <i>American Journal of Medical Genetics Part A</i> , 1995, 56, 413-419.	2.4	15
81	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, 373-380.	1.7	15
82	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-bat036.	3.0	15
83	The UMD-APC Database, a Model of Nation-Wide Knowledge Base: Update with Data from 3,581 Variations. <i>Human Mutation</i> , 2014, 35, 532-536.	2.5	15
84	Partial duplications of the MSH2 and MLH1 genes in hereditary nonpolyposis colorectal cancer. <i>European Journal of Human Genetics</i> , 2007, 15, 383-386.	2.8	14
85	Y253H mutation appearing in a t(8;16)(p11;p13) BCR-ABL (e19a2) CML. <i>Leukemia Research</i> , 2008, 32, 361-362.	0.8	13
86	New types of MYST3-CBP and CBP-MYST3 fusion transcripts in t(8;16)(p11;p13) acute myeloid leukemias. <i>Haematologica</i> , 2007, 92, 262-263.	3.5	12
87	Value of microsatellite instability typing in detecting hereditary non-polyposis colorectal cancer. <i>Gastroenterologie Clinique Et Biologique</i> , 2005, 29, 667-675.	0.9	11
88	Is the controversy on breast cancer as part of the Lynch-related tumor spectrum still open?. <i>Familial Cancer</i> , 2012, 11, 681-683.	1.9	11
89	Improving Mutation Screening in Patients with Colorectal Cancer Predisposition Using Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2017, 19, 589-601.	2.8	11
90	Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2). <i>European Journal of Human Genetics</i> , 2010, 18, 1071-1071.	2.8	10

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91	Clinical utility gene card for: Familial adenomatous polyposis (FAP) and attenuated FAP (AFAP). European Journal of Human Genetics, 2011, 19, 832-832.	2.8	10
92	Characterisation of heterozygous <i>PMS2</i> variants in French patients with Lynch syndrome. Journal of Medical Genetics, 2020, 57, 487-499.	3.2	10
93	High-resolution Genotyping of Chromosome 8 in Colon Adenocarcinomas Reveals Recurrent Break Point But No Gene Mutation in the 8p21 Region. Diagnostic Molecular Pathology, 2008, 17, 90-93.	2.1	9
94	Duodenal adenocarcinoma and Mut Y human homologue-associated polyposis. European Journal of Gastroenterology and Hepatology, 2008, 20, 1024-1027.	1.6	9
95	Clinical Utility Gene Card for: Familial adenomatous polyposis (FAP) and attenuated FAP (AFAP) - update 2014. European Journal of Human Genetics, 2015, 23, 889-889.	2.8	9
96	Retention of chromosome arm 5q in stage II colon cancers identifies 83% of liver metastasis occurrences. Genes Chromosomes and Cancer, 2006, 45, 94-102.	2.8	8
97	SMAD4 protein expression and cell proliferation in colorectal adenocarcinomas. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2011, 459, 511-519.	2.8	7
98	Detection by DGGE of a new polymorphism closely linked to the adenomatous polyposis coli region. Human Genetics, 1992, 88, 658-660.	3.8	6
99	SMAD4 Germinal Mosaicism in a Family with Juvenile Polyposis and Hypertrophic Osteoarthropathy. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 117-120.	1.8	6
100	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. Human Mutation, 2011, 32, 696-697.	2.5	6
101	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	2.5	6
102	Reliability of presymptomatic test for adenomatous polyposis coli. Lancet, The, 1991, 337, 1171-1172.	13.7	5
103	Statistical inference on the penetrances of rare genetic mutations based on a case-family design. Biostatistics, 2010, 11, 519-532.	1.5	5
104	Age-Dependent Cancer Risk Is Not Different in between <i>MSH2</i> and <i>MLH1</i> Mutation Carriers. Journal of Cancer Epidemiology, 2009, 2009, 1-6.	1.1	4
105	KRAS Mutation Spectrum Notably Diverges between Non-small Cell Lung and Colorectal Carcinomas. Journal of Thoracic Oncology, 2012, 7, 773-774.	1.1	4
106	Gastric Adenocarcinoma in Familial Adenomatous Polyposis Can Occur Without Previous Lesions. Journal of Gastrointestinal Cancer, 2014, 45, 377-379.	1.3	4
107	High-resolution analysis of DNA copy number alterations in rectal cancer. Strahlentherapie Und Onkologie, 2014, 190, 1028-1036.	2.0	4
108	Design of a Core Classification Process for DNA Mismatch Repair Variations of A Priori Unknown Functional Significance. Human Mutation, 2013, 34, 920-922.	2.5	3

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109	Frequent intragenic rearrangements of DPYD in colorectal tumours. <i>Pharmacogenomics Journal</i> , 2015, 15, 211-218.	2.0	3
110	Germ-line mutations of the APC gene in patients with FAP associated hepatoblastoma are located out of the mutation cluster region (codons 1286-1513). <i>Gastroenterology</i> , 2000, 118, A45.	1.3	2
111	Analysis of Candidate Genes in Occurrence and Growth of Colorectal Adenomas. <i>Journal of Oncology</i> , 2009, 2009, 1-9.	1.3	2
112	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.	1.5	2
113	Genomic variations integrated database for <i>MUTYH</i> -associated adenomatous polyposis. <i>Journal of Medical Genetics</i> , 2015, 52, 25-27.	3.2	2
114	Chronic Iron deficiency Anemia Caused by a Jejunojejunal Intussusception on a Solitary Hamartomatous Polyp. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2010, 50, 450-452.	1.8	1
115	Abstract 5555: Pharmacogenetics-pharmacokinetics study of bevacizumab in mCRC patients treated with Avastin-Folfiri regimen: Search for predictive markers of response and study of the pharmacokinetics variability. , 2014, , .		1
116	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. <i>Cancéro Digest</i> , 2009, , 7.	0.0	0
117	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.	0.6	0
118	Expression phénotypique de la polyposse adénomateuse familiale. <i>Medecine/Sciences</i> , 1994, 10, 454.	0.2	0