

# Stéphane Richard

## List of Publications by Year in descending order

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119  
papers

12,438  
citations

36303

51  
h-index

24258

110  
g-index

132  
all docs

132  
docs citations

132  
times ranked

12125  
citing authors

#	ARTICLE	IF	CITATIONS
1	Germline and somatic mutations in the tyrosine kinase domain of the MET proto-oncogene in papillary renal carcinomas. <i>Nature Genetics</i> , 1997, 16, 68-73.	21.4	1,461
2	Exome sequencing identifies frequent mutation of the SWI/SNF complex gene PBRM1 in renal carcinoma. <i>Nature</i> , 2011, 469, 539-542.	27.8	1,127
3	Genetic Testing in Pheochromocytoma or Functional Paraganglioma. <i>Journal of Clinical Oncology</i> , 2005, 23, 8812-8818.	1.6	612
4	von Hippel-Lindau disease: A clinical and scientific review. <i>European Journal of Human Genetics</i> , 2011, 19, 617-623.	2.8	588
5	Birt-Hogg-Dubois syndrome: diagnosis and management. <i>Lancet Oncology</i> , The, 2009, 10, 1199-1206.	10.7	509
6	Novel mutations of the MET proto-oncogene in papillary renal carcinomas. <i>Oncogene</i> , 1999, 18, 2343-2350.	5.9	487
7	A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. <i>Nature</i> , 2011, 480, 94-98.	27.8	466
8	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
9	Pancreatic involvement in von Hippel-Lindau disease. <i>Gastroenterology</i> , 2000, 119, 1087-1095.	1.3	374
10	An Antioxidant Response Phenotype Shared between Hereditary and Sporadic Type 2 Papillary Renal Cell Carcinoma. <i>Cancer Cell</i> , 2011, 20, 511-523.	16.8	347
11	<i>PHD2</i> Mutation and Congenital Erythrocytosis with Paraganglioma. <i>New England Journal of Medicine</i> , 2008, 359, 2685-2692.	27.0	284
12	Hereditary leiomyomatosis and renal cell cancer (HLRCC): renal cancer risk, surveillance and treatment. <i>Familial Cancer</i> , 2014, 13, 637-644.	1.9	251
13	Germline BAP1 Mutations Predispose to Renal Cell Carcinomas. <i>American Journal of Human Genetics</i> , 2013, 92, 974-980.	6.2	239
14	Microsporidia Infection in Patients with the Human Immunodeficiency Virus and Unexplained Cholangitis. <i>New England Journal of Medicine</i> , 1993, 328, 95-99.	27.0	230
15	The Y Deletion <i>gr/gr</i> and Susceptibility to Testicular Germ Cell Tumor. <i>American Journal of Human Genetics</i> , 2005, 77, 1034-1043.	6.2	197
16	Protection of p27Kip1 mRNA by quaking RNA binding proteins promotes oligodendrocyte differentiation. <i>Nature Neuroscience</i> , 2005, 8, 27-33.	14.8	151
17	Somatic inactivation of the VHL gene in Von Hippel-Lindau disease tumors. <i>American Journal of Human Genetics</i> , 1997, 60, 765-71.	6.2	149
18	Central nervous system hemangioblastomas, endolymphatic sac tumors, and von Hippel-Lindau disease. <i>Neurosurgical Review</i> , 2000, 23, 1-22.	2.4	147

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19	Genome-wide linkage screen for testicular germ cell tumour susceptibility loci. <i>Human Molecular Genetics</i> , 2006, 15, 443-451.	2.9	138
20	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. <i>Journal of Clinical Oncology</i> , 2016, 34, 2172-2181.	1.6	132
21	Mutations of the VHL gene in sporadic renal cell carcinoma: Definition of a risk factor for VHL patients to develop an RCC. <i>Human Mutation</i> , 1999, 13, 464-475.	2.5	126
22	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> , 2011, 48, 226-234.	3.2	116
23	Renal Cell Carcinoma Programmed Death-ligand 1, a New Direct Target of Hypoxia-inducible Factor-2 Alpha, is Regulated by von Hippel-Lindau Gene Mutation Status. <i>European Urology</i> , 2016, 70, 623-632.	1.9	115
24	Head and Neck Paragangliomas in Von Hippel-Lindau Disease and Multiple Endocrine Neoplasia Type 2. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 1938-1944.	3.6	112
25	Treatment of von Hippel-Lindau retinal hemangioblastoma by the vascular endothelial growth factor receptor inhibitor SU5416 is more effective for associated macular edema than for hemangioblastomas. <i>American Journal of Ophthalmology</i> , 2003, 136, 194-196.	3.3	109
26	Renal involvement in von Hippel-Lindau disease. <i>Kidney International</i> , 1996, 50, 944-951.	5.2	107
27	Von Hippel-Lindau disease. <i>Lancet</i> , The, 2004, 363, 1231-1234.	13.7	106
28	Reassessing the clinical spectrum associated with hereditary leiomyomatosis and renal cell carcinoma syndrome in French <i>fh</i> mutation carriers. <i>Clinical Genetics</i> , 2017, 92, 606-615.	2.0	103
29	Genetic Basis of Congenital Erythrocytosis: Mutation Update and Online Databases. <i>Human Mutation</i> , 2014, 35, 15-26.	2.5	101
30	Inactivation of BHD in sporadic renal tumors. <i>Cancer Research</i> , 2003, 63, 4583-7.	0.9	96
31	A new locus-specific database (LSDB) for mutations in the folliculin ( <i>FLCN</i> ) gene. <i>Human Mutation</i> , 2010, 31, E1043-E1051.	2.5	93
32	Von Hippel-Lindau: How a rare disease illuminates cancer biology. <i>Seminars in Cancer Biology</i> , 2013, 23, 26-37.	9.6	93
33	Retinal hemangioblastoma in von Hippel-Lindau disease: a clinical and molecular study. <i>Investigative Ophthalmology and Visual Science</i> , 2002, 43, 3067-74.	3.3	91
34	Germline mutation profile of the VHL gene in von Hippel-Lindau disease and in sporadic hemangioblastoma. <i>Human Mutation</i> , 1998, 12, 424-430.	2.5	89
35	Genomic expression and single-nucleotide polymorphism profiling discriminates chromophobe renal cell carcinoma and oncocytoma. <i>BMC Cancer</i> , 2010, 10, 196.	2.6	86
36	Somatic mutations of KIT in familial testicular germ cell tumours. <i>British Journal of Cancer</i> , 2004, 90, 2397-2401.	6.4	85

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37	Genotype-phenotype correlation in von Hippel-Lindau families with renal lesions. <i>Human Mutation</i> , 2004, 24, 215-224.	2.5	81
38	Renal cell tumour characteristics in patients with the Birt-Hogg-Dubé cancer susceptibility syndrome: a retrospective, multicentre study. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 163.	2.7	78
39	Endocrine Pancreatic Tumors in von Hippel-Lindau Disease. <i>Pancreas</i> , 2008, 37, 85-93.	1.1	75
40	Vitreoretinal Surgery for Severe Retinal Capillary Hemangiomas in Von Hippel-Lindau Disease. <i>Ophthalmology</i> , 2011, 118, 142-149.	5.2	73
41	Identification of a new VHL exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. <i>Blood</i> , 2018, 132, 469-483.	1.4	70
42	Birt-Hogg-Dubé renal tumors are genetically distinct from other renal neoplasias and are associated with up-regulation of mitochondrial gene expression. <i>BMC Medical Genomics</i> , 2010, 3, 59.	1.5	68
43	Mutations in BHD and TP53 genes, but not in HNF1 $\beta$ gene, in a large series of sporadic chromophobe renal cell carcinoma. <i>British Journal of Cancer</i> , 2007, 96, 336-340.	6.4	65
44	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> , 2018, 31, 974-983.	5.5	65
45	Familial Non-VHL Clear Cell (Conventional) Renal Cell Carcinoma: Clinical Features, Segregation Analysis, and Mutation Analysis of <i>FLCN</i> . <i>Clinical Cancer Research</i> , 2008, 14, 5925-5930.	7.0	64
46	Molecular Profiling of Pancreatic Neuroendocrine Tumors in Sporadic and Von Hippel-Lindau Patients. <i>Clinical Cancer Research</i> , 2012, 18, 2838-2849.	7.0	61
47	Pancreatic Endocrine Microadenomatosis in Patients With von Hippel-Lindau Disease. <i>American Journal of Surgical Pathology</i> , 2009, 33, 739-748.	3.7	60
48	Enterocytozoon bienewsi infection in acquired immunodeficiency syndrome-related sclerosing cholangitis. <i>Gastroenterology</i> , 1992, 102, 1778-1781.	1.3	59
49	Somatic Pairing of Chromosome 19 in Renal Oncocytoma Is Associated with Deregulated ELGN2-Mediated Oxygen-Sensing Response. <i>PLoS Genetics</i> , 2008, 4, e1000176.	3.5	58
50	Radiofrequency ablation of renal tumours: diagnostic accuracy of contrast-enhanced ultrasound for early detection of residual tumour. <i>European Radiology</i> , 2010, 20, 1812-1821.	4.5	53
51	Paradoxical secondary polycythemia in von Hippel-Lindau patients treated with anti-vascular endothelial growth factor receptor therapy. <i>Blood</i> , 2002, 99, 3851-3853.	1.4	50
52	Distinct deregulation of the hypoxia inducible factor by PHD2 mutants identified in germline DNA of patients with polycythemia. <i>Haematologica</i> , 2012, 97, 9-14.	3.5	50
53	Spectrum of Abdominal Imaging Findings in von Hippel-Lindau Disease. <i>American Journal of Roentgenology</i> , 2003, 181, 1049-1054.	2.2	49
54	Characterization of endolymphatic sac tumors and von Hippel-Lindau disease in the International Endolymphatic Sac Tumor Registry. <i>Head and Neck</i> , 2016, 38, E673-9.	2.0	48

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55	Long-term Prognosis of Resected Pancreatic Neuroendocrine Tumors in von Hippel-Lindau Disease Is Favorable and Not Influenced by Small Tumors Left in Place. <i>Annals of Surgery</i> , 2015, 262, 384-388.	4.2	46
56	Natural History of Supratentorial Hemangioblastomas in von Hippel-Lindau Disease. <i>Neurosurgery</i> , 2010, 67, 577-587.	1.1	42
57	The International Testicular Cancer Linkage Consortium: A clinicopathologic descriptive analysis of 461 familial malignant testicular germ cell tumor kindred. <i>Urologic Oncology: Seminars and Original Investigations</i> , 2010, 28, 492-499.	1.6	42
58	Combined <i>Vhlh</i> and <i>Pten</i> Mutation Causes Genital Tract Cystadenoma and Squamous Metaplasia. <i>Molecular and Cellular Biology</i> , 2008, 28, 4536-4548.	2.3	41
59	Local Recurrence After Nephron-Sparing Surgery in von Hippel-Lindau Disease. <i>Urology</i> , 2007, 70, 435-439.	1.0	40
60	The role of PHD2 mutations in the pathogenesis of erythrocytosis. <i>Hypoxia (Auckland, N Z)</i> , 2014, 2, 71.	1.9	39
61	Progress in Nephron Sparing Therapy for Renal Cell Carcinoma and von Hippel-Lindau Disease. <i>Journal of Urology</i> , 2011, 185, 2056-2060.	0.4	38
62	A germline mutation in <i>PBRM1</i> predisposes to renal cell carcinoma. <i>Journal of Medical Genetics</i> , 2015, 52, 426-430.	3.2	38
63	Coexpression of erythropoietin and erythropoietin receptor in von Hippel-Lindau disease-associated renal cysts and renal cell carcinoma. <i>Clinical Cancer Research</i> , 2005, 11, 1059-64.	7.0	38
64	Nephron Sparing Surgery for Renal Cell Carcinoma and von Hippel-Lindau Disease: A Single Center Experience. <i>Journal of Urology</i> , 2003, 170, 1752-1755.	0.4	37
65	Analysis of the <i>DND1</i> gene in men with sporadic and familial testicular germ cell tumors. <i>Genes Chromosomes and Cancer</i> , 2008, 47, 247-252.	2.8	37
66	Results of microsurgical treatment of medulla oblongata and spinal cord hemangioblastomas: a comparison of two distinct clinical patient groups. <i>Journal of Neuro-Oncology</i> , 2009, 93, 133-137.	2.9	37
67	Management of Endolymphatic Sac Tumors. <i>Otology and Neurotology</i> , 2014, 35, 899-904.	1.3	35
68	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
69	Genetic Evidence of a Precisely Tuned Dysregulation in the Hypoxia Signaling Pathway during Oncogenesis. <i>Cancer Research</i> , 2014, 74, 6554-6564.	0.9	32
70	Allelic loss on chromosome 22 correlates with histopathological predictors of recurrence of meningiomas. <i>International Journal of Cancer</i> , 1992, 50, 391-394.	5.1	30
71	A novel familial germline mutation in the initiator codon of the BHD gene in a patient with Birt-Hogg-Dubé syndrome. <i>British Journal of Dermatology</i> , 2006, 155, 1067-1069.	1.5	29
72	No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. <i>Familial Cancer</i> , 2010, 9, 245-251.	1.9	26

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73	Biphasic Squamoid Alveolar Renal Cell Carcinoma: 2 Cases in a Family Supporting a Continuous Spectrum With Papillary Type I Renal Cell Carcinoma. <i>American Journal of Surgical Pathology</i> , 2017, 41, 1011-1012.	3.7	26
74	Trichloroethylene exposure and somatic mutations of the VHL gene in patients with Renal Cell Carcinoma. <i>Journal of Occupational Medicine and Toxicology</i> , 2007, 2, 13.	2.2	23
75	Axitinib Induces Paradoxical Erythropoietin Synthesis in Metastatic Renal Cell Carcinoma. <i>Journal of Clinical Oncology</i> , 2009, 27, 472-473.	1.6	23
76	iPSC-Derived Embryoid Bodies as Models of c-Met-Mutated Hereditary Papillary Renal Cell Carcinoma. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4867.	4.1	23
77	Attitudes of von Hippel-Lindau disease patients towards presymptomatic genetic diagnosis in children and prenatal diagnosis. <i>Journal of Medical Genetics</i> , 2000, 37, 476-479.	3.2	22
78	Sunitinib for the treatment of benign and malignant neoplasms from von Hippel-Lindau disease: A single-arm, prospective phase II clinical study from the PREDIR group. <i>Oncotarget</i> , 2016, 7, 85306-85317.	1.8	22
79	von Hippel-Lindau disease: recent advances and therapeutic perspectives. <i>Expert Review of Anticancer Therapy</i> , 2003, 3, 215-233.	2.4	21
80	The growing family of hereditary renal cell carcinoma. <i>Nephrology Dialysis Transplantation</i> , 2004, 19, 2954-2958.	0.7	21
81	Somatic von Hippel-Lindau (VHL) gene analysis and clinical outcome under antiangiogenic treatment in metastatic renal cell carcinoma: preliminary results. <i>Targeted Oncology</i> , 2007, 2, 3-6.	3.6	20
82	Del cell line: A "malignant histiocytosis" CD30 + T(5;6)(Q35;P21) cell line. <i>International Journal of Cancer</i> , 1990, 45, 546-553.	5.1	19
83	Telomere crisis in kidney epithelial cells promotes the acquisition of a microRNA signature retrieved in aggressive renal cell carcinomas. <i>Carcinogenesis</i> , 2013, 34, 1173-1180.	2.8	19
84	Long polymerase chain reaction in detection of germline deletions in the von Hippel-Lindau tumour suppressor gene. <i>Human Genetics</i> , 1999, 105, 333-336.	3.8	18
85	Laser Photocoagulation for Peripheral Retinal Capillary Hemangioblastoma in von Hippel-Lindau Disease. <i>Ophthalmology Retina</i> , 2017, 1, 59-67.	2.4	18
86	Response to systemic therapy in fumarate hydratase-deficient renal cell carcinoma. <i>European Journal of Cancer</i> , 2021, 151, 106-114.	2.8	18
87	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> , 2019, 21, 462-470.	2.8	16
88	MET alterations in biphasic squamoid alveolar papillary renal cell carcinomas and clinicopathological features. <i>Modern Pathology</i> , 2021, 34, 647-659.	5.5	16
89	High incidence of renal tumours in vitamins A and E synthesis workers: A new cause of occupational cancer?. <i>International Journal of Cancer</i> , 2004, 108, 942-944.	5.1	14
90	Von Hippel-Lindau disease and aggressive GH-PRL pituitary adenoma in a young boy. <i>Annales D'Endocrinologie</i> , 2012, 73, 37-42.	1.4	14

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91	Fumarate Hydratase-deficient Cell Line NCCFH1 as a New In Vitro Model of Hereditary Papillary Renal Cell Carcinoma Type 2. <i>Anticancer Research</i> , 2015, 35, 6639-53.	1.1	14
92	Novel somatic mutations of the VHL gene in an erythropoietin-producing renal carcinoma associated with secondary polycythemia and elevated circulating endothelial progenitor cells. <i>American Journal of Hematology</i> , 2008, 83, 155-158.	4.1	13
93	Endolymphatic Sac Tumors in von Hippel-Lindau Disease. <i>Otology and Neurotology</i> , 2010, 31, 660-664.	1.3	13
94	Surgical resection of medulla oblongata hemangioblastomas: outcome and complications. <i>Acta Neurochirurgica</i> , 2016, 158, 1333-1341.	1.7	12
95	Identification of a new aggressive axis driven by ciliogenesis and absence of VDAC1- $\beta$ in clear cell Renal Cell Carcinoma patients. <i>Theranostics</i> , 2020, 10, 2696-2713.	10.0	12
96	Germline mutations in the new E1 <sup>TM</sup> cryptic exon of the <i>VHL</i> gene in patients with tumours of von Hippel-Lindau disease spectrum or with paraganglioma. <i>Journal of Medical Genetics</i> , 2020, 57, 752-759.	3.2	12
97	Congenital Ciliary Aplasia in Two Siblings. <i>Pathology Research and Practice</i> , 1989, 185, 181-183.	2.3	10
98	Conservative management of endolymphatic sac tumors in von Hippel-Lindau disease: case report. <i>Acta Neurochirurgica</i> , 2011, 153, 42-47.	1.7	10
99	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> , 2018, 53, 1455-1468.	3.3	9
100	Genotypic differences in hemangiopericytic meningioma. <i>Human Pathology</i> , 1991, 22, 402.	2.0	8
101	Novel <i>FH</i> 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> , 2010, 163, 1337-1339.	1.5	8
102	Novel germline <i>MET</i> pathogenic variants in French patients with papillary renal cell carcinomas type I. <i>Human Mutation</i> , 2022, 43, 316-327.	2.5	8
103	Difficult Diagnosis of Atypical Cystic Pancreatic Lesions in von Hippel-Lindau Disease. <i>Journal of Computer Assisted Tomography</i> , 2010, 34, 140-145.	0.9	7
104	Pathological heterogeneity in sporadic synchronous renal tumors: Is the histological concordance predictable?. <i>Urologic Oncology: Seminars and Original Investigations</i> , 2018, 36, 11.e7-11.e12.	1.6	7
105	Germline mutation in the NBR1 gene involved in autophagy detected in a family with renal tumors. <i>Cancer Genetics</i> , 2021, 258-259, 51-56.	0.4	5
106	Involvement of PBRM1 in VHL disease-associated clear cell renal cell carcinoma and its putative relationship with the HIF pathway. <i>Oncology Letters</i> , 2021, 22, 835.	1.8	5
107	Diagnosis of pheochromocytoma and laparoscopic adrenalectomy in two anephric patients with von hippel-lindau disease. <i>American Journal of Kidney Diseases</i> , 2002, 39, e6.1-e6.4.	1.9	4
108	Supratentorial Hemangioblastoma in the Neonatal Period. <i>Pediatric Neurosurgery</i> , 2009, 45, 155-156.	0.7	4

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109	Letter to the Editor: Pregnancy and von Hippel-Lindau disease. Journal of Neurosurgery, 2013, 118, 1380-1382.	1.6	4
110	Pancreatic Involvement in Von Hippel-Lindau Disease. , 2004, , 144-152.		4
111	A type 2B von Hippel-Lindau family masquerading as a metastatic sporadic renal cell carcinoma. BJU International, 2003, 91, 425-426.	2.5	3
112	A comparison study reveals important features of agreement and disagreement between summarized DNA and RNA data obtained from renal cell carcinoma. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2008, 657, 77-83.	1.7	3
113	Intermitochondrial junctions in a subpopulation of peripheral blood lymphocytes from healthy subjects*. Biology of the Cell, 1990, 70, 27-32.	2.0	2
114	Radiology Quiz Case. JAMA Otolaryngology, 2002, 128, 855.	1.2	2
115	Clear cell and papillary renal cell carcinomas in hereditary papillary renal cell carcinoma (HPRCC) syndrome: a case report. Diagnostic Pathology, 2021, 16, 107.	2.0	2
116	Congenital Soft Tissue Dysplasias: A Morphological and Biochemical Study. Pediatric Pathology, 1994, 14, 873-894.	0.5	1
117	Familial cancer syndromes. Lancet, The, 1994, 343, 1222.	13.7	1
118	Maladie de von Hippel-Lindau. , 2009, , 179-182.		0
119	Abstract 3820: Deregulation of KEAP1-NRF axis in phenotypically type 2 papillary renal cell carcinoma. , 2011, , .		0