Julian R Sampson

List of Publications by Year
in descending order

112 papers	15,261 citations	28274 55 h-index	24982 109 g-index
121 all docs	121 docs citations	121 times ranked	12167 citing authors

#	Article	IF	Citations
1	Identification of the Tuberous Sclerosis Gene TSC1 on Chromosome 9q34. Science, 1997, 277, 805-808.	12.6	1,550
2	Inherited variants of MYH associated with somatic G:Câ†'T:A mutations in colorectal tumors. Nature Genetics, 2002, 30, 227-232.	21.4	1,239
3	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
4	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
5	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. Gut, 2013, 62, 812-823.	12.1	630
6	Tuberous sclerosis complex. Nature Reviews Disease Primers, 2016, 2, 16035.	30.5	473
7	Deletion of the TSC2 and PKD1 genes associated with severe infantile polycystic kidney disease â€" a contiguous gene syndrome. Nature Genetics, 1994, 8, 328-332.	21.4	466
8	Comprehensive Mutation Analysis of TSC1 and TSC2â€"and Phenotypic Correlations in 150 Families with Tuberous Sclerosis. American Journal of Human Genetics, 1999, 64, 1305-1315.	6.2	453
9	Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. Lancet, The, 2003, 362, 39-41.	13.7	421
10	Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. Gut, 2017, 66, 464-472.	12.1	411
11	Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut, 2018, 67, 1306-1316.	12.1	410
12	Biallelic germline mutations in MYH predispose to multiple colorectal adenoma and somatic G:C->T:A mutations. Human Molecular Genetics, 2002, 11, 2961-2967.	2.9	365
13	Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genetics in Medicine, 2020, 22, 15-25.	2.4	365
14	Renal Cystic Disease in Tuberous Sclerosis: Role of the Polycystic Kidney Disease 1 Gene. American Journal of Human Genetics, 1997, 61, 843-851.	6.2	331
15	Molecular genetic advances in tuberous sclerosis. Human Genetics, 2000, 107, 97-114.	3.8	323
16	Expanded Extracolonic Tumor Spectrum in MUTYH-Associated Polyposis. Gastroenterology, 2009, 137, 1976-1985.e10.	1.3	295
17	Sirolimus Therapy for Angiomyolipoma in Tuberous Sclerosis and Sporadic Lymphangioleiomyomatosis: A Phase 2 Trial. Clinical Cancer Research, 2011, 17, 4071-4081.	7.0	278
18	Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66.	2.1	230

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19	The tuberous sclerosis-1 (TSC1) gene product hamartin suppresses cell growth and augments the expression of the TSC2 product tuberin by inhibiting its ubiquitination. Oncogene, 2000, 19, 6306-6316.	5.9	227
20	Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled trial. Lancet, The, 2020, 395, 1855-1863.	13.7	220
21	Sirolimus Therapy in Tuberous Sclerosis or Sporadic Lymphangioleiomyomatosis. New England Journal of Medicine, 2008, 358, 200-203.	27.0	208
22	Tuberous sclerosis complex tumor suppressorâ€“mediated S6 kinase inhibition by phosphatidylinositide-3-OH kinase is mTOR independent. Journal of Cell Biology, 2002, 159, 217-224.	5.2	199
23	Early onset seizures and Rett-like features associated with mutations in CDKL5. European Journal of Human Genetics, 2005, 13, 1113-1120.	2.8	160
24	Analysis of MUTYH Genotypes and Colorectal Phenotypes in Patients With MUTYH-Associated Polyposis. Gastroenterology, 2009, 136, 471-476.	1.3	149
25	Characterization of GATA3 Mutations in the Hypoparathyroidism, Deafness, and Renal Dysplasia (HDR) Syndrome. Journal of Biological Chemistry, 2004, 279, 22624-22634.	3.4	145
26	MUTYH-associated polyposisâ€”From defect in base excision repair to clinical genetic testing. DNA Repair, 2007, 6, 274-279.	2.8	135
27	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. Gut, 2017, 66, 1657-1664.	12.1	127
28	People of the British Isles: preliminary analysis of genotypes and surnames in a UK-control population. European Journal of Human Genetics, 2012, 20, 203-210.	2.8	126
29	Germline APC variants in patients with multiple colorectal adenomas, with evidence for the particular importance of E1317Q. Human Molecular Genetics, 2000, 9, 2215-2221.	2.9	125
30	Characterization of large rearrangements in autosomal dominant polycystic kidney disease and the PKD1/TSC2 contiguous gene syndrome. Kidney International, 2008, 74, 1468-1479.	5.2	120
31	Comprehensive analysis of the contribution of germlineMYH variation to early-onset colorectal cancer. International Journal of Cancer, 2004, 109, 554-558.	5.1	114
32	Increased Colorectal Cancer Incidence in Obligate Carriers of Heterozygous Mutations in MUTYH. Gastroenterology, 2009, 137, 489-494.e1.	1.3	114
33	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56.	5.2	110
34	The Tuberous Sclerosis 2000 Study: presentation, initial assessments and implications for diagnosis and management. Archives of Disease in Childhood, 2011, 96, 1020-1025.	1.9	104
35	Therapeutic targeting of mTOR in tuberous sclerosis. Biochemical Society Transactions, 2009, 37, 259-264.	3.4	99
36	Randomized Trial of a Specialist Genetic Assessment Service for Familial Breast Cancer. Journal of the National Cancer Institute, 2000, 92, 1345-1351.	6.3	89

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37	Spectrum of mutations in the renin-angiotensin system genes in autosomal recessive renal tubular dysgenesis. Human Mutation, 2012, 33, 316-326.	2.5	86
38	Correlation between clinical severity in patients with Rett syndrome with a p.R168X or p.T158M MECP2 mutation, and the direction and degree of skewing of X-chromosome inactivation. Journal of Medical Genetics, 2006, 44, 148-152.	3.2	83
39	Detailed mapping of germline deletions of the von Hippelâ€"Lindau disease tumour suppressor gene. Human Molecular Genetics, 1994, 3, 595-598.	2.9	81
40	Extensive Telomere Erosion in the Initiation of Colorectal Adenomas and Its Association With Chromosomal Instability. Journal of the National Cancer Institute, 2013, 105, 1202-1211.	6.3	81
41	Exposing the MYtH about base excision repair and human inherited disease. Human Molecular Genetics, 2003, 12, R159-R165.	2.9	80
42	Multiple Rare Nonsynonymous Variants in the Adenomatous Polyposis Coli Gene Predispose to Colorectal Adenomas. Cancer Research, 2008, 68, 358-363.	0.9	77
43	The role of mTOR signalling in neurogenesis, insights from tuberous sclerosis complex. Seminars in Cell and Developmental Biology, 2016, 52, 12-20.	5.0	74
44	Functional assessment of variants in the TSC1 and TSC2 genes identified in individuals with Tuberous Sclerosis Complex. Human Mutation, 2011, 32, 424-435.	2.5	73
45	Leiden open variation database of the MUTYH gene. Human Mutation, 2010, 31, 1205-1215.	2.5	72
46	Cross-Species Comparison of Human and Mouse Intestinal Polyps Reveals Conserved Mechanisms in Adenomatous Polyposis Coli (APC)-Driven Tumorigenesis. American Journal of Pathology, 2008, 172, 1363-1380.	3.8	71
47	Comparative Analysis and Genomic Structure of the Tuberous Sclerosis 2 (TSC2) Gene in Human and Pufferfish. Human Molecular Genetics, 1996, 5, 131-137.	2.9	66
48	G3BPs tether the TSC complex to lysosomes and suppress mTORC1 signaling. Cell, 2021, 184, 655-674.e27.	28.9	65
49	Alternative Splicing of the Tuberous Sclerosis 2 (TSC2) Gene in Human and Mouse Tissues. Genomics, 1995, 27, 475-480.	2.9	64
50	A mouse model of tuberous sclerosis 1 showing background specific early post-natal mortality and metastatic renal cell carcinoma. Human Molecular Genetics, 2005, 14, 1839-1850.	2.9	63
51	Chromosomal Instability in MYH- and APC-Mutant Adenomatous Polyps. Cancer Research, 2006, 66, 2514-2519.	0.9	62
52	Functional characterization of two human MutY homolog (hMYH) missense mutations (R227W and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf Nucleic Acids Research, 2005, 33, 597-604.	14.5	61
53	Survival in women with MMR mutations and ovarian cancer: a multicentre study in Lynch syndrome kindreds. Journal of Medical Genetics, 2010, 47, 99-102.	3.2	61
54	MUTYH-associated polyposis. Bailliere's Best Practice and Research in Clinical Gastroenterology, 2009, 23, 209-218.	2.4	60

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55	Application and evaluation of denaturing HPLC for molecular genetic analysis in tuberous sclerosis. Human Genetics, 2000, 106, 663-668.	3.8	58
56	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	10.7	58
57	The clinical profile of tuberous sclerosis complex (TSC) in the United Kingdom: A retrospective cohort study in the Clinical Practice Research Datalink (CPRD). European Journal of Paediatric Neurology, 2016, 20, 296-308.	1.6	56
58	The â€œunnaturalâ€•history of colorectal cancer in Lynch syndrome: Lessons from colonoscopy surveillance. International Journal of Cancer, 2021, 148, 800-811.	5.1	55
59	Epilepsy in Tuberous Sclerosis: Phenotypes, Mechanisms, and Treatments. Seminars in Neurology, 2015, 35, 269-276.	1.4	54
60	Low level mosaicism detectable by DHPLC but not by direct sequencing. Human Mutation, 2001, 17, 233-234.	2.5	53
61	Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. Hereditary Cancer in Clinical Practice, 2017, 15, 18.	1.5	49
62	Tsc1 Haploinsufficiency without Mammalian Target of Rapamycin Activation Is Sufficient for Renal Cyst Formation in Tsc1+/â^' Mice. Cancer Research, 2006, 66, 7934-7938.	0.9	47
63	Tuberous sclerosis causing mutants of the TSC2 gene product affect proliferation and p27 expression. Oncogene, 2001, 20, 4904-4909.	5.9	46
64	Evidence for pericyte origin of TSC-associated renal angiomyolipomas and implications for angiotensin receptor inhibition therapy. American Journal of Physiology - Renal Physiology, 2014, 307, F560-F570.	2.7	44
65	Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 8.	1.5	42
66	Molecular analysis of the TSC1 and TSC2 tumour suppressor genes in sporadic glial and glioneuronal tumours. Human Genetics, 2000, 107, 350-356.	3.8	41
67	Survival of MUTYH-Associated Polyposis Patients With Colorectal Cancer and Matched Control Colorectal Cancer Patients. Journal of the National Cancer Institute, 2010, 102, 1724-1730.	6.3	40
68	Adenoma development in familial adenomatous polyposis and <scp>MUTYH</scp>â€associated polyposis: somatic landscape and driver genes. Journal of Pathology, 2016, 238, 98-108.	4.5	39
69	Different combinations of biallelic APC mutation confer different growth advantages in colorectal tumours. Cancer Research, 2002, 62, 363-6.	0.9	32
70	Identification of a leader exon and a core promoter for the rat tuberous sclerosis 2 (Tsc2) gene and structural comparison with the human homolog. Mammalian Genome, 1997, 8, 554-558.	2.2	30
71	Endoplasmic reticulum stress and cell death in mTORC1â€overactive cells is induced by nelfinavir andÂenhanced by chloroquine. Molecular Oncology, 2015, 9, 675-688.	4.6	30
72	Inherited MUTYH mutations cause elevated somatic mutation rates and distinctive mutational signatures in normal human cells. Nature Communications, 2022, 13, .	12.8	30

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73	Response to everolimus is seen in TSC-associated SEGAs and angiomyolipomas independent of mutation type and site in TSC1 and TSC2. European Journal of Human Genetics, 2015, 23, 1665-1672.	2.8	29
74	Risk-reducing hysterectomy and bilateral salpingo-oophorectomy in female heterozygotes of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. Genetics in Medicine, 2021, 23, 705-712.	2.4	28
75	Survival by colon cancer stage and screening interval in Lynch syndrome: a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 28.	1.5	27
76	Burden and Profile of Somatic Mutation in Duodenal Adenomas from Patients with Familial Adenomatous- and MUTYH-associated Polyposis. Clinical Cancer Research, 2017, 23, 6721-6732.	7.0	24
77	The impact of chromoendoscopy for surveillance of the duodenum in patients with MUTYH-associated polyposis and familial adenomatous polyposis. Gastrointestinal Endoscopy, 2018, 88, 665-673.	1.0	24
78	Germline MBD4 deficiency causes a multi-tumor predisposition syndrome. American Journal of Human Genetics, 2022, 109, 953-960.	6.2	23
79	Gorlin syndrome: Identification of 4 novel germ-line mutations of the human patched (PTCH) gene. Human Mutation, 1998, 11, 480-480.	2.5	21
80	The economic burden of tuberous sclerosis complex in the UK: A retrospective cohort study in the Clinical Practice Research Datalink. Journal of Medical Economics, 2016, 19, 1087-1098.	2.1	21
81	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
82	Duodenal Adenomas and Cancer in MUTYH-associated Polyposis: An International Cohort Study. Gastroenterology, 2021, 160, 952-954.e4.	1.3	20
83	Prospective observational data informs understanding and future management of Lynch syndrome: insights from the Prospective Lynch Syndrome Database (PLSD). Familial Cancer, 2021, 20, 35-39.	1.9	19
84	Inherited predisposition to colorectal cancer: towards a more complete picture. Journal of Medical Genetics, 2015, 52, 791-796.	3.2	17
85	Refining the Primrose syndrome phenotype: A study of five patients with ZBTB20 de novo variants and a review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 344-349.	1.2	16
86	Intellectual ability in tuberous sclerosis complex correlates with predicted effects of mutations on TSC1 and TSC2 proteins. Journal of Medical Genetics, 2015, 52, 815-822.	3.2	15
87	Feasibility and economic assessment of chromocolonoscopy for detection of proximal serrated neoplasia within a population-based colorectal cancer screening programme (CONSCOP): an open-label, randomised controlled non-inferiority trial. The Lancet Gastroenterology and Hepatology, 2019, 4, 364-375.	8.1	15
88	Application and evaluation of denaturing HPLC for molecular genetic analysis in tuberous sclerosis. Human Genetics, 2000, 106, 663-668.	3.8	15
89	Efficacy of Dual Inhibition of Glycolysis and Glutaminolysis for Therapy of Renal Lesions in Tsc2+/â^' Mice. Neoplasia, 2019, 21, 230-238.	5.3	13
90	Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database. Journal of Clinical Medicine, 2020, 9, 2290.	2.4	12

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91	The use of everolimus in the treatment of neurocognitive problems in tuberous sclerosis (TRON): study protocol for a randomised controlled trial. Trials, 2016, 17, 398.	1.6	11
92	Tuberous Sclerosis Complex (TSC): Expert Recommendations for Provision of Coordinated Care. Frontiers in Neurology, 2019, 10, 1116.	2.4	11
93	Uptake of hysterectomy and bilateral salpingo-oophorectomy in carriers of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. European Journal of Cancer, 2021, 148, 124-133.	2.8	11
94	No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study. Journal of Clinical Medicine, 2021, 10, 2856.	2.4	11
95	Loss of tuberous sclerosis complex 2 sensitizes tumors to nelfinavirâˆ’bortezomib therapy to intensify endoplasmic reticulum stress-induced cell death. Oncogene, 2018, 37, 5913-5925.	5.9	10
96	Allosteric and ATP-Competitive Inhibitors of mTOR Effectively Suppress Tumor Progression-Associated Epithelial-Mesenchymal Transition in the Kidneys of Tsc2+/âˆ’ Mice. Neoplasia, 2019, 21, 731-739.	5.3	9
97	Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum. International Journal of Cancer, 2021, 148, 512-513.	5.1	9
98	TheAPCVariant p.Glu1317Gln predisposes to colorectal adenomas by a novel mechanism of relaxing the target for tumorigenic somaticAPCmutations. Human Mutation, 2009, 30, 1412-1418.	2.5	8
99	The dual PI3K/mTOR inhibitor GSK2126458 is effective for treating solid renal tumours in Tsc2+/- mice through suppression of cell proliferation and induction of apoptosis. Oncotarget, 2017, 8, 58504-58512.	1.8	8
100	Phenotypes associated with inherited and developmental somatic mutations in genes encoding mTOR pathway components. Seminars in Cell and Developmental Biology, 2014, 36, 140-146.	5.0	7
101	Combination of Everolimus with Sorafenib for Solid Renal Tumors in Tsc2+/âˆ’ Mice Is Superior to Everolimus Alone. Neoplasia, 2017, 19, 112-120.	5.3	6
102	Genomic organization and comparative analysis of the mouse tuberous sclerosis 1 (Tsc1) locus. Mammalian Genome, 2000, 11, 1135-1138.	2.2	5
103	APC transcription studies and molecular diagnosis of familial adenomatous polyposis. European Journal of Human Genetics, 2020, 28, 118-121.	2.8	5
104	Using data from the 100,000 Genomes Project to resolve conflicting interpretations of a recurrent TUBB2A mutation. Journal of Medical Genetics, 2021, , jmedgenet-2020-107528.	3.2	3
105	Assessment of Response of Kidney Tumors to Rapamycin and Atorvastatin in Tsc1 +/âˆ’ Mice. Translational Oncology, 2017, 10, 793-799.	3.7	2
106	Gorlin syndrome: Identification of 4 novel germâ€line mutations of the human patched (PTCH) gene. Human Mutation, 1998, 11, 480-480.	2.5	2
107	Letter to the Editor-Recent advances in Lynch syndrome. Familial Cancer, 2021, 20, 117-118.	1.9	1
108	Gastrointestinal polyposis syndromes for the general gastroenterologist. Frontline Gastroenterology, 2014, 5, 68-76.	1.8	0

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109	Response to Tolva et al.. Genetics in Medicine, 2020, 22, 813-814.	2.4	0
110	Peter Harper. European Journal of Human Genetics, 2021, 29, 1168-1170.	2.8	0
111	Towards evidence-based personalised precision medicine for Lynch syndrome. Lancet Oncology, The, 2021, 22, e383.	10.7	0
112	Response to Chambuso etÂal. Genetics in Medicine, 2022, , .	2.4	0