Ann-Marie Patch

List of Publications by Year
in descending order

77 papers	20,005 citations	66315 42 h-index	69214 77 g-index
84 all docs	84 docs citations	84 times ranked	31059 citing authors

#	Article	IF	Citations
1	A Common Variant in the FTO Gene Is Associated with Body Mass Index and Predisposes to Childhood and Adult Obesity. Science, 2007, 316, 889-894.	6.0	3,884
2	Genomic analyses identify molecular subtypes of pancreatic cancer. Nature, 2016, 531, 47-52.	13.7	2,700
3	Whole genomes redefine the mutational landscape of pancreatic cancer. Nature, 2015, 518, 495-501.	13.7	2,132
4	Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes. Nature, 2012, 491, 399-405.	13.7	1,741
5	Wholeâ€“genome characterization of chemoresistant ovarian cancer. Nature, 2015, 521, 489-494.	13.7	1,206
6	Whole-genome landscapes of major melanoma subtypes. Nature, 2017, 545, 175-180.	13.7	1,068
7	Whole-genome landscape of pancreatic neuroendocrine tumours. Nature, 2017, 543, 65-71.	13.7	716
8	Insulin gene mutations as a cause of permanent neonatal diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15040-15044.	3.3	494
9	Insulin Mutation Screening in 1,044 Patients With Diabetes. Diabetes, 2008, 57, 1034-1042.	0.3	347
10	Using SIFT and PolyPhen to Predict Loss-of-Function and Gain-of-Function Mutations. Genetic Testing and Molecular Biomarkers, 2010, 14, 533-537.	0.3	330
11	Mutations in ATP-Sensitive K+ Channel Genes Cause Transient Neonatal Diabetes and Permanent Diabetes in Childhood or Adulthood. Diabetes, 2007, 56, 1930-1937.	0.3	320
12	Rfx6 directs islet formation and insulin production in mice and humans. Nature, 2010, 463, 775-780.	13.7	300
13	A2AR Adenosine Signaling Suppresses Natural Killer Cell Maturation in the Tumor Microenvironment. Cancer Research, 2018, 78, 1003-1016.	0.4	269
14	A comprehensive assessment of somatic mutation detection in cancer using whole-genome sequencing. Nature Communications, 2015, 6, 10001.	5.8	266
15	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	9.4	255
16	Effective Treatment With Oral Sulfonylureas in Patients With Diabetes Due to Sulfonylurea Receptor 1 (SUR1) Mutations. Diabetes Care, 2008, 31, 204-209.	4.3	239
17	Genomic catastrophes frequently arise in esophageal adenocarcinoma and drive tumorigenesis. Nature Communications, 2014, 5, 5224.	5.8	236
18	Partial lipodystrophy and insulin resistant diabetes in a patient with a homozygous nonsense mutation in CIDEC. EMBO Molecular Medicine, 2009, 1, 280-287.	3.3	235

#	Article	IF	Citations
19	Whole-genome landscape of mucosal melanoma reveals diverse drivers and therapeutic targets. Nature Communications, 2019, 10, 3163.	5.8	205
20	Point Mutations in Exon 1B of APC Reveal Gastric Adenocarcinoma and Proximal Polyposis of the Stomach as a Familial Adenomatous Polyposis Variant. American Journal of Human Genetics, 2016, 98, 830-842.	2.6	201
21	Permanent Neonatal Diabetes Caused by Dominant, Recessive, or Compound Heterozygous SUR1 Mutations with Opposite Functional Effects. American Journal of Human Genetics, 2007, 81, 375-382.	2.6	194
22	Genomeâ€wide DNA methylation patterns in pancreatic ductal adenocarcinoma reveal epigenetic deregulation of SLITâ€ROBO, ITGA2 and MET signaling. International Journal of Cancer, 2014, 135, 1110-1118.	2.3	192
23	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 3105-3110.	3.3	185
24	Hypermutation In Pancreatic Cancer. Gastroenterology, 2017, 152, 68-74.e2.	0.6	174
25	Wolcott-Rallison Syndrome Is the Most Common Genetic Cause of Permanent Neonatal Diabetes in Consanguineous Families. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4162-4170.	1.8	127
26	Analysis of Transcription Factors Key for Mouse Pancreatic Development Establishes NKX2-2 and MNX1 Mutations as Causes of Neonatal Diabetes in Man. Cell Metabolism, 2014, 19, 146-154.	7.2	123
27	Whole-genome sequencing of acral melanoma reveals genomic complexity and diversity. Nature Communications, 2020, 11, 5259.	5.8	102
28	Integrated genomic and transcriptomic analysis of human brain metastases identifies alterations of potential clinical significance. Journal of Pathology, 2015, 237, 363-378.	2.1	98
29	Clinical and molecular characterization of HER2 amplified-pancreatic cancer. Genome Medicine, 2013, 5, 78.	3.6	97
30	Targeting DNA Damage Response and Replication Stress in Pancreatic Cancer. Gastroenterology, 2021, 160, 362-377.e13.	0.6	90
31	Novel GLIS3 mutations demonstrate an extended multisystem phenotype. European Journal of Endocrinology, 2011, 164, 437-443.	1.9	86
32	Homologous Recombination DNA Repair Pathway Disruption and Retinoblastoma Protein Loss Are Associated with Exceptional Survival in High-Grade Serous Ovarian Cancer. Clinical Cancer Research, 2018, 24, 569-580.	3.2	79
33	HNF4A and GATA6 Loss Reveals Therapeutically Actionable Subtypes in Pancreatic Cancer. Cell Reports, 2020, 31, 107625.	2.9	78
34	Long Noncoding RNAs CUPID1 and CUPID2 Mediate Breast Cancer Risk at 11q13 by Modulating the Response to DNA Damage. American Journal of Human Genetics, 2017, 101, 255-266.	2.6	77
35	Somatic Point Mutation Calling in Low Cellularity Tumors. PLoS ONE, 2013, 8, e74380.	1.1	67
36	Whole-genome sequencing reveals clinically relevant insights into the aetiology of familial breast cancers. Annals of Oncology, 2019, 30, 1071-1079.	0.6	64

#	Article	IF	Citations
37	Recessive SLC19A2 mutations are a cause ofÂneonatal diabetes mellitus inÂthiamine-responsive megaloblastic anaemia. Pediatric Diabetes, 2012, 13, 314-321.	1.2	57
38	Germline and somatic variant identification using BGISEQ-500 and HiSeq X Ten whole genome sequencing. PLoS ONE, 2018, 13, e0190264.	1.1	57
39	EIF1AX and NRAS Mutations Co-occur and Cooperate in Low-Grade Serous Ovarian Carcinomas. Cancer Research, 2017, 77, 4268-4278.	0.4	56
40	Whole genome sequencing of melanomas in adolescent and young adults reveals distinct mutation landscapes and the potential role of germline variants in disease susceptibility. International Journal of Cancer, 2019, 144, 1049-1060.	2.3	54
41	Increased ATPase activity produced by mutations at arginine-1380 in nucleotide-binding domain 2 of ABCC8 causes neonatal diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 18988-18992.	3.3	51
42	Genome-Wide Homozygosity Analysis Reveals HADH Mutations as a Common Cause of Diazoxide-Responsive Hyperinsulinemic-Hypoglycemia in Consanguineous Pedigrees. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E498-E502.	1.8	51
43	Identification of the CIMP-like subtype and aberrant methylation of members of the chromosomal segregation and spindle assembly pathways in esophageal adenocarcinoma. Carcinogenesis, 2016, 37, 356-365.	1.3	46
44	Integrative Genome-Scale DNA Methylation Analysis of a Large and Unselected Cohort Reveals 5 Distinct Subtypes of Colorectal Adenocarcinomas. Cellular and Molecular Gastroenterology and Hepatology, 2019, 8, 269-290.	2.3	42
45	Unexpected UVR and non-UVR mutation burden in some acral and cutaneous melanomas. Laboratory Investigation, 2017, 97, 130-145.	1.7	40
46	Telomere sequence content can be used to determine ALT activity in tumours. Nucleic Acids Research, 2018, 46, 4903-4918.	6.5	40
47	Intratumoural Heterogeneity Underlies Distinct Therapy Responses and Treatment Resistance in Glioblastoma. Cancers, 2019, 11, 190.	1.7	39
48	Mutations in the ABCC8 (SUR1 subunit of the K_{ATP} channel) gene are associated with a variable clinical phenotype. Clinical Endocrinology, 2009, 71, 358-362.	1.2	35
49	Whole exome sequencing of an asbestos-induced wild-type murine model of malignant mesothelioma. BMC Cancer, 2017, 17, 396.	1.1	30
50	APC Mutation Marks an Aggressive Subtype of BRAF Mutant Colorectal Cancers. Cancers, 2020, 12, 1171.	1.7	28
51	Lost in translation: returning germline genetic results in genome-scale cancer research. Genome Medicine, 2017, 9, 41.	3.6	27
52	DNA methylation patterns identify subgroups of pancreatic neuroendocrine tumors with clinical association. Communications Biology, 2021, 4, 155.	2.0	26
53	Genomic and Molecular Analyses Identify Molecular Subtypes of Pancreatic Cancer Recurrence. Gastroenterology, 2022, 162, 320-324.e4.	0.6	26
54	Comprehensive genomic and tumour immune profiling reveals potential therapeutic targets in malignant pleural mesothelioma. Genome Medicine, 2022, 14, .	3.6	24

#	Article	IF	Citations
55	Malignant cells from pleural fluids in malignant mesothelioma patients reveal novel mutations. Lung Cancer, 2018, 119, 64-70.	0.9	23
56	In vitro expression of NGN3 identifies RAB3B as the predominant Ras-associated GTP-binding protein 3 family member in human islets. Journal of Endocrinology, 2010, 207, 151-161.	1.2	22
57	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. Genetics in Medicine, 2019, 21, 982-986.	1.1	22
58	Mutation load in melanoma is affected by <scp>MC</scp>1R genotype. Pigment Cell and Melanoma Research, 2017, 30, 255-258.	1.5	19
59	BRAF Mutations in Low-Grade Serous Ovarian Cancer and Response to BRAF Inhibition. JCO Precision Oncology, 2018, 2, 1-14.	1.5	19
60	Complex structural rearrangements are present in high-grade dysplastic Barrettâ€™s oesophagus samples. BMC Medical Genomics, 2019, 12, 31.	0.7	19
61	Patient-derived xenograft models capture genomic heterogeneity in endometrial cancer. Genome Medicine, 2022, 14, 3.	3.6	16
62	Alterations in signaling pathways that accompany spontaneous transition to malignancy in a mouse model of BRAF mutant microsatellite stable colorectal cancer. Neoplasia, 2020, 22, 120-128.	2.3	14
63	Copy number profiles of paired primary and metastatic colorectal cancers. Oncotarget, 2018, 9, 3394-3405.	0.8	14
64	Evaluation of the contribution of germline variants in BRCA1 and BRCA2 to uveal and cutaneous melanoma. Melanoma Research, 2019, 29, 483-490.	0.6	13
65	Injection site vaccinology of a recombinant vaccinia-based vector reveals diverse innate immune signatures. PLoS Pathogens, 2021, 17, e1009215.	2.1	13
66	Neonatal diabetes mellitus due to pancreas agenesis: a new case report and review of the literature. Pediatric Diabetes, 2009, 10, 487-491.	1.2	10
67	FGFR2c Mesenchymal Isoform Expression Is Associated with Poor Prognosis and Further Refines Risk Stratification within Endometrial Cancer Molecular Subtypes. Clinical Cancer Research, 2020, 26, 4569-4580.	3.2	10
68	Evaluation of Crizotinib Treatment in a Patient With Unresectable GOPC-ROS1 Fusion Agminated Spitz Nevi. JAMA Dermatology, 2021, 157, 836-841.	2.0	9
69	Sequencing of candidate genes selected by beta cell experts in monogenic diabetes of unknown aetiology. JOP: Journal of the Pancreas, 2010, 11, 14-7.	1.5	8
70	The Impact of Next Generation Sequencing in Cancer Research. Cancers, 2020, 12, 2928.	1.7	7
71	Fingerprinting fission yeast: polymorphic markers for molecular genetic analysis of Schizosaccharomyces pombe strains. Microbiology (United Kingdom), 2007, 153, 887-897.	0.7	6
72	Identification of a Locus Near ULK1 Associated With Progression-Free Survival in Ovarian Cancer. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 1669-1680.	1.1	5

#	Article	IF	Citations
73	qmotif: determination of telomere content from whole-genome sequence data. Bioinformatics Advances, 2022, 2, .	0.9	5
74	ROR1 and ROR2 expression in pancreatic cancer. BMC Cancer, 2021, 21, 1199.	1.1	4
75	Neoantigens â€“ the next frontier in precision immunotherapy for B-cell lymphoproliferative disorders. Blood Reviews, 2022, 56, 100969.	2.8	2
76	Neoantigens Are Typically Associated with Intact HLA Class I Presentation in Early-Stage Follicular Lymphoma. Blood, 2020, 136, 37-38.	0.6	1
77	A workflow to increase verification rate of chromosomal structural rearrangements using high-throughput next-generation sequencing. BioTechniques, 2014, 57, 31-38.	0.8	0