Janine Altmller

List of Publications by Citations

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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.

95 papers	4,000 citations	28 h-index	62 g-index
112 ext. papers	5,500 ext. citations	9.4 avg, IF	4.49 L-index

#	Paper	IF	Citations
95	Comprehensive genomic profiles of small cell lung cancer. *Nature*, **2015**, 524, 47-53	50.4	1061
94	Multiancestry association study identifies new asthma risk loci that colocalize with immune-cell enhancer marks. *Nature Genetics*, **2018**, 50, 42-53	36.3	246
93	Mutational dynamics between primary and relapse neuroblastomas. *Nature Genetics*, **2015**, 47, 872-7	36.3	191
92	Integrative genomic profiling of large-cell neuroendocrine carcinomas reveals distinct subtypes of high-grade neuroendocrine lung tumors. *Nature Communications*, **2018**, 9, 1048	17.4	152
91	Attenuated BMP1 function compromises osteogenesis, leading to bone fragility in humans and zebrafish. *American Journal of Human Genetics*, **2012**, 90, 661-74	11	152
90	A truncating mutation of CEP135 causes primary microcephaly and disturbed centrosomal function. *American Journal of Human Genetics*, **2012**, 90, 871-8	11	140
89	Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. *American Journal of Human Genetics*, **2016**, 99, 337-51	11	139
88	Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. *Nature Communications*, **2018**, 9, 727	17.4	116
87	A mechanistic classification of clinical phenotypes in neuroblastoma. *Science*, **2018**, 362, 1165-1170	33.3	115
86	Linear mitochondrial DNA is rapidly degraded by components of the replication machinery. *Nature Communications*, **2018**, 9, 1727	17.4	87
85	HMGB2 Loss upon Senescence Entry Disrupts Genomic Organization and Induces CTCF Clustering across Cell Types. *Molecular Cell*, **2018**, 70, 730-744.e6	17.6	83
84	Loss-of-Function GAS8 Mutations Cause Primary Ciliary Dyskinesia and Disrupt the Nexin-Dynein Regulatory Complex. *American Journal of Human Genetics*, **2015**, 97, 546-54	11	80
83	Prevalence of deleterious germline variants in risk genes including BRCA1/2 in consecutive ovarian cancer patients (AGO-TR-1). *PLoS ONE*, **2017**, 12, e0186043	3.7	74
82	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. *American Journal of Human Genetics*, **2011**, 89, 668-74	11	74
81	Mutations in Three Genes Encoding Proteins Involved in Hair Shaft Formation Cause Uncombable Hair Syndrome. *American Journal of Human Genetics*, **2016**, 99, 1292-1304	11	72
80	BRF1 mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. *Genome Research*, **2015**, 25, 155-66	9.7	68
79	De Novo Mutations in FOXJ1 Result in a Motile Ciliopathy with Hydrocephalus and Randomization of Left/Right Body Asymmetry. *American Journal of Human Genetics*, **2019**, 105, 1030-1039	11	68

78	Nonsense mutations in SMPX, encoding a protein responsive to physical force, result in X-chromosomal hearing loss. *American Journal of Human Genetics*, **2011**, 88, 621-7	11	60
77	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. *Nature Genetics*, **2016**, 48, 36-43	36.3	53
76	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. *Nature Genetics*, **2017**, 49, 537-549	36.3	52
75	De novo FUS mutations are the most frequent genetic cause in early-onset German ALS patients. *Neurobiology of Aging*, **2015**, 36, 3117.e1-3117.e6	5.6	48
74	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. *American Journal of Human Genetics*, **2017**, 101, 833-843	11	37
73	Floral Induction in Arabidopsis by FLOWERING LOCUS T Requires Direct Repression of BLADE-ON-PETIOLE Genes by the Homeodomain Protein PENNYWISE. *Plant Physiology*, **2015**, 169, 2187-99	6.6	32
72	How to make a tumour: cell type specific dissection of Ustilago maydis-induced tumour development in maize leaves. *New Phytologist*, **2018**, 217, 1681-1695	9.8	31
71	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. *Journal of Medical Genetics*, **2018**, 55, 837-846	5.8	31
70	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. *American Journal of Human Genetics*, **2014**, 95, 763-70	11	30
69	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. *American Journal of Human Genetics*, **2018**, 103, 777-785	11	29
68	Mutations in CKAP2L, the human homolog of the mouse Radmis gene, cause Filippi syndrome. *American Journal of Human Genetics*, **2014**, 95, 622-32	11	28
67	Enrichment of target sequences for next-generation sequencing applications in research and diagnostics. *Biological Chemistry*, **2014**, 395, 231-7	4.5	28
66	Exon Junction Complexes Suppress Spurious Splice Sites to Safeguard Transcriptome Integrity. *Molecular Cell*, **2018**, 72, 482-495.e7	17.6	28
65	Long-lived macrophage reprogramming drives spike protein-mediated inflammasome activation in COVID-19. *EMBO Molecular Medicine*, **2021**, 13, e14150	12	27
64	Assessing the enrichment performance in targeted resequencing experiments. *Human Mutation*, **2012**, 33, 635-41	4.7	25
63	Antagonistic modulation of NPY/AgRP and POMC neurons in the arcuate nucleus by noradrenalin. *ELife*, **2017**, 6,	8.9	25
62	Tissue-infiltrating macrophages mediate an exosome-based metabolic reprogramming upon DNA damage. *Nature Communications*, **2020**, 11, 42	17.4	25
61	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. *Human Molecular Genetics*, **2015**, 24, 3708-17	5.6	23

60	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. *Human Molecular Genetics*, **2015**, 24, 2594-603	5.6	23
59	Pathogenicity of POFUT1 in Dowling-Degos disease: additional mutations and clinical overlap with reticulate acropigmentation of kitamura. *Journal of Investigative Dermatology*, **2015**, 135, 615-618	4.3	22
58	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca-Activated K Channel SK3 Cause Zimmermann-Laband Syndrome. *American Journal of Human Genetics*, **2019**, 104, 1139-1157	11	20
57	The genomic and clinical landscape of fetal akinesia. *Genetics in Medicine*, **2020**, 22, 511-523	8.1	19
56	IG- neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. *Blood*, **2018**, 132, 2280-2285	2.2	19
55	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. *Molecular and Cellular Probes*, **2015**, 29, 330-4	3.3	18
54	Homozygous missense mutation of NDUFV1 as the cause of infantile bilateral striatal necrosis. *Neurogenetics*, **2013**, 14, 85-7	3	18
53	Human brain organoids assemble functionally integrated bilateral optic vesicles. *Cell Stem Cell*, **2021**, 28, 1740-1757.e8	18	18
52	Copy number increases of transposable elements and protein-coding genes in an invasive fish of hybrid origin. *Molecular Ecology*, **2017**, 26, 4712-4724	5.7	17
51	Mechanism suppressing H3K9 trimethylation in pluripotent stem cells and its demise by polyQ-expanded huntingtin mutations. *Human Molecular Genetics*, **2018**, 27, 4117-4134	5.6	17
50	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. *Genetics in Medicine*, **2019**, 21, 1832-1841	8.1	16
49	Confirmation of CAGSSS syndrome as a distinct entity in a Danish patient with a novel homozygous mutation in IARS2. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1102-1108	2.5	15
48	cfNOMe -A single assay for comprehensive epigenetic analyses of cell-free DNA. *Genome Medicine*, **2020**, 12, 54	14.4	15
47	A large deletion in RPGR causes XLPRA in Weimaraner dogs. *Canine Genetics and Epidemiology*, **2016**, 3, 7	2.8	15
46	Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. *Journal of Human Genetics*, **2019**, 64, 803-813	4.3	14
45	Novel IFT122 mutations in three Argentinian patients with cranioectodermal dysplasia: Expanding the mutational spectrum. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1295-301	2.5	14
44	CASC3 promotes transcriptome-wide activation of nonsense-mediated decay by the exon junction complex. *Nucleic Acids Research*, **2020**, 48, 8626-8644	20.1	13
43	A systematic comparison of two new releases of exome sequencing products: the aim of use determines the choice of product. *Biological Chemistry*, **2016**, 397, 791-801	4.5	12

42	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. *American Journal of Human Genetics*, **2019**, 104, 994-1006	11	11
41	Genome-wide patterns of transposon proliferation in an evolutionary young hybrid fish. *Molecular Ecology*, **2019**, 28, 1491-1505	5.7	11
40	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. *Scientific Reports*, **2018**, 8, 7907	4.9	11
39	A syndrome of microcephaly, short stature, polysyndactyly, and dental anomalies caused by a homozygous KATNB1 mutation. *American Journal of Medical Genetics, Part A*, **2016**, 170, 728-33	2.5	10
38	Novel PNKP mutations causing defective DNA strand break repair and PARP1 hyperactivity in MCSZ. *Neurology: Genetics*, **2019**, 5, e320	3.8	9
37	Homozygosity for the c.428delG variant in in a healthy individual: implications for molecular testing in patients with Joubert syndrome. *Journal of Medical Genetics*, **2019**, 56, 261-264	5.8	8
36	SMG5-SMG7 authorize nonsense-mediated mRNA decay by enabling SMG6 endonucleolytic activity. *Nature Communications*, **2021**, 12, 3965	17.4	8
35	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. *Genetics in Medicine*, **2021**, 23, 341-351	8.1	8
34	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. *Nature Genetics*, **2021**, 53, 1673-1685	36.3	7
33	Reconstruction of rearranged T-cell receptor loci by whole genome and transcriptome sequencing gives insights into the initial steps of T-cell prolymphocytic leukemia. *Genes Chromosomes and Cancer*, **2020**, 59, 261-267	5	7
32	Ultra-rapid emergency genomic diagnosis of Donahue syndrome in a preterm infant within 17 hours. *American Journal of Medical Genetics, Part A*, **2021**, 185, 90-96	2.5	7
31	A new CUL4B variant associated with a mild phenotype and an exceptional pattern of leukoencephalopathy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2803-2807	2.5	5
30	Ectodysplasin signalling genes and phenotypic evolution in sculpins (Cottus). *Proceedings of the Royal Society B: Biological Sciences*, **2015**, 282,	4.4	5
29	R-loops trigger the release of cytoplasmic ssDNAs leading to chronic inflammation upon DNA damage. *Science Advances*, **2021**, 7, eabj5769	14.3	5
28	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. *Human Mutation*, **2020**, 41, 591-599	4.7	5
27	The splicing factor XAB2 interacts with ERCC1-XPF and XPG for R-loop processing. *Nature Communications*, **2021**, 12, 3153	17.4	5
26	A novel homozygous PAM16 mutation in a patient with a milder phenotype and longer survival. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2436-9	2.5	5
25	Exome sequencing in 38 patients with intracranial aneurysms and subarachnoid hemorrhage. *Journal of Neurology*, **2020**, 267, 2533-2545	5.5	4

24	RNA polymerase II is required for spatial chromatin reorganization following exit from mitosis. *Science Advances*, **2021**, 7, eabg8205	14.3	4
23	Single cell transcriptome sequencing on the Nanopore platform with ScNapBar. *Rna*, **2021**,	5.8	3
22	Transposable elements and introgression introduce genetic variation in the invasive ant Cardiocondyla obscurior. *Molecular Ecology*, **2021**, 30, 6211-6228	5.7	3
21	Micro-RNA networks in T-cell prolymphocytic leukemia reflect T-cell activation and shape DNA damage response and survival pathways. *Haematologica*, **2020**,	6.6	2
20	Nonsense-mediated mRNA decay relies on Ewo-factor authentication by SMG5-SMG7		2
19	Single cell transcriptome sequencing on the Nanopore platform with ScNapBar		2
18	A Homozygous Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. *Genes*, **2021**, 12,	4.2	2
17	Metatarsal bony syndactyly in 2 fetuses with Smith-Lemli-Opitz syndrome: An under-recognized part of the clinical spectrum. *Clinical Genetics*, **2017**, 92, 342-343	4	1
16	Variant profiling of colorectal adenomas from three patients of two families with MSH3-related adenomatous polyposis. *PLoS ONE*, **2021**, 16, e0259185	3.7	1
15	A novel remitting leukodystrophy associated with a variant in. *Brain Communications*, **2021**, 3, fcab036	4.5	1
14	Clinical and genetic characterization of PYROXD1-related myopathy patients from Turkey. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1678-1690	2.5	1
13	Modifier Genes in Microcephaly: A Report on , , and Variants Exacerbating Disease Caused by Biallelic Mutations of and. *Genes*, **2021**, 12,	4.2	1
12	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. *Genetics in Medicine*, **2021**, 23, 2138-2149	8.1	1
11	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. *American Journal of Human Genetics*, **2021**, 108, 357-367	11	1
10	Senior-Løken syndrome with IQCB1 mutation in Taiwan. *Kaohsiung Journal of Medical Sciences*, **2018**, 34, 588-589	2.4	1
9	Mitochondrial respiratory chain function promotes extracellular matrix integrity in cartilage. *Journal of Biological Chemistry*, **2021**, 297, 101224	5.4	1
8	Biallelic variants in YRDC cause a developmental disorder with progeroid features. *Human Genetics*, **2021**, 140, 1679-1693	6.3	1
7	Single Cell- and Spatial Omics revolutionizes Physiology. *Acta Physiologica*,	5.6	1

6	A novel missense variant of SCN4A co-segregates with congenital essential tremor in a consanguineous Kurdish family.. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
5	MAGED2 controls vasopressin-induced aquaporin-2 expression in collecting duct cells. *Journal of Proteomics*, **2021**, 252, 104424	3.9	0
4	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. *Neurogenetics*, **2021**, 22, 263-269	3	0
3	Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. *Frontiers in Genetics*, **2021**, 12, 689940	4.5	0
2	variants of cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway.. *Human Genetics and Genomics Advances*, **2022**, 3, 100111	0.8	0
1	Sequenzierung in der nächsten Generation Forschung und Diagnostik. *BioSpektrum*, **2012**, 18, 499-503	0.1	