

Janine Altmüller

List of Publications by Year in Descending Order

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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	variants of cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway.. <i>Human Genetics and Genomics Advances</i> , 2022 , 3, 100111	0.8	0
94	A novel missense variant of SCN4A co-segregates with congenital essential tremor in a consanguineous Kurdish family.. <i>American Journal of Medical Genetics, Part A</i> , 2021 ,	2.5	0
93	MAGED2 controls vasopressin-induced aquaporin-2 expression in collecting duct cells. <i>Journal of Proteomics</i> , 2021 , 252, 104424	3.9	0
92	R-loops trigger the release of cytoplasmic ssDNAs leading to chronic inflammation upon DNA damage. <i>Science Advances</i> , 2021 , 7, eabj5769	14.3	5
91	Variant profiling of colorectal adenomas from three patients of two families with MSH3-related adenomatous polyposis. <i>PLoS ONE</i> , 2021 , 16, e0259185	3.7	1
90	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. <i>Nature Genetics</i> , 2021 , 53, 1673-1685	36.3	7
89	RNA polymerase II is required for spatial chromatin reorganization following exit from mitosis. <i>Science Advances</i> , 2021 , 7, eabg8205	14.3	4
88	A novel remitting leukodystrophy associated with a variant in. <i>Brain Communications</i> , 2021 , 3, fcab036	4.5	1
87	Clinical and genetic characterization of PYROXD1-related myopathy patients from Turkey. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1678-1690	2.5	1
86	Single cell transcriptome sequencing on the Nanopore platform with ScNapBar. <i>Rna</i> , 2021 ,	5.8	3
85	The splicing factor XAB2 interacts with ERCC1-XPF and XPG for R-loop processing. <i>Nature Communications</i> , 2021 , 12, 3153	17.4	5
84	Modifier Genes in Microcephaly: A Report on , , and Variants Exacerbating Disease Caused by Biallelic Mutations of and. <i>Genes</i> , 2021 , 12,	4.2	1
83	Long-lived macrophage reprogramming drives spike protein-mediated inflammasome activation in COVID-19. <i>EMBO Molecular Medicine</i> , 2021 , 13, e14150	12	27
82	SMG5-SMG7 authorize nonsense-mediated mRNA decay by enabling SMG6 endonucleolytic activity. <i>Nature Communications</i> , 2021 , 12, 3965	17.4	8
81	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. <i>Neurogenetics</i> , 2021 , 22, 263-269	3	0
80	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. <i>Genetics in Medicine</i> , 2021 , 23, 2138-2149	8.1	1
79	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. <i>Genetics in Medicine</i> , 2021 , 23, 341-351	8.1	8

78	Ultra-rapid emergency genomic diagnosis of Donahue syndrome in a preterm infant within 17 hours. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 90-96	2.5	7
77	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021 , 108, 357-367	11	1
76	Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. <i>Frontiers in Genetics</i> , 2021 , 12, 689940	4.5	0
75	Human brain organoids assemble functionally integrated bilateral optic vesicles. <i>Cell Stem Cell</i> , 2021 , 28, 1740-1757.e8	18	18
74	Transposable elements and introgression introduce genetic variation in the invasive ant <i>Cardiocondyla obscurior</i> . <i>Molecular Ecology</i> , 2021 , 30, 6211-6228	5.7	3
73	Mitochondrial respiratory chain function promotes extracellular matrix integrity in cartilage. <i>Journal of Biological Chemistry</i> , 2021 , 297, 101224	5.4	1
72	A Homozygous Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. <i>Genes</i> , 2021 , 12,	4.2	2
71	Biallelic variants in YRDC cause a developmental disorder with progeroid features. <i>Human Genetics</i> , 2021 , 140, 1679-1693	6.3	1
70	Exome sequencing in 38 patients with intracranial aneurysms and subarachnoid hemorrhage. <i>Journal of Neurology</i> , 2020 , 267, 2533-2545	5.5	4
69	cfNOMe -TA single assay for comprehensive epigenetic analyses of cell-free DNA. <i>Genome Medicine</i> , 2020 , 12, 54	14.4	15
68	CASC3 promotes transcriptome-wide activation of nonsense-mediated decay by the exon junction complex. <i>Nucleic Acids Research</i> , 2020 , 48, 8626-8644	20.1	13
67	Micro-RNA networks in T-cell prolymphocytic leukemia reflect T-cell activation and shape DNA damage response and survival pathways. <i>Haematologica</i> , 2020 ,	6.6	2
66	Reconstruction of rearranged T-cell receptor loci by whole genome and transcriptome sequencing gives insights into the initial steps of T-cell prolymphocytic leukemia. <i>Genes Chromosomes and Cancer</i> , 2020 , 59, 261-267	5	7
65	The genomic and clinical landscape of fetal akinesia. <i>Genetics in Medicine</i> , 2020 , 22, 511-523	8.1	19
64	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. <i>Human Mutation</i> , 2020 , 41, 591-599	4.7	5
63	Tissue-infiltrating macrophages mediate an exosome-based metabolic reprogramming upon DNA damage. <i>Nature Communications</i> , 2020 , 11, 42	17.4	25
62	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. <i>Genetics in Medicine</i> , 2019 , 21, 1832-1841	8.1	16
61	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca-Activated K Channel SK3 Cause Zimmermann-Laband Syndrome. <i>American Journal of Human Genetics</i> , 2019 , 104, 1139-1157	11	20

60	Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. <i>Journal of Human Genetics</i> , 2019 , 64, 803-813	4.3	14
59	Novel PNKP mutations causing defective DNA strand break repair and PARP1 hyperactivity in MCSZ. <i>Neurology: Genetics</i> , 2019 , 5, e320	3.8	9
58	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. <i>American Journal of Human Genetics</i> , 2019 , 104, 994-1006	11	11
57	Homozygosity for the c.428delG variant in a healthy individual: implications for molecular testing in patients with Joubert syndrome. <i>Journal of Medical Genetics</i> , 2019 , 56, 261-264	5.8	8
56	De Novo Mutations in FOXJ1 Result in a Motile Ciliopathy with Hydrocephalus and Randomization of Left/Right Body Asymmetry. <i>American Journal of Human Genetics</i> , 2019 , 105, 1030-1039	11	68
55	Genome-wide patterns of transposon proliferation in an evolutionary young hybrid fish. <i>Molecular Ecology</i> , 2019 , 28, 1491-1505	5.7	11
54	Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. <i>Nature Communications</i> , 2018 , 9, 727	17.4	116
53	Multiancestry association study identifies new asthma risk loci that colocalize with immune-cell enhancer marks. <i>Nature Genetics</i> , 2018 , 50, 42-53	36.3	246
52	How to make a tumour: cell type specific dissection of <i>Ustilago maydis</i> -induced tumour development in maize leaves. <i>New Phytologist</i> , 2018 , 217, 1681-1695	9.8	31
51	Linear mitochondrial DNA is rapidly degraded by components of the replication machinery. <i>Nature Communications</i> , 2018 , 9, 1727	17.4	87
50	HMGB2 Loss upon Senescence Entry Disrupts Genomic Organization and Induces CTCF Clustering across Cell Types. <i>Molecular Cell</i> , 2018 , 70, 730-744.e6	17.6	83
49	Integrative genomic profiling of large-cell neuroendocrine carcinomas reveals distinct subtypes of high-grade neuroendocrine lung tumors. <i>Nature Communications</i> , 2018 , 9, 1048	17.4	152
48	Mechanism suppressing H3K9 trimethylation in pluripotent stem cells and its demise by polyQ-expanded huntingtin mutations. <i>Human Molecular Genetics</i> , 2018 , 27, 4117-4134	5.6	17
47	A mechanistic classification of clinical phenotypes in neuroblastoma. <i>Science</i> , 2018 , 362, 1165-1170	33.3	115
46	IG- neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. <i>Blood</i> , 2018 , 132, 2280-2285	2.2	19
45	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. <i>American Journal of Human Genetics</i> , 2018 , 103, 777-785	11	29
44	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. <i>Journal of Medical Genetics</i> , 2018 , 55, 837-846	5.8	31
43	Exon Junction Complexes Suppress Spurious Splice Sites to Safeguard Transcriptome Integrity. <i>Molecular Cell</i> , 2018 , 72, 482-495.e7	17.6	28

42	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. <i>Scientific Reports</i> , 2018 , 8, 7907	4.9	11
41	Senior-Lièen syndrome with IQCB1 mutation in Taiwan. <i>Kaohsiung Journal of Medical Sciences</i> , 2018 , 34, 588-589	2.4	1
40	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017 , 49, 537-549	36.3	52
39	Confirmation of CAGSSS syndrome as a distinct entity in a Danish patient with a novel homozygous mutation in IARS2. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1102-1108	2.5	15
38	Metatarsal bony syndactyly in 2 fetuses with Smith-Lemli-Opitz syndrome: An under-recognized part of the clinical spectrum. <i>Clinical Genetics</i> , 2017 , 92, 342-343	4	1
37	Copy number increases of transposable elements and protein-coding genes in an invasive fish of hybrid origin. <i>Molecular Ecology</i> , 2017 , 26, 4712-4724	5.7	17
36	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017 , 101, 833-843	11	37
35	Prevalence of deleterious germline variants in risk genes including BRCA1/2 in consecutive ovarian cancer patients (AGO-TR-1). <i>PLoS ONE</i> , 2017 , 12, e0186043	3.7	74
34	A new CUL4B variant associated with a mild phenotype and an exceptional pattern of leukoencephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 2803-2807	2.5	5
33	Antagonistic modulation of NPY/AgRP and POMC neurons in the arcuate nucleus by noradrenalin. <i>ELife</i> , 2017 , 6,	8.9	25
32	A large deletion in RPGR causes XLPR in Weimaraner dogs. <i>Canine Genetics and Epidemiology</i> , 2016 , 3, 7	2.8	15
31	Mutations in Three Genes Encoding Proteins Involved in Hair Shaft Formation Cause Uncombable Hair Syndrome. <i>American Journal of Human Genetics</i> , 2016 , 99, 1292-1304	11	72
30	Novel IFT122 mutations in three Argentinian patients with cranioectodermal dysplasia: Expanding the mutational spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 1295-301	2.5	14
29	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. <i>Nature Genetics</i> , 2016 , 48, 36-43	36.3	53
28	A syndrome of microcephaly, short stature, polysyndactyly, and dental anomalies caused by a homozygous KATNB1 mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 728-33	2.5	10
27	A systematic comparison of two new releases of exome sequencing products: the aim of use determines the choice of product. <i>Biological Chemistry</i> , 2016 , 397, 791-801	4.5	12
26	A novel homozygous PAM16 mutation in a patient with a milder phenotype and longer survival. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2436-9	2.5	5
25	Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. <i>American Journal of Human Genetics</i> , 2016 , 99, 337-51	11	139

24	BRF1 mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. <i>Genome Research</i> , 2015 , 25, 155-66	9.7	68
23	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. <i>Molecular and Cellular Probes</i> , 2015 , 29, 330-4	3.3	18
22	Comprehensive genomic profiles of small cell lung cancer. <i>Nature</i> , 2015 , 524, 47-53	50.4	1061
21	Mutational dynamics between primary and relapse neuroblastomas. <i>Nature Genetics</i> , 2015 , 47, 872-7	36.3	191
20	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. <i>Human Molecular Genetics</i> , 2015 , 24, 3708-17	5.6	23
19	Floral Induction in Arabidopsis by FLOWERING LOCUS T Requires Direct Repression of BLADE-ON-PETIOLE Genes by the Homeodomain Protein PENNYWISE. <i>Plant Physiology</i> , 2015 , 169, 2187-93	6.6	32
18	De novo FUS mutations are the most frequent genetic cause in early-onset German ALS patients. <i>Neurobiology of Aging</i> , 2015 , 36, 3117.e1-3117.e6	5.6	48
17	Loss-of-Function GAS8 Mutations Cause Primary Ciliary Dyskinesia and Disrupt the Nexin-Dynein Regulatory Complex. <i>American Journal of Human Genetics</i> , 2015 , 97, 546-54	11	80
16	Ectodysplasin signalling genes and phenotypic evolution in sculpins (Cottus). <i>Proceedings of the Royal Society B: Biological Sciences</i> , 2015 , 282,	4.4	5
15	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. <i>Human Molecular Genetics</i> , 2015 , 24, 2594-603	5.6	23
14	Pathogenicity of POFUT1 in Dowling-Degos disease: additional mutations and clinical overlap with reticulate acropigmentation of kitamura. <i>Journal of Investigative Dermatology</i> , 2015 , 135, 615-618	4.3	22
13	Mutations in CKAP2L, the human homolog of the mouse Radmis gene, cause Filippi syndrome. <i>American Journal of Human Genetics</i> , 2014 , 95, 622-32	11	28
12	Enrichment of target sequences for next-generation sequencing applications in research and diagnostics. <i>Biological Chemistry</i> , 2014 , 395, 231-7	4.5	28
11	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. <i>American Journal of Human Genetics</i> , 2014 , 95, 763-70	11	30
10	Homozygous missense mutation of NDUFV1 as the cause of infantile bilateral striatal necrosis. <i>Neurogenetics</i> , 2013 , 14, 85-7	3	18
9	Assessing the enrichment performance in targeted resequencing experiments. <i>Human Mutation</i> , 2012 , 33, 635-41	4.7	25
8	Sequenzierung in der nächsten Generation [Forschung und Diagnostik. <i>BioSpektrum</i> , 2012 , 18, 499-503	0.1	
7	Attenuated BMP1 function compromises osteogenesis, leading to bone fragility in humans and zebrafish. <i>American Journal of Human Genetics</i> , 2012 , 90, 661-74	11	152

6	A truncating mutation of CEP135 causes primary microcephaly and disturbed centrosomal function. <i>American Journal of Human Genetics</i> , 2012 , 90, 871-8	11	140
5	Nonsense mutations in SMPX, encoding a protein responsive to physical force, result in X-chromosomal hearing loss. <i>American Journal of Human Genetics</i> , 2011 , 88, 621-7	11	60
4	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. <i>American Journal of Human Genetics</i> , 2011 , 89, 668-74	11	74
3	Nonsense-mediated mRNA decay relies on Ewo-factor authentication by SMG5-SMG7		2
2	Single cell transcriptome sequencing on the Nanopore platform with ScNapBar		2
1	Single Cell- and Spatial Omics revolutionizes Physiology. <i>Acta Physiologica</i> ,	5.6	1