Janine AltmÃ¼ller

List of Publications by Year
in descending order

101 papers	6,728 citations	109137 35 h-index	74018 75 g-index
112 all docs	112 docs citations	112 times ranked	14154 citing authors

#	Article	IF	Citations
1	Loss-of-function variants in DNM1 cause a specific form of developmental and epileptic encephalopathy only in biallelic state. Journal of Medical Genetics, 2022, 59, 549-553.	1.5	9
2	Micro-RNA networks in T-cell prolymphocytic leukemia reflect T-cell activation and shape DNA damage response and survival pathways. Haematologica, 2022, 107, 187-200.	1.7	10
3	MAGED2 controls vasopressin-induced aquaporin-2 expression in collecting duct cells. Journal of Proteomics, 2022, 252, 104424.	1.2	1
4	Claudin-10a Deficiency Shifts Proximal Tubular Cl- Permeability to Cation Selectivity via Claudin-2 Redistribution. Journal of the American Society of Nephrology: JASN, 2022, 33, 699-717.	3.0	20
5	Biallelic PAN2 variants in individuals with a syndromic neurodevelopmental disorder and multiple congenital anomalies. European Journal of Human Genetics, 2022, 30, 611-618.	1.4	4
6	Unraveling Structural Rearrangements of the CFH Gene Cluster in Atypical Hemolytic Uremic Syndrome Patients Using Molecular Combing and Long-Fragment Targeted Sequencing. Journal of Molecular Diagnostics, 2022, 24, 619-631.	1.2	5
7	De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 3, 100111.	1.0	7
8	Single cellâ€•and spatial â€˜Omics revolutionize physiology. Acta Physiologica, 2022, 235, .	1.8	8
9	Deregulation and epigenetic modification of BCL2-family genes cause resistance to venetoclax in hematologic malignancies. Blood, 2022, 140, 2113-2126.	0.6	24
10	WARS1 and SARS1 : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471.	1.1	5
11	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	1.1	16
12	Ultraâ€•rapid emergency genomic diagnosis of Donahue syndrome in a preterm infant within 17â€‰hours. American Journal of Medical Genetics, Part A, 2021, 185, 90-96.	0.7	14
13	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.	2.6	14
14	A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036.	1.5	2
15	Clinical and genetic characterization of <scp>PYROXD1</scp>â€•related myopathy patients from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 1678-1690.	0.7	5
16	Single-cell transcriptome sequencing on the Nanopore platform with ScNapBar. Rna, 2021, 27, 763-770.	1.6	12
17	The splicing factor XAB2 interacts with ERCC1-XPF and XPG for R-loop processing. Nature Communications, 2021, 12, 3153.	5.8	27
18	Modifier Genes in Microcephaly: A Report on WDR62, CEP63, RAD50 and PCNT Variants Exacerbating Disease Caused by Biallelic Mutations of ASPM and CENPJ. Genes, 2021, 12, 731.	1.0	8

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19	Longâ€lived macrophage reprogramming drives spike proteinâ€mediated inflammasome activation in COVIDâ€19. EMBO Molecular Medicine, 2021, 13, e14150.	3.3	98
20	SMG5-SMG7 authorize nonsense-mediated mRNA decay by enabling SMG6 endonucleolytic activity. Nature Communications, 2021, 12, 3965.	5.8	54
21	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269.	0.7	8
22	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	1.1	11
23	Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. Frontiers in Genetics, 2021, 12, 689940.	1.1	5
24	Human brain organoids assemble functionally integrated bilateral optic vesicles. Cell Stem Cell, 2021, 28, 1740-1757.e8.	5.2	77
25	Transposable elements and introgression introduce genetic variation in the invasive ant Cardiocondyla obscurior. Molecular Ecology, 2021, 30, 6211-6228.	2.0	20
26	Mitochondrial respiratory chain function promotes extracellular matrix integrity in cartilage. Journal of Biological Chemistry, 2021, 297, 101224.	1.6	16
27	A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 2021, 12, 1494.	1.0	3
28	Biallelic variants in YRDC cause a developmental disorder with progeroid features. Human Genetics, 2021, 140, 1679-1693.	1.8	3
29	RNA polymerase II is required for spatial chromatin reorganization following exit from mitosis. Science Advances, 2021, 7, eabg8205.	4.7	70
30	Familial cleft tongue caused by a unique translation initiation codon variant in TP63. European Journal of Human Genetics, 2021, , .	1.4	7
31	R-loops trigger the release of cytoplasmic ssDNAs leading to chronic inflammation upon DNA damage. Science Advances, 2021, 7, eabj5769.	4.7	30
32	Variant profiling of colorectal adenomas from three patients of two families with MSH3-related adenomatous polyposis. PLoS ONE, 2021, 16, e0259185.	1.1	5
33	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. Nature Genetics, 2021, 53, 1673-1685.	9.4	61
34	A novel missense variant of SCN4A coâ€segregates with congenital essential tremor in a consanguineous Kurdish family. American Journal of Medical Genetics, Part A, 2021, , .	0.7	1
35	Reconstruction of rearranged Tâ€cell receptor loci by whole genome and transcriptome sequencing gives insights into the initial steps of Tâ€cell prolymphocytic leukemia. Genes Chromosomes and Cancer, 2020, 59, 261-267.	1.5	16
36	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	1.1	35

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37	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	1.1	6
38	Tissue-infiltrating macrophages mediate an exosome-based metabolic reprogramming upon DNA damage. Nature Communications, 2020, 11, 42.	5.8	44
39	Exome sequencing in 38 patients with intracranial aneurysms and subarachnoid hemorrhage. Journal of Neurology, 2020, 267, 2533-2545.	1.8	14
40	cfNOMe â€"ÂA single assay for comprehensive epigenetic analyses of cell-free DNA. Genome Medicine, 2020, 12, 54.	3.6	39
41	CASC3 promotes transcriptome-wide activation of nonsense-mediated decay by the exon junction complex. Nucleic Acids Research, 2020, 48, 8626-8644.	6.5	35
42	Homozygosity for the c.428delG variant in KIAA0586 in a healthy individual: implications for molecular testing in patients with Joubert syndrome. Journal of Medical Genetics, 2019, 56, 261-264.	1.5	15
43	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.	2.6	129
44	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.	1.1	26
45	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca2+-Activated K+ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	2.6	45
46	Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. Journal of Human Genetics, 2019, 64, 803-813.	1.1	25
47	Novel PNKP mutations causing defective DNA strand break repair and PARP1 hyperactivity in MCSZ. Neurology: Genetics, 2019, 5, e320.	0.9	15
48	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	2.6	47
49	Genomeâ€wide patterns of transposon proliferation in an evolutionary young hybrid fish. Molecular Ecology, 2019, 28, 1491-1505.	2.0	18
50	Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. Nature Communications, 2018, 9, 727.	5.8	160
51	Multiancestry association study identifies new asthma risk loci that colocalize with immune-cell enhancer marks. Nature Genetics, 2018, 50, 42-53.	9.4	426
52	How to make a tumour: cell type specific dissection of Ustilago maydisâ€induced tumour development in maize leaves. New Phytologist, 2018, 217, 1681-1695.	3.5	55
53	Linear mitochondrial DNA is rapidly degraded by components of the replication machinery. Nature Communications, 2018, 9, 1727.	5.8	151
54	HMGB2 Loss upon Senescence Entry Disrupts Genomic Organization and Induces CTCF Clustering across Cell Types. Molecular Cell, 2018, 70, 730-744.e6.	4.5	164

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55	Integrative genomic profiling of large-cell neuroendocrine carcinomas reveals distinct subtypes of high-grade neuroendocrine lung tumors. Nature Communications, 2018, 9, 1048.	5.8	254
56	A mechanistic classification of clinical phenotypes in neuroblastoma. Science, 2018, 362, 1165-1170.	6.0	213
57	IG-MYC+ neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. Blood, 2018, 132, 2280-2285.	0.6	50
58	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. American Journal of Human Genetics, 2018, 103, 777-785.	2.6	55
59	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	1.5	44
60	Exon Junction Complexes Suppress Spurious Splice Sites to Safeguard Transcriptome Integrity. Molecular Cell, 2018, 72, 482-495.e7.	4.5	61
61	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. Scientific Reports, 2018, 8, 7907.	1.6	16
62	Seniorâ€Ã¸ken syndrome with IQCB1 mutation in Taiwan. Kaohsiung Journal of Medical Sciences, 2018, 34, 588-589.	0.8	2
63	Mechanism suppressing H3K9 trimethylation in pluripotent stem cells and its demise by polyQ-expanded huntingtin mutations. Human Molecular Genetics, 2018, 27, 4117-4134.	1.4	21
64	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	9.4	81
65	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.	0.7	17
66	Metatarsal bony syndactyly in 2 fetuses with Smithâ€Lemliâ€Opitz syndrome: An underâ€recognized part of the clinical spectrum. Clinical Genetics, 2017, 92, 342-343.	1.0	1
67	Copy number increases of transposable elements and proteinâ€coding genes in an invasive fish of hybrid origin. Molecular Ecology, 2017, 26, 4712-4724.	2.0	28
68	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.	2.6	56
69	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.	0.7	7
70	P385â€…Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , .		0
71	Prevalence of deleterious germline variants in risk genes including BRCA1/2 in consecutive ovarian cancer patients (AGO-TR-1). PLoS ONE, 2017, 12, e0186043.	1.1	105
72	Antagonistic modulation of NPY/AgRP and POMC neurons in the arcuate nucleus by noradrenalin. ELife, 2017, 6, .	2.8	35

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73	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, 728-733.	0.7	13
74	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.	1.2	15
75	A novel homozygous PAM16 mutation in a patient with a milder phenotype and longer survival. American Journal of Medical Genetics, Part A, 2016, 170, 2436-2439.	0.7	7
76	Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. American Journal of Human Genetics, 2016, 99, 337-351.	2.6	198
77	A large deletion in RPGR causes XLPRA in Weimaraner dogs. Canine Genetics and Epidemiology, 2016, 3, 7.	2.9	18
78	Mutations in Three Genes Encoding Proteins Involved in Hair Shaft Formation Cause Uncombable Hair Syndrome. American Journal of Human Genetics, 2016, 99, 1292-1304.	2.6	127
79	Cover Image, Volume 170A, Number 9, September 2016. , 2016, 170, i-i.		0
80	Novel IFT122 mutations in three Argentinian patients with cranioectodermal dysplasia: Expanding the mutational spectrum. American Journal of Medical Genetics, Part A, 2016, 170, 1295-1301.	0.7	17
81	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	9.4	74
82	Ectodysplasin signalling genes and phenotypic evolution in sculpins (Cottus). Proceedings of the Royal Society B: Biological Sciences, 2015, 282, 20150746.	1.2	9
83	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Human Molecular Genetics, 2015, 24, 2594-2603.	1.4	32
84	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.	0.3	25
85	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	2.4	85
86	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. Molecular and Cellular Probes, 2015, 29, 330-334.	0.9	22
87	Comprehensive genomic profiles of small cell lung cancer. Nature, 2015, 524, 47-53.	13.7	1,634
88	Mutational dynamics between primary and relapse neuroblastomas. Nature Genetics, 2015, 47, 872-877.	9.4	253
89	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	1.4	26
90	Floral induction in Arabidopsis thaliana by FLOWERING LOCUS T requires direct repression of BLADE-ON-PETIOLE genes by homeodomain protein PENNYWISE. Plant Physiology, 2015, 169, pp.00960.2015.	2.3	51

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91	De novo FUS mutations are the most frequent genetic cause inÂearly-onset German ALS patients. Neurobiology of Aging, 2015, 36, 3117.e1-3117.e6.	1.5	59
92	Loss-of-Function GAS8 Mutations Cause Primary Ciliary Dyskinesia and Disrupt the Nexin-Dynein Regulatory Complex. American Journal of Human Genetics, 2015, 97, 546-554.	2.6	107
93	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	2.6	37
94	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	2.6	34
95	Enrichment of target sequences for next-generation sequencing applications in research and diagnostics. Biological Chemistry, 2014, 395, 231-237.	1.2	32
96	Homozygous missense mutation of NDUFV1 as the cause of infantile bilateral striatal necrosis. Neurogenetics, 2013, 14, 85-87.	0.7	140
97	Attenuated BMP1 Function Compromises Osteogenesis, Leading to Bone Fragility in Humans and Zebrafish. American Journal of Human Genetics, 2012, 90, 661-674.	2.6	192
98	A Truncating Mutation of CEP135 Causes Primary Microcephaly and Disturbed Centrosomal Function. American Journal of Human Genetics, 2012, 90, 871-878.	2.6	153
99	Assessing the Enrichment Performance in Targeted Resequencing Experiments. Human Mutation, 2012, 33, 635-641.	1.1	27
100	Nonsense Mutations in SMPX, Encoding a Protein Responsive to Physical Force, Result in X-Chromosomal Hearing Loss. American Journal of Human Genetics, 2011, 88, 621-627.	2.6	70
101	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. American Journal of Human Genetics, 2011, 89, 668-674.	2.6	89