# Janine AltmÃ¼ller

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | Comprehensive genomic profiles of small cell lung cancer. Nature, 2015, 524, 47-53. | 13.7 | 1,634 |
| 2 | Multiancestry association study identifies new asthma risk loci that colocalize with immune-cell enhancer marks. Nature Genetics, 2018, 50, 42-53. | 9.4 | 426 |
| 3 | 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 |
| 4 | Mutational dynamics between primary and relapse neuroblastomas. Nature Genetics, 2015, 47, 872-877. | 9.4 | 253 |
| 5 | A mechanistic classification of clinical phenotypes in neuroblastoma. Science, 2018, 362, 1165-1170. | 6.0 | 213 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. Nature Communications, 2018, 9, 727. | 5.8 | 160 |
| 10 | A Truncating Mutation of CEP135 Causes Primary Microcephaly and Disturbed Centrosomal Function. American Journal of Human Genetics, 2012, 90, 871-878. | 2.6 | 153 |
| 11 | Linear mitochondrial DNA is rapidly degraded by components of the replication machinery. Nature Communications, 2018, 9, 1727. | 5.8 | 151 |
| 12 | Homozygous missense mutation of NDUFV1 as the cause of infantile bilateral striatal necrosis. Neurogenetics, 2013, 14, 85-87. | 0.7 | 140 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Longâ€lived macrophage reprogramming drives spike proteinâ€mediated inflammasome activation in COVIDâ€19. EMBO Molecular Medicine, 2021, 13, e14150. | 3.3 | 98 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166. | 2.4 | 85 |
| 20 | Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549. | 9.4 | 81 |
| 21 | Human brain organoids assemble functionally integrated bilateral optic vesicles. Cell Stem Cell, 2021, 28, 1740-1757.e8. | 5.2 | 77 |
| 22 | TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43. | 9.4 | 74 |
| 23 | 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 |
| 24 | RNA polymerase II is required for spatial chromatin reorganization following exit from mitosis. Science Advances, 2021, 7, eabg8205. | 4.7 | 70 |
| 25 | Exon Junction Complexes Suppress Spurious Splice Sites to Safeguard Transcriptome Integrity. Molecular Cell, 2018, 72, 482-495.e7. | 4.5 | 61 |
| 26 | Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. Nature Genetics, 2021, 53, 1673-1685. | 9.4 | 61 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | SMG5-SMG7 authorize nonsense-mediated mRNA decay by enabling SMG6 endonucleolytic activity. Nature Communications, 2021, 12, 3965. | 5.8 | 54 |
| 32 | 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 |
| 33 | IG-MYC+ neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. Blood, 2018, 132, 2280-2285. | 0.6 | 50 |
| 34 | 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 |
| 35 | 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 |
| 36 | Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846. | 1.5 | 44 |

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|---|---|---|---|
| 37 | Tissue-infiltrating macrophages mediate an exosome-based metabolic reprogramming upon DNA damage. Nature Communications, 2020, 11, 42. | 5.8 | 44 |
| 38 | cfNOMe â€”ÂA single assay for comprehensive epigenetic analyses of cell-free DNA. Genome Medicine, 2020, 12, 54. | 3.6 | 39 |
| 39 | Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770. | 2.6 | 37 |
| 40 | The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523. | 1.1 | 35 |
| 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 | Antagonistic modulation of NPY/AgRP and POMC neurons in the arcuate nucleus by noradrenalin. ELife, 2017, 6, . | 2.8 | 35 |
| 43 | 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 |
| 44 | Enrichment of target sequences for next-generation sequencing applications in research and diagnostics. Biological Chemistry, 2014, 395, 231-237. | 1.2 | 32 |
| 45 | 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 |
| 46 | R-loops trigger the release of cytoplasmic ssDNAs leading to chronic inflammation upon DNA damage. Science Advances, 2021, 7, eabj5769. | 4.7 | 30 |
| 47 | 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 |
| 48 | Assessing the Enrichment Performance in Targeted Resequencing Experiments. Human Mutation, 2012, 33, 635-641. | 1.1 | 27 |
| 49 | The splicing factor XAB2 interacts with ERCC1-XPF and XPG for R-loop processing. Nature Communications, 2021, 12, 3153. | 5.8 | 27 |
| 50 | Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17. | 1.4 | 26 |
| 51 | 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 |
| 52 | 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 |
| 53 | Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. Journal of Human Genetics, 2019, 64, 803-813. | 1.1 | 25 |
| 54 | Deregulation and epigenetic modification of BCL2-family genes cause resistance to venetoclax in hematologic malignancies. Blood, 2022, 140, 2113-2126. | 0.6 | 24 |

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|---|---|---|---|
| 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, 330-334. | 0.9 | 22 |
| 56 | 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 |
| 57 | Transposable elements and introgression introduce genetic variation in the invasive ant Cardiocondyla obscurior. Molecular Ecology, 2021, 30, 6211-6228. | 2.0 | 20 |
| 58 | 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 |
| 59 | A large deletion in RPGR causes XLPRA in Weimaraner dogs. Canine Genetics and Epidemiology, 2016, 3, 7. | 2.9 | 18 |
| 60 | Genomeâ€wide patterns of transposon proliferation in an evolutionary young hybrid fish. Molecular Ecology, 2019, 28, 1491-1505. | 2.0 | 18 |
| 61 | 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 |
| 62 | 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 |
| 63 | Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. Scientific Reports, 2018, 8, 7907. | 1.6 | 16 |
| 64 | 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 |
| 65 | Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351. | 1.1 | 16 |
| 66 | Mitochondrial respiratory chain function promotes extracellular matrix integrity in cartilage. Journal of Biological Chemistry, 2021, 297, 101224. | 1.6 | 16 |
| 67 | 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 |
| 68 | 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 |
| 69 | Novel PNKP mutations causing defective DNA strand break repair and PARP1 hyperactivity in MCSZ. Neurology: Genetics, 2019, 5, e320. | 0.9 | 15 |
| 70 | Exome sequencing in 38 patients with intracranial aneurysms and subarachnoid hemorrhage. Journal of Neurology, 2020, 267, 2533-2545. | 1.8 | 14 |
| 71 | 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 |
| 72 | 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 |

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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 | Single-cell transcriptome sequencing on the Nanopore platform with ScNapBar. Rna, 2021, 27, 763-770. | 1.6 | 12 |
| 75 | 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 |
| 76 | 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 |
| 77 | Ectodysplasin signalling genes and phenotypic evolution in sculpins ( Cottus ). Proceedings of the Royal Society B: Biological Sciences, 2015, 282, 20150746. | 1.2 | 9 |
| 78 | 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 |
| 79 | 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 |
| 80 | ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269. | 0.7 | 8 |
| 81 | Single cellâ€•and spatial â€˜Omics revolutionize physiology. Acta Physiologica, 2022, 235, . | 1.8 | 8 |
| 82 | 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 |
| 83 | 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 |
| 84 | Familial cleft tongue caused by a unique translation initiation codon variant in TP63. European Journal of Human Genetics, 2021, , . | 1.4 | 7 |
| 85 | 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 |
| 86 | The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599. | 1.1 | 6 |
| 87 | 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 |
| 88 | 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 |
| 89 | Variant profiling of colorectal adenomas from three patients of two families with MSH3-related adenomatous polyposis. PLoS ONE, 2021, 16, e0259185. | 1.1 | 5 |
| 90 | 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 |

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|---|---|---|---|
| 91 | WARS1 and SARS1 : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471. | 1.1 | 5 |
| 92 | 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 |
| 93 | A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 2021, 12, 1494. | 1.0 | 3 |
| 94 | Biallelic variants in YRDC cause a developmental disorder with progeroid features. Human Genetics, 2021, 140, 1679-1693. | 1.8 | 3 |
| 95 | Seniorâ€LÃ¸ken syndrome with IQCB1 mutation in Taiwan. Kaohsiung Journal of Medical Sciences, 2018, 34, 588-589. | 0.8 | 2 |
| 96 | A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036. | 1.5 | 2 |
| 97 | 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 |
| 98 | MAGED2 controls vasopressin-induced aquaporin-2 expression in collecting duct cells. Journal of Proteomics, 2022, 252, 104424. | 1.2 | 1 |
| 99 | 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 |
| 100 | Cover Image, Volume 170A, Number 9, September 2016. , 2016, 170, i-i. | | 0 |
| 101 | P385â€…Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , . | | 0 |