Daniel Nilsson

List of Publications by Year
in descending order

76 papers	5,007 citations	159585 30 h-index	95266 68 g-index
81 all docs	81 docs citations	81 times ranked	7730 citing authors

#	Article	IF	Citations
1	The Genome Sequence of Trypanosoma cruzi , Etiologic Agent of Chagas Disease. Science, 2005, 309, 409-415.	12.6	1,273
2	Comparative Genomics of Trypanosomatid Parasitic Protozoa. Science, 2005, 309, 404-409.	12.6	713
3	Selective Charging of tRNA Isoacceptors Explains Patterns of Codon Usage. Science, 2003, 300, 1718-1722.	12.6	228
4	Autoregulation of the nonsense-mediated mRNA decay pathway in human cells. Rna, 2011, 17, 2108-2118.	3.5	221
5	Spliced Leader Trapping Reveals Widespread Alternative Splicing Patterns in the Highly Dynamic Transcriptome of Trypanosoma brucei. PLoS Pathogens, 2010, 6, e1001037.	4.7	165
6	SweGen: a whole-genome data resource of genetic variability in a cross-section of the Swedish population. European Journal of Human Genetics, 2017, 25, 1253-1260.	2.8	148
7	The genome of the heartworm, Dirofilaria immitis , reveals drug and vaccine targets. FASEB Journal, 2012, 26, 4650-4661.	0.5	124
8	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
9	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	6.2	110
10	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations inPIGT. Journal of Medical Genetics, 2013, 50, 521-528.	3.2	108
11	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
12	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	8.2	88
13	PfEMP1-DBL1Î± amino acid motifs in severe disease states of Plasmodium falciparum malaria. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15835-15840.	7.1	83
14	Genetic Spectrum of Idiopathic Restrictive Cardiomyopathy Uncovered by Next-Generation Sequencing. PLoS ONE, 2016, 11, e0163362.	2.5	78
15	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	76
16	Messenger RNA processing sites in Trypanosoma brucei. Molecular and Biochemical Parasitology, 2005, 143, 125-134.	1.1	73
17	Repetitive DNA is associated with centromeric domains in Trypanosoma brucei but not Trypanosoma cruzi. Genome Biology, 2007, 8, R37.	9.6	67
18	Proteomics in Trypanosoma cruzi â€“ localization of novel proteins to various organelles. Proteomics, 2008, 8, 2735-2749.	2.2	60

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19	Phylogenomics of Ligand-Gated Ion Channels Predicts Monepantel Effect. PLoS Pathogens, 2010, 6, e1001091.	4.7	59
20	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	2.5	58
21	The Trypanosoma brucei MitoCarta and its regulation and splicing pattern during development. Nucleic Acids Research, 2010, 38, 7378-7387.	14.5	57
22	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	3.2	57
23	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	51
24	Genome-Wide Identification of Molecular Mimicry Candidates in Parasites. PLoS ONE, 2011, 6, e17546.	2.5	49
25	Database of Trypanosoma cruzi repeated genes: 20 000 additional gene variants. BMC Genomics, 2007, 8, 391.	2.8	47
26	Epigenetic Regulation of Transcription and Virulence in Trypanosoma cruzi by O-Linked Thymine Glucosylation of DNA. Molecular and Cellular Biology, 2011, 31, 1690-1700.	2.3	40
27	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	3.5	36
28	A Solanesyl-diphosphate Synthase Localizes in Glycosomes of Trypanosoma cruzi. Journal of Biological Chemistry, 2006, 281, 39339-39348.	3.4	35
29	CTNND2â€"a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	3.2	35
30	The Short Non-Coding Transcriptome of the Protozoan Parasite Trypanosoma cruzi. PLoS Neglected Tropical Diseases, 2011, 5, e1283.	3.0	35
31	Exome sequencing in one family with gastric- and rectal cancer. BMC Genetics, 2016, 17, 41.	2.7	31
32	Whole-exome sequencing of Ethiopian patients with ichthyosis vulgaris and atopic dermatitis. Journal of Allergy and Clinical Immunology, 2015, 136, 507-509.e19.	2.9	30
33	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€"Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	2.8	29
34	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. Clinical Genetics, 2018, 94, 528-537.	2.0	29
35	Further evidence for specific IFIH1 mutation as a cause of Singletonâ€"Merten syndrome with phenotypic heterogeneity. American Journal of Medical Genetics, Part A, 2017, 173, 1396-1399.	1.2	28
36	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	3.5	28

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37	Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30.	8.9	25
38	Expressed sequence tag analysis of Sarcoptes scabiei. Parasitology, 2003, 127, 139-145.	1.5	24
39	Identification of putative pathogenic single nucleotide variants (SNVs) in genes associated with heart disease in 290 cases of stillbirth. PLoS ONE, 2019, 14, e0210017.	2.5	24
40	var gene transcription dynamics in Plasmodium falciparum patient isolates. Molecular and Biochemical Parasitology, 2010, 170, 74-83.	1.1	23
41	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	2.9	23
42	A Large Inversion Involving GNAS Exon A/B and All Exons Encoding GsÎ± Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2017, 32, 776-783.	2.8	22
43	An N-Terminal Missense Mutation in STX11 Causative of FHL4 Abrogates Syntaxin-11 Binding to Munc18-2. Frontiers in Immunology, 2014, 4, 515.	4.8	20
44	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
45	AMYCNE: Confident copy number assessment using whole genome sequencing data. PLoS ONE, 2018, 13, e0189710.	2.5	19
46	Identification of three novelFGF16mutations in Xâ€linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Molecular Genetics & Genomic Medicine, 2014, 2, 402-411.	1.2	17
47	Atherosclerosis Susceptibility in Mice Is Independent of the V1 Immunoglobulin Heavy Chain Gene. Arteriosclerosis, Thrombosis, and Vascular Biology, 2016, 36, 25-36.	2.4	17
48	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	2.5	16
49	Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. PLoS ONE, 2021, 16, e0245488.	2.5	13
50	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	2.5	12
51	The Positive Inside Rule Is Stronger When Followed by a Transmembrane Helix. Journal of Molecular Biology, 2014, 426, 2982-2991.	4.2	11
52	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. Neurogenetics, 2021, 22, 71-79.	1.4	11
53	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. PLoS ONE, 2018, 13, e0193928.	2.5	11
54	V374A KCND3 Pathogenic Variant Associated With Paroxysmal Ataxia Exacerbations. Neurology: Genetics, 2021, 7, e546.	1.9	10

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55	A novel stop mutation in the EDNRB gene in a family with Hirschsprungâ€™s disease associated with Multiple Sclerosis. Journal of Pediatric Surgery, 2014, 49, 622-625.	1.6	9
56	Evaluation of the ISL1 gene in the pathogenesis of bladder exstrophy in a Swedish cohort. Human Genome Variation, 2018, 5, 18009.	0.7	9
57	Further support linking the 22q11.2 microduplication to an increased risk of bladder exstrophy and highlighting LZTR1 as a candidate gene. Molecular Genetics & Genomic Medicine, 2019, 7, e666.	1.2	9
58	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. Human Genetics, 2021, 140, 775-790.	3.8	9
59	Strand asymmetry patterns in trypanosomatid parasites. Experimental Parasitology, 2005, 109, 143-149.	1.2	8
60	Comparative genomics of metabolic networks of free-living and parasitic eukaryotes. BMC Genomics, 2010, 11, 217.	2.8	8
61	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	2.3	7
62	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. Leukemia and Lymphoma, 2020, 61, 604-613.	1.3	7
63	pyCancerSig: subclassifying human cancer with comprehensive single nucleotide, structural and microsatellite mutational signature deconstruction from whole genome sequencing. BMC Bioinformatics, 2020, 21, 128.	2.6	7
64	Severe congenital neutropeniaâ€associated JAGN1 mutations unleash a calpainâ€dependent cell death programme in myeloid cells. British Journal of Haematology, 2021, 192, 200-211.	2.5	7
65	Characterization of a Trypanosoma cruzi acetyltransferase: cellular location, activity and structureâ˜†. Molecular and Biochemical Parasitology, 2007, 152, 123-131.	1.1	5
66	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	2.6	5
67	PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , .	2.5	5
68	Two novel colorectal cancer risk loci in the region on chromosome 9q22.32. Oncotarget, 2018, 9, 11170-11179.	1.8	4
69	Heterozygous variants in DCC. Neurology: Genetics, 2020, 6, e526.	1.9	4
70	Whole-genome Linkage Analysis and Sequence Analysis of Candidate Loci in Familial Breast Cancer. Anticancer Research, 2015, 35, 3155-65.	1.1	4
71	GRATâ€”genome-scale rapid alignment tool. Computer Methods and Programs in Biomedicine, 2007, 86, 87-92.	4.7	3
72	Massive parallel sequencing in individuals with multiple primary tumours reveals the benefit of re-analysis. Hereditary Cancer in Clinical Practice, 2021, 19, 46.	1.5	3

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73	A graphical tool for parasite genome annotation. Computer Methods and Programs in Biomedicine, 2004, 73, 55-60.	4.7	2
74	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
75	Adult-Onset Ataxia With Neuropathy and White Matter Abnormalities Due to a Novel SAMD9L Variant. Neurology: Genetics, 2021, 7, e628.	1.9	1
76	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	2.3	1