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	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
2	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
3	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. Neurogenetics, 2021, 22, 71-79.	1.4	11
4	V374A KCND3 Pathogenic Variant Associated With Paroxysmal Ataxia Exacerbations. Neurology: Genetics, 2021, 7, e546.	1.9	10
5	Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. PLoS ONE, 2021, 16, e0245488.	2.5	13
6	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
7	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
8	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
9	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
10	Adult-Onset Ataxia With Neuropathy and White Matter Abnormalities Due to a Novel SAMD9L Variant. Neurology: Genetics, 2021, 7, e628.	1.9	1
11	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
12	Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30.	8.9	25
13	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. Leukemia and Lymphoma, 2020, 61, 604-613.	1.3	7
14	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	2.5	12
15	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	2.6	5
16	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
17	Heterozygous variants in DCC. Neurology: Genetics, 2020, 6, e526.	1.9	4
18	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	2.3	7

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19	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
20	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
21	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	3.5	36
22	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
23	Evaluation of the ISL1 gene in the pathogenesis of bladder exstrophy in a Swedish cohort. Human Genome Variation, 2018, 5, 18009.	0.7	9
24	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
25	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
26	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	2.5	16
27	AMYCNE: Confident copy number assessment using whole genome sequencing data. PLoS ONE, 2018, 13, e0189710.	2.5	19
28	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. PLoS ONE, 2018, 13, e0193928.	2.5	11
29	Two novel colorectal cancer risk loci in the region on chromosome 9q22.32. Oncotarget, 2018, 9, 11170-11179.	1.8	4
30	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
31	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
32	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
33	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
34	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	76
35	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	51
36	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

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37	Exome sequencing in one family with gastric- and rectal cancer. BMC Genetics, 2016, 17, 41.	2.7	31
38	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
39	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
40	Genetic Spectrum of Idiopathic Restrictive Cardiomyopathy Uncovered by Next-Generation Sequencing. PLoS ONE, 2016, 11, e0163362.	2.5	78
41	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	2.9	23
42	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
43	CTNND2â€”a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	3.2	35
44	Whole-genome Linkage Analysis and Sequence Analysis of Candidate Loci in Familial Breast Cancer. Anticancer Research, 2015, 35, 3155-65.	1.1	4
45	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
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	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
48	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
49	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	3.2	57
50	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
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	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
53	The genome of the heartworm, Dirofilaria immitis , reveals drug and vaccine targets. FASEB Journal, 2012, 26, 4650-4661.	0.5	124
54	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

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55	Autoregulation of the nonsense-mediated mRNA decay pathway in human cells. Rna, 2011, 17, 2108-2118.	3.5	221
56	The Short Non-Coding Transcriptome of the Protozoan Parasite Trypanosoma cruzi. PLoS Neglected Tropical Diseases, 2011, 5, e1283.	3.0	35
57	Genome-Wide Identification of Molecular Mimicry Candidates in Parasites. PLoS ONE, 2011, 6, e17546.	2.5	49
58	var gene transcription dynamics in Plasmodium falciparum patient isolates. Molecular and Biochemical Parasitology, 2010, 170, 74-83.	1.1	23
59	Comparative genomics of metabolic networks of free-living and parasitic eukaryotes. BMC Genomics, 2010, 11, 217.	2.8	8
60	The Trypanosoma brucei MitoCarta and its regulation and splicing pattern during development. Nucleic Acids Research, 2010, 38, 7378-7387.	14.5	57
61	Phylogenomics of Ligand-Gated Ion Channels Predicts Monepantel Effect. PLoS Pathogens, 2010, 6, e1001091.	4.7	59
62	Spliced Leader Trapping Reveals Widespread Alternative Splicing Patterns in the Highly Dynamic Transcriptome of Trypanosoma brucei. PLoS Pathogens, 2010, 6, e1001037.	4.7	165
63	Proteomics in Trypanosoma cruzi â€“ localization of novel proteins to various organelles. Proteomics, 2008, 8, 2735-2749.	2.2	60
64	Repetitive DNA is associated with centromeric domains in Trypanosoma brucei but not Trypanosoma cruzi. Genome Biology, 2007, 8, R37.	9.6	67
65	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
66	Database of Trypanosoma cruzi repeated genes: 20 000 additional gene variants. BMC Genomics, 2007, 8, 391.	2.8	47
67	GRATâ€”genome-scale rapid alignment tool. Computer Methods and Programs in Biomedicine, 2007, 86, 87-92.	4.7	3
68	Characterization of a Trypanosoma cruzi acetyltransferase: cellular location, activity and structureâ˜†. Molecular and Biochemical Parasitology, 2007, 152, 123-131.	1.1	5
69	A Solanesyl-diphosphate Synthase Localizes in Glycosomes of Trypanosoma cruzi. Journal of Biological Chemistry, 2006, 281, 39339-39348.	3.4	35
70	Strand asymmetry patterns in trypanosomatid parasites. Experimental Parasitology, 2005, 109, 143-149.	1.2	8
71	Messenger RNA processing sites in Trypanosoma brucei. Molecular and Biochemical Parasitology, 2005, 143, 125-134.	1.1	73
72	The Genome Sequence of Trypanosoma cruzi , Etiologic Agent of Chagas Disease. Science, 2005, 309, 409-415.	12.6	1,273

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73	Comparative Genomics of Trypanosomatid Parasitic Protozoa. Science, 2005, 309, 404-409.	12.6	713
74	A graphical tool for parasite genome annotation. Computer Methods and Programs in Biomedicine, 2004, 73, 55-60.	4.7	2
75	Selective Charging of tRNA Isoacceptors Explains Patterns of Codon Usage. Science, 2003, 300, 1718-1722.	12.6	228
76	Expressed sequence tag analysis of Sarcoptes scabiei. Parasitology, 2003, 127, 139-145.	1.5	24