

Cas Simons

List of Publications by Year in descending order

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83
papers

5,189
citations

126708
33
h-index

95083
68
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92
all docs

92
docs citations

92
times ranked

8820
citing authors

#	ARTICLE	IF	CITATIONS
1	Long noncoding RNAs in mouse embryonic stem cell pluripotency and differentiation. <i>Genome Research</i> , 2008, 18, 1433-1445.	2.4	698
2	The transcriptional network that controls growth arrest and differentiation in a human myeloid leukemia cell line. <i>Nature Genetics</i> , 2009, 41, 553-562.	9.4	408
3	Tiny RNAs associated with transcription start sites in animals. <i>Nature Genetics</i> , 2009, 41, 572-578.	9.4	327
4	Expression of distinct RNAs from 3' untranslated regions. <i>Nucleic Acids Research</i> , 2011, 39, 2393-2403.	6.5	185
5	Effect of 5'UTR introns on gene expression in <i>Arabidopsis thaliana</i> . <i>BMC Genomics</i> , 2006, 7, 120.	1.2	184
6	A De Novo Mutation in the β -Tubulin Gene TUBB4A Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. <i>American Journal of Human Genetics</i> , 2013, 92, 767-773.	2.6	174
7	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephalopathies. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 501-515.	0.5	163
8	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. <i>Brain</i> , 2014, 137, 1921-1930.	3.7	161
9	Patient-iPSC-Derived Kidney Organoids Show Functional Validation of a Ciliopathic Renal Phenotype and Reveal Underlying Pathogenetic Mechanisms. <i>American Journal of Human Genetics</i> , 2018, 102, 816-831.	2.6	157
10	Mutations in DARS Cause Hypomyelination with Brain Stem and Spinal Cord Involvement and Leg Spasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 774-780.	2.6	151
11	Nuclear-localized tiny RNAs are associated with transcription initiation and splice sites in metazoans. <i>Nature Structural and Molecular Biology</i> , 2010, 17, 1030-1034.	3.6	146
12	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. <i>Nature Genetics</i> , 2015, 47, 73-77.	9.4	130
13	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. <i>Nature Communications</i> , 2015, 6, 7623.	5.8	127
14	Whole exome sequencing in patients with white matter abnormalities. <i>Annals of Neurology</i> , 2016, 79, 1031-1037.	2.8	116
15	Transposon-free regions in mammalian genomes. <i>Genome Research</i> , 2005, 16, 164-172.	2.4	102
16	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. <i>American Journal of Human Genetics</i> , 2019, 104, 925-935.	2.6	92
17	Mural lymphatic endothelial cells regulate meningeal angiogenesis in the zebrafish. <i>Nature Neuroscience</i> , 2017, 20, 774-783.	7.1	91
18	Evolution, biogenesis and function of promoter-associated RNAs. <i>Cell Cycle</i> , 2009, 8, 2332-2338.	1.3	89

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19	Loss-of-Function Alanyl-tRNA Synthetase Mutations Cause an Autosomal-Recessive Early-Onset Epileptic Encephalopathy with Persistent Myelination Defect. American Journal of Human Genetics, 2015, 96, 675-681.	2.6	84
20	Whole exome sequencing in family trios reveals <i>de novo</i> mutations in <i>PURA</i> as a cause of severe neurodevelopmental delay and learning disability. Journal of Medical Genetics, 2014, 51, 806-813.	1.5	73
21	Exome sequencing in developmental eye disease leads to identification of causal variants in <i>CJA8</i> , <i>CRYGC</i> , <i>PAX6</i> and <i>CYP1B1</i> . European Journal of Human Genetics, 2014, 22, 907-915.	1.4	66
22	A recurrent <i>de novo</i> mutation in <i>TMEM106B</i> causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	3.7	64
23	<i>mafba</i> is a downstream transcriptional effector of <i>Vegfc</i> signaling essential for embryonic lymphangiogenesis in zebrafish. Genes and Development, 2015, 29, 1618-1630.	2.7	63
24	<i>Tmem2</i> Regulates Embryonic <i>Vegf</i> Signaling by Controlling Hyaluronic Acid Turnover. Developmental Cell, 2017, 40, 123-136.	3.1	63
25	Identification of a Novel <i>de Novo</i> p.Phe932Ile <i>KCNT1</i> Mutation in a Patient With Leukoencephalopathy and Severe Epilepsy. Pediatric Neurology, 2014, 50, 112-114.	1.0	62
26	<i>DARS</i> -associated leukoencephalopathy can mimic a steroid-responsive neuroinflammatory disorder. Neurology, 2015, 84, 226-230.	1.5	61
27	<i>TUBB4A</i> mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. Human Molecular Genetics, 2017, 26, 4506-4518.	1.4	59
28	Bi-allelic Mutations in <i>EPRS</i> , Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2018, 102, 676-684.	2.6	58
29	Chronic non-freezing cold injury results in neuropathic pain due to a sensory neuropathy. Brain, 2017, 140, 2557-2569.	3.7	54
30	<i>TUBB4A</i> <i>de novo</i> mutations cause isolated hypomyelination. Neurology, 2014, 83, 898-902.	1.5	52
31	Heterozygous Variants in the Mechanosensitive Ion Channel <i>TMEM63A</i> Result in Transient Hypomyelination during Infancy. American Journal of Human Genetics, 2019, 105, 996-1004.	2.6	52
32	<i>De Novo</i> Variants in <i>WDR37</i> Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
33	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in <i>AIFM1</i> . Neurogenetics, 2017, 18, 185-194.	0.7	38
34	The clinical, biochemical and genetic features associated with <i>RMND1</i> -related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	1.5	35
35	The Alternative Splicing Regulator <i>Nova2</i> Constrains Vascular <i>Erk</i> Signaling to Limit Specification of the Lymphatic Lineage. Developmental Cell, 2019, 49, 279-292.e5.	3.1	35
36	Orthologous MicroRNA Genes Are Located in Cancer-Associated Genomic Regions in Human and Mouse. PLoS ONE, 2007, 2, e1133.	1.1	34

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37	Magnetic resonance imaging spectrum of succinate dehydrogenase-related infantile leukoencephalopathy. <i>Annals of Neurology</i> , 2016, 79, 379-386.	2.8	34
38	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. <i>Med</i> , 2021, 2, 49-73.e10.	2.2	33
39	Biallelic variants in <i>LARS2</i> and <i>KARS</i> cause deafness and (ovario)leukodystrophy. <i>Neurology</i> , 2019, 92, e1225-e1237.	1.5	32
40	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 86.	1.2	29
41	Identification of Candidate Genes for Mayer-Rokitansky-Kuster-Hauser Syndrome Using Genomic Approaches. <i>Sexual Development</i> , 2019, 13, 26-34.	1.1	29
42	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. <i>Human Molecular Genetics</i> , 2020, 29, 1568-1579.	1.4	29
43	Maintenance of transposon-free regions throughout vertebrate evolution. <i>BMC Genomics</i> , 2007, 8, 470.	1.2	28
44	A mutation affecting laminin alpha 5 polymerisation gives rise to a syndromic developmental disorder. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	28
45	Genome sequencing in persistently unsolved white matter disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 144-152.	1.7	26
46	Utilising polymorphisms to achieve allele-specific genome editing in zebrafish. <i>Biology Open</i> , 2017, 6, 125-131.	0.6	19
47	Expression Pattern of the Aspartyl-tRNA Synthetase DARS in the Human Brain. <i>Frontiers in Molecular Neuroscience</i> , 2018, 11, 81.	1.4	19
48	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. <i>Brain</i> , 2017, 140, 2550-2556.	3.7	18
49	Cerebral hypomyelination associated with biallelic variants of <i>FIG4</i> . <i>Human Mutation</i> , 2019, 40, 619-630.	1.1	18
50	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. <i>Annals of Neurology</i> , 2020, 88, 264-273.	2.8	17
51	Multomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFBI0. <i>Human Mutation</i> , 2021, 42, 19-24.	1.1	17
52	Biallelic <i>PI4KA</i> variants cause neurological, intestinal and immunological disease. <i>Brain</i> , 2021, 144, 3597-3610.	3.7	17
53	The RNA helicase Ddx21 controls Vegf-driven developmental lymphangiogenesis by balancing endothelial cell ribosome biogenesis and p53 function. <i>Nature Cell Biology</i> , 2021, 23, 1136-1147.	4.6	17
54	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. <i>PLoS ONE</i> , 2017, 12, e0178125.	1.1	16

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55	Mutations in <i>SZT2</i> result in early-onset epileptic encephalopathy and leukoencephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 1443-1448.	0.7	15
56	Leukoencephalopathy due to variants in <i>GFPT1</i> associated congenital myasthenic syndrome. Neurology, 2019, 92, e587-e593.	1.5	14
57	Type II Alexander disease caused by splicing errors and aberrant overexpression of an uncharacterized GFAP isoform. Human Mutation, 2020, 41, 1131-1137.	1.1	14
58	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16 579 Newborns by Using a Novel Genomic Workflow. JAMA Network Open, 2022, 5, e2141911.	2.8	14
59	Gene sequences from New Zealand's extinct huia. Journal of the Royal Society of New Zealand, 2002, 32, 327-335.	1.0	12
60	Tmem2 Regulates Embryonic Vegf Signaling by Controlling Hyaluronic Acid Turnover. Developmental Cell, 2017, 40, 421.	3.1	12
61	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss—Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	1.0	12
62	The zebrafish <i>grime</i> mutant uncovers an evolutionarily conserved role for Tmem161b in the control of cardiac rhythm. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	12
63	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
64	Absence of Axoglial Paranodal Junctions in a Child With <i>CNTNAP1</i> Mutations, Hypomyelination, and Arthrogryposis. Journal of Child Neurology, 2018, 33, 642-650.	0.7	11
65	Severe Leukoencephalopathy with Clinical Recovery Caused by Recessive BOLA3 Mutations. JIMD Reports, 2018, 43, 63-70.	0.7	10
66	Recessive mutations in <i>NDUFA2</i> cause mitochondrial leukoencephalopathy. Clinical Genetics, 2018, 93, 396-400.	1.0	10
67	<i>MAFB</i> modulates the maturation of lymphatic vascular networks in mice. Developmental Dynamics, 2020, 249, 1201-1216.	0.8	10
68	Isolated proteinuria due to CUBN homozygous mutation “challenging the investigative paradigm. BMC Nephrology, 2019, 20, 330.	0.8	9
69	Lessons learnt from multifaceted diagnostic approaches to the first 150 families in Victoria's Undiagnosed Diseases Program. Journal of Medical Genetics, 2022, 59, 748-758.	1.5	9
70	A protocol for the identification and validation of novel genetic causes of kidney disease. BMC Nephrology, 2015, 16, 152.	0.8	8
71	Adult-Diagnosed Nonsyndromic Nephronophthisis in Australian Families Caused by Biallelic NPHP4 Variants. American Journal of Kidney Diseases, 2020, 76, 282-287.	2.1	8
72	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	1.4	8

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73	Expanded phenotype of AARS1-related white matter disease. <i>Genetics in Medicine</i> , 2021, 23, 2352-2359.	1.1	8
74	Expanding the genotypic spectrum of <i>CCBE1</i> mutations in Hennekam syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2694-2697.	0.7	7
75	Localised Collagen2a1 secretion supports lymphatic endothelial cell migration in the zebrafish embryo. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	7
76	Myosin Vb is required for correct trafficking of N-cadherin and cardiac chamber ballooning. <i>Developmental Dynamics</i> , 2019, 248, 284-295.	0.8	6
77	Rapid Identification of a Novel Complex I MT-ND3 m.10134C>A Mutation in a Leigh Syndrome Patient. <i>PLoS ONE</i> , 2014, 9, e104879.	1.1	5
78	Precision Medicine Diagnostics for Rare Kidney Disease: Twitter as a Tool in Clinical Genomic Translation. <i>Kidney Medicine</i> , 2019, 1, 315-318.	1.0	4
79	Early-Onset Vascular Leukoencephalopathy Caused by Bi-Allelic NOTCH3 Variants. <i>Neuropediatrics</i> , 2022, 53, 115-121.	0.3	4
80	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. <i>Journal of Child Neurology</i> , 2021, 36, 133-140.	0.7	3
81	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. <i>European Journal of Medical Genetics</i> , 2021, 64, 104259.	0.7	2
82	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. <i>Pediatric Neurology</i> , 2021, 121, 11-19.	1.0	2
83	Reply: The recurrent mutation in TMEM106B also causes hypomyelinating leukodystrophy in China and is a CpG hotspot. <i>Brain</i> , 2018, 141, e37-e37.	3.7	0