Cas Simons

List of Publications by Citations

81 papers	3,843 citations	31 h-index	61 g-index
92 ext. papers	4,604 ext. citations	8.3 avg, IF	4.49 L-index

#	Paper	IF	Citations
81	Long noncoding RNAs in mouse embryonic stem cell pluripotency and differentiation. *Genome Research*, **2008**, 18, 1433-45	9.7	608
80	The transcriptional network that controls growth arrest and differentiation in a human myeloid leukemia cell line. *Nature Genetics*, **2009**, 41, 553-62	36.3	356
79	Tiny RNAs associated with transcription start sites in animals. *Nature Genetics*, **2009**, 41, 572-8	36.3	302
78	Expression of distinct RNAs from 3Ɯntranslated regions. *Nucleic Acids Research*, **2011**, 39, 2393-403	20.1	153
77	Effect of 5ꓦTR introns on gene expression in Arabidopsis thaliana. *BMC Genomics*, **2006**, 7, 120	4.5	136
76	Nuclear-localized tiny RNAs are associated with transcription initiation and splice sites in metazoans. *Nature Structural and Molecular Biology*, **2010**, 17, 1030-4	17.6	134
75	A de novo mutation in the Ɛtubulin gene TUBB4A results in the leukoencephalopathy hypomyelination with atrophy of the basal ganglia and cerebellum. *American Journal of Human Genetics*, **2013**, 92, 767-73	11	133
74	Mutations in DARS cause hypomyelination with brain stem and spinal cord involvement and leg spasticity. *American Journal of Human Genetics*, **2013**, 92, 774-80	11	127
73	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephelopathies. *Molecular Genetics and Metabolism*, **2015**, 114, 501-515	3.7	126
72	Patient-iPSC-Derived Kidney Organoids Show Functional Validation of a Ciliopathic Renal Phenotype and Reveal Underlying Pathogenetic Mechanisms. *American Journal of Human Genetics*, **2018**, 102, 816-831	11	93
71	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. *Nature Communications*, **2015**, 6, 7623	17.4	92
70	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. *Nature Genetics*, **2015**, 47, 73-7	36.3	91
69	Transposon-free regions in mammalian genomes. *Genome Research*, **2006**, 16, 164-72	9.7	89
68	Whole exome sequencing in patients with white matter abnormalities. *Annals of Neurology*, **2016**, 79, 1031-1037	9.4	86
67	Evolution, biogenesis and function of promoter-associated RNAs. *Cell Cycle*, **2009**, 8, 2332-8	4.7	83
66	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, 675-81	11	72
65	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. *Brain*, **2014**, 137, 1921-30	11.2	72

64	Mural lymphatic endothelial cells regulate meningeal angiogenesis in the zebrafish. *Nature Neuroscience*, **2017**, 20, 774-783	25.5	62
63	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. *American Journal of Human Genetics*, **2019**, 104, 925-935	11	56
62	Whole exome sequencing in family trios reveals de novo mutations in PURA as a cause of severe neurodevelopmental delay and learning disability. *Journal of Medical Genetics*, **2014**, 51, 806-13	5.8	55
61	Exome sequencing in developmental eye disease leads to identification of causal variants in GJA8, CRYGC, PAX6 and CYP1B1. *European Journal of Human Genetics*, **2014**, 22, 907-15	5.3	52
60	Identification of a novel de novo p.Phe932Ile KCNT1 mutation in a patient with leukoencephalopathy and severe epilepsy. *Pediatric Neurology*, **2014**, 50, 112-4	2.9	49
59	DARS-associated leukoencephalopathy can mimic a steroid-responsive neuroinflammatory disorder. *Neurology*, **2015**, 84, 226-30	6.5	48
58	Chronic non-freezing cold injury results in neuropathic pain due to a sensory neuropathy. *Brain*, **2017**, 140, 2557-2569	11.2	42
57	mafba is a downstream transcriptional effector of Vegfc signaling essential for embryonic lymphangiogenesis in zebrafish. *Genes and Development*, **2015**, 29, 1618-30	12.6	42
56	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. *Brain*, **2017**, 140, 3105-3111	11.2	42
55	TUBB4A de novo mutations cause isolated hypomyelination. *Neurology*, **2014**, 83, 898-902	6.5	41
54	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. *American Journal of Human Genetics*, **2018**, 102, 676-684	11	38
53	Tmem2 Regulates Embryonic Vegf Signaling by Controlling Hyaluronic Acid Turnover. *Developmental Cell*, **2017**, 40, 123-136	10.2	33
52	TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. *Human Molecular Genetics*, **2017**, 26, 4506-4518	5.6	32
51	Orthologous microRNA genes are located in cancer-associated genomic regions in human and mouse. *PLoS ONE*, **2007**, 2, e1133	3.7	32
50	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. *Neurogenetics*, **2017**, 18, 185-194	3	28
49	The clinical, biochemical and genetic features associated with -related mitochondrial disease. *Journal of Medical Genetics*, **2016**, 53, 768-775	5.8	27
48	Magnetic resonance imaging spectrum of succinate dehydrogenase-related infantile leukoencephalopathy. *Annals of Neurology*, **2016**, 79, 379-86	9.4	26
47	Maintenance of transposon-free regions throughout vertebrate evolution. *BMC Genomics*, **2007**, 8, 470	4.5	23

46	Biallelic variants in and cause deafness and (ovario)leukodystrophy. *Neurology*, **2019**, 92, e1225-e1237	6.5	21
45	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. *American Journal of Human Genetics*, **2019**, 105, 413-424	11	19
44	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. *Orphanet Journal of Rare Diseases*, **2018**, 13, 86	4.2	16
43	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. *Brain*, **2017**, 140, 2550-2556	11.2	15
42	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. *Med*, **2021**, 2, 49-73	31.7	15
41	Heterozygous Variants in the Mechanosensitive Ion Channel TMEM63A Result in Transient Hypomyelination during Infancy. *American Journal of Human Genetics*, **2019**, 105, 996-1004	11	13
40	The Alternative Splicing Regulator Nova2 Constrains Vascular Erk Signaling to Limit Specification of the Lymphatic Lineage. *Developmental Cell*, **2019**, 49, 279-292.e5	10.2	13
39	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. *PLoS ONE*, **2017**, 12, e0178125	3.7	13
38	Genome sequencing in persistently unsolved white matter disorders. *Annals of Clinical and Translational Neurology*, **2020**, 7, 144-152	5.3	13
37	Utilising polymorphisms to achieve allele-specific genome editing in zebrafish. *Biology Open*, **2017**, 6, 125-131	2.2	12
36	Identification of Candidate Genes for Mayer-Rokitansky-Küster-Hauser Syndrome Using Genomic Approaches. *Sexual Development*, **2019**, 13, 26-34	1.6	12
35	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. *Human Molecular Genetics*, **2020**, 29, 1568-1579	5.6	11
34	Expression Pattern of the Aspartyl-tRNA Synthetase DARS in the Human Brain. *Frontiers in Molecular Neuroscience*, **2018**, 11, 81	6.1	11
33	A mutation affecting laminin alpha 5 polymerisation gives rise to a syndromic developmental disorder. *Development (Cambridge)*, **2020**, 147,	6.6	11
32	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss-Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. *Pediatric Neurology*, **2017**, 66, 59-62	2.9	10
31	Gene sequences from New Zealand's extinct huia. *Journal of the Royal Society of New Zealand*, **2002**, 32, 327-335	2	10
30	Mutations in SZT2 result in early-onset epileptic encephalopathy and leukoencephalopathy. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1443-1448	2.5	9
29	Multiomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. *Human Mutation*, **2021**, 42, 19-24	4.7	9

28	Recessive mutations in NDUFA2 cause mitochondrial leukoencephalopathy. *Clinical Genetics*, **2018**, 93, 396-400	4	8
27	Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogryposis. *Journal of Child Neurology*, **2018**, 33, 642-650	2.5	8
26	Cerebral hypomyelination associated with biallelic variants of FIG4. *Human Mutation*, **2019**, 40, 619-630	4.7	8
25	Lysosomal dysfunction in hypomyelinating leukodystrophy. *Neurology: Genetics*, **2018**, 4, e288	3.8	8
24	Tmem2 Regulates Embryonic Vegf Signaling by Controlling Hyaluronic Acid Turnover. *Developmental Cell*, **2017**, 40, 421	10.2	7
23	Type II Alexander disease caused by splicing errors and aberrant overexpression of an uncharacterized GFAP isoform. *Human Mutation*, **2020**, 41, 1131-1137	4.7	7
22	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. *Annals of Neurology*, **2020**, 88, 264-273	9.4	7
21	Leukoencephalopathy due to variants in associated congenital myasthenic syndrome. *Neurology*, **2019**, 92, e587-e593	6.5	7
20	Expanding the genotypic spectrum of CCBE1 mutations in Hennekam syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2694-7	2.5	6
19	A protocol for the identification and validation of novel genetic causes of kidney disease. *BMC Nephrology*, **2015**, 16, 152	2.7	6
18	Adult-Diagnosed Nonsyndromic Nephronophthisis in Australian Families Caused by Biallelic NPHP4 Variants. *American Journal of Kidney Diseases*, **2020**, 76, 282-287	7.4	6
17	MAFB modulates the maturation of lymphatic vascular networks in mice. *Developmental Dynamics*, **2020**, 249, 1201-1216	2.9	5
16	Severe Leukoencephalopathy with Clinical Recovery Caused by Recessive BOLA3 Mutations. *JIMD Reports*, **2019**, 43, 63-70	1.9	5
15	Rapid identification of a novel complex I MT-ND3 m.10134C>A mutation in a Leigh syndrome patient. *PLoS ONE*, **2014**, 9, e104879	3.7	5
14	Localised Collagen2a1 secretion supports lymphatic endothelial cell migration in the zebrafish embryo. *Development (Cambridge)*, **2020**, 147,	6.6	5
13	The RNA helicase Ddx21 controls Vegfc-driven developmental lymphangiogenesis by balancing endothelial cell ribosome biogenesis and p53 function. *Nature Cell Biology*, **2021**, 23, 1136-1147	23.4	4
12	The zebrafish 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,	11.5	4
11	Isolated proteinuria due to CUBN homozygous mutation - challenging the investigative paradigm. *BMC Nephrology*, **2019**, 20, 330	2.7	3

10	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. *European Journal of Human Genetics*, **2021**, 29, 271-279	5.3	3
9	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16 579 Newborns by Using a Novel Genomic Workflow.. *JAMA Network Open*, **2022**, 5, e2141911	10.4	2
8	Expanded phenotype of AARS1-related white matter disease. *Genetics in Medicine*, **2021**, 23, 2352-2359	8.1	2
7	Precision Medicine Diagnostics for Rare Kidney Disease: Twitter as a Tool in Clinical Genomic Translation. *Kidney Medicine*, **2019**, 1, 315-318	2.8	1
6	Myosin Vb is required for correct trafficking of N-cadherin and cardiac chamber ballooning. *Developmental Dynamics*, **2019**, 248, 284-295	2.9	1
5	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. *Brain*, **2021**,	11.2	1
4	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. *European Journal of Medical Genetics*, **2021**, 64, 104259	2.6	0
3	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. *Journal of Child Neurology*, **2021**, 36, 133-140	2.5	0
2	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. *Pediatric Neurology*, **2021**, 121, 11-19	2.9	0
1	Reply: The recurrent mutation in TMEM106B also causes hypomyelinating leukodystrophy in China and is a CpG hotspot. *Brain*, **2018**, 141, e37	11.2	