

Cas Simons

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16 579 Newborns by Using a Novel Genomic Workflow.. <i>JAMA Network Open</i> , 2022 , 5, e2141911	10.4	2
80	The RNA helicase Ddx21 controls Vegfc-driven developmental lymphangiogenesis by balancing endothelial cell ribosome biogenesis and p53 function. <i>Nature Cell Biology</i> , 2021 , 23, 1136-1147	23.4	4
79	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104259	2.6	0
78	Multimic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. <i>Human Mutation</i> , 2021 , 42, 19-24	4.7	9
77	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. <i>Journal of Child Neurology</i> , 2021 , 36, 133-140	2.5	0
76	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. <i>Med</i> , 2021 , 2, 49-73	31.7	15
75	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. <i>European Journal of Human Genetics</i> , 2021 , 29, 271-279	5.3	3
74	The zebrafish mutant uncovers an evolutionarily conserved role for Tmem161b in the control of cardiac rhythm. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021 , 118,	11.5	4
73	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. <i>Brain</i> , 2021 ,	11.2	1
72	Expanded phenotype of AARS1-related white matter disease. <i>Genetics in Medicine</i> , 2021 , 23, 2352-2359	8.1	2
71	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. <i>Pediatric Neurology</i> , 2021 , 121, 11-19	2.9	0
70	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. <i>Human Molecular Genetics</i> , 2020 , 29, 1568-1579	5.6	11
69	MAFB modulates the maturation of lymphatic vascular networks in mice. <i>Developmental Dynamics</i> , 2020 , 249, 1201-1216	2.9	5
68	Type II Alexander disease caused by splicing errors and aberrant overexpression of an uncharacterized GFAP isoform. <i>Human Mutation</i> , 2020 , 41, 1131-1137	4.7	7
67	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. <i>Annals of Neurology</i> , 2020 , 88, 264-273	9.4	7
66	A mutation affecting laminin alpha 5 polymerisation gives rise to a syndromic developmental disorder. <i>Development (Cambridge)</i> , 2020 , 147,	6.6	11
65	Genome sequencing in persistently unsolved white matter disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 144-152	5.3	13

64	Adult-Diagnosed Nonsyndromic Nephronophthisis in Australian Families Caused by Biallelic NPHP4 Variants. <i>American Journal of Kidney Diseases</i> , 2020 , 76, 282-287	7.4	6
63	Localised Collagen2a1 secretion supports lymphatic endothelial cell migration in the zebrafish embryo. <i>Development (Cambridge)</i> , 2020 , 147,	6.6	5
62	Isolated proteinuria due to CUBN homozygous mutation - challenging the investigative paradigm. <i>BMC Nephrology</i> , 2019 , 20, 330	2.7	3
61	Precision Medicine Diagnostics for Rare Kidney Disease: Twitter as a Tool in Clinical Genomic Translation. <i>Kidney Medicine</i> , 2019 , 1, 315-318	2.8	1
60	Heterozygous Variants in the Mechanosensitive Ion Channel TMEM63A Result in Transient Hypomyelination during Infancy. <i>American Journal of Human Genetics</i> , 2019 , 105, 996-1004	11	13
59	The Alternative Splicing Regulator Nova2 Constrains Vascular Erk Signaling to Limit Specification of the Lymphatic Lineage. <i>Developmental Cell</i> , 2019 , 49, 279-292.e5	10.2	13
58	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	11	56
57	Biallelic variants in and cause deafness and (ovario)leukodystrophy. <i>Neurology</i> , 2019 , 92, e1225-e1237	6.5	21
56	Myosin Vb is required for correct trafficking of N-cadherin and cardiac chamber ballooning. <i>Developmental Dynamics</i> , 2019 , 248, 284-295	2.9	1
55	Severe Leukoencephalopathy with Clinical Recovery Caused by Recessive BOLA3 Mutations. <i>JIMD Reports</i> , 2019 , 43, 63-70	1.9	5
54	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019 , 105, 413-424	11	19
53	Cerebral hypomyelination associated with biallelic variants of FIG4. <i>Human Mutation</i> , 2019 , 40, 619-630	4.7	8
52	Leukoencephalopathy due to variants in associated congenital myasthenic syndrome. <i>Neurology</i> , 2019 , 92, e587-e593	6.5	7
51	Identification of Candidate Genes for Mayer-Rokitansky-Küster-Hauser Syndrome Using Genomic Approaches. <i>Sexual Development</i> , 2019 , 13, 26-34	1.6	12
50	Reply: The recurrent mutation in TMEM106B also causes hypomyelinating leukodystrophy in China and is a CpG hotspot. <i>Brain</i> , 2018 , 141, e37	11.2	
49	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	11	93
48	Mutations in SZT2 result in early-onset epileptic encephalopathy and leukoencephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1443-1448	2.5	9
47	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. <i>American Journal of Human Genetics</i> , 2018 , 102, 676-684	11	38

46	Recessive mutations in NDUFA2 cause mitochondrial leukoencephalopathy. <i>Clinical Genetics</i> , 2018 , 93, 396-400	4	8
45	Expression Pattern of the Aspartyl-tRNA Synthetase DARS in the Human Brain. <i>Frontiers in Molecular Neuroscience</i> , 2018 , 11, 81	6.1	11
44	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	4.2	16
43	Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogyrosis. <i>Journal of Child Neurology</i> , 2018 , 33, 642-650	2.5	8
42	Lysosomal dysfunction in hypomyelinating leukodystrophy. <i>Neurology: Genetics</i> , 2018 , 4, e288	3.8	8
41	Tmem2 Regulates Embryonic Vegf Signaling by Controlling Hyaluronic Acid Turnover. <i>Developmental Cell</i> , 2017 , 40, 123-136	10.2	33
40	Tmem2 Regulates Embryonic Vegf Signaling by Controlling Hyaluronic Acid Turnover. <i>Developmental Cell</i> , 2017 , 40, 421	10.2	7
39	Mural lymphatic endothelial cells regulate meningeal angiogenesis in the zebrafish. <i>Nature Neuroscience</i> , 2017 , 20, 774-783	25.5	62
38	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. <i>Brain</i> , 2017 , 140, 2550-2556	11.2	15
37	TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. <i>Human Molecular Genetics</i> , 2017 , 26, 4506-4518	5.6	32
36	Chronic non-freezing cold injury results in neuropathic pain due to a sensory neuropathy. <i>Brain</i> , 2017 , 140, 2557-2569	11.2	42
35	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. <i>Neurogenetics</i> , 2017 , 18, 185-194	3	28
34	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss-Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. <i>Pediatric Neurology</i> , 2017 , 66, 59-62	2.9	10
33	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. <i>Brain</i> , 2017 , 140, 3105-3111	11.2	42
32	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. <i>PLoS ONE</i> , 2017 , 12, e0178125	3.7	13
31	Utilising polymorphisms to achieve allele-specific genome editing in zebrafish. <i>Biology Open</i> , 2017 , 6, 125-131	2.2	12
30	Expanding the genotypic spectrum of CCBE1 mutations in Hennekam syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2694-7	2.5	6
29	Magnetic resonance imaging spectrum of succinate dehydrogenase-related infantile leukoencephalopathy. <i>Annals of Neurology</i> , 2016 , 79, 379-86	9.4	26

28	Whole exome sequencing in patients with white matter abnormalities. <i>Annals of Neurology</i> , 2016 , 79, 1031-1037	9.4	86
27	The clinical, biochemical and genetic features associated with -related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016 , 53, 768-775	5.8	27
26	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephalopathies. <i>Molecular Genetics and Metabolism</i> , 2015 , 114, 501-515	3.7	126
25	Loss-of-function alanyl-tRNA synthetase mutations cause an autosomal-recessive early-onset epileptic encephalopathy with persistent myelination defect. <i>American Journal of Human Genetics</i> , 2015 , 96, 675-81	11	72
24	Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. <i>Nature Communications</i> , 2015 , 6, 7623	17.4	92
23	mafba is a downstream transcriptional effector of Vegfc signaling essential for embryonic lymphangiogenesis in zebrafish. <i>Genes and Development</i> , 2015 , 29, 1618-30	12.6	42
22	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. <i>Nature Genetics</i> , 2015 , 47, 73-7	36.3	91
21	A protocol for the identification and validation of novel genetic causes of kidney disease. <i>BMC Nephrology</i> , 2015 , 16, 152	2.7	6
20	DARS-associated leukoencephalopathy can mimic a steroid-responsive neuroinflammatory disorder. <i>Neurology</i> , 2015 , 84, 226-30	6.5	48
19	TUBB4A de novo mutations cause isolated hypomyelination. <i>Neurology</i> , 2014 , 83, 898-902	6.5	41
18	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. <i>Brain</i> , 2014 , 137, 1921-30	11.2	72
17	Identification of a novel de novo p.Phe932Ile KCNT1 mutation in a patient with leukoencephalopathy and severe epilepsy. <i>Pediatric Neurology</i> , 2014 , 50, 112-4	2.9	49
16	Exome sequencing in developmental eye disease leads to identification of causal variants in GJA8, CRYGC, PAX6 and CYP1B1. <i>European Journal of Human Genetics</i> , 2014 , 22, 907-15	5.3	52
15	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	3.7	5
14	Whole exome sequencing in family trios reveals de novo mutations in PURA as a cause of severe neurodevelopmental delay and learning disability. <i>Journal of Medical Genetics</i> , 2014 , 51, 806-13	5.8	55
13	Mutations in DARS cause hypomyelination with brain stem and spinal cord involvement and leg spasticity. <i>American Journal of Human Genetics</i> , 2013 , 92, 774-80	11	127
12	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, 767-73	11	133
11	Expression of distinct RNAs from 3' untranslated regions. <i>Nucleic Acids Research</i> , 2011 , 39, 2393-403	20.1	153

10	Nuclear-localized tiny RNAs are associated with transcription initiation and splice sites in metazoans. <i>Nature Structural and Molecular Biology</i> , 2010 , 17, 1030-4	17.6	134
9	Evolution, biogenesis and function of promoter-associated RNAs. <i>Cell Cycle</i> , 2009 , 8, 2332-8	4.7	83
8	Tiny RNAs associated with transcription start sites in animals. <i>Nature Genetics</i> , 2009 , 41, 572-8	36.3	302
7	The transcriptional network that controls growth arrest and differentiation in a human myeloid leukemia cell line. <i>Nature Genetics</i> , 2009 , 41, 553-62	36.3	356
6	Long noncoding RNAs in mouse embryonic stem cell pluripotency and differentiation. <i>Genome Research</i> , 2008 , 18, 1433-45	9.7	608
5	Orthologous microRNA genes are located in cancer-associated genomic regions in human and mouse. <i>PLoS ONE</i> , 2007 , 2, e1133	3.7	32
4	Maintenance of transposon-free regions throughout vertebrate evolution. <i>BMC Genomics</i> , 2007 , 8, 470	4.5	23
3	Effect of 5ΨTR introns on gene expression in <i>Arabidopsis thaliana</i> . <i>BMC Genomics</i> , 2006 , 7, 120	4.5	136
2	Transposon-free regions in mammalian genomes. <i>Genome Research</i> , 2006 , 16, 164-72	9.7	89
1	Gene sequences from New Zealand's extinct huia. <i>Journal of the Royal Society of New Zealand</i> , 2002 , 32, 327-335	2	10