

# Paul N Valdmanis

## List of Publications by Year in descending order

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Version: 2024-02-01

47  
papers

6,124  
citations

201575

27  
h-index

214721

47  
g-index

49  
all docs

49  
docs citations

49  
times ranked

7644  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the <i>FUS/TLS</i> Gene on Chromosome 16 Cause Familial Amyotrophic Lateral Sclerosis. <i>Science</i> , 2009, 323, 1205-1208.	6.0	2,302
2	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2008, 40, 572-574.	9.4	1,371
3	The Loop Position of shRNAs and Pre-miRNAs Is Critical for the Accuracy of Dicer Processing In Vivo. <i>Cell</i> , 2012, 151, 900-911.	13.5	266
4	Promoterless gene targeting without nucleases ameliorates haemophilia B in mice. <i>Nature</i> , 2015, 517, 360-364.	13.7	226
5	Genetics of familial amyotrophic lateral sclerosis. <i>Neurology</i> , 2008, 70, 144-152.	1.5	189
6	Mutations in the KIAA0196 Gene at the SPG8 Locus Cause Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2007, 80, 152-161.	2.6	168
7	Contribution of TARDBP mutations to sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2008, 46, 112-114.	1.5	162
8	Oxidized/misfolded superoxide dismutase-1: the cause of all amyotrophic lateral sclerosis?. <i>Annals of Neurology</i> , 2007, 62, 553-559.	2.8	137
9	From animal models to human disease: a genetic approach for personalized medicine in ALS. <i>Acta Neuropathologica Communications</i> , 2016, 4, 70.	2.4	115
10	Three Families With Amyotrophic Lateral Sclerosis and Frontotemporal Dementia With Evidence of Linkage to Chromosome 9p. <i>Archives of Neurology</i> , 2007, 64, 240.	4.9	111
11	Recent advances in the genetics of amyotrophic lateral sclerosis. <i>Current Neurology and Neuroscience Reports</i> , 2009, 9, 198-205.	2.0	103
12	Investigating the contribution of VAPB/ALS8 loss of function in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2013, 22, 2350-2360.	1.4	75
13	Integrative gene-tissue microarray-based approach for identification of human disease biomarkers: application to amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2010, 19, 3233-3253.	1.4	58
14	Four familial ALS pedigrees discordant for two SOD1 mutations: are all SOD1 mutations pathogenic?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 572-577.	0.9	57
15	Resequencing of 29 Candidate Genes in Patients With Familial and Sporadic Amyotrophic Lateral Sclerosis. <i>Archives of Neurology</i> , 2011, 68, 587-93.	4.9	52
16	A tRNA-Derived Small RNA Regulates Ribosomal Protein S28 Protein Levels after Translation Initiation in Humans and Mice. <i>Cell Reports</i> , 2019, 29, 3816-3824.e4.	2.9	52
17	miR-122 removal in the liver activates imprinted microRNAs and enables more effective microRNA-mediated gene repression. <i>Nature Communications</i> , 2018, 9, 5321.	5.8	48
18	Upregulation of the microRNA cluster at the Dlk1-Dio3 locus in lung adenocarcinoma. <i>Oncogene</i> , 2015, 34, 94-103.	2.6	46

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19	Future of rAAV Gene Therapy: Platform for RNAi, Gene Editing, and Beyond. <i>Human Gene Therapy</i> , 2017, 28, 361-372.	1.4	40
20	Evolution of a Human-Specific Tandem Repeat Associated with ALS. <i>American Journal of Human Genetics</i> , 2020, 107, 445-460.	2.6	39
21	Expression determinants of mammalian argonaute proteins in mediating gene silencing. <i>Nucleic Acids Research</i> , 2012, 40, 3704-3713.	6.5	35
22	rAAV-Mediated Tumorigenesis: Still Unresolved After an AAV Assault. <i>Molecular Therapy</i> , 2012, 20, 2014-2017.	3.7	33
23	A Mutation that Creates a Pseudoexon in <i>SOD1</i> Causes Familial ALS. <i>Annals of Human Genetics</i> , 2009, 73, 652-657.	0.3	32
24	A mutation in the RNF170 gene causes autosomal dominant sensory ataxia. <i>Brain</i> , 2011, 134, 602-607.	3.7	32
25	RNA interference-induced hepatotoxicity results from loss of the first synthesized isoform of microRNA-122 in mice. <i>Nature Medicine</i> , 2016, 22, 557-562.	15.2	32
26	50bp deletion in the promoter for superoxide dismutase 1 (SOD1) reduces SOD1 expression in vitro and may correlate with increased age of onset of sporadic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 229-237.	2.3	29
27	Alternative splicing in a presenilin 2 variant associated with Alzheimer disease. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 762-777.	1.7	29
28	Autosomal dominant primary lateral sclerosis. <i>Neurology</i> , 2007, 68, 1156-1157.	1.5	28
29	The Expanding Repertoire of Circular RNAs. <i>Molecular Therapy</i> , 2013, 21, 1112-1114.	3.7	28
30	Regulation of microRNA-mediated gene silencing by microRNA precursors. <i>Nature Structural and Molecular Biology</i> , 2014, 21, 825-832.	3.6	23
31	The proportion of mutations predicted to have a deleterious effect differs between gain and loss of function genes in neurodegenerative disease. <i>Human Mutation</i> , 2009, 30, E481-E489.	1.1	20
32	Tracking Adeno-Associated Virus Capsid Evolution by High-Throughput Sequencing. <i>Human Gene Therapy</i> , 2020, 31, 553-564.	1.4	19
33	A novel TYMP mutation in a French Canadian patient with mitochondrial neurogastrointestinal encephalomyopathy. <i>Clinical Neurology and Neurosurgery</i> , 2009, 111, 691-694.	0.6	18
34	Analysis of <i>DPP6</i> and <i>FGGY</i> as candidate genes for amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 389-391.	2.3	18
35	Characterizing nucleotide variation and expansion dynamics in human-specific variable number tandem repeats. <i>Genome Research</i> , 2021, 31, 1313-1324.	2.4	15
36	Progress in Amyotrophic Lateral Sclerosis Gene Discovery. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	15

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37	Human Induced Pluripotent Stem Cell-Derived TDP-43 Mutant Neurons Exhibit Consistent Functional Phenotypes Across Multiple Gene Edited Lines Despite Transcriptomic and Splicing Discrepancies. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 728707.	1.8	13
38	Autosomal dominant sensory ataxia: a neuroaxonal dystrophy. <i>Acta Neuropathologica</i> , 2008, 116, 331-336.	3.9	12
39	A Novel Mutation in a Large French-Canadian Family with LGMD1B. <i>Canadian Journal of Neurological Sciences</i> , 2008, 35, 331-334.	0.3	12
40	A Locus for Primary Lateral Sclerosis on Chromosome 4ptel-4p16.1. <i>Archives of Neurology</i> , 2008, 65, 383-6.	4.9	12
41	No TARDBP Mutations in a French Canadian Population of Patients With Parkinson Disease. <i>Archives of Neurology</i> , 2009, 66, 281-2.	4.9	12
42	Prophylactic <i>In Vivo</i> Hematopoietic Stem Cell Gene Therapy with an Immune Checkpoint Inhibitor Reverses Tumor Growth in Syngeneic Mouse Tumor Models. <i>Cancer Research</i> , 2020, 80, 549-560.	0.4	12
43	ALS predisposition modifiers: Knock NOX, who's there? SOD1 mice still are. <i>European Journal of Human Genetics</i> , 2008, 16, 140-142.	1.4	11
44	Expression of an alternatively spliced variant of SORL1 in neuronal dendrites is decreased in patients with Alzheimer's disease. <i>Acta Neuropathologica Communications</i> , 2021, 9, 43.	2.4	7
45	Endogenous MicroRNA Competition as a Mechanism of shRNA-Induced Cardiotoxicity. <i>Molecular Therapy - Nucleic Acids</i> , 2020, 19, 572-580.	2.3	4
46	Amyotrophic Lateral Sclerosis: Genome-wide association studies in amyotrophic lateral sclerosis. <i>European Journal of Human Genetics</i> , 2009, 17, 137-138.	1.4	3
47	A Complete Pipeline for Isolating and Sequencing MicroRNAs, and Analyzing Them Using Open Source Tools. <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	2