

Jan Senderek

List of Publications by Year in descending order

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101
papers

8,318
citations

50276

46
h-index

46799

89
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107
all docs

107
docs citations

107
times ranked

9961
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. <i>Nature Genetics</i> , 2004, 36, 449-451.	21.4	1,391
2	PKHD1, the Polycystic Kidney and Hepatic Disease 1 Gene, Encodes a Novel Large Protein Containing Multiple Immunoglobulin-Like Plexin-Transcription Factor Domains and Parallel Beta-Helix 1 Repeats. <i>American Journal of Human Genetics</i> , 2002, 70, 1305-1317.	6.2	445
3	Loss of Nephrocystin-3 Function Can Cause Embryonic Lethality, Meckel-Gruber-like Syndrome, Situs Inversus, and Renal-Hepatic-Pancreatic Dysplasia. <i>American Journal of Human Genetics</i> , 2008, 82, 959-970.	6.2	294
4	Clinical consequences of PKHD1 mutations in 164 patients with autosomal-recessive polycystic kidney disease (ARPKD). <i>Kidney International</i> , 2005, 67, 829-848.	5.2	277
5	Mutations in SIL1 cause Marinesco-Sjogren syndrome, a cerebellar ataxia with cataract and myopathy. <i>Nature Genetics</i> , 2005, 37, 1312-1314.	21.4	232
6	Mutation of the SBF2 gene, encoding a novel member of the myotubularin family, in Charcot-Marie-Tooth neuropathy type 4B/11p15. <i>Human Molecular Genetics</i> , 2003, 12, 349-356.	2.9	230
7	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. <i>Nature Genetics</i> , 2010, 42, 160-164.	21.4	228
8	Spectrum of Mutations in the Gene for Autosomal Recessive Polycystic Kidney Disease (ARPKD/PKHD1). <i>Journal of the American Society of Nephrology: JASN</i> , 2003, 14, 76-89.	6.1	226
9	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. <i>Nature Genetics</i> , 2012, 44, 704-708.	21.4	216
10	Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. <i>Nature Genetics</i> , 2009, 41, 1179-1181.	21.4	205
11	Mutations in a Gene Encoding a Novel SH3/TPR Domain Protein Cause Autosomal Recessive Charcot-Marie-Tooth Type 4C Neuropathy. <i>American Journal of Human Genetics</i> , 2003, 73, 1106-1119.	6.2	185
12	Autosomal-Dominant Distal Myopathy Associated with a Recurrent Missense Mutation in the Gene Encoding the Nuclear Matrix Protein, Matrin 3. <i>American Journal of Human Genetics</i> , 2009, 84, 511-518.	6.2	161
13	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. <i>American Journal of Human Genetics</i> , 2011, 88, 162-172.	6.2	153
14	Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS): expanding the genetic, clinical and imaging spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 41.	2.7	147
15	PKHD1 mutations in autosomal recessive polycystic kidney disease (ARPKD). <i>Human Mutation</i> , 2004, 23, 453-463.	2.5	145
16	Transcriptional regulator PRDM12 is essential for human pain perception. <i>Nature Genetics</i> , 2015, 47, 803-808.	21.4	137
17	Mutations of the <i>CEP290</i> gene encoding a centrosomal protein cause Meckel-Gruber syndrome. <i>Human Mutation</i> , 2008, 29, 45-52.	2.5	131
18	Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. <i>American Journal of Human Genetics</i> , 2007, 81, 158-164.	6.2	128

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19	Mutations in the ganglioside-induced differentiation-associated protein-1 (GDAP1) gene in intermediate type autosomal recessive Charcot-Marie-Tooth neuropathy. <i>Brain</i> , 2003, 126, 642-649.	7.6	115
20	Milder Presentation of Recessive Polycystic Kidney Disease Requires Presence of Amino Acid Substitution Mutations. <i>Journal of the American Society of Nephrology: JASN</i> , 2003, 14, 2004-2014.	6.1	113
21	Transient, Recurrent, White Matter Lesions in X-linked Charcot-Marie-Tooth Disease With Novel Connexin 32 Mutation. <i>Archives of Neurology</i> , 2003, 60, 605.	4.5	111
22	Oligophrenin 1 (OPHN1) gene mutation causes syndromic X-linked mental retardation with epilepsy, rostral ventricular enlargement and cerebellar hypoplasia. <i>Brain</i> , 2003, 126, 1537-1544.	7.6	106
23	Identification and Characterization of Pkhd1, the Mouse Orthologue of the Human ARPKD Gene. <i>Journal of the American Society of Nephrology: JASN</i> , 2002, 13, 2246-2258.	6.1	104
24	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. <i>American Journal of Human Genetics</i> , 2016, 99, 735-743.	6.2	99
25	SH3TC2/KIAA1985 protein is required for proper myelination and the integrity of the node of Ranvier in the peripheral nervous system. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 17528-17533.	7.1	97
26	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. <i>Brain</i> , 2015, 138, 845-861.	7.6	94
27	Loss of tubulin deglutamylase <sc>CCP</sc> 1 causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	7.8	86
28	X-linked dominant Charcot-Marie-Tooth neuropathy: clinical, electrophysiological, and morphological phenotype in four families with different connexin32 mutations. <i>Journal of the Neurological Sciences</i> , 1999, 167, 90-101.	0.6	85
29	A mouse model for cystic biliary dysgenesis in autosomal recessive polycystic kidney disease (ARPKD). <i>Hepatology</i> , 2005, 41, 1113-1121.	7.3	84
30	Pontocerebellar hypoplasia type 1. <i>Neurology</i> , 2013, 80, 438-446.	1.1	84
31	SH3TC2, a protein mutant in Charcot-Marie-Tooth neuropathy, links peripheral nerve myelination to endosomal recycling. <i>Brain</i> , 2010, 133, 2462-2474.	7.6	82
32	Charcot-Marie-Tooth Neuropathy Type 2 and P0 Point Mutations: Two Novel Amino Acid Substitutions (Asp61Gly; Tyr119Cys) and a Possible "Hotspot" on Thr124Met. <i>Brain Pathology</i> , 2000, 10, 235-248.	4.1	74
33	PKHD1 mutations in families requesting prenatal diagnosis for autosomal recessive polycystic kidney disease (ARPKD). <i>Human Mutation</i> , 2004, 23, 487-495.	2.5	74
34	Phenotype of matrin-related distal myopathy in 16 German patients. <i>Annals of Neurology</i> , 2014, 76, 669-680.	5.3	74
35	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. <i>Journal of Neurology</i> , 2012, 259, 838-850.	3.6	72
36	Loss of function mutations in <i>HARS</i> cause a spectrum of inherited peripheral neuropathies. <i>Brain</i> , 2015, 138, 2161-2172.	7.6	71

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37	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 75-90.	2.6	69
38	Mutations in INPP5K , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. <i>American Journal of Human Genetics</i> , 2017, 100, 523-536.	6.2	67
39	Algorithm for efficientPKHD1mutation screening in autosomal recessive polycystic kidney disease (ARPKD). <i>Human Mutation</i> , 2005, 25, 225-231.	2.5	65
40	SIL1 mutations and clinical spectrum in patients with Marinesco-Sjögren syndrome. <i>Brain</i> , 2013, 136, 3634-3644.	7.6	65
41	Fibulin-5 mutations link inherited neuropathies, age-related macular degeneration and hyperelastic skin. <i>Brain</i> , 2011, 134, 1839-1852.	7.6	64
42	Myelin is dependent on the Charcot-Marie-Tooth Type 4H disease culprit protein FRABIN/FGD4 in Schwann cells. <i>Brain</i> , 2012, 135, 3567-3583.	7.6	63
43	Vascular changes in the periosteum of congenital pseudarthrosis of the tibia. <i>Pathology Research and Practice</i> , 2005, 201, 305-312.	2.3	56
44	Autosomal recessive postlingual hearing loss (DFNB8): compound heterozygosity for two novel TMPRSS3 mutations in German siblings. <i>Journal of Medical Genetics</i> , 2007, 44, e81-e81.	3.2	53
45	Myopathy in Marinesco-Sjögren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. <i>Acta Neuropathologica</i> , 2014, 127, 761-777.	7.7	51
46	Whole-exome sequencing in patients with inherited neuropathies: outcome and challenges. <i>Journal of Neurology</i> , 2014, 261, 970-982.	3.6	50
47	Towards a functional pathology of hereditary neuropathies. <i>Acta Neuropathologica</i> , 2017, 133, 493-515.	7.7	48
48	50 years to diagnosis: Autosomal dominant tubular aggregate myopathy caused by a novel STIM1 mutation. <i>Neuromuscular Disorders</i> , 2015, 25, 577-584.	0.6	47
49	Rare Variants in MME, Encoding Metalloprotease Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. <i>American Journal of Human Genetics</i> , 2016, 99, 607-623.	6.2	47
50	Identification of mutations in the <i>MYO9A</i> gene in patients with congenital myasthenic syndrome. <i>Brain</i> , 2016, 139, 2143-2153.	7.6	45
51	<i>HSJ1</i> -related hereditary neuropathies. <i>Neurology</i> , 2014, 83, 1726-1732.	1.1	42
52	Autosomal recessive polycystic kidney disease (ARPKD). <i>Journal of Nephrology</i> , 2003, 16, 453-8.	2.0	41
53	PRDM12 Is Required for Initiation of the Nociceptive Neuron Lineage during Neurogenesis. <i>Cell Reports</i> , 2019, 26, 3484-3492.e4.	6.4	40
54	RSPO4 Is the Major Gene in Autosomal-Recessive Anonychia and Mutations Cluster in the Furin-Like Cysteine-Rich Domains of the Wnt Signaling Ligand R-spondin 4. <i>Journal of Investigative Dermatology</i> , 2008, 128, 791-796.	0.7	39

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55	Sh3tc2 deficiency affects neuregulin/ErbB signaling. <i>Glia</i> , 2013, 61, 1041-1051.	4.9	39
56	Founder SH3TC2 mutations are responsible for a CMT4C French-Canadians cluster. <i>Neuromuscular Disorders</i> , 2008, 18, 483-492.	0.6	35
57	Aberrant splicing is a common mutational mechanism inMKS1, a key player in Meckel-Gruber syndrome. <i>Human Mutation</i> , 2007, 28, 638-639.	2.5	34
58	Mutations of the LMNA gene can mimic autosomal dominant proximal spinal muscular atrophy. <i>Neurogenetics</i> , 2007, 8, 137-142.	1.4	33
59	A 3'-UTR mutation creates a microRNA target site in the GFPT1 gene of patients with congenital myasthenic syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 3418-3426.	2.9	32
60	Overlap between VACTERL and hemifacial microsomia illustrating a spectrum of malformations seen in axial mesodermal dysplasia complex (AMDC). <i>American Journal of Medical Genetics, Part A</i> , 2003, 121A, 151-155.	1.2	31
61	PLEKHG5 deficiency leads to an intermediate form of autosomal-recessive Charcot-Marie-Tooth disease. <i>Human Molecular Genetics</i> , 2013, 22, 4224-4232.	2.9	31
62	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. <i>Brain</i> , 2019, 142, 1547-1560.	7.6	30
63	SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. <i>Human Genetics</i> , 2018, 137, 911-919.	3.8	29
64	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	12.8	28
65	A novel nonsense mutation in the ABC1 gene causes a severe syringomyelia-like phenotype of Tangier disease. <i>Brain</i> , 2003, 126, 920-927.	7.6	26
66	Recessive variants of <i>MuSK</i> are associated with late onset CMS and predominant limb girdle weakness. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1594-1601.	1.2	25
67	Pontocerebellar hypoplasia with spinal muscular atrophy (PCH1): identification of SLC25A46 mutations in the original Dutch PCH1 family. <i>Brain</i> , 2017, 140, e46-e46.	7.6	23
68	Functional analysis of PKHD1 splicing in autosomal recessive polycystic kidney disease. <i>Journal of Human Genetics</i> , 2006, 51, 788-793.	2.3	22
69	Recessive truncating <i>IGHMBP2</i> mutations presenting as axonal sensorimotor neuropathy. <i>Neurology</i> , 2015, 84, 523-531.	1.1	22
70	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. <i>Brain</i> , 2021, 144, 1422-1434.	7.6	22
71	Facioscapulohumeral muscular dystrophy and Charcot-Marie-Tooth neuropathy 1A - evidence for "double trouble" overlapping syndromes. <i>BMC Medical Genetics</i> , 2013, 14, 92.	2.1	21
72	Inverted formin 2-related Charcot-Marie-Tooth disease: extension of the mutational spectrum and pathological findings in Schwann cells and axons. <i>Journal of the Peripheral Nervous System</i> , 2015, 20, 52-59.	3.1	21

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73	Clinical and morphological variability of the E396K mutation in the neurofilament light chain gene in patients with Charcot-Marie-Tooth disease type 2E. , 2014, 33, 335-343.		20
74	Becker muscular dystrophy combined with x-linked Charcot-Marie-Tooth neuropathy. , 2000, 23, 818-823.		19
75	SNP array-based whole genome homozygosity mapping as the first step to a molecular diagnosis in patients with Charcot-Marie-Tooth disease. Journal of Neurology, 2012, 259, 515-523.	3.6	19
76	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.1	19
77	Phenotypic variation of a novel nonsense mutation in the P0 intracellular domain. Journal of the Neurological Sciences, 2001, 192, 49-51.	0.6	18
78	Genetic pain loss disorders. Nature Reviews Disease Primers, 2022, 8, .	30.5	18
79	MPV17 mutations in juvenile and adult onset axonal sensorimotor polyneuropathy. Clinical Genetics, 2019, 95, 182-186.	2.0	16
80	In-depth phenotyping of lymphoblastoid cells suggests selective cellular vulnerability in Marinesco-Sjögren syndrome. Oncotarget, 2017, 8, 68493-68516.	1.8	16
81	Facioaudiosymphalangism syndrome and growth acceleration associated with a heterozygous <i>NOG</i> mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 1540-1544.	1.2	15
82	Congenital myasthenic syndrome caused by novel COL13A1 mutations. Journal of Neurology, 2019, 266, 1107-1112.	3.6	14
83	Association of A Novel Splice Site Mutation in P/Q-Type Calcium Channels with Childhood Epilepsy and Late-Onset Slowly Progressive Non-Episodic Cerebellar Ataxia. International Journal of Molecular Sciences, 2020, 21, 3810.	4.1	14
84	Diagnosis, Pathogenesis, and Treatment Prospects in Cystic Kidney Disease. Molecular Diagnosis and Therapy, 2006, 10, 163-174.	3.8	13
85	Behr syndrome with homozygous C19ORF12 mutation. Journal of the Neurological Sciences, 2015, 357, 115-118.	0.6	12
86	Hereditary Neuropathies: Update 2017. Neuropediatrics, 2017, 48, 282-293.	0.6	11
87	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.6	11
88	Small Rho GTPases are key regulators of peripheral nerve biology in health and disease. Journal of the Peripheral Nervous System, 2008, 13, 188-199.	3.1	10
89	Warburg micro syndrome type 1 associated with peripheral neuropathy and cardiomyopathy. Folia Neuropathologica, 2016, 3, 273-281.	1.2	10
90	A 10.7 Mb interstitial deletion of 13q21 without phenotypic effect defines a further non-pathogenic euchromatic variant. American Journal of Medical Genetics, Part A, 2008, 146A, 2417-2420.	1.2	9

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91	Charcot-Marie-Tooth disease and hereditary motor neuropathies – Update 2020. <i>Medizinische Genetik</i> , 2020, 32, 207-219.	0.2	9
92	A point mutation in the human connexin32 promoter P2 does not correlate with X-linked dominant Charcot-Marie-Tooth neuropathy in Germany. <i>Molecular Brain Research</i> , 2001, 88, 183-185.	2.3	8
93	Myotilin is not the Causative Gene for Vocal Cord and Pharyngeal Weakness with Distal Myopathy (VCPDM). <i>Annals of Human Genetics</i> , 2008, 70, 414-416.	0.8	6
94	The Caveolin-3 G56S sequence variant of unknown significance: Muscle biopsy findings and functional cell biological analysis. <i>Proteomics - Clinical Applications</i> , 2017, 11, 1600007.	1.6	6
95	Cloning, expression and characterization of the murine orthologue of SBF2, the gene mutated in Charcot-Marie-Tooth disease type 4B2. <i>Gene Expression Patterns</i> , 2006, 6, 978-984.	0.8	3
96	Light Microscopic, Immunohistochemical, and Ultrastructural Findings in Congenital Fibular Aplasia or Hypoplasia (FAH). <i>Pediatric and Developmental Pathology</i> , 2005, 8, 474-482.	1.0	2
97	Introduction to the hereditary neuropathies. , 2014, , 59-61.		2
98	SIL1-negative Marinesco-Sjögren syndrome: First report of two sibs from India. <i>Journal of Pediatric Neurosciences</i> , 2014, 9, 291.	0.3	2
99	Search for cryptic subtelomeric aberrations in patients with non-classical Marinesco-Sjögren phenotype. <i>Journal of Pediatric Neurology</i> , 2015, 10, 167-172.	0.2	1
100	PLEKHG5: Merging phenotypes and disease mechanisms in Charcot-Marie-Tooth neuropathy and lower motor neuron disease. <i>European Journal of Neurology</i> , 2021, 28, 1106-1107.	3.3	0
101	A polymorphic AT-repeat causes frequent allele dropout for an <i>MME</i> mutational hotspot exon. <i>Journal of Medical Genetics</i> , 2022, , jmedgenet-2021-108281.	3.2	0