Jan Senderek

List of Publications by Year
in descending order

101 papers	8,318 citations	50276 46 h-index	46799 89 g-index
107 all docs	107 docs citations	107 times ranked	9961 citing authors

#	Article	IF	Citations
1	A polymorphic AT-repeat causes frequent allele dropout for an MME mutational hotspot exon. Journal of Medical Genetics, 2022, , jmedgenet-2021-108281.	3.2	0
2	Genetic pain loss disorders. Nature Reviews Disease Primers, 2022, 8, .	30.5	18
3	PLEKHG5: Merging phenotypes and disease mechanisms in Charcotâ€Marieâ€Tooth neuropathy and lower motor neuron disease. European Journal of Neurology, 2021, 28, 1106-1107.	3.3	0
4	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
5	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
6	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.1	19
7	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
8	Charcot-Marie-Tooth disease and hereditary motor neuropathies â€“ Update 2020. Medizinische Genetik, 2020, 32, 207-219.	0.2	9
9	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. Brain, 2019, 142, 1547-1560.	7.6	30
10	PRDM12 Is Required for Initiation of the Nociceptive Neuron Lineage during Neurogenesis. Cell Reports, 2019, 26, 3484-3492.e4.	6.4	40
11	Congenital myasthenic syndrome caused by novel COL13A1 mutations. Journal of Neurology, 2019, 266, 1107-1112.	3.6	14
12	MPV17 mutations in juvenileâ€•and adultâ€•onset axonal sensorimotor polyneuropathy. Clinical Genetics, 2019, 95, 182-186.	2.0	16
13	Recessive variants of MuSK are associated with late onset CMS and predominant limb girdle weakness. American Journal of Medical Genetics, Part A, 2018, 176, 1594-1601.	1.2	25
14	SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. Human Genetics, 2018, 137, 911-919.	3.8	29
15	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€•onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
16	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.6	11
17	Mutations in INPP5K , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. American Journal of Human Genetics, 2017, 100, 523-536.	6.2	67
18	Hereditary Neuropathies: Update 2017. Neuropediatrics, 2017, 48, 282-293.	0.6	11

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19	Pontocerebellar hypoplasia with spinal muscular atrophy (PCH1): identification of SLC25A46 mutations in the original Dutch PCH1 family. Brain, 2017, 140, e46-e46.	7.6	23
20	The Caveolin-3 G56S sequence variant of unknown significance: Muscle biopsy findings and functional cell biological analysis. Proteomics - Clinical Applications, 2017, 11, 1600007.	1.6	6
21	Towards a functional pathology of hereditary neuropathies. Acta Neuropathologica, 2017, 133, 493-515.	7.7	48
22	In-depth phenotyping of lymphoblastoid cells suggests selective cellular vulnerability in Marinesco-SjÃ¶gren syndrome. Oncotarget, 2017, 8, 68493-68516.	1.8	16
23	Warburg micro syndrome type 1 associated with peripheral neuropathy and cardiomyopathy. Folia Neuropathologica, 2016, 3, 273-281.	1.2	10
24	Rare Variants in MME, Encoding Metalloprotease Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. American Journal of Human Genetics, 2016, 99, 607-623.	6.2	47
25	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
26	Identification of mutations in theMYO9Agene in patients with congenital myasthenic syndrome. Brain, 2016, 139, 2143-2153.	7.6	45
27	Inverted formin 2â€related Charcotâ€Marieâ€Tooth disease: extension of the mutational spectrum and pathological findings in Schwann cells and axons. Journal of the Peripheral Nervous System, 2015, 20, 52-59.	3.1	21
28	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	21.4	137
29	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. Neurology, 2015, 84, 523-531.	1.1	22
30	50 years to diagnosis: Autosomal dominant tubular aggregate myopathy caused by a novel STIM1 mutation. Neuromuscular Disorders, 2015, 25, 577-584.	0.6	47
31	Behr syndrome with homozygous C19ORF12 mutation. Journal of the Neurological Sciences, 2015, 357, 115-118.	0.6	12
32	Loss of function mutations inHARScause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	7.6	71
33	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. Brain, 2015, 138, 845-861.	7.6	94
34	A 3'-UTR mutation creates a microRNA target site in the GFPT1 gene of patients with congenital myasthenic syndrome. Human Molecular Genetics, 2015, 24, 3418-3426.	2.9	32
35	Search for cryptic subtelomeric aberrations in patients with non-classical Marinesco-SjÃ¶gren phenotype. Journal of Pediatric Neurology, 2015, 10, 167-172.	0.2	1
36	Phenotype of matrinâ€3â€“related distal myopathy in 16 <scp>G</scp>erman patients. Annals of Neurology, 2014, 76, 669-680.	5.3	74

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37	Introduction to the hereditary neuropathies. , 2014, , 59-61.		2
38	Myopathy in Marinescoâ€“SjÃ¶gren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. Acta Neuropathologica, 2014, 127, 761-777.	7.7	51
39	Whole-exome sequencing in patients with inherited neuropathies: outcome and challenges. Journal of Neurology, 2014, 261, 970-982.	3.6	50
40	HSJ1 -related hereditary neuropathies. Neurology, 2014, 83, 1726-1732.	1.1	42
41	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
42	SIL1-negative Marinesco-SjÃ¶gren syndrome: First report of two sibs from India. Journal of Pediatric Neurosciences, 2014, 9, 291.	0.3	2
43	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
44	Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS): expanding the genetic, clinical and imaging spectrum. Orphanet Journal of Rare Diseases, 2013, 8, 41.	2.7	147
45	Facioscapulohumeral muscular dystrophy and Charcot-Marie-Tooth neuropathy 1A - evidence for â€œdouble troubleâ€•overlapping syndromes. BMC Medical Genetics, 2013, 14, 92.	2.1	21
46	SIL1 mutations and clinical spectrum in patients with Marinesco-SjÃ¶gren syndrome. Brain, 2013, 136, 3634-3644.	7.6	65
47	PLEKHG5 deficiency leads to an intermediate form of autosomal-recessive Charcotâ€“Marieâ€“Tooth disease. Human Molecular Genetics, 2013, 22, 4224-4232.	2.9	31
48	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
49	Sh3tc2 deficiency affects neuregulinâ€1/ErbB signaling. Glia, 2013, 61, 1041-1051.	4.9	39
50	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature Genetics, 2012, 44, 704-708.	21.4	216
51	Myelin is dependent on the Charcotâ€“Marieâ€“Tooth Type 4H disease culprit protein FRABIN/FGD4 in Schwann cells. Brain, 2012, 135, 3567-3583.	7.6	63
52	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
53	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
54	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153

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55	Fibulin-5 mutations link inherited neuropathies, age-related macular degeneration and hyperelastic skin. Brain, 2011, 134, 1839-1852.	7.6	64
56	Facioaudiosymphalangism syndrome and growth acceleration associated with a heterozygous NOG mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 1540-1544.	1.2	15
57	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	21.4	228
58	SH3TC2, a protein mutant in Charcotâ€“Marieâ€“Tooth neuropathy, links peripheral nerve myelination to endosomal recycling. Brain, 2010, 133, 2462-2474.	7.6	82
59	SH3TC2/KIAA1985 protein is required for proper myelination and the integrity of the node of Ranvier in the peripheral nervous system. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 17528-17533.	7.1	97
60	Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. Nature Genetics, 2009, 41, 1179-1181.	21.4	205
61	Autosomal-Dominant Distal Myopathy Associated with a Recurrent Missense Mutation in the Gene Encoding the Nuclear Matrix Protein, Matrin 3. American Journal of Human Genetics, 2009, 84, 511-518.	6.2	161
62	Mutations of theCEP290gene encoding a centrosomal protein cause Meckel-Gruber syndrome. Human Mutation, 2008, 29, 45-52.	2.5	131
63	A 10.7 Mb interstitial deletion of 13q21 without phenotypic effect defines a further nonâ€pathogenic euchromatic variant. American Journal of Medical Genetics, Part A, 2008, 146A, 2417-2420.	1.2	9
64	Loss of Nephrocystin-3 Function Can Cause Embryonic Lethality,ÂMeckel-Gruber-like Syndrome, Situs Inversus, and Renal-Hepatic-Pancreatic Dysplasia. American Journal of Human Genetics, 2008, 82, 959-970.	6.2	294
65	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. Journal of Investigative Dermatology, 2008, 128, 791-796.	0.7	39
66	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
67	Myotilin is not the Causative Gene for Vocal Cord and Pharyngeal Weakness with Distal Myopathy (VCPDM). Annals of Human Genetics, 2008, 70, 414-416.	0.8	6
68	Founder SH3TC2 mutations are responsible for a CMT4C French-Canadians cluster. Neuromuscular Disorders, 2008, 18, 483-492.	0.6	35
69	Autosomal recessive postlingual hearing loss (DFNB8): compound heterozygosity for two novel TMPRSS3 mutations in German siblings. Journal of Medical Genetics, 2007, 44, e81-e81.	3.2	53
70	Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. American Journal of Human Genetics, 2007, 81, 158-164.	6.2	128
71	Aberrant splicing is a common mutational mechanism inMKS1, a key player in Meckel-Gruber syndrome. Human Mutation, 2007, 28, 638-639.	2.5	34
72	Mutations of the LMNA gene can mimic autosomal dominant proximal spinal muscular atrophy. Neurogenetics, 2007, 8, 137-142.	1.4	33

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73	Diagnosis, Pathogenesis, and Treatment Prospects in Cystic Kidney Disease. Molecular Diagnosis and Therapy, 2006, 10, 163-174.	3.8	13
74	Functional analysis of PKHD1 splicing in autosomal recessive polycystic kidney disease. Journal of Human Genetics, 2006, 51, 788-793.	2.3	22
75	Cloning, expression and characterization of the murine orthologue of SBF2, the gene mutated in Charcot-Marie-Tooth disease type 4B2. Gene Expression Patterns, 2006, 6, 978-984.	0.8	3
76	Clinical consequences of PKHD1 mutations in 164 patients with autosomal-recessive polycystic kidney disease (ARPKD). Kidney International, 2005, 67, 829-848.	5.2	277
77	Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. Nature Genetics, 2005, 37, 1312-1314.	21.4	232
78	A mouse model for cystic biliary dysgenesis in autosomal recessive polycystic kidney disease (ARPKD). Hepatology, 2005, 41, 1113-1121.	7.3	84
79	Algorithm for efficientPKHD1mutation screening in autosomal recessive polycystic kidney disease (ARPKD). Human Mutation, 2005, 25, 225-231.	2.5	65
80	Light Microscopic, Immunohistochemical, and Ultrastructural Findings in Congenital Fibular Aplasia or Hypoplasia (FAH). Pediatric and Developmental Pathology, 2005, 8, 474-482.	1.0	2
81	Vascular changes in the periosteum of congenital pseudarthrosis of the tibia. Pathology Research and Practice, 2005, 201, 305-312.	2.3	56
82	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451.	21.4	1,391
83	PKHD1mutations in families requesting prenatal diagnosis for autosomal recessive polycystic kidney disease (ARPKD). Human Mutation, 2004, 23, 487-495.	2.5	74
84	PKHD1mutations in autosomal recessive polycystic kidney disease (ARPKD). Human Mutation, 2004, 23, 453-463.	2.5	145
85	Overlap between VACTERL and hemifacial microsomia illustrating a spectrum of malformations seen in axial mesodermal dysplasia complex (AMDC). American Journal of Medical Genetics, Part A, 2003, 121A, 151-155.	1.2	31
86	Mutations in the ganglioside-induced differentiation-associated protein-1 (GDAP1) gene in intermediate type autosomal recessive Charcot-Marie-Tooth neuropathy. Brain, 2003, 126, 642-649.	7.6	115
87	Mutations in a Gene Encoding a Novel SH3/TPR Domain Protein Cause Autosomal Recessive Charcot-Marie-Tooth Type 4C Neuropathy. American Journal of Human Genetics, 2003, 73, 1106-1119.	6.2	185
88	Mutation of the SBF2 gene, encoding a novel member of the myotubularin family, in Charcot-Marie-Tooth neuropathy type 4B2/11p15. Human Molecular Genetics, 2003, 12, 349-356.	2.9	230
89	Oligophrenin 1 (OPHN1) gene mutation causes syndromic X-linked mental retardation with epilepsy, rostral ventricular enlargement and cerebellar hypoplasia. Brain, 2003, 126, 1537-1544.	7.6	106
90	Transient, Recurrent, White Matter Lesions in X-linked Charcot-Marie-Tooth Disease With Novel Connexin 32 Mutation. Archives of Neurology, 2003, 60, 605.	4.5	111

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91	Spectrum of Mutations in the Gene for Autosomal Recessive Polycystic Kidney Disease (ARPKD/PKHD1). Journal of the American Society of Nephrology: JASN, 2003, 14, 76-89.	6.1	226
92	Milder Presentation of Recessive Polycystic Kidney Disease Requires Presence of Amino Acid Substitution Mutations. Journal of the American Society of Nephrology: JASN, 2003, 14, 2004-2014.	6.1	113
93	A novel nonsense mutation in the ABC1 gene causes a severe syringomyelia-like phenotype of Tangier disease. Brain, 2003, 126, 920-927.	7.6	26
94	Autosomal recessive polycystic kidney disease (ARPKD). Journal of Nephrology, 2003, 16, 453-8.	2.0	41
95	Identification and Characterization of Pkhd1, the Mouse Orthologue of the Human ARPKD Gene. Journal of the American Society of Nephrology: JASN, 2002, 13, 2246-2258.	6.1	104
96	PKHD1, the Polycystic Kidney and Hepatic Disease 1 Gene, Encodes a Novel Large Protein Containing Multiple Immunoglobulin-Like Plexin-Transcriptionâ€“Factor Domains and Parallel Beta-Helix 1 Repeats. American Journal of Human Genetics, 2002, 70, 1305-1317.	6.2	445
97	A point mutation in the human connexin32 promoter P2 does not correlate with X-linked dominant Charcot-Marie-Tooth neuropathy in Germany. Molecular Brain Research, 2001, 88, 183-185.	2.3	8
98	Phenotypic variation of a novel nonsense mutation in the P0 intracellular domain. Journal of the Neurological Sciences, 2001, 192, 49-51.	0.6	18
99	Becker muscular dystrophy combined with x-linked Charcot-Marie-Tooth neuropathy. , 2000, 23, 818-823.		19
100	Charcotâ€Marieâ€Tooth Neuropathy Type 2 and P0 Point Mutations: Two Novel Amino Acid Substitutions (Asp61Gly; Tyr119Cys) and a Possible â€œHotspotâ€•on Thr124Met. Brain Pathology, 2000, 10, 235-248.	4.1	74
101	X-linked dominant Charcotâ€“Marieâ€“Tooth neuropathy: clinical, electrophysiological, and morphological phenotype in four families with different connexin32 mutations. Journal of the Neurological Sciences, 1999, 167, 90-101.	0.6	85