Susan J Hayflick

List of Publications by Year
in descending order

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53 papers	5,729 citations	126907 33 h-index	175258 52 g-index
53 all docs	53 docs citations	53 times ranked	6665 citing authors

#	Article	IF	Citations
1	A novel pantothenate kinase gene (PANK2) is defective in Hallervorden-Spatz syndrome. Nature Genetics, 2001, 28, 345-349.	21.4	721
2	Genetic, Clinical, and Radiographic Delineation of Hallervordenâ€“Spatz Syndrome. New England Journal of Medicine, 2003, 348, 33-40.	27.0	671
3	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	21.4	497
4	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	6.2	309
5	Mutations in GJB6 cause hidrotic ectodermal dysplasia. Nature Genetics, 2000, 26, 142-144.	21.4	270
6	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	6.2	207
7	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	7.6	203
8	Neurodegeneration with Brain Iron Accumulation: Genetic Diversity and Pathophysiological Mechanisms. Annual Review of Genomics and Human Genetics, 2015, 16, 257-279.	6.2	195
9	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	6.2	176
10	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	21.4	158
11	Neurodegeneration with brain iron accumulation. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 293-305.	1.8	153
12	New NBIA subtype. Neurology, 2013, 80, 268-275.	1.1	151
13	Pantethine rescues a Drosophila model for pantothenate kinaseâ€“associated neurodegeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 6988-6993.	7.1	132
14	Deficiency of pantothenate kinase 2 (Pank2) in mice leads to retinal degeneration and azoospermia. Human Molecular Genetics, 2005, 14, 49-57.	2.9	120
15	Novel histopathologic findings in molecularly-confirmed pantothenate kinase-associated neurodegeneration. Brain, 2011, 134, 947-958.	7.6	117
16	Mitochondrial Localization of Human PANK2 and Hypotheses of Secondary Iron Accumulation in Pantothenate Kinase-Associated Neurodegeneration. Annals of the New York Academy of Sciences, 2004, 1012, 282-298.	3.8	106
17	Unraveling the Hallervorden-Spatz syndrome: pantothenate kinase???associated neurodegeneration is the name . . .. Current Opinion in Pediatrics, 2003, 15, 572-577.	2.0	105
18	Safety and efficacy of deferiprone for pantothenate kinase-associated neurodegeneration: a randomised, double-blind, controlled trial and an open-label extension study. Lancet Neurology, The, 2019, 18, 631-642.	10.2	102

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19	Extracellular 4â€²-phosphopantetheine is a source for intracellular coenzyme A synthesis. Nature Chemical Biology, 2015, 11, 784-792.	8.0	98
20	Neurodegeneration With Brain Iron Accumulation: From Genes to Pathogenesis. Seminars in Pediatric Neurology, 2006, 13, 182-185.	2.0	92
21	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	6.2	91
22	Neuro-Ophthalmologic and Electroretinographic Findings in Pantothenate Kinase-Associated Neurodegeneration (formerly Hallervorden-Spatz Syndrome). American Journal of Ophthalmology, 2005, 140, 267.e1-267.e9.	3.3	87
23	Pantothenate kinase-associated neurodegeneration: altered mitochondria membrane potential and defective respiration in Pank2 knock-out mouse model. Human Molecular Genetics, 2012, 21, 5294-5305.	2.9	87
24	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
25	Impaired Coenzyme A metabolism affects histone and tubulin acetylation in Drosophila and human cell models of pantothenate kinase associated neurodegeneration. EMBO Molecular Medicine, 2011, 3, 755-766.	6.9	71
26	Pantothenate Kinase-Associated Neurodegeneration (PKAN) and PLA2G6-Associated Neurodegeneration (PLAN). International Review of Neurobiology, 2013, 110, 49-71.	2.0	68
27	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	1.1	64
28	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
29	4â€²â€Phosphopantetheine corrects CoA, iron, and dopamine metabolic defects in mammalian models of <scp>PKAN</scp>. EMBO Molecular Medicine, 2019, 11, e10489.	6.9	53
30	CoAâ€dependent activation of mitochondrial acyl carrier protein links four neurodegenerative diseases. EMBO Molecular Medicine, 2019, 11, e10488.	6.9	46
31	Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. European Journal of Human Genetics, 2000, 8, 372-380.	2.8	43
32	Autosomal dominant mitochondrial membrane proteinâ€associated neurodegeneration (MPAN). Molecular Genetics & Genomic Medicine, 2019, 7, e00736.	1.2	40
33	Pantothenate kinase-associated neurodegeneration (formerly Hallervordenâ€“Spatz Syndrome). Journal of the Neurological Sciences, 2003, 207, 106-107.	0.6	36
34	Novel founder intronic variant in SLC39A14 in two families causing Manganism and potential treatment strategies. Molecular Genetics and Metabolism, 2018, 124, 161-167.	1.1	36
35	Changes in Red Blood Cell membrane lipid composition: A new perspective into the pathogenesis of PKAN. Molecular Genetics and Metabolism, 2017, 121, 180-189.	1.1	34
36	Late diagnosis of fucosidosis in a child with progressive fixed dystonia, bilateral pallidal lesions and red spots on the skin. European Journal of Paediatric Neurology, 2014, 18, 516-519.	1.6	31

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37	Acetyl-4â€²-phosphopantetheine is stable in serum and prevents phenotypes induced by pantothenate kinase deficiency. Scientific Reports, 2017, 7, 11260.	3.3	27
38	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25
39	Novel WDR45 Mutation and Pathognomonic BPAN Imaging in a Young Female With Mild Cognitive Delay. Pediatrics, 2015, 136, e714-e717.	2.1	25
40	Brain MRI Pattern Recognition in Neurodegeneration With Brain Iron Accumulation. Frontiers in Neurology, 2020, 11, 1024.	2.4	20
41	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
42	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	2.1	17
43	Looking Deep into the Eye-of-the-Tiger in Pantothenate Kinaseâ€“Associated Neurodegeneration. American Journal of Neuroradiology, 2018, 39, 583-588.	2.4	16
44	Pallidal neuronal apolipoprotein E in pantothenate kinase-associated neurodegeneration recapitulates ischemic injury to the globus pallidus. Molecular Genetics and Metabolism, 2015, 116, 289-297.	1.1	15
45	Mitochondrial <scp>D</scp><scp>NA</scp> Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. Annals of Neurology, 2021, 89, 1240-1247.	5.3	12
46	De novo apparent loss-of-function mutations in PRR12 in three patients with intellectual disability and iris abnormalities. Human Genetics, 2018, 137, 257-264.	3.8	8
47	Pantothenate kinase 2 interacts with PINK1 to regulate mitochondrial quality control via acetyl-CoA metabolism. Nature Communications, 2022, 13, 2412.	12.8	8
48	Towards Precision Therapies for Inherited Disorders of Neurodegeneration with Brain Iron Accumulation. Tremor and Other Hyperkinetic Movements, 2021, 11, 51.	2.0	7
49	Phase unwrapping and background correction in MRI. , 2008, , .		6
50	Coenzyme A precursors flow from mother to zygote and from microbiome to host. Molecular Cell, 2022, 82, 2650-2665.e12.	9.7	6
51	A new NBIA patient from Turkey with homozygous C19ORF12 mutation. Acta Neurologica Belgica, 2019, 119, 623-625.	1.1	4
52	Net uptake of catecholamines into isolated chromaffin granules demonstrated by a novel polarographic technique. FEBS Letters, 1982, 141, 63-67.	2.8	2
53	Cannabis Use in Children With Pantothenate Kinaseâ€“Associated Neurodegeneration. Journal of Child Neurology, 2020, 35, 259-264.	1.4	2