Susan J Hayflick

List of Publications by Year
in descending order

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	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
2	Pantothenate kinase 2 interacts with PINK1 to regulate mitochondrial quality control via acetyl-CoA metabolism. Nature Communications, 2022, 13, 2412.	12.8	8
3	Coenzyme A precursors flow from mother to zygote and from microbiome to host. Molecular Cell, 2022, 82, 2650-2665.e12.	9.7	6
4	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
5	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	2.1	17
6	Towards Precision Therapies for Inherited Disorders of Neurodegeneration with Brain Iron Accumulation. Tremor and Other Hyperkinetic Movements, 2021, 11, 51.	2.0	7
7	Cannabis Use in Children With Pantothenate Kinaseâ€“Associated Neurodegeneration. Journal of Child Neurology, 2020, 35, 259-264.	1.4	2
8	Brain MRI Pattern Recognition in Neurodegeneration With Brain Iron Accumulation. Frontiers in Neurology, 2020, 11, 1024.	2.4	20
9	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
10	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
11	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
12	Autosomal dominant mitochondrial membrane proteinâ€associated neurodegeneration (MPAN). Molecular Genetics & Genomic Medicine, 2019, 7, e00736.	1.2	40
13	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
14	CoAâ€dependent activation of mitochondrial acyl carrier protein links four neurodegenerative diseases. EMBO Molecular Medicine, 2019, 11, e10488.	6.9	46
15	A new NBIA patient from Turkey with homozygous C19ORF12 mutation. Acta Neurologica Belgica, 2019, 119, 623-625.	1.1	4
16	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
17	Looking Deep into the Eye-of-the-Tiger in Pantothenate Kinaseâ€“Associated Neurodegeneration. American Journal of Neuroradiology, 2018, 39, 583-588.	2.4	16
18	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

#	Article	IF	Citations
19	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
20	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
21	Consensus clinical management guideline for pantothenate kinase-associated neurodegeneration (PKAN). Molecular Genetics and Metabolism, 2017, 120, 278-287.	1.1	64
22	Acetyl-4â€²-phosphopantetheine is stable in serum and prevents phenotypes induced by pantothenate kinase deficiency. Scientific Reports, 2017, 7, 11260.	3.3	27
23	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
24	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
25	Neurodegeneration with Brain Iron Accumulation: Genetic Diversity and Pathophysiological Mechanisms. Annual Review of Genomics and Human Genetics, 2015, 16, 257-279.	6.2	195
26	Novel WDR45 Mutation and Pathognomonic BPAN Imaging in a Young Female With Mild Cognitive Delay. Pediatrics, 2015, 136, e714-e717.	2.1	25
27	Extracellular 4â€²-phosphopantetheine is a source for intracellular coenzyme A synthesis. Nature Chemical Biology, 2015, 11, 784-792.	8.0	98
28	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
29	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
30	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
31	Pantothenate Kinase-Associated Neurodegeneration (PKAN) and PLA2G6-Associated Neurodegeneration (PLAN). International Review of Neurobiology, 2013, 110, 49-71.	2.0	68
32	New NBIA subtype. Neurology, 2013, 80, 268-275.	1.1	151
33	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	7.6	203
34	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25
35	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
36	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

#	Article	IF	Citations
37	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
38	Novel histopathologic findings in molecularly-confirmed pantothenate kinase-associated neurodegeneration. Brain, 2011, 134, 947-958.	7.6	117
39	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
40	Phase unwrapping and background correction in MRI. , 2008, , .		6
41	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nature Genetics, 2006, 38, 752-754.	21.4	497
42	Neurodegeneration With Brain Iron Accumulation: From Genes to Pathogenesis. Seminars in Pediatric Neurology, 2006, 13, 182-185.	2.0	92
43	Deficiency of pantothenate kinase 2 (Pank2) in mice leads to retinal degeneration and azoospermia. Human Molecular Genetics, 2005, 14, 49-57.	2.9	120
44	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
45	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
46	Pantothenate kinase-associated neurodegeneration (formerly Hallervordenâ€“Spatz Syndrome). Journal of the Neurological Sciences, 2003, 207, 106-107.	0.6	36
47	Genetic, Clinical, and Radiographic Delineation of Hallervordenâ€“Spatz Syndrome. New England Journal of Medicine, 2003, 348, 33-40.	27.0	671
48	Unraveling the Hallervorden-Spatz syndrome: pantothenate kinase???associated neurodegeneration is the name . . .. Current Opinion in Pediatrics, 2003, 15, 572-577.	2.0	105
49	A novel pantothenate kinase gene (PANK2) is defective in Hallervorden-Spatz syndrome. Nature Genetics, 2001, 28, 345-349.	21.4	721
50	Mutations in GJB6 cause hidrotic ectodermal dysplasia. Nature Genetics, 2000, 26, 142-144.	21.4	270
51	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
52	Homozygosity mapping of Hallervordenâ€“Spatz syndrome to chromosome 20p12.3â€“p13. Nature Genetics, 1996, 14, 479-481.	21.4	158
53	Net uptake of catecholamines into isolated chromaffin granules demonstrated by a novel polarographic technique. FEBS Letters, 1982, 141, 63-67.	2.8	2