Darius Ebrahimi-Fakhari

List of Publications by Year
in descending order

75 papers	8,255 citations	159585 30 h-index	79698 73 g-index
81 all docs	81 docs citations	81 times ranked	19466 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Distinct RolesIn Vivofor the Ubiquitinâ€“Proteasome System and the Autophagyâ€“Lysosomal Pathway in the Degradation of Î±-Synuclein. Journal of Neuroscience, 2011, 31, 14508-14520.	3.6	311
3	The Circadian Protein BMAL1 Regulates Translation in Response to S6K1-Mediated Phosphorylation. Cell, 2015, 161, 1138-1151.	28.9	270
4	<scp>N</scp>omenclature of genetic movement disorders: <scp>R</scp>ecommendations of the international <scp>P</scp>arkinson and movement disorder society task force. Movement Disorders, 2016, 31, 436-457.	3.9	228
5	The evolving spectrum ofPRRT2-associated paroxysmal diseases. Brain, 2015, 138, 3476-3495.	7.6	218
6	Protein degradation pathways in Parkinsonâ€™s disease: curse or blessing. Acta Neuropathologica, 2012, 124, 153-172.	7.7	213
7	Autism and the synapse. Current Opinion in Neurology, 2015, 28, 91-102.	3.6	156
8	Abnormal mTOR Activation in Autism. Annual Review of Neuroscience, 2018, 41, 1-23.	10.7	152
9	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. Brain, 2016, 139, 317-337.	7.6	126
10	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. Cell Reports, 2016, 17, 1053-1070.	6.4	125
11	Tuberous Sclerosis Complex. Pediatric Clinics of North America, 2015, 62, 633-648.	1.8	119
12	Alpha-synuclein aggregation involves a bafilomycin A₁-sensitive autophagy pathway. Autophagy, 2012, 8, 754-766.	9.1	111
13	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
14	Incidence of tuberous sclerosis and age at first diagnosis: new data and emerging trends from a national, prospective surveillance study. Orphanet Journal of Rare Diseases, 2018, 13, 117.	2.7	86
15	Reduction of TMEM97 increases NPC1 protein levels and restores cholesterol trafficking in Niemann-pick type C1 disease cells. Human Molecular Genetics, 2016, 25, 3588-3599.	2.9	74
16	Molecular chaperones and protein folding as therapeutic targets in Parkinsonâ€™s disease and other synucleinopathies. Acta Neuropathologica Communications, 2013, 1, 79.	5.2	73
17	Molecular Chaperones in Parkinson's Disease â€“ Present and Future. Journal of Parkinson's Disease, 2011, 1, 299-320.	2.8	63
18	Movement Disorders in Treatable Inborn Errors of Metabolism. Movement Disorders, 2019, 34, 598-613.	3.9	60

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19	Alpha-synucleinâ€™s degradation in vivo. Autophagy, 2012, 8, 281-283.	9.1	50
20	Direct detection of alpha synuclein oligomers in vivo. Acta Neuropathologica Communications, 2013, 1, 6.	5.2	49
21	p62/SQSTM1 Cooperates with Hyperactive mTORC1 to Regulate Glutathione Production, Maintain Mitochondrial Integrity, and Promote Tumorigenesis. Cancer Research, 2017, 77, 3255-3267.	0.9	49
22	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force â€“ An Update. Movement Disorders, 2022, 37, 905-935.	3.9	49
23	Molecular Chaperones and Co-Chaperones in Parkinson Disease. Neuroscientist, 2012, 18, 589-601.	3.5	47
24	Clinical and genetic characterization of AP4B1â€associated SPG47. American Journal of Medical Genetics, Part A, 2018, 176, 311-318.	1.2	47
25	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2017, 40, 631-640.	3.6	46
26	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
27	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. Human Molecular Genetics, 2020, 29, 320-334.	2.9	45
28	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	2.3	42
29	The Stress-Induced Atf3-Gelsolin Cascade Underlies Dendritic Spine Deficits in Neuronal Models of Tuberous Sclerosis Complex. Journal of Neuroscience, 2015, 35, 10762-10772.	3.6	40
30	Chronic Treatment with Novel Small Molecule Hsp90 Inhibitors Rescues Striatal Dopamine Levels but Not Î±-Synuclein-Induced Neuronal Cell Loss. PLoS ONE, 2014, 9, e86048.	2.5	35
31	Novel insights into the clinical and molecular spectrum of congenital disorders of autophagy. Journal of Inherited Metabolic Disease, 2020, 43, 51-62.	3.6	31
32	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
33	Molecular chaperones in Parkinson's disease--present and future. Journal of Parkinson's Disease, 2011, 1, 299-320.	2.8	29
34	The Spectrum of Movement Disorders in Childhoodâ€onset Lysosomal Storage Diseases. Movement Disorders Clinical Practice, 2018, 5, 149-155.	1.5	27
35	Familial Mediterranean fever in Germany: clinical presentation and amyloidosis risk. Scandinavian Journal of Rheumatology, 2013, 42, 52-58.	1.1	23
36	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. Annals of Clinical and Translational Neurology, 2022, 9, 193-205.	3.7	23

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37	AP-4-mediated axonal transport controls endocannabinoid production in neurons. Nature Communications, 2022, 13, 1058.	12.8	19
38	Using tuberous sclerosis complex to understand the impact of MTORC1 signaling on mitochondrial dynamics and mitophagy in neurons. Autophagy, 2017, 13, 754-756.	9.1	18
39	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
40	Proteotoxicity and Cardiac Dysfunction. New England Journal of Medicine, 2013, 368, 1754-1755.	27.0	17
41	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. Scientific Reports, 2018, 8, 7907.	3.3	16
42	Clinical Manifestations and Longterm Followup of a Patient with CINCA/NOMID Syndrome: Figure 1.. Journal of Rheumatology, 2010, 37, 2196-2197.	2.0	15
43	Congenital Disorders of Autophagy: What a Pediatric Neurologist Should Know. Neuropediatrics, 2018, 49, 018-025.	0.6	15
44	Loss of ap4s1 in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. Annals of Clinical and Translational Neurology, 2020, 7, 584-589.	3.7	15
45	Generation and characterization of six human induced pluripotent stem cell lines (iPSC) from three families with AP4B1-associated hereditary spastic paraplegia (SPG47). Stem Cell Research, 2019, 40, 101575.	0.7	14
46	Child Neurology: PRRT2 -associated movement disorders and differential diagnoses. Neurology, 2014, 83, 1680-1683.	1.1	13
47	Disruption of SOX6 Is Associated With a Rapid-Onset Dopa-Responsive Movement Disorder, Delayed Development, andÂDysmorphic Features. Pediatric Neurology, 2015, 52, 115-118.	2.1	12
48	Expansion of the genetic landscape of ERLIN2â€related disorders. Annals of Clinical and Translational Neurology, 2020, 7, 573-578.	3.7	12
49	Childhood-onset hereditary spastic paraplegia and its treatable mimics. Molecular Genetics and Metabolism, 2022, 137, 436-444.	1.1	11
50	High-throughput imaging of ATG9A distribution as a diagnostic functional assay for adaptor protein complex 4-associated hereditary spastic paraplegia. Brain Communications, 2021, 3, fcab221.	3.3	11
51	Restoring impaired protein metabolism in Parkinson's disease â€" TFEBâ€mediated autophagy as a novel therapeutic target. Movement Disorders, 2013, 28, 1346-1346.	3.9	10
52	Combination Clearance Therapy and Barbiturate Coma for Severe Carbamazepine Overdose. Pediatrics, 2017, 139, .	2.1	10
53	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	3.3	10
54	Systematic Analysis of Brain MRI Findings in Adaptor Protein Complex 4â€"Associated Hereditary Spastic Paraplegia. Neurology, 2021, 97, e1942-e1954.	1.1	10

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55	International electives in the final year of German medical school education--a student's perspective. GMS Zeitschrift FÃ¼r Medizinische Ausbildung, 2014, 31, Doc26.	1.2	10
56	An 8-year old boy with continuous spikes and waves during slow sleep presenting with positive onconeuronal antibodies. European Journal of Paediatric Neurology, 2015, 19, 257-261.	1.6	9
57	Congenital Chylothorax as the Initial Presentation of PTPN11-Associated Noonan Syndrome. Journal of Pediatrics, 2017, 185, 248-248.e1.	1.8	9
58	Generation and characterization of six human induced pluripotent stem cell lines (iPSC) from three families with AP4M1-associated hereditary spastic paraplegia (SPG50). Stem Cell Research, 2021, 53, 102335.	0.7	9
59	Recurrent Stroke-Like Episodes in FBXL4-Associated Early-Onset Mitochondrial Encephalomyopathy. Pediatric Neurology, 2015, 53, 549-550.	2.1	8
60	Tuberous Sclerosis Complex Associated Neuropsychiatric Disorders and Parental Stress: Findings from a National, Prospective TSC Surveillance Study. Neuropediatrics, 2019, 50, 294-299.	0.6	7
61	Blended Phenotype of Silver-Russell Syndrome and SPG50 Caused by Maternal Isodisomy of Chromosome 7. Neurology: Genetics, 2021, 7, e544.	1.9	7
62	EPG5 Variants with Modest Functional Impact Result in an Ameliorated and Primarily Neurological Phenotype in a 3.5-Year-Old Patient with Vici Syndrome. Neuropediatrics, 2019, 50, 257-261.	0.6	5
63	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy â€“ a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	9.1	5
64	Studying protein degradation pathways in vivo using a cranial window-based approach. Methods, 2011, 53, 194-200.	3.8	4
65	Autophagy and neurodegeneration â€" genetic findings in SENDA syndrome, a subtype of neurodegeneration with brain iron accumulation, provide a novel link. Movement Disorders, 2013, 28, 1050-1050.	3.9	4
66	Novel <scp>CAPN1</scp> missense variants in complex hereditary spastic paraplegia with earlyâ€onset psychosis. Annals of Clinical and Translational Neurology, 2022, 9, 570-576.	3.7	4
67	Disease Severity and Motor Impairment Correlate With Health-Related Quality of Life in AP-4-Associated Hereditary Spastic Paraplegia. Neurology: Genetics, 2021, 7, e605.	1.9	2
68	Social Deficits and Cerebellar Degeneration in Purkinje Cell Scn8a Knockout Mice. Frontiers in Molecular Neuroscience, 2022, 15, 822129.	2.9	2
69	Parkinson's disease: A disorder of axonal mitophagy?. Movement Disorders, 2014, 29, 1582-1582.	3.9	1
70	Reply letter to Jinnah â€œLocus pocusâ€•and Albanese â€œComplex dystonia is not a category in the new 2013 consensus classificationâ€• Necessary evolution, no magic!. Movement Disorders, 2016, 31, 1760-1762.	3.9	1
71	A special issue on childhoodâ€onset movement disorders. Movement Disorders, 2019, 34, 595-597.	3.9	1
72	Modeling Parkinson's disease in a dish â€" A story of yeast and men. Movement Disorders, 2014, 29, 34-34.	3.9	0

#	Article	IF	Citations
73	Resident and Fellow Section in Neuropediatrics. Neuropediatrics, 2018, 49, 229-230.	0.6	0
74	Startle Epilepsy Triggered By Maternal Cough. Neuropediatrics, 2021, 52, 341-342.	0.6	0
75	High-Throughput Imaging of ATG9A Distribution as a Diagnostic Functional Assay for Adaptor Protein Complex 4: Associated Hereditary Spastic Paraplegia (AP-4-HSP). , 2021, 52, .		0