

# Darius Ebrahimi-Fakhari

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Childhood-onset hereditary spastic paraplegia and its treatable mimics. <i>Molecular Genetics and Metabolism</i> , 2022, 137, 436-444.	1.1	11
2	Quantitative retrospective natural history modeling of <i>WDR45</i> -related developmental and epileptic encephalopathy – a systematic cross-sectional analysis of 160 published cases. <i>Autophagy</i> , 2022, 18, 1715-1727.	9.1	5
3	Mendelian etiologies identified with whole exome sequencing in cerebral palsy. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 193-205.	3.7	23
4	AP-4-mediated axonal transport controls endocannabinoid production in neurons. <i>Nature Communications</i> , 2022, 13, 1058.	12.8	19
5	Novel <i>CAPN1</i> missense variants in complex hereditary spastic paraplegia with early-onset psychosis. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 570-576.	3.7	4
6	Social Deficits and Cerebellar Degeneration in Purkinje Cell <i>Scn8a</i> Knockout Mice. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, 822129.	2.9	2
7	Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force – An Update. <i>Movement Disorders</i> , 2022, 37, 905-935.	3.9	49
8	Homozygous missense <i>WIPI2</i> variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. <i>Brain Communications</i> , 2021, 3, fcab183.	3.3	10
9	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> – associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	2.5	18
10	Generation and characterization of six human induced pluripotent stem cell lines (iPSC) from three families with AP4M1-associated hereditary spastic paraplegia (SPG50). <i>Stem Cell Research</i> , 2021, 53, 102335.	0.7	9
11	Startle Epilepsy Triggered By Maternal Cough. <i>Neuropediatrics</i> , 2021, 52, 341-342.	0.6	0
12	Disease Severity and Motor Impairment Correlate With Health-Related Quality of Life in AP-4-Associated Hereditary Spastic Paraplegia. <i>Neurology: Genetics</i> , 2021, 7, e605.	1.9	2
13	Systematic Analysis of Brain MRI Findings in Adaptor Protein Complex 4 – Associated Hereditary Spastic Paraplegia. <i>Neurology</i> , 2021, 97, e1942-e1954.	1.1	10
14	High-throughput imaging of ATG9A distribution as a diagnostic functional assay for adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain Communications</i> , 2021, 3, fcab221.	3.3	11
15	Blended Phenotype of Silver-Russell Syndrome and SPG50 Caused by Maternal Isodisomy of Chromosome 7. <i>Neurology: Genetics</i> , 2021, 7, e544.	1.9	7
16	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
17	Novel insights into the clinical and molecular spectrum of congenital disorders of autophagy. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 51-62.	3.6	31
18	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. <i>Human Molecular Genetics</i> , 2020, 29, 320-334.	2.9	45

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19	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain</i> , 2020, 143, 2929-2944.	7.6	29
20	Expansion of the genetic landscape of <i>ERLIN2</i> -related disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 573-578.	3.7	12
21	Loss of <i>ap4s1</i> in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 584-589.	3.7	15
22	EPG5 Variants with Modest Functional Impact Result in an Ameliorated and Primarily Neurological Phenotype in a 3.5-Year-Old Patient with Vici Syndrome. <i>Neuropediatrics</i> , 2019, 50, 257-261.	0.6	5
23	Tuberous Sclerosis Complex Associated Neuropsychiatric Disorders and Parental Stress: Findings from a National, Prospective TSC Surveillance Study. <i>Neuropediatrics</i> , 2019, 50, 294-299.	0.6	7
24	Generation and characterization of six human induced pluripotent stem cell lines (iPSC) from three families with AP4B1-associated hereditary spastic paraplegia (SPG47). <i>Stem Cell Research</i> , 2019, 40, 101575.	0.7	14
25	A special issue on childhood-onset movement disorders. <i>Movement Disorders</i> , 2019, 34, 595-597.	3.9	1
26	Movement Disorders in Treatable Inborn Errors of Metabolism. <i>Movement Disorders</i> , 2019, 34, 598-613.	3.9	60
27	Abnormal mTOR Activation in Autism. <i>Annual Review of Neuroscience</i> , 2018, 41, 1-23.	10.7	152
28	The Spectrum of Movement Disorders in Childhood-onset Lysosomal Storage Diseases. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 149-155.	1.5	27
29	Clinical and genetic characterization of <i>AP4B1</i> -associated SPG47. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 311-318.	1.2	47
30	Congenital Disorders of Autophagy: What a Pediatric Neurologist Should Know. <i>Neuropediatrics</i> , 2018, 49, 018-025.	0.6	15
31	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. <i>Scientific Reports</i> , 2018, 8, 7907.	3.3	16
32	Resident and Fellow Section in Neuropediatrics. <i>Neuropediatrics</i> , 2018, 49, 229-230.	0.6	0
33	Incidence of tuberous sclerosis and age at first diagnosis: new data and emerging trends from a national, prospective surveillance study. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 117.	2.7	86
34	Using tuberous sclerosis complex to understand the impact of MTORC1 signaling on mitochondrial dynamics and mitophagy in neurons. <i>Autophagy</i> , 2017, 13, 754-756.	9.1	18
35	Congenital Chylothorax as the Initial Presentation of PTPN11-Associated Noonan Syndrome. <i>Journal of Pediatrics</i> , 2017, 185, 248-248.e1.	1.8	9
36	Combination Clearance Therapy and Barbiturate Coma for Severe Carbamazepine Overdose. <i>Pediatrics</i> , 2017, 139, .	2.1	10

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37	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 631-640.	3.6	46
38	p62/SQSTM1 Cooperates with Hyperactive mTORC1 to Regulate Glutathione Production, Maintain Mitochondrial Integrity, and Promote Tumorigenesis. <i>Cancer Research</i> , 2017, 77, 3255-3267.	0.9	49
39	<sc>N</sc>omenclature of genetic movement disorders: <sc>R</sc>ecommendations of the international <sc>P</sc>arkinson and movement disorder society task force. <i>Movement Disorders</i> , 2016, 31, 436-457.	3.9	228
40	Reduction of TMEM97 increases NPC1 protein levels and restores cholesterol trafficking in Niemann-pick type C1 disease cells. <i>Human Molecular Genetics</i> , 2016, 25, 3588-3599.	2.9	74
41	Reply letter to Jinnah "Locus pocus" and Albanese "Complex dystonia is not a category in the new 2013 consensus classification" Necessary evolution, no magic!. <i>Movement Disorders</i> , 2016, 31, 1760-1762.	3.9	1
42	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. <i>Cell Reports</i> , 2016, 17, 1053-1070.	6.4	125
43	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
44	<i>EPG5</i>-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	7.6	99
45	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. <i>Brain</i> , 2016, 139, 317-337.	7.6	126
46	An 8-year old boy with continuous spikes and waves during slow sleep presenting with positive onconeural antibodies. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 257-261.	1.6	9
47	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to <i>FBXL4</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 905-914.	3.6	45
48	The Circadian Protein BMAL1 Regulates Translation in Response to S6K1-Mediated Phosphorylation. <i>Cell</i> , 2015, 161, 1138-1151.	28.9	270
49	Tuberous Sclerosis Complex. <i>Pediatric Clinics of North America</i> , 2015, 62, 633-648.	1.8	119
50	Autism and the synapse. <i>Current Opinion in Neurology</i> , 2015, 28, 91-102.	3.6	156
51	Recurrent Stroke-Like Episodes in FBXL4-Associated Early-Onset Mitochondrial Encephalomyopathy. <i>Pediatric Neurology</i> , 2015, 53, 549-550.	2.1	8
52	The Stress-Induced Atf3-Gelsolin Cascade Underlies Dendritic Spine Deficits in Neuronal Models of Tuberous Sclerosis Complex. <i>Journal of Neuroscience</i> , 2015, 35, 10762-10772.	3.6	40
53	The evolving spectrum of <i>PRRT2</i>-associated paroxysmal diseases. <i>Brain</i> , 2015, 138, 3476-3495.	7.6	218
54	Disruption of SOX6 Is Associated With a Rapid-Onset Dopa-Responsive Movement Disorder, Delayed Development, and Dysmorphic Features. <i>Pediatric Neurology</i> , 2015, 52, 115-118.	2.1	12

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55	Chronic Treatment with Novel Small Molecule Hsp90 Inhibitors Rescues Striatal Dopamine Levels but Not $\alpha$ -Synuclein-Induced Neuronal Cell Loss. PLoS ONE, 2014, 9, e86048.	2.5	35
56	Parkinson's disease: A disorder of axonal mitophagy?. Movement Disorders, 2014, 29, 1582-1582.	3.9	1
57	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	2.3	42
58	Modeling Parkinson's disease in a dish – A story of yeast and men. Movement Disorders, 2014, 29, 34-34.	3.9	0
59	Child Neurology: <i>PRRT2</i> -associated movement disorders and differential diagnoses. Neurology, 2014, 83, 1680-1683.	1.1	13
60	International electives in the final year of German medical school education—a student's perspective. GMS Zeitschrift für Medizinische Ausbildung, 2014, 31, Doc26.	1.2	10
61	Direct detection of alpha synuclein oligomers in vivo. Acta Neuropathologica Communications, 2013, 1, 6.	5.2	49
62	Restoring impaired protein metabolism in Parkinson's disease – TFE $\beta$ -mediated autophagy as a novel therapeutic target. Movement Disorders, 2013, 28, 1346-1346.	3.9	10
63	Molecular chaperones and protein folding as therapeutic targets in Parkinson's disease and other synucleinopathies. Acta Neuropathologica Communications, 2013, 1, 79.	5.2	73
64	Familial Mediterranean fever in Germany: clinical presentation and amyloidosis risk. Scandinavian Journal of Rheumatology, 2013, 42, 52-58.	1.1	23
65	Proteotoxicity and Cardiac Dysfunction. New England Journal of Medicine, 2013, 368, 1754-1755.	27.0	17
66	Autophagy and neurodegeneration – 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
67	Alpha-synuclein's degradation in vivo. Autophagy, 2012, 8, 281-283.	9.1	50
68	Alpha-synuclein aggregation involves a bafilomycin A <sub>1</sub> -sensitive autophagy pathway. Autophagy, 2012, 8, 754-766.	9.1	111
69	Molecular Chaperones and Co-Chaperones in Parkinson Disease. Neuroscientist, 2012, 18, 589-601.	3.5	47
70	Protein degradation pathways in Parkinson's disease: curse or blessing. Acta Neuropathologica, 2012, 124, 153-172.	7.7	213
71	Studying protein degradation pathways in vivo using a cranial window-based approach. Methods, 2011, 53, 194-200.	3.8	4
72	Molecular Chaperones in Parkinson's Disease – Present and Future. Journal of Parkinson's Disease, 2011, 1, 299-320.	2.8	63

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73	Distinct Roles <i>In Vivo</i> for the Ubiquitin-Proteasome System and the Autophagy-Lysosomal Pathway in the Degradation of $\alpha$ -Synuclein. <i>Journal of Neuroscience</i> , 2011, 31, 14508-14520.	3.6	311
74	Molecular chaperones in Parkinson's disease--present and future. <i>Journal of Parkinson's Disease</i> , 2011, 1, 299-320.	2.8	29
75	Clinical Manifestations and Longterm Followup of a Patient with CINCA/NOMID Syndrome: Figure 1.. <i>Journal of Rheumatology</i> , 2010, 37, 2196-2197.	2.0	15