Edor Kabashi

List of Publications by Year
in descending order

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70 papers	5,826 citations	117571 34 h-index	95218 68 g-index
73 all docs	73 docs citations	73 times ranked	8124 citing authors

#	Article	IF	Citations
1	Novel genome-editing-based approaches to treat motor neuron diseases: Promises and challenges. Molecular Therapy, 2022, 30, 47-53.	3.7	13
2	Autophagy and ALS: mechanistic insights and therapeutic implications. Autophagy, 2022, 18, 254-282.	4.3	66
3	Functional characterisation of the amyotrophic lateral sclerosis risk locus GPX3/TNIP1. Genome Medicine, 2022, 14, 7.	3.6	12
4	TDP-43 Regulation of AChE Expression Can Mediate ALS-Like Phenotype in Zebrafish. Cells, 2021, 10, 221.	1.8	16
5	Synaptic disruption and CREBâ€regulated transcription are restored by K ⁺ channel blockers in ALS. EMBO Molecular Medicine, 2021, 13, e13131.	3.3	22
6	Deep phenotyping unstructured data mining in an extensive pediatric database to unravel a common KCNA2 variant in neurodevelopmental syndromes. Genetics in Medicine, 2021, 23, 968-971.	1.1	9
7	Behavioral And Physiological Analysis In A Zebrafish Model Of Epilepsy. Journal of Visualized Experiments, 2021, , .	0.2	1
8	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Human Mutation, 2020, 41, 69-80.	1.1	33
9	Freezing activity brief data from a new FUS mutant zebrafish line. Data in Brief, 2020, 31, 105921.	0.5	2
10	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	2.6	17
11	Functional Characterization of Neurofilament Light Splicing and Misbalance in Zebrafish. Cells, 2020, 9, 1238.	1.8	3
12	Functional characterization of a FUS mutant zebrafish line as a novel genetic model for ALS. Neurobiology of Disease, 2020, 142, 104935.	2.1	18
13	Diagnostic Challenge and Neuromuscular Junction Contribution to ALS Pathogenesis. Frontiers in Neurology, 2019, 10, 68.	1.1	27
14	Association of Rare Genetic Variants in Opioid Receptors with Tourette Syndrome. Tremor and Other Hyperkinetic Movements, 2019, 9, .	1.1	13
15	Depdc5 knockdown causes <scp>mTOR</scp>â€dependent motor hyperactivity in zebrafish. Annals of Clinical and Translational Neurology, 2018, 5, 510-523.	1.7	32
16	Transcriptomic Analysis of Zebrafish TDP-43 Transgenic Lines. Frontiers in Molecular Neuroscience, 2018, 11, 463.	1.4	17
17	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	1.5	86
18	Neuroleptics as therapeutic compounds stabilizing neuromuscular transmission in amyotrophic lateral sclerosis. JCI Insight, 2017, 2, .	2.3	83

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19	Neuromuscular Junction Impairment in Amyotrophic Lateral Sclerosis: Reassessing the Role of Acetylcholinesterase. Frontiers in Molecular Neuroscience, 2016, 9, 160.	1.4	49
20	ALSUntangled No. 35: Hyperbaric Oxygen Therapy*. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 622-624.	1.1	0
21	The most prevalent genetic cause of ALS-FTD, C9orf72 synergizes the toxicity of ATXN2 intermediate polyglutamine repeats through the autophagy pathway. Autophagy, 2016, 12, 1406-1408.	4.3	35
22	Loss of C9 <scp>ORF</scp> 72 impairs autophagy and synergizes with polyQ Ataxinâ€2 to induce motor neuron dysfunction and cell death. EMBO Journal, 2016, 35, 1276-1297.	3.5	343
23	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	2.6	333
24	Deciphering spreading mechanisms in amyotrophic lateral sclerosis. Current Opinion in Neurology, 2015, 28, 455-461.	1.8	17
25	ALSUntangled No. 30: Methylcobalamin. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 536-539.	1.1	7
26	ALSUntangled No. 28: Acupuncture. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 286-289.	1.1	1
27	ALSUntangled No. 27: Precision Stem Cell. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 282-285.	1.1	2
28	ALSUntangled No. 29: MitoQ. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 427-429.	1.1	1
29	Defining the genetic connection linking amyotrophic lateral sclerosis (ALS) with frontotemporal dementia (FTD). Trends in Genetics, 2015, 31, 263-273.	2.9	106
30	Abnormal splicing switch of DMDâ€™s penultimate exon compromises muscle fibre maintenance in myotonic dystrophy. Nature Communications, 2015, 6, 7205.	5.8	76
31	Two novel COLVI long chains in zebrafish that are essential for muscle development. Human Molecular Genetics, 2015, 24, 6624-6639.	1.4	18
32	Sqstm1 knock-down causes a locomotor phenotype ameliorated by rapamycin in a zebrafish model of ALS/FTLD. Human Molecular Genetics, 2015, 24, 1682-1690.	1.4	69
33	Fishing for causes and cures of motor neuron disorders. DMM Disease Models and Mechanisms, 2014, 7, 799-809.	1.2	60
34	ALSUntangled No. 26: Lunasin. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 622-626.	1.1	7
35	Contribution of ATXN2 intermediary polyQ expansions in a spectrum of neurodegenerative disorders. Neurology, 2014, 83, 990-995.	1.5	70
36	Defining the association of TMEM106B variants among frontotemporal lobar degeneration patients with GRN mutations andÂ C9orf72 repeat expansions. Neurobiology of Aging, 2014, 35, 2658.e1-2658.e5.	1.5	33

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37	hnRNPA2B1 and hnRNPA1 mutations are rare in patients with â€œmultisystem proteinopathyâ€•and frontotemporal lobar degeneration phenotypes. Neurobiology of Aging, 2014, 35, 934.e5-934.e6.	1.5	47
38	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. Neurobiology of Aging, 2014, 35, 2419.e23-2419.e25.	1.5	84
39	Loss of Function of Glucocerebrosidase GBA2 Is Responsible for Motor Neuron Defects in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 238-244.	2.6	154
40	TARDBPandFUSMutations Associated with Amyotrophic Lateral Sclerosis: Summary and Update. Human Mutation, 2013, 34, 812-826.	1.1	216
41	Pharmacological reduction of ER stress protects against TDP-43 neuronal toxicity in vivo. Neurobiology of Disease, 2013, 55, 64-75.	2.1	113
42	Mutations in the PFN1 gene are not a common cause in patients with amyotrophic lateral sclerosis and frontotemporal lobar degeneration in France. Neurobiology of Aging, 2013, 34, 1709.e1-1709.e2.	1.5	21
43	Screening UBQLN-2 in French frontotemporal lobar degeneration and frontotemporal lobar degenerationâ€“amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2013, 34, 2078.e5-2078.e6.	1.5	6
44	TREM2 mutations are rare in a French cohort of patients with frontotemporal dementia. Neurobiology of Aging, 2013, 34, 2443.e1-2443.e2.	1.5	35
45	Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. Annals of Neurology, 2013, 74, 180-187.	2.8	284
46	ALS Untangled No. 21: Fecal transplants. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 482-485.	1.1	5
47	ALS Untangled No. 20: The Deanna Protocol. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 319-323.	1.1	19
48	SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10.	4.5	153
49	Investigating the contribution of VAPB/ALS8 loss of function in amyotrophic lateral sclerosis. Human Molecular Genetics, 2013, 22, 2350-2360.	1.4	75
50	ALS Untangled No. 17: â€œWhen ALS Is Lymeâ€• Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 487-491.	2.3	6
51	Spatacsin and spastizin act in the same pathway required for proper spinal motor neuron axon outgrowth in zebrafish. Neurobiology of Disease, 2012, 48, 299-308.	2.1	42
52	Impaired proteasome function in sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 367-371.	2.3	54
53	Methylene Blue Protects against TDP-43 and FUS Neuronal Toxicity in C. elegans and D. rerio. PLoS ONE, 2012, 7, e42117.	1.1	88
54	Zebrafish models for the functional genomics of neurogenetic disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 335-345.	1.8	95

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55	FUS and TARDBP but Not SOD1 Interact in Genetic Models of Amyotrophic Lateral Sclerosis. PLoS Genetics, 2011, 7, e1002214.	1.5	167
56	In the swim of things: recent insights to neurogenetic disorders from zebrafish. Trends in Genetics, 2010, 26, 373-381.	2.9	45
57	Gain and loss of function of ALS-related mutations of TARDBP (TDP-43) cause motor deficits in vivo. Human Molecular Genetics, 2010, 19, 671-683.	1.4	350
58	No TARDBP Mutations in a French Canadian Population of Patients With Parkinson Disease. Archives of Neurology, 2009, 66, 281-2.	4.9	12
59	TARDBP mutations in individuals with sporadic and familial amyotrophic lateral sclerosis. Nature Genetics, 2008, 40, 572-574.	9.4	1,371
60	ALS predisposition modifiers: Knock NOX, who's there? SOD1 mice still are. European Journal of Human Genetics, 2008, 16, 140-142.	1.4	11
61	Proteasomes remain intact, but show early focal alteration in their composition in a mouse model of amyotrophic lateral sclerosis. Journal of Neurochemistry, 2008, 105, 2353-2366.	2.1	31
62	Contribution of TARDBP mutations to sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2008, 46, 112-114.	1.5	162
63	Als2 mRNA splicing variants detected in KO mice rescue severe motor dysfunction phenotype in Als2 knock-down zebrafish. Human Molecular Genetics, 2008, 17, 2691-2702.	1.4	48
64	Tryptophan 32 Potentiates Aggregation and Cytotoxicity of a Copper/Zinc Superoxide Dismutase Mutant Associated with Familial Amyotrophic Lateral Sclerosis. Journal of Biological Chemistry, 2007, 282, 16329-16335.	1.6	67
65	G.P.18.11 Functional characterization of strumpellin, mutated in hereditary spastic paraplegia. Neuromuscular Disorders, 2007, 17, 893.	0.3	0
66	Oxidized/misfolded superoxide dismutaseâ€1: the cause of all amyotrophic lateral sclerosis?. Annals of Neurology, 2007, 62, 553-559.	2.8	137
67	Failure of protein quality control in amyotrophic lateral sclerosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 1038-1050.	1.8	78
68	Motor Neuron Disease. , 2006, , 247-264.		0
69	Proteasome activity or expression is not altered by activation of the heat shock transcription factor Hsf1 in cultured fibroblasts or myoblasts. Cell Stress and Chaperones, 2005, 10, 230.	1.2	10
70	Focal dysfunction of the proteasome: a pathogenic factor in a mouse model of amyotrophic lateral sclerosis. Journal of Neurochemistry, 2004, 89, 1325-1335.	2.1	141