Wan Hee Yoon

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
2	Loss of Nardilysin, a Mitochondrial Co-chaperone for \hat{l}_{\pm} -Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
3	A TRPV Channel in Drosophila Motor Neurons Regulates Presynaptic Resting Ca2+ Levels, Synapse Growth, and Synaptic Transmission. Neuron, 2014, 84, 764-777.	8.1	68
4	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
5	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	6.2	33
6	A biallelic pathogenic variant in the <scp><i>OGDH</i></scp> gene results in a neurological disorder with features of a mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 388-400.	3.6	24
7	Neuronal overexpression of human VAPB slows motor impairment and neuromuscular denervation in a mouse model of ALS. Human Molecular Genetics, 2016, 25, ddw294.	2.9	19
8	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	8.2	16
9	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
10	Knockdown of genes involved in axonal transport enhances the toxicity of human neuromuscular diseaseâ€inked MATR3 mutations in ⟨i⟩Drosophila⟨/i⟩. FEBS Letters, 2020, 594, 2800-2818.	2.8	9
11	CRISPR/Cas9-mediated tissue-specific knockout and cDNA rescue using sgRNAs that target exon-intron junctions in Drosophila melanogaster. STAR Protocols, 2022, 3, 101465.	1.2	5