

Heterozygous de-novo mutations in ATP1A3 in patients childhood: a whole-exome sequencing gene-identificati

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Citation Report

#	ARTICLE	IF	CITATIONS
1	High-Throughput Sequencing and Rare Genetic Diseases. <i>Molecular Syndromology</i> , 2012, 3, 197-203.	0.3	6
3	Clinical spectrum of disease associated with ATP1A3 mutations. <i>Lancet Neurology</i> , The, 2012, 11, 741-743.	4.9	26
5	Episodic Movement Disorders: From Phenotype to Genotype and Back. <i>Current Neurology and Neuroscience Reports</i> , 2013, 13, 379.	2.0	9
6	Genetics in Dystonia: An Update. <i>Current Neurology and Neuroscience Reports</i> , 2013, 13, 410.	2.0	13
7	Quantifying Long-Term Scientific Impact. <i>Science</i> , 2013, 342, 127-132.	6.0	604
8	From the periphery to centre stage: de novo single nucleotide variants play a key role in human genetic disease. <i>Journal of Medical Genetics</i> , 2013, 50, 203-211.	1.5	33
9	A Specific and Essential Role for Na,K-ATPase β 3 in Neurons Co-expressing β 1 and β 3. <i>Journal of Biological Chemistry</i> , 2013, 288, 2734-2743.	1.6	105
10	Functional studies and proteomics in platelets and fibroblasts reveal a lysosomal defect with increased cathepsin-dependent apoptosis in ATP1A3 defective alternating hemiplegia of childhood. <i>Journal of Proteomics</i> , 2013, 86, 53-69.	1.2	15
11	Crystal Structure of Na ⁺ , K ⁺ -ATPase in the Na ⁺ -Bound State. <i>Science</i> , 2013, 342, 123-127.	6.0	168
12	Zebrafish as a novel model to assess Na ⁺ /K ⁺ -ATPase-related neurological disorders. <i>Neuroscience and Biobehavioral Reviews</i> , 2013, 37, 2774-2787.	2.9	17
13	Next-generation sequencing in understanding complex neurological disease. <i>Expert Review of Neurotherapeutics</i> , 2013, 13, 215-227.	1.4	18
14	Personalized Medicine: Impact on Patient Care in Genetic Counseling. <i>Current Genetic Medicine Reports</i> , 2013, 1, 129-134.	1.9	7
15	The Neurogenomics View of Neurological Diseases. <i>JAMA Neurology</i> , 2013, 70, 689.	4.5	19
16	Pearls and pitfalls in genetic studies of migraine. <i>Cephalalgia</i> , 2013, 33, 614-625.	1.8	38
17	The multiple faces of the <i>ATP1A3</i> -related dystonic movement disorder. <i>Movement Disorders</i> , 2013, 28, 1457-1459.	2.2	62
18	The genetics of dystonia: new twists in an old tale. <i>Brain</i> , 2013, 136, 2017-2037.	3.7	102
19	Dystonia. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2013, 19, 1225-1241.	0.4	11
20	Primary and secondary dystonic syndromes. <i>Current Opinion in Neurology</i> , 2013, 26, 406-412.	1.8	13

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21	Na ⁺ /K ⁺ ATPase $\hat{\alpha}$ 1 and $\hat{\alpha}$ 3 Isoforms Are Differentially Expressed in $\hat{\alpha}$ - and $\hat{\alpha}$ -Motoneurons. <i>Journal of Neuroscience</i> , 2013, 33, 9913-9919.	1.7	61
22	Enhanced inhibitory neurotransmission in the cerebellar cortex of <i>Atp1a3</i> deficient heterozygous mice. <i>Journal of Physiology</i> , 2013, 591, 3433-3449.	1.3	57
23	No Mutation in the SLC2A3 Gene in Cohorts of GLUT1 Deficiency Syndrome—Like Patients Negative for SLC2A1 and in Patients with AHC Negative for ATP1A3. <i>JIMD Reports</i> , 2013, 12, 115-120.	0.7	4
24	Commentary. <i>Movement Disorders</i> , 2013, 28, 1939-1939.	2.2	4
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26	Ionic leakage underlies a gain-of-function effect of dominant disease mutations affecting diverse P-type ATPases. <i>Nature Genetics</i> , 2014, 46, 144-151.	9.4	39
27	Heterozygous mice deficient in <i>Atp1a3</i> exhibit motor deficits by chronic restraint stress. <i>Behavioural Brain Research</i> , 2014, 272, 100-110.	1.2	28
28	Application of Magnetic Resonance Spectroscopy in Patients with Alternating Hemiplegia of Childhood: Findings on Metabolic Dysfunctions. <i>Neuropediatrics</i> , 2014, 45, 162-168.	0.3	0
29	Genotype—phenotype correlations in alternating hemiplegia of childhood. <i>Neurology</i> , 2014, 82, 482-490.	1.5	93
30	Whole-Exome Sequencing of Patients with Severe Disorders of Insulin Action. <i>Frontiers in Diabetes</i> , 2014, , 87-101.	0.4	0
31	<i>ATP1A3</i> mutations. <i>Neurology</i> , 2014, 82, 468-469.	1.5	9
32	Intermediate form between alternating hemiplegia of childhood and rapid-onset dystonia—parkinsonism. <i>Movement Disorders</i> , 2014, 29, 153-154.	2.2	21
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41	The promise of whole-exome sequencing in medical genetics. <i>Journal of Human Genetics</i> , 2014, 59, 5-15.	1.1	404
42	Origins and functional diversification of salinity-responsive Na ⁺ , K ⁺ ATPase \pm 1 paralogs in salmonids. <i>Molecular Ecology</i> , 2014, 23, 3483-3503.	2.0	15
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44	Clinical and genetic analysis in alternating hemiplegia of childhood: Ten new patients from Southern Europe. <i>Journal of the Neurological Sciences</i> , 2014, 344, 37-42.	0.3	19
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52	The expanding clinical and genetic spectrum of ATP1A3-related disorders. <i>Neurology</i> , 2014, 82, 945-955.	1.5	98
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54	Alternating hemiplegia of childhood: New diagnostic options. <i>Neurologia I Neurochirurgia Polska</i> , 2014, 48, 130-135.	0.6	9
55	Alternating Hemiplegia of Childhood With a de Novo Mutation in ATP1A3 and Changes in SLC2A1 Responsive to a Ketogenic Diet. <i>Pediatric Neurology</i> , 2014, 50, 377-379.	1.0	27
56	Alternating hemiplegia of childhood and rapid-onset dystonia-parkinsonism are both ATP1A3-related disorders. <i>Molecular and Cellular Pediatrics</i> , 2014, 1, A15.	1.0	0
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79	The Expanding Spectrum of Neurological Phenotypes in Children With ATP1A3 Mutations, Alternating Hemiplegia of Childhood, Rapid-onset Dystonia-Parkinsonism, CAPOS and Beyond. <i>Pediatric Neurology</i> , 2015, 52, 56-64.	1.0	119
80	A novel <i>SLC2A1</i> mutation linking hemiplegic migraine with alternating hemiplegia of childhood. <i>Cephalalgia</i> , 2015, 35, 10-15.	1.8	28
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90	A novel de novo mutation in <i>ATP1A3</i> and childhood-onset schizophrenia. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001008.	0.5	46
91	De novo p.Arg756Cys mutation of ATP1A3 causes an atypical form of alternating hemiplegia of childhood with prolonged paralysis and choreoathetosis. <i>BMC Neurology</i> , 2016, 16, 174.	0.8	21
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126	Mechanisms of increased hippocampal excitability in the <i>Mash1</i> ^{+/Δ} mouse model of Na ⁺ /K ⁺ -ATPase dysfunction. <i>Epilepsia</i> , 2018, 59, 1455-1468.	2.6	38
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136	A case of early onset life-threatening epilepsy associated with a novel ATP1A3 gene variant. <i>Brain and Development</i> , 2019, 41, 285-291.	0.6	10
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155	Effect of Flunarizine on Alternating Hemiplegia of Childhood in a Patient with the p.E815K Mutation in ATP1A3: A Case Report. <i>Case Reports in Neurology</i> , 2021, 12, 299-306.	0.3	4
156	Combined dystonias: clinical and genetic updates. <i>Journal of Neural Transmission</i> , 2021, 128, 417-429.	1.4	12
158	Alternating hemiplegia of childhood presenting as recurrent apnoea in a term newborn infant. <i>Annals of the Academy of Medicine, Singapore</i> , 2021, 50, 174-176.	0.2	0
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