

# Exome sequencing identifies truncating mutations in P kinesigenic dyskinesia

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Citation Report

#	ARTICLE	IF	CITATIONS
1	<i>PRRT2</i> mutations cause hemiplegic migraine. <i>Neurology</i> , 2012, 79, 2122-2124.	1.5	118
2	A 600-kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. <i>Journal of Medical Genetics</i> , 2012, 49, 660-668.	1.5	251
3	<i>PRRT2</i> mutations in familial infantile seizures, paroxysmal dyskinesia, and hemiplegic migraine. <i>Neurology</i> , 2012, 79, 2109-2114.	1.5	106
4	Mild paroxysmal kinesigenic dyskinesia caused by <i>PRRT2</i> missense mutation with reduced penetrance. <i>Neurology</i> , 2012, 79, 946-948.	1.5	23
5	<i>PRRT2</i> mutations. <i>Neurology</i> , 2012, 79, 170-174.	1.5	98
6	Genetics of Parkinson disease and other movement disorders. <i>Current Opinion in Neurology</i> , 2012, 25, 466-474.	1.8	59
7	Discovery of epilepsy susceptibility genes: implications for therapy development and pharmacogenomics. <i>Pharmacogenomics</i> , 2012, 13, 731-734.	0.6	6
8	<i>PRRT2</i> mutation causes benign familial infantile convulsions. <i>Neurology</i> , 2012, 79, 2154-2155.	1.5	22
9	<i>PRRT2</i> links infantile convulsions and paroxysmal dyskinesia with migraine. <i>Neurology</i> , 2012, 79, 2097-2103.	1.5	90
10	<i>PRRT2</i> gene mutations. <i>Neurology</i> , 2012, 79, 2115-2121.	1.5	159
11	The Genetics of Dystonias. <i>Advances in Genetics</i> , 2012, 79, 35-85.	0.8	31
12	Mutations in <i>PRRT2</i> result in paroxysmal dyskinesias with marked variability in clinical expression. <i>Journal of Medical Genetics</i> , 2012, 49, 79-82.	1.5	91
13	<i>PRRT2</i> phenotypes and penetrance of paroxysmal kinesigenic dyskinesia and infantile convulsions. <i>Neurology</i> , 2012, 79, 777-784.	1.5	79
14	Journal Watch: Our panel of experts highlight the most important research articles across the spectrum of topics relevant to the field of neurodegenerative disease management. <i>Neurodegenerative Disease Management</i> , 2012, 2, 569-572.	1.2	0
15	The Use of Next-Generation Sequencing in Movement Disorders. <i>Frontiers in Genetics</i> , 2012, 3, 75.	1.1	21
18	Paroxysmal disorders associated with <i>PRRT2</i> mutations shake up expectations on ion channel genes. <i>Neurology</i> , 2012, 79, 2086-2088.	1.5	18
19	Review: Genetics and neuropathology of primary pure dystonia. <i>Neuropathology and Applied Neurobiology</i> , 2012, 38, 520-534.	1.8	35
20	Familial <i>PRRT2</i> mutation with heterogeneous paroxysmal disorders including paroxysmal torticollis and hemiplegic migraine. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 958-960.	1.1	100

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21	Homozygous c.649dupC mutation in <i>PRRT2</i> worsens the BFIS/PKD phenotype with mental retardation, episodic ataxia, and absences. <i>Epilepsia</i> , 2012, 53, e196-9.	2.6	78
22	Exacerbation of idiopathic paroxysmal kinesigenic dyskinesia in remission state caused by secondary hypoparathyroidism with hypocalcemia after thyroidectomy: Evidence for ion channelopathy. <i>Brain and Development</i> , 2012, 34, 840-843.	0.6	8
23	Identification of a novel <i>PRRT2</i> mutation in patients with paroxysmal kinesigenic dyskinesias and c.649dupC as a mutation hot-spot. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 704-706.	1.1	51
24	Microdeletions detected using chromosome microarray in children with suspected genetic movement disorders: a single-centre study. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 618-623.	1.1	68
25	<i>PRRT2</i> phenotypic spectrum includes sporadic and fever-related infantile seizures. <i>Neurology</i> , 2012, 79, 2104-2108.	1.5	75
26	Novel <i>PRRT2</i> mutation in an African-American family with paroxysmal kinesigenic dyskinesia. <i>BMC Neurology</i> , 2012, 12, 93.	0.8	24
27	Two Faces of the Same Coin: Benign Familial Infantile Seizures and Paroxysmal Kinesigenic Dyskinesia Caused by <i>PRRT2</i> Mutations. <i>Archives of Neurology</i> , 2012, 69, 668.	4.9	28
28	Next-generation sequencing: impact of exome sequencing in characterizing Mendelian disorders. <i>Journal of Human Genetics</i> , 2012, 57, 621-632.	1.1	177
29	Next Generation Sequencing Methodologies - An Overview. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 1-26.	1.0	21
30	Genetic diagnosis of hyperkinetic movement disorders. <i>Expert Opinion on Medical Diagnostics</i> , 2012, 6, 439-447.	1.6	0
31	The <i>PRRT2</i> mutation c.649dupC is the so far most frequent cause of benign familial infantile convulsions. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2012, 21, 740-742.	0.9	17
32	Mutations in <i>PRRT2</i> responsible for paroxysmal kinesigenic dyskinesias also cause benign familial infantile convulsions. <i>Journal of Human Genetics</i> , 2012, 57, 338-341.	1.1	82
33	Next Generation Sequencing in the Clinical Domain: Clinical Advantages, Practical, and Ethical Challenges. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 27-63.	1.0	21
34	<i>PRRT2</i> Mutations in Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions in a Taiwanese Cohort. <i>PLoS ONE</i> , 2012, 7, e38543.	1.1	46
35	New algorithm for the diagnosis of hereditary dystonia. <i>Arquivos De Neuro-Psiquiatria</i> , 2012, 70, 715-717.	0.3	3
36	Identification of <i>PRRT2</i> as the causative gene of paroxysmal kinesigenic dyskinesia. <i>Movement Disorders</i> , 2012, 27, 707-707.	2.2	10
37	<i>PRRT2</i> Mutations are the major cause of benign familial infantile seizures. <i>Human Mutation</i> , 2012, 33, 1439-1443.	1.1	93
38	<i>PRRT2</i> Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 152-160.	2.6	234

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39	Clinical and polygraphic study of familial paroxysmal kinesigenic dyskinesia with <i>PRRT2</i> mutation. <i>Epileptic Disorders</i> , 2013, 15, 123-127.	0.7	11
40	Episodic Neurologic Disorders: Syndromes, Genes, and Mechanisms. <i>Annual Review of Neuroscience</i> , 2013, 36, 25-50.	5.0	23
41	Assessment of patients with isolated or combined dystonia: An update on dystonia syndromes. <i>Movement Disorders</i> , 2013, 28, 889-898.	2.2	88
42	Genetics of dystonia: What's known? What's new? What's next?. <i>Movement Disorders</i> , 2013, 28, 899-905.	2.2	78
43	PRRT2-related disorders: further PKD and ICCA cases and review of the literature. <i>Journal of Neurology</i> , 2013, 260, 1234-1244.	1.8	63
44	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. <i>Journal of Neurology</i> , 2013, 260, 656-660.	1.8	17
46	Episodic Movement Disorders: From Phenotype to Genotype and Back. <i>Current Neurology and Neuroscience Reports</i> , 2013, 13, 379.	2.0	9
48	Paroxysmale Dyskinesien. <i>Medizinische Genetik</i> , 2013, 25, 228-231.	0.1	0
49	Phenotypic overlap among paroxysmal dyskinesia subtypes: Lesson from a family with PRRT2 gene mutation. <i>Brain and Development</i> , 2013, 35, 664-666.	0.6	27
50	IFITMs Restrict the Replication of Multiple Pathogenic Viruses. <i>Journal of Molecular Biology</i> , 2013, 425, 4937-4955.	2.0	196
51	Genetics in Dystonia: An Update. <i>Current Neurology and Neuroscience Reports</i> , 2013, 13, 410.	2.0	13
52	Recent advances in the molecular genetics of epilepsy. <i>Journal of Medical Genetics</i> , 2013, 50, 271-279.	1.5	111
53	Benign infantile convulsions (IC) and subsequent paroxysmal kinesigenic dyskinesia (PKD) in a patient with 16p11.2 microdeletion syndrome. <i>Neurogenetics</i> , 2013, 14, 251-253.	0.7	29
54	Phenotypes and PRRT2 mutations in Chinese families with benign familial infantile epilepsy and infantile convulsions with paroxysmal choreoathetosis. <i>BMC Neurology</i> , 2013, 13, 209.	0.8	13
57	Altered intrinsic brain activity in patients with paroxysmal kinesigenic dyskinesia by PRRT2 mutation. <i>Neurological Sciences</i> , 2013, 34, 1925-1931.	0.9	21
58	Defining and refining the phenotype of <i>PRRT2</i> mutations. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 297-297.	1.1	6
59	Non-Parkinson movement disorders. <i>Neurology: Clinical Practice</i> , 2013, 3, 22-29.	0.8	4
60	Genetic and phenotypic heterogeneity in sporadic and familial forms of paroxysmal dyskinesia. <i>Journal of Neurology</i> , 2013, 260, 93-99.	1.8	48

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61	Genetic analysis of PRRT2 for benign infantile epilepsy, infantile convulsions with choreoathetosis syndrome, and benign convulsions with mild gastroenteritis. <i>Brain and Development</i> , 2013, 35, 524-530.	0.6	27
62	PRRT2 mutation in Japanese children with benign infantile epilepsy. <i>Brain and Development</i> , 2013, 35, 641-646.	0.6	31
64	PRRT2 is mutated in familial and non-familial benign infantile seizures. <i>European Journal of Paediatric Neurology</i> , 2013, 17, 77-81.	0.7	22
65	A novel mutation and functional implications of 5 variants in the PRRT2 gene in paroxysmal kinesigenic dyskinesia pedigrees. <i>Parkinsonism and Related Disorders</i> , 2013, 19, 639-642.	1.1	8
66	Mutations in PRRT2 result in familial infantile seizures with heterogeneous phenotypes including febrile convulsions and probable SUDEP. <i>Epilepsy Research</i> , 2013, 104, 280-284.	0.8	29
67	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. <i>Epilepsy and Behavior</i> , 2013, 26, 241-246.	0.9	6
68	PRRT2 mutation causes paroxysmal kinesigenic dyskinesia and hemiplegic migraine in monozygotic twins. <i>European Journal of Paediatric Neurology</i> , 2013, 17, 254-258.	0.7	19
69	Missense mutations of the proline-rich transmembrane protein 2 gene cosegregate with mild paroxysmal kinesigenic dyskinesia and infantile convulsions in a Chinese pedigree. <i>Parkinsonism and Related Disorders</i> , 2013, 19, 402-403.	1.1	7
70	PRRT2 c.649dupC Mutation Derived from De Novo in Paroxysmal Kinesigenic Dyskinesia. <i>CNS Neuroscience and Therapeutics</i> , 2013, 19, 61-65.	1.9	21
71	Clinical features of childhood-onset paroxysmal kinesigenic dyskinesia with PRRT2 gene mutations. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 327-334.	1.1	39
72	Novel PRRT2 mutations in paroxysmal dyskinesia patients with variant inheritance and phenotypes. <i>Genes, Brain and Behavior</i> , 2013, 12, 234-240.	1.1	42
74	Role of PRRT2 in common paroxysmal neurological disorders: a gene with remarkable pleiotropy. <i>Journal of Medical Genetics</i> , 2013, 50, 133-139.	1.5	88
75	The genetics of dystonia: new twists in an old tale. <i>Brain</i> , 2013, 136, 2017-2037.	3.7	102
76	PRRT2 mutation correlated with phenotype of paroxysmal kinesigenic dyskinesia and drug response. <i>Neurology</i> , 2013, 80, 1534-1535.	1.5	55
77	Genetic Issues in the Diagnosis of Dystonias. <i>Frontiers in Neurology</i> , 2013, 4, 34.	1.1	27
78	Assessing association between protein truncating variants and quantitative traits. <i>Bioinformatics</i> , 2013, 29, 2419-2426.	1.8	12
79	Pediatric epilepsy genetics. <i>Current Opinion in Neurology</i> , 2013, 26, 137-145.	1.8	17
80	Genetics of the epilepsies. <i>Current Opinion in Neurology</i> , 2013, 26, 179-185.	1.8	75

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81	Primary and secondary dystonic syndromes. <i>Current Opinion in Neurology</i> , 2013, 26, 406-412.	1.8	13
82	<scp> <i>PRRT2</i> </scp> mutations and paroxysmal disorders. <i>European Journal of Neurology</i> , 2013, 20, 872-878.	1.7	95
83	Recent advances in the management of choreas. <i>Therapeutic Advances in Neurological Disorders</i> , 2013, 6, 117-127.	1.5	14
86	<i>PRRT2</i> gene mutations in familial and sporadic paroxysmal kinesigenic dyskinesia cases. <i>Movement Disorders</i> , 2013, 28, 1313-1314.	2.2	5
87	Mutations of prolineâ€rich transmembrane proteinâ€2 and paroxysmal kinesigenic dyskinesia in Taiwan. <i>Movement Disorders</i> , 2013, 28, 1459-1460.	2.2	3
88	Altered inhibitory modulation of somatosensory cortices in paroxysmal kinesigenic dyskinesia. <i>Movement Disorders</i> , 2013, 28, 1728-1731.	2.2	17
90	A Mutation Hot-Spot for Benign Infantile Epilepsy. <i>Epilepsy Currents</i> , 2013, 13, 20-21.	0.4	1
91	Successful control with carbamazepine of family with paroxysmal kinesigenic dyskinesia of PRRT2		

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103	Evaluation of <i>SLC20A2</i> mutations that cause idiopathic basal ganglia calcification in Japan. <i>Neurology</i> , 2014, 82, 705-712.	1.5	71
104	Common DNA methylation alterations in multiple brain regions in autism. <i>Molecular Psychiatry</i> , 2014, 19, 862-871.	4.1	279
105	Primary dystonias and genetic disorders with dystonia as clinical feature of the disease. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 79-105.	0.7	14
106	Psychogenic paroxysmal movement disorders – Clinical features and diagnostic clues. <i>Parkinsonism and Related Disorders</i> , 2014, 20, 41-46.	1.1	77
107	Genotype–phenotype correlation in a cohort of paroxysmal kinesigenic dyskinesia cases. <i>Journal of the Neurological Sciences</i> , 2014, 340, 91-93.	0.3	21
108	Genetics in dystonia. <i>Parkinsonism and Related Disorders</i> , 2014, 20, S137-S142.	1.1	79
109	Genetics of Huntington's disease and related disorders. <i>Drug Discovery Today</i> , 2014, 19, 985-989.	3.2	13
110	Exome sequencing greatly expedites the progressive research of Mendelian diseases. <i>Frontiers of Medicine</i> , 2014, 8, 42-57.	1.5	48
111	Genetic Biomarkers in Epilepsy. <i>Neurotherapeutics</i> , 2014, 11, 324-333.	2.1	26
112	The Phenomenology of Functional (Psychogenic) Dystonia. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 36-44.	0.8	40
113	Genetic Epilepsy Syndromes Without Structural Brain Abnormalities: Clinical Features and Experimental Models. <i>Neurotherapeutics</i> , 2014, 11, 269-285.	2.1	51
114	The hidden genetics of epilepsy – a clinically important new paradigm. <i>Nature Reviews Neurology</i> , 2014, 10, 283-292.	4.9	232
115	Girl with a <i>PRRT2</i> mutation and infantile focal epilepsy with bilateral spikes. <i>Brain and Development</i> , 2014, 36, 342-345.	0.6	12
116	Genomic biomarkers of SUDEP in brain and heart. <i>Epilepsy and Behavior</i> , 2014, 38, 172-179.	0.9	66
117	<i>PRRT2</i> mutations: exploring the phenotypical boundaries. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 462-465.	0.9	27
118	Mutations in <i>STX1B</i> , encoding a presynaptic protein, cause fever-associated epilepsy syndromes. <i>Nature Genetics</i> , 2014, 46, 1327-1332.	9.4	178
119	Child Neurology: <i>PRRT2</i> -associated movement disorders and differential diagnoses. <i>Neurology</i> , 2014, 83, 1680-1683.	1.5	13
120	<i>PRRT2</i> : A major cause of infantile epilepsy and other paroxysmal disorders of childhood. <i>Progress in Brain Research</i> , 2014, 213, 141-158.	0.9	27

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121	Next Generation Sequencing and the Future of Genetic Diagnosis. <i>Neurotherapeutics</i> , 2014, 11, 699-707.	2.1	126
122	Benign infantile convulsion as a diagnostic clue of paroxysmal kinesigenic dyskinesia: a case series. <i>Journal of Medical Case Reports</i> , 2014, 8, 174.	0.4	4
123	New technologies in molecular genetics. <i>Progress in Brain Research</i> , 2014, 213, 253-278.	0.9	6
124	Re-evaluation of PRRT2 mutations in paroxysmal disorders. <i>Journal of Neurology</i> , 2014, 261, 951-953.	1.8	6
125	Recent Advances in the Genetics of Dystonia. <i>Current Neurology and Neuroscience Reports</i> , 2014, 14, 462.	2.0	19
126	The expanding universe of disorders of the basal ganglia. <i>Lancet, The</i> , 2014, 384, 523-531.	6.3	155
127	Epilepsy: Old Syndromes, New Genes. <i>Current Neurology and Neuroscience Reports</i> , 2014, 14, 447.	2.0	17
128	Paroxysmal dyskinesias revisited: A review of 500 genetically proven cases and a new classification. <i>Movement Disorders</i> , 2014, 29, 1108-1116.	2.2	224
129	Atypical course in individuals from Spanish families with benign familial infantile seizures and mutations in the PRRT2 gene. <i>Epilepsy Research</i> , 2014, 108, 1274-1278.	0.8	15
131	Clinicogenetic comparisons of paroxysmal kinesigenic dyskinesia patients with and without PRRT2 mutations. <i>European Journal of Neurology</i> , 2014, 21, 674-678.	1.7	32
133	Paroxysmal Dyskinesias. <i>Journal of Pediatric Neurology</i> , 2015, 13, 225-230.	0.0	2
135	Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. <i>PLoS ONE</i> , 2015, 10, e0118946.	1.1	13
136	Genetics of Dystonia. , 2015, , 27-48.		0
137	Severe phenotypic spectrum of biallelic mutations in PRRT2 gene. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 782-785.	0.9	72
138	The contribution of next generation sequencing to epilepsy genetics. <i>Expert Review of Molecular Diagnostics</i> , 2015, 15, 1531-1538.	1.5	68
139	Urine-derived induced pluripotent stem cells as a modeling tool for paroxysmal kinesigenic dyskinesia. <i>Biology Open</i> , 2015, 4, 1744-1752.	0.6	24
141	Diagnosis and Treatment of Chorea Syndromes. <i>Current Neurology and Neuroscience Reports</i> , 2015, 15, 514.	2.0	80
142	Five cases of paroxysmal kinesigenic dyskinesia by genetic diagnosis. <i>Experimental and Therapeutic Medicine</i> , 2015, 9, 909-912.	0.8	7



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143	Epilepsy genetics: The ongoing revolution. <i>Revue Neurologique</i> , 2015, 171, 539-557.	0.6	10
144	Paroxysmal Kinesigenic Dyskinesia: Seeing Is Believing. <i>Pediatric Neurology</i> , 2015, 53, 369-370.	1.0	3
145	PRRT2 Mutant Leads to Dysfunction of Glutamate Signaling. <i>International Journal of Molecular Sciences</i> , 2015, 16, 9134-9151.	1.8	64
146	Mouse Models of Dystonia. , 2015, , 465-481.		3
147	Dystonia and Dystonic Syndromes. , 2015, , .		3
149	Increased interhemispheric resting-state functional connectivity in paroxysmal kinesigenic dyskinesia: A resting-state fMRI study. <i>Journal of the Neurological Sciences</i> , 2015, 351, 93-98.	0.3	21
150	Treatment of Paroxysmal Dyskinesias in Children. <i>Current Treatment Options in Neurology</i> , 2015, 17, 350.	0.7	18
151	16p11.2 Microdeletion/Microduplication Syndrome and Benign Infantile Epilepsy. <i>Journal of Pediatric Epilepsy</i> , 2015, 04, 035-040.	0.1	0
152	Paroxysmal kinesigenic dyskinesia. <i>Neurology</i> , 2015, 85, 1546-1553.	1.5	72
153	Reduced Penetrance of <i>PRRT2</i> Mutation in a Chinese Family With Infantile Convulsion and Choreoathetosis Syndrome. <i>Journal of Child Neurology</i> , 2015, 30, 1263-1269.	0.7	13
154	Genetics of Paroxysmal Dyskinesia. , 2015, , 191-211.		1
155	Our panel of experts highlight the most important research articles across the spectrum of topics relevant to the field of neurodegenerative disease management. <i>Neurodegenerative Disease Management</i> , 2015, 5, 279-281.	1.2	0
156	The evolving spectrum of <i>PRRT2</i> -associated paroxysmal diseases. <i>Brain</i> , 2015, 138, 3476-3495.	3.7	218
157	Characteristics of patients with benign partial epilepsy in infancy without <i>PRRT2</i> mutations. <i>Epilepsy Research</i> , 2015, 118, 10-13.	0.8	8
158	The clinical and genetic heterogeneity of paroxysmal dyskinesias. <i>Brain</i> , 2015, 138, 3567-3580.	3.7	129
159	Paroxysmal Movement Disorders. , 2015, , 767-778.		4
161	Genetics of Huntington Disease (HD), HD-Like Disorders, and Other Choreiform Disorders. , 2015, , 519-532.		0
162	Episodic and Electrical Nervous System Disorders Caused by Nonchannel Genes. <i>Annual Review of Physiology</i> , 2015, 77, 525-541.	5.6	9

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163	Mutation Analysis of MR-1, SLC2A1, and CLCN1 in 28 PRRT2-negative Paroxysmal Kinesigenic Dyskinesia Patients. Chinese Medical Journal, 2016, 129, 1017-1021.	0.9	21
164	Paroxysmal Dyskinesias. , 2016, , 127-139.		0
165	PT608. Analysis of glutamate-induced processing of proline-rich transmembrane protein 2 (PRRT2). International Journal of Neuropsychopharmacology, 2016, 19, 23-23.	1.0	0
166	Genetics of movement disorders in the next generation sequencing era. Movement Disorders, 2016, 31, 458-470.	2.2	34
167	<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.	2.2	228
168	Benign infantile seizures and paroxysmal dyskinesia caused by an <i>SCN8A</i> mutation. Annals of Neurology, 2016, 79, 428-436.	2.8	159
170	Medical treatment of dystonia. Journal of Clinical Movement Disorders, 2016, 3, 19.	2.2	55
171	Novel Locus for Paroxysmal Kinesigenic Dyskinesia Mapped to Chromosome 3q28-29. Scientific Reports, 2016, 6, 25790.	1.6	5
172	Frequency-Specific Local Synchronization Changes in Paroxysmal Kinesigenic Dyskinesia. Medicine (United States), 2016, 95, e3293.	0.4	9
173	Paroxysmal hypnogenic dyskinesia is associated with mutations in the <i>PRRT2</i> gene. Neurology: Genetics, 2016, 2, e66.	0.9	31
174	PRRT2 Is a Key Component of the Ca <sup>2+</sup> -Dependent Neurotransmitter Release Machinery. Cell Reports, 2016, 15, 117-131.	2.9	121
175	Paroxysmal movement disorders: An update. Revue Neurologique, 2016, 172, 433-445.	0.6	51
176	Progressive ataxia related to PRRT2 gene mutation. Journal of the Neurological Sciences, 2016, 367, 220-221.	0.3	5
177	PRRT2: from Paroxysmal Disorders to Regulation of Synaptic Function. Trends in Neurosciences, 2016, 39, 668-679.	4.2	68
178	Genetic insights into migraine and glutamate: a protagonist driving the headache. Journal of the Neurological Sciences, 2016, 367, 258-268.	0.3	19
179	Intronic PRRT2 mutation generates novel splice acceptor site and causes paroxysmal kinesigenic dyskinesia with infantile convulsions (PKD/IC) in a three generation family. BMC Medical Genetics, 2016, 17, 16.	2.1	5
181	Identification of a Premature Termination Mutation in the Proline-Rich Transmembrane Protein 2 Gene in a Chinese Family with Febrile Seizures. Molecular Neurobiology, 2016, 53, 835-841.	1.9	20
182	A Novel Topology of Proline-rich Transmembrane Protein 2 (PRRT2). Journal of Biological Chemistry, 2016, 291, 6111-6123.	1.6	59

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183	Exome sequencing explained: a practical guide to its clinical application. <i>Briefings in Functional Genomics</i> , 2016, 15, 374-384.	1.3	58
184	Focal seizures and epileptic spasms in a child with Down syndrome from a family with a PRRT2 mutation. <i>Brain and Development</i> , 2016, 38, 597-600.	0.6	7
185	Expanding phenotype of PRRT2 gene mutations: A new case with epilepsy and benign myoclonus of early infancy. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 454-456.	0.7	21
186	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 152-157.	0.7	14
187	Mutation screening of the PRRT2 gene for benign epilepsy with centrotemporal spikes in Chinese mainland population. <i>International Journal of Neuroscience</i> , 2017, 127, 10-13.	0.8	2
188	The PRRT2 knockout mouse recapitulates the neurological diseases associated with PRRT2 mutations. <i>Neurobiology of Disease</i> , 2017, 99, 66-83.	2.1	72
189	Pharmacokinetics and Pharmacogenetics of Carbamazepine in Children. <i>European Journal of Drug Metabolism and Pharmacokinetics</i> , 2017, 42, 729-744.	0.6	23
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191	Munchausen syndrome by genetics: Next-generation challenges for clinicians. <i>Neurology</i> , 2017, 88, 1000-1001.	1.5	9
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