Naomichi Matsumoto

List of Publications by Year
in descending order

207	8,186	61857	64668
papers	citations	43 h-index	79 g-index

217	217	217	14982
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	1.5	4
2	Gait disturbance in a patient with de novo 1.0-kb SOX2 microdeletion. Brain and Development, 2022, 44, 68-72.	0.6	1
3	Expanding the phenotypic spectrum of cardiospondylocarpofacial syndrome: From a detailed clinical and radiological observation of a boy with a novel missense variant in <scp>MAP3K7</scp>. American Journal of Medical Genetics, Part A, 2022, 188, 350-356.	0.7	3
4	Biallelic null variants in ZNF142 cause global developmental delay with familial epilepsy and dysmorphic features. Journal of Human Genetics, 2022, 67, 169-173.	1.1	7
5	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome. Brain, 2022, 145, 1139-1150.	3.7	19
6	Duplications in the G3 domain or switch II region in HRAS identified in patients with Costello syndrome. Human Mutation, 2022, 43, 3-15.	1.1	7
7	GGC Repeat Expansion of NOTCH2NLC in Taiwanese Patients With Inherited Neuropathies. Neurology, 2022, 98, .	1.5	24
8	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
9	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant. Human Genetics, 2022, 141, 283-293.	1.8	6
10	Severe cardiac defect in Cornelia de Lange syndrome from a novel SMC1A variant. Pediatrics International, 2022, 64, e15031.	0.2	1
11	A case of <scp>VEXAS</scp> syndrome with Sweet's disease and pulmonary involvement. Journal of Dermatology, 2022, 49, .	0.6	4
12	Six yearsâ€™ accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. Journal of Human Genetics, 2022, 67, 505-513.	1.1	17
13	Hornerin deposits in neuronal intranuclear inclusion disease: direct identification of proteins with compositionally biased regions in inclusions. Acta Neuropathologica Communications, 2022, 10, 28.	2.4	4
14	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
15	BehÃ§et's disease with a somatic UBA1 variant:Expanding spectrum of autoinflammatory phenotypes of VEXAS syndrome. Clinical Immunology, 2022, 238, 108996.	1.4	20
16	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
17	A homozygous <scp>ABHD16A</scp> variant causes a complex hereditary spastic paraplegia with developmental delay, absent speech, and characteristic face. Clinical Genetics, 2022, 101, 359-363.	1.0	2
18	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	3.7	11

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19	Neuronal intranuclear inclusion disease in patients with adult-onset non-vascular leukoencephalopathy. Brain, 2022, 145, 3010-3021.	3.7	28
20	Large-scale discovery of novel neurodevelopmental disorder-related genes through a unified analysis of single-nucleotide and copy number variants. Genome Medicine, 2022, 14, 40.	3.6	13
21	Long-term course of early onset developmental and epileptic encephalopathy associated with 2q24.3 microduplication. Epilepsy and Behavior Reports, 2022, 19, 100547.	0.5	0
22	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	1.8	2
23	Perampanel markedly improved clinical seizures in a patient with a Rettâ€like phenotype and 960â€kb deletion on chromosome 9q34.11 including the STXBP1. Clinical Case Reports (discontinued), 2022, 10, .	0.2	2
24	Clinical course of a Japanese patient with developmental delay linked to a small 6q16.1 deletion. Human Genome Variation, 2022, 9, 14.	0.4	2
25	Genetic and Imaging Characteristics of a Family With Neuronal Intranuclear Inclusion Disease.		

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37	Preliminary report for Epilepsia Open A case of West syndrome with severe global developmental delay and confirmed KIF5A gene variant. Epilepsia Open, 2021, 6, 230-234.	1.3	0
38	Association of early-onset epileptic encephalopathy with involuntary movements â€“ Case series and literature review. Epilepsy and Behavior Reports, 2021, 15, 100417.	0.5	4
39	Genome-wide survey of tandem repeats by nanopore sequencing shows that disease-associated repeats are more polymorphic in the general population. BMC Medical Genomics, 2021, 14, 17.	0.7	9
40	Comprehensive Genetic Analysis of Non-syndromic Autism Spectrum Disorder in Clinical Settings. Journal of Autism and Developmental Disorders, 2021, 51, 4655-4662.	1.7	4
41	Limb-clasping, cognitive deficit and increased vulnerability to kainic acid-induced seizures in neuronal glycosylphosphatidylinositol deficiency mouse models. Human Molecular Genetics, 2021, 30, 758-770.	1.4	7
42	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	1.8	4
43	Pathogenic UBA1 variants associated with VEXAS syndrome in Japanese patients with relapsing polychondritis. Annals of the Rheumatic Diseases, 2021, 80, 1057-1061.	0.5	88
44	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	4.7	13
45	Complete sequencing of expandedSAMD12repeats by long-read sequencing and Cas9-mediated enrichment. Brain, 2021, 144, 1103-1117.	3.7	25
46	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. Nature Communications, 2021, 12, 2046.	5.8	7
47	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. Nature Communications, 2021, 12, 2107.	5.8	30
48	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. Brain and Development, 2021, 43, 505-514.	0.6	14
49	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	1.8	18
50	<scp>COG1â€</scp>congenital disorders of glycosylation: Milder presentation and review. Clinical Genetics, 2021, 100, 318-323.	1.0	5
51	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. Med, 2021, 2, 611-632.e9.	2.2	1
52	Cerebrovascular diseases in two patients with entire NSD1 deletion. Human Genome Variation, 2021, 8, 20.	0.4	2
53	De novo pathogenic <scp>DHX30</scp> variants in two cases. Clinical Genetics, 2021, 100, 350-351.	1.0	1
54	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. Nature Communications, 2021, 12, 3750.	5.8	15

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55	Progressive cerebral atrophies in three children with COL4A1 mutations. Brain and Development, 2021, 43, 1033-1038.	0.6	4
56	A 23-year follow-up report of juvenile-onset Sandhoff disease presenting with a motor neuron disease phenotype and a novel variant. Brain and Development, 2021, 43, 1029-1032.	0.6	1
57	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	1.1	4
58	A novel LRP6 variant in a Japanese family with oligodontia. Human Genome Variation, 2021, 8, 30.	0.4	7
59	Clinical course of epilepsy and white matter abnormality linked to a novel DYRK1A variant. Human Genome Variation, 2021, 8, 26.	0.4	1
60	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	0.7	6
61	Intellectual disability and microcephaly associated with a novel CHAMP1 mutation. Human Genome Variation, 2021, 8, 34.	0.4	3
62	Expanding the phenotypic spectrum of TNFRSF11A-associated dysosteosclerosis: a case with intracranial extramedullary hematopoiesis. Journal of Human Genetics, 2021, 66, 607-611.	1.1	6
63	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	4.7	25
64	Long-read whole-genome sequencing identified a partial MBD5 deletion in an exome-negative patient with neurodevelopmental disorder. Journal of Human Genetics, 2021, 66, 697-705.	1.1	8
65	Multiple alterations in glutamatergic transmission and dopamine D2 receptor splicing in induced pluripotent stem cell-derived neurons from patients with familial schizophrenia. Translational Psychiatry, 2021, 11, 548.	2.4	6
66	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ^'Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	1.5	18
67	Father-to-offspring transmission of extremely long NOTCH2NLC repeat expansions with contractions: genetic and epigenetic profiling with long-read sequencing. Clinical Epigenetics, 2021, 13, 204.	1.8	22
68	Long-read sequencing for rare human genetic diseases. Journal of Human Genetics, 2020, 65, 11-19.	1.1	72
69	An atypical case of KMT2Bâ€related dystonia manifesting asterixis and effect of deep brain stimulation of the globus pallidus. Neurology and Clinical Neuroscience, 2020, 8, 36-38.	0.2	4
70	Two males with sick sinus syndrome in a family with 0.6â€¯kb deletions involving major domains in MECP2. European Journal of Medical Genetics, 2020, 63, 103769.	0.7	0
71	Phenotypeâ€“genotype correlations in patients with GNB1 gene variants, including the first three reported Japanese patients to exhibit spastic diplegia, dyskinetic quadriplegia, and infantile spasms. Brain and Development, 2020, 42, 199-204.	0.6	16
72	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. American Journal of Human Genetics, 2020, 106, 13-25.	2.6	25

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73	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	1.1	6
74	Cerebrospinal fluid abnormalities in developmental and epileptic encephalopathy with a de novo CDK19 variant. Neurology: Genetics, 2020, 6, e527.	0.9	1
75	Clinical and genetic characteristics of patients with Doose syndrome. Epilepsia Open, 2020, 5, 442-450.	1.3	8
76	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. Journal of the Neurological Sciences, 2020, 416, 117047.	0.3	8
77	A 2â€yearâ€old patient with a diffuse intrinsic pontine glioma and radiationâ€induced moyamoya syndrome. Pediatric Blood and Cancer, 2020, 67, e28618.	0.8	0
78	A pipeline for complete characterization of complex germline rearrangements from long DNA reads. Genome Medicine, 2020, 12, 67.	3.6	27
79	GNAO1organizes the cytoskeletal remodeling and firing of developing neurons. FASEB Journal, 2020, 34, 16601-16621.	0.2	14
80	De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107137.	1.5	3
81	Leggâ€CalvÃ©â€Perthes disease in a patient with Bardetâ€Biedl syndrome: A case report of a novelMKKS/BBS6mutation. Clinical Case Reports (discontinued), 2020, 8, 3110-3115.	0.2	1
82	Clonazepam as an Effective Treatment for Epilepsy in a Female Patient with <i>NEXMIF</i> Mutation: Case Report. Molecular Syndromology, 2020, 11, 232-238.	0.3	6
83	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	1.1	13
84	Reply to â€œ<scp>GGC</scp> Repeat Expansion of <scp>NOTCH2NLC</scp> is Rare in European Leukoencephalopathyâ€• Annals of Neurology, 2020, 88, 642-643.	2.8	2
85	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
86	De Novo Truncating Variants in the Last Exon of SEMA6B Cause Progressive Myoclonic Epilepsy. American Journal of Human Genetics, 2020, 106, 549-558.	2.6	32
87	Delineation of musculocontractural Ehlersâ€“Danlos Syndrome caused by dermatan sulfate epimerase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1197.	0.6	18
88	Fifteen-year follow-up of a patient with a DHDDS variant with non-progressive early onset myoclonic tremor and rare generalized epilepsy. Brain and Development, 2020, 42, 696-699.	0.6	14
89	Long-read sequencing identifies the pathogenic nucleotide repeat expansion in RFC1 in a Japanese case of CANVAS. Journal of Human Genetics, 2020, 65, 475-480.	1.1	35
90	A message for 2020. Journal of Human Genetics, 2020, 65, 351-353.	1.1	0

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91	Long-read DNA sequencing fully characterized chromothripsis in a patient with Langerâ€“Giedion syndrome and Cornelia de Lange syndrome-4. Journal of Human Genetics, 2020, 65, 667-674.	1.1	24
92	Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. Science Advances, 2020, 6, .	4.7	39
93	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. Neurology: Genetics, 2020, 6, e531.	0.9	20
94	A novel homozygous mutation of CLCN2 in a patient with characteristic brain MRI images â€“ A first case of CLCN2-related leukoencephalopathy in Japan. Brain and Development, 2019, 41, 101-105.	0.6	9
95	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98
96	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. Journal of Human Genetics, 2019, 64, 1005-1014.	1.1	9
97	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	1.1	11
98	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. Nature Genetics, 2019, 51, 1215-1221.	9.4	328
99	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	1.1	43
100	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	5.8	21
101	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. Genetics in Medicine, 2019, 21, 2734-2743.	1.1	33
102	Recurrent NUS1 canonical splice donor site mutation in two unrelated individuals with epilepsy, myoclonus, ataxia and scoliosis - a case report. BMC Neurology, 2019, 19, 253.	0.8	20
103	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
104	Different types of suppression-burst patterns in patients with epilepsy of infancy with migrating focal seizures (EIMFS). Seizure: the Journal of the British Epilepsy Association, 2019, 65, 118-123.	0.9	5
105	Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. Journal of Human Genetics, 2019, 64, 955-960.	1.1	28
106	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	5.8	46
107	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	2.6	35
108	Rapid progression of a walking disability in a 5-year-old boy with a CLN6 mutation. Brain and Development, 2019, 41, 726-730.	0.6	7

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109	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	3.8	103
110	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
111	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	2.6	92
112	Recurrent de novo MAPK8IP3 variants cause neurological phenotypes. Annals of Neurology, 2019, 85, 927-933.	2.8	34
113	A 12-kb structural variation in progressive myoclonic epilepsy was newly identified by long-read whole-genome sequencing. Journal of Human Genetics, 2019, 64, 359-368.	1.1	48
114	Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.	0.9	10
115	Detecting a long insertion variant in SAMD12 by SMRT sequencing: implications of long-read whole-genome sequencing for repeat expansion diseases. Journal of Human Genetics, 2019, 64, 191-197.	1.1	33
116	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
117	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. Journal of Human Genetics, 2019, 64, 313-322.	1.1	51
118	Cancer Management in Kabuki Syndrome: The First Case of Wilms Tumor and a Literature Review. Journal of Pediatric Hematology/Oncology, 2018, 40, 391-394.	0.3	13
119	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	2.8	60
120	De novo variants in <scp>CAMK</scp>2A and <scp>CAMK</scp>2B cause neurodevelopmental disorders. Annals of Clinical and Translational Neurology, 2018, 5, 280-296.	1.7	65
121	Earlyâ€onset epileptic encephalopathy and severe developmental delay in an association with de novo double mutations in NF1 and MAGEL2. Epilepsia Open, 2018, 3, 81-85.	1.3	4
122	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	3.7	69
123	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. Brain and Development, 2018, 40, 493-497.	0.6	11
124	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	1.4	36
125	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
126	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. Human Genetics, 2018, 137, 95-104.	1.8	60

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127	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation. Brain and Development, 2018, 40, 728-732.	0.6	13
128	Dysosteosclerosis is also caused by TNFRSF11A mutation. Journal of Human Genetics, 2018, 63, 769-774.	1.1	21
129	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies. Brain and Development, 2018, 40, 406-409.	0.6	12
130	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
131	GRIN2D variants in three cases of developmental and epileptic encephalopathy. Clinical Genetics, 2018, 94, 538-547.	1.0	17
132	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. Journal of Human Genetics, 2018, 63, 1223-1229.	1.1	16
133	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25
134	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
135	A recurrent homozygous NHLRC1 variant in siblings with Lafora disease. Human Genome Variation, 2018, 5, 16.	0.4	13
136	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
137	A novel CYCS mutation in the Î±â€helix of the CYCS Câ€terminal domain causes nonâ€syndromic thrombocytopenia. Clinical Genetics, 2018, 94, 548-553.	1.0	20
138	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
139	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	1.0	28
140	Dystonia due to bilateral caudate hemorrhage associated with a COL4A1 mutation. Parkinsonism and Related Disorders, 2017, 40, 80-82.	1.1	4
141	A severe pulmonary complication in a patient with COL4A1 -related disorder: A case report. European Journal of Medical Genetics, 2017, 60, 169-171.	0.7	11
142	Three Cases of KCNT1 Mutations: Malignant Migrating Partial Seizures in Infancy with Massive Systemic to Pulmonary Collateral Arteries. Journal of Pediatrics, 2017, 191, 270-274.	0.9	27
143	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9
144	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28

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145	The first report of Japanese patients with asparagine synthetase deficiency. Brain and Development, 2017, 39, 236-242.	0.6	25
146	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
147	Ineffective quinidine therapy in early onset epileptic encephalopathy with <scp>KCNT</scp>1 mutation. Annals of Neurology, 2016, 79, 502-503.	2.8	68
148	Dermatan 4â€Oâ€sulfotransferase 1â€deficient Ehlersâ€“Danlos syndrome complicated by a large subcutaneous hematoma on the back. Journal of Dermatology, 2016, 43, 832-833.	0.6	15
149	Ultraâ€“sensitive droplet digital PCR for detecting a lowâ€“prevalence somatic GNAQ mutation in Sturgeâ€“Weber syndrome. Scientific Reports, 2016, 6, 22985.	1.6	60
150	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
151	WDR45 mutations in three male patients with West syndrome. Journal of Human Genetics, 2016, 61, 653-661.	1.1	39
152	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. European Journal of Paediatric Neurology, 2016, 20, 412-417.	0.7	25
153	Two cases of early-onset myoclonic seizures with continuous parietal delta activity caused by EEF1A2 mutations. Brain and Development, 2016, 38, 520-524.	0.6	32
154	A female case of aromatic l-amino acid decarboxylase deficiency responsive to MAO-B inhibition. Brain and Development, 2016, 38, 959-963.	0.6	12
155	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	1.1	270
156	Novel COL4A1 mutation in an infant with severe dysmorphic syndrome with schizencephaly, periventricular calcifications, and cataract resembling congenital infection. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 304-307.	1.6	19
157	Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. European Journal of Human Genetics, 2016, 24, 129-134.	1.4	98
158	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
159	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30
160	High prevalence of genetic alterations in early-onset epileptic encephalopathies associated with infantile movement disorders. Brain and Development, 2016, 38, 285-292.	0.6	54
161	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. Journal of Human Genetics, 2016, 61, 199-206.	1.1	91
162	<scp>GRIN</scp>1 mutations cause encephalopathy with infantileâ€onset epilepsy, and hyperkinetic and stereotyped movement disorders. Epilepsia, 2015, 56, 841-848.	2.6	76

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163	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
164	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. Human Genome Variation, 2015, 2, 15024.	0.4	11
165	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). Human Genome Variation, 2015, 2, 15012.	0.4	7
166	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	2.6	95
167	De novo SHANK3 mutation causes Rett syndromeâ€like phenotype in a female patient. American Journal of Medical Genetics, Part A, 2015, 167, 1593-1596.	0.7	26
168	Somatic Mutations in the <scp>MTOR</scp> gene cause focal cortical dysplasia type <scp>II</scp>b. Annals of Neurology, 2015, 78, 375-386.	2.8	169
169	Electroclinical features of epileptic encephalopathy caused by <scp>SCN8A</scp> mutation. Pediatrics International, 2015, 57, 758-762.	0.2	13
170	A Japanese case of cerebellar ataxia, spastic paraparesis and deep sensory impairment associated with a novel homozygous TTC19 mutation. Journal of Human Genetics, 2015, 60, 187-191.	1.1	14
171	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca2+ channels. Human Molecular Genetics, 2015, 24, 637-648.	1.4	132
172	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an â€˜exome-firstâ€™ approach. Journal of Human Genetics, 2015, 60, 175-182.	1.1	54
173	A case of autism spectrum disorder arising from a de novo missense mutation in POGZ. Journal of Human Genetics, 2015, 60, 277-279.	1.1	42
174	A Novel Mutation inELOVL4Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia. JAMA Neurology, 2015, 72, 797.	4.5	79
175	DNA methylation and gene expression dynamics during spermatogonial stem cell differentiation in the early postnatal mouse testis. BMC Genomics, 2015, 16, 624.	1.2	112
176	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. Journal of Human Genetics, 2015, 60, 739-742.	1.1	58
177	Whole Exome Analysis Identifies Frequent CNGA1 Mutations in Japanese Population with Autosomal Recessive Retinitis Pigmentosa. PLoS ONE, 2014, 9, e108721.	1.1	56
178	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
179	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. Journal of Human Genetics, 2014, 59, 649-654.	1.1	46
180	Characteristic MRI findings in beta-propeller protein-associated neurodegeneration (BPAN). Neurology: Clinical Practice, 2014, 4, 175-177.	0.8	27

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