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	Haploinsufficiency of NSD1 causes Sotos syndrome. Nature Genetics, 2002, 30, 365-366.	9.4	560
2	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nature Genetics, 2008, 40, 782-788.	9.4	498
3	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	9.4	435
4	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. Nature Genetics, 2013, 45, 445-449.	9.4	396
5	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. Nature Genetics, 2019, 51, 1215-1221.	9.4	328
6	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
7	De Novo Mutations in GNAO1, Encoding a GÎ±o Subunit of Heterotrimeric G Proteins, Cause Epileptic Encephalopathy. American Journal of Human Genetics, 2013, 93, 496-505.	2.6	187
8	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
9	Phenotypic Spectrum of COL4A1 Mutations: Porencephaly to Schizencephaly. Annals of Neurology, 2013, 73, 48-57.	2.8	143
10	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. Human Mutation, 2010, 31, 966-974.	1.1	137
11	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
12	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
13	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 16139-16144.	3.3	115
14	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
15	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	3.8	103
16	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
17	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
18	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98

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19	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	2.6	95
20	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
21	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. Journal of Human Genetics, 2016, 61, 199-206.	1.1	91
22	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
23	Molecular characterization of NSD1, a human homologue of the mouse Nsd1 gene. Gene, 2001, 279, 197-204.	1.0	82
24	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
25	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76
26	<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
27	Long-read sequencing for rare human genetic diseases. Journal of Human Genetics, 2020, 65, 11-19.	1.1	72
28	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	2.6	71
29	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	3.7	69
30	Ineffective quinidine therapy in early onset epileptic encephalopathy with <scp>KCNT</scp>1 mutation. Annals of Neurology, 2016, 79, 502-503.	2.8	68
31	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
32	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
33	Delineation of dermatan 4â€Oâ€sulfotransferase 1 deficient Ehlersâ€“Danlos syndrome: Observation of two additional patients and comprehensive review of 20 reported patients. American Journal of Medical Genetics, Part A, 2011, 155, 1949-1958.	0.7	60
34	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
35	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	2.8	60
36	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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37	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
38	Whole Exome Analysis Identifies Frequent CNGA1 Mutations in Japanese Population with Autosomal Recessive Retinitis Pigmentosa. PLoS ONE, 2014, 9, e108721.	1.1	56
39	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
40	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
41	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
42	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. Journal of Human Genetics, 2014, 59, 292-295.	1.1	49
43	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
44	Narrowing candidate region for monosomy 9p syndrome to a 4.7-Mb segment at 9p22.2-p23. American Journal of Medical Genetics, Part A, 2006, 140A, 373-377.	0.7	46
45	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. Journal of Human Genetics, 2014, 59, 649-654.	1.1	46
46	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	5.8	46
47	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	1.1	43
48	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
49	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
50	<scp>SCN3A</scp>â€Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. Annals of Neurology, 2020, 88, 348-362.	2.8	42
51	WDR45 mutations in three male patients with West syndrome. Journal of Human Genetics, 2016, 61, 653-661.	1.1	39
52	Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. Science Advances, 2020, 6, .	4.7	39
53	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	1.4	36
54	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36

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55	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	2.6	35
56	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
57	Recurrent de novo MAPK8IP3 variants cause neurological phenotypes. Annals of Neurology, 2019, 85, 927-933.	2.8	34
58	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
59	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
60	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
61	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
62	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slitâ€Robo Rho GTPase activating protein 2 (SRGAP2). American Journal of Medical Genetics, Part A, 2012, 158A, 199-205.	0.7	31
63	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30
64	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
65	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
66	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
67	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. European Journal of Paediatric Neurology, 2014, 18, 567-571.	0.7	28
68	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
69	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28
70	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
71	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
72	Neuronal intranuclear inclusion disease in patients with adult-onset non-vascular leukoencephalopathy. Brain, 2022, 145, 3010-3021.	3.7	28

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73	Characteristic MRI findings in beta-propeller protein-associated neurodegeneration (BPAN). Neurology: Clinical Practice, 2014, 4, 175-177.	0.8	27
74	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
75	A pipeline for complete characterization of complex germline rearrangements from long DNA reads. Genome Medicine, 2020, 12, 67.	3.6	27
76	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
77	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. European Journal of Paediatric Neurology, 2016, 20, 412-417.	0.7	25
78	The first report of Japanese patients with asparagine synthetase deficiency. Brain and Development, 2017, 39, 236-242.	0.6	25
79	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25
80	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
81	Complete sequencing of expandedSAMD12repeats by long-read sequencing and Cas9-mediated enrichment. Brain, 2021, 144, 1103-1117.	3.7	25
82	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	4.7	25
83	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
84	GGC Repeat Expansion of NOTCH2NLC in Taiwanese Patients With Inherited Neuropathies. Neurology, 2022, 98, .	1.5	24
85	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	1.5	22
86	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
87	A response to: Loss of dermatan-4-sulfotransferase 1 (D4ST1/CHST14) function represents the first dermatan sulfate biosynthesis defect, â€œdermatan sulfate-deficient Adducted Thumb-Clubfoot Syndromeâ€• Which name is appropriate, â€œAdducted Thumb-Clubfoot Synd. Human Mutation, 2011, 32, 1507-1509.	1.1	21
88	Different patterns of cerebellar abnormality and hypomyelination between POLR3A and POLR3B mutations. Brain and Development, 2014, 36, 259-263.	0.6	21
89	Dysosteosclerosis is also caused by TNFRSF11A mutation. Journal of Human Genetics, 2018, 63, 769-774.	1.1	21
90	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	5.8	21

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91	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
92	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
93	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. Neurology: Genetics, 2020, 6, e531.	0.9	20
94	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
95	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
96	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
97	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome. Brain, 2022, 145, 1139-1150.	3.7	19
98	Breakpoint determination of X;autosome balanced translocations in four patients with premature ovarian failure. Journal of Human Genetics, 2011, 56, 156-160.	1.1	18
99	Delineation of musculocontractural Ehlersâ€“Danlos Syndrome caused by dermatan sulfate epimerase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1197.	0.6	18
100	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	1.1	18
101	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
102	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
103	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
104	Characterization of the complex 7q21.3 rearrangement in a patient with bilateral splitâ€foot malformation and hearing loss. American Journal of Medical Genetics, Part A, 2009, 149A, 1224-1230.	0.7	17
105	GRIN2D variants in three cases of developmental and epileptic encephalopathy. Clinical Genetics, 2018, 94, 538-547.	1.0	17
106	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
107	Vanoxerine, a New Drug for Terminating Atrial Fibrillation and Flutter. Journal of Cardiovascular Electrophysiology, 2010, 21, 311-319.	0.8	16
108	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. Journal of Human Genetics, 2018, 63, 1223-1229.	1.1	16

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109	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
110	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	1.1	16
111	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
112	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
113	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
114	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	1.1	15
115	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. Nature Communications, 2021, 12, 3750.	5.8	15
116	Actin-binding protein filamin-A drives tau aggregation and contributes to progressive supranuclear palsy pathology. Science Advances, 2022, 8, .	4.7	15
117	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
118	GNAO1organizes the cytoskeletal remodeling and firing of developing neurons. FASEB Journal, 2020, 34, 16601-16621.	0.2	14
119	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
120	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. Brain and Development, 2021, 43, 505-514.	0.6	14
121	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
122	A unique case of de novo 5q33.3â€“q34 triplication with uniparental isodisomy of 5q34â€“qter. American Journal of Medical Genetics, Part A, 2013, 161, 1904-1909.	0.7	13
123	Electroclinical features of epileptic encephalopathy caused by <scp>SCN8A</scp> mutation. Pediatrics International, 2015, 57, 758-762.	0.2	13
124	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
125	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation. Brain and Development, 2018, 40, 728-732.	0.6	13
126	A recurrent homozygous NHLRC1 variant in siblings with Lafora disease. Human Genome Variation, 2018, 5, 16.	0.4	13

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127	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	1.1	13
128	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	4.7	13
129	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
130	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
131	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
132	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. Human Genome Variation, 2015, 2, 15024.	0.4	11
133	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
134	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. Brain and Development, 2018, 40, 493-497.	0.6	11
135	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
136	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	3.7	11
137	Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.	0.9	10
138	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
139	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9
140	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
141	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. Journal of Human Genetics, 2019, 64, 1005-1014.	1.1	9
142	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
143	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
144	Clinical and genetic characteristics of patients with Doose syndrome. Epilepsia Open, 2020, 5, 442-450.	1.3	8

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145	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. Journal of the Neurological Sciences, 2020, 416, 117047.	0.3	8
146	The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. Journal of Human Genetics, 2021, 66, 371-377.	1.1	8
147	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
148	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). Human Genome Variation, 2015, 2, 15012.	0.4	7
149	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
150	Clinical variations of epileptic syndrome associated with PACS2 variant. Brain and Development, 2021, 43, 343-347.	0.6	7
151	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
152	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. Nature Communications, 2021, 12, 2046.	5.8	7
153	A novel LRP6 variant in a Japanese family with oligodontia. Human Genome Variation, 2021, 8, 30.	0.4	7
154	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
155	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
156	Case Report: Coexistence of Multiple Myeloma and Auricular Chondritis in VEXAS Syndrome. Frontiers in Immunology, 0, 13, .	2.2	7
157	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	1.1	6
158	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
159	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	0.7	6
160	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
161	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
162	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

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163	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
164	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. Brain and Development, 2021, 43, 475-481.	0.6	5
165	<scp>COG1â€</scp>congenital disorders of glycosylation: Milder presentation and review. Clinical Genetics, 2021, 100, 318-323.	1.0	5
166	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
167	Dystonia due to bilateral caudate hemorrhage associated with a COL4A1 mutation. Parkinsonism and Related Disorders, 2017, 40, 80-82.	1.1	4
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