Naomichi Matsumoto

List of Publications by Citations

199 papers	5,489 citations	38 h-index	69 g-index
217 ext. papers	7,064 ext. citations	6 avg, IF	5.15 L-index

#	Paper	IF	Citations
199	Haploinsufficiency of NSD1 causes Sotos syndrome. *Nature Genetics*, **2002**, 30, 365-6	36.3	453
198	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. *Nature Genetics*, **2008**, 40, 782-8	36.3	416
197	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. *Nature Genetics*, **2012**, 44, 376-8	36.3	350
196	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. *Nature Genetics*, **2013**, 45, 445-9, 449e1	36.3	330
195	Human genetic variation database, a reference database of genetic variations in the Japanese population. *Journal of Human Genetics*, **2016**, 61, 547-53	4.3	212
194	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. *Nature Genetics*, **2019**, 51, 1215-1221	36.3	164
193	De Novo mutations in GNAO1, encoding a Gβ subunit of heterotrimeric G proteins, cause epileptic encephalopathy. *American Journal of Human Genetics*, **2013**, 93, 496-505	11	137
192	Somatic Mutations in the MTOR gene cause focal cortical dysplasia type IIb. *Annals of Neurology*, **2015**, 78, 375-86	9.4	129
191	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. *Human Mutation*, **2010**, 31, 966-74	4.7	119
190	Phenotypic spectrum of COL4A1 mutations: porencephaly to schizencephaly. *Annals of Neurology*, **2013**, 73, 48-57	9.4	117
189	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca□+ channels. *Human Molecular Genetics*, **2015**, 24, 637-48	5.6	104
188	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, 16139-44	11.5	104
187	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. *Cell Reports*, **2018**, 22, 734-747	10.6	86
186	DNA methylation and gene expression dynamics during spermatogonial stem cell differentiation in the early postnatal mouse testis. *BMC Genomics*, **2015**, 16, 624	4.5	84
185	Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. *European Journal of Human Genetics*, **2016**, 24, 129-34	5.3	72
184	De novo KCNT1 mutations in early-onset epileptic encephalopathy. *Epilepsia*, **2015**, 56, e121-8	6.4	66
183	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia*, **2013**, 54, 1262-9	6.4	64

182	Molecular characterization of NSD1, a human homologue of the mouse Nsd1 gene. *Gene*, **2001**, 279, 197-204	3.2	63
181	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. *American Journal of Human Genetics*, **2016**, 98, 615-26	11	59
180	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	11	56
179	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
178	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. *Genome Biology*, **2019**, 20, 58	18.3	53
177	A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia: A Broadened Spectrum of SCA34. *JAMA Neurology*, **2015**, 72, 797-805	17.2	53
176	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. *Journal of Human Genetics*, **2016**, 61, 199-206	4.3	52
175	Delineation of dermatan 4-O-sulfotransferase 1 deficient Ehlers-Danlos syndrome: observation of two additional patients and comprehensive review of 20 reported patients. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1949-58	2.5	52
174	Ineffective quinidine therapy in early onset epileptic encephalopathy with KCNT1 mutation. *Annals of Neurology*, **2016**, 79, 502-3	9.4	52
173	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. *Scientific Reports*, **2016**, 6, 30072	4.9	52
172	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. *Annals of Neurology*, **2019**, 86, 962-968	9.4	49
171	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. *Brain*, **2018**, 141, 1703-1718	11.2	44
170	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. *Journal of Human Genetics*, **2014**, 59, 292-5	4.3	44
169	Whole exome analysis identifies frequent CNGA1 mutations in Japanese population with autosomal recessive retinitis pigmentosa. *PLoS ONE*, **2014**, 9, e108721	3.7	44
168	Biallelic mutations in the 3′exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464	36.3	43
167	Ultra-sensitive droplet digital PCR for detecting a low-prevalence somatic GNAQ mutation in Sturge-Weber syndrome. *Scientific Reports*, **2016**, 6, 22985	4.9	43
166	High prevalence of genetic alterations in early-onset epileptic encephalopathies associated with infantile movement disorders. *Brain and Development*, **2016**, 38, 285-92	2.2	42
165	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an ‘exome-first’ approach. *Journal of Human Genetics*, **2015**, 60, 175-82	4.3	41

164	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**, 140, 373-7	2.5	41
163	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. *Journal of Human Genetics*, **2014**, 59, 649-54	4.3	40
162	Long-read sequencing for rare human genetic diseases. *Journal of Human Genetics*, **2020**, 65, 11-19	4.3	40
161	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. *Journal of Human Genetics*, **2015**, 60, 739-42	4.3	37
160	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37
159	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. *Human Genetics*, **2018**, 137, 95-104	6.3	36
158	WDR45 mutations in three male patients with West syndrome. *Journal of Human Genetics*, **2016**, 61, 653-61	4.3	36
157	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	4.3	36
156	A case of autism spectrum disorder arising from a de novo missense mutation in POGZ. *Journal of Human Genetics*, **2015**, 60, 277-9	4.3	33
155	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. *Journal of Human Genetics*, **2019**, 64, 313-322	4.3	29
154	variants in and cause neurodevelopmental disorders. *Annals of Clinical and Translational Neurology*, **2018**, 5, 280-296	5.3	27
153	Two cases of early-onset myoclonic seizures with continuous parietal delta activity caused by EEF1A2 mutations. *Brain and Development*, **2016**, 38, 520-4	2.2	27
152	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. *Journal of Human Genetics*, **2019**, 64, 967-978	4.3	25
151	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slit-Robo Rho GTPase activating protein 2 (SRGAP2). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 199-205	2.5	25
150	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. *Journal of Human Genetics*, **2016**, 61, 527-31	4.3	24
149	Characteristic MRI findings in beta-propeller protein-associated neurodegeneration (BPAN). *Neurology: Clinical Practice*, **2014**, 4, 175-177	1.7	24
148	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. *Human Molecular Genetics*, **2018**, 27, 1421-1433	5.6	23
147	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. *European Journal of Paediatric Neurology*, **2014**, 18, 567-71	3.8	23

146	Pathogenic variants associated with VEXAS syndrome in Japanese patients with relapsing polychondritis. *Annals of the Rheumatic Diseases*, **2021**,	2.4	23
145	A novel mutation in SLC1A3 causes episodic ataxia. *Journal of Human Genetics*, **2018**, 63, 207-211	4.3	23
144	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. *Nature Communications*, **2019**, 10, 2506	17.4	22
143	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. *American Journal of Human Genetics*, **2019**, 104, 1233-1240	11	22
142	A novel SLC9A1 mutation causes cerebellar ataxia. *Journal of Human Genetics*, **2018**, 63, 1049-1054	4.3	22
141	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	4.3	22
140	De novo SHANK3 mutation causes Rett syndrome-like phenotype in a female patient. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1593-6	2.5	21
139	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics* , **2019**, 64, 1173-1186	4.3	20
138	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. *Journal of Medical Genetics*, **2019**, 56, 396-407	5.8	20
137	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	4.3	20
136	Identification of novel SNORD118 mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. *Clinical Genetics*, **2017**, 92, 180-187	4	19
135	SCN3A-Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. *Annals of Neurology*, **2020**, 88, 348-362	9.4	19
134	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in thePISD gene. *Genetics in Medicine*, **2019**, 21, 2734-2743	8.1	19
133	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. *Human Mutation*, **2017**, 38, 1542-1554	4.7	19
132	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. *European Journal of Paediatric Neurology*, **2016**, 20, 412-7	3.8	19
131	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	4.3	18
130	A response to: loss of dermatan-4-sulfotransferase 1 (D4ST1/CHST14) function represents the first dermatan sulfate biosynthesis defect, "dermatan sulfate-deficient Adducted Thumb-Clubfoot Syndrome". Which name is appropriate, "Adducted Thumb-Clubfoot Syndrome" or "Ehlers-Danlos syndrome"?. *Human Mutation*, **2011**, 32, 1507-9	4.7	18
129	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	3.6	17

128	Different patterns of cerebellar abnormality and hypomyelination between POLR3A and POLR3B mutations. *Brain and Development*, **2014**, 36, 259-63	2.2	17
127	The first report of Japanese patients with asparagine synthetase deficiency. *Brain and Development*, **2017**, 39, 236-242	2.2	17
126	Breakpoint determination of X;autosome balanced translocations in four patients with premature ovarian failure. *Journal of Human Genetics*, **2011**, 56, 156-60	4.3	17
125	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, 304-7		16
124	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. *Scientific Reports*, **2014**, 4, 7132	4.9	16
123	Recurrent de novo MAPK8IP3 variants cause neurological phenotypes. *Annals of Neurology*, **2019**, 85, 927-933	9.4	15
122	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. *Journal of Human Genetics*, **2018**, 63, 263-270	4.3	15
121	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. *Nature Communications*, **2019**, 10, 2884	17.4	15
120	A novel CYCS mutation in the helix of the CYCS C-terminal domain causes non-syndromic thrombocytopenia. *Clinical Genetics*, **2018**, 94, 548-553	4	14
119	Characterization of the complex 7q21.3 rearrangement in a patient with bilateral split-foot malformation and hearing loss. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1224-30	2.5	14
118	Digenic mutations in and impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. *Science Advances*, **2020**, 6,	14.3	14
117	Nonsense variants in STAG2 result in distinct sex-dependent phenotypes. *Journal of Human Genetics*, **2019**, 64, 487-492	4.3	14
116	De novo variants in RHOBTB2, an atypical Rho GTPase gene, cause epileptic encephalopathy. *Human Mutation*, **2018**, 39, 1070-1075	4.7	14
115	Dysosteosclerosis is also caused by TNFRSF11A mutation. *Journal of Human Genetics*, **2018**, 63, 769-774	4.3	13
114	Vanoxerine, a new drug for terminating atrial fibrillation and flutter. *Journal of Cardiovascular Electrophysiology*, **2010**, 21, 311-9	2.7	13
113	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2662-70	2.5	13
112	Long-read DNA sequencing fully characterized chromothripsis in a patient with Langer-Giedion syndrome and Cornelia de Lange syndrome-4. *Journal of Human Genetics*, **2020**, 65, 667-674	4.3	13
111	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	3.1	12

110	Electroclinical features of epileptic encephalopathy caused by SCN8A mutation. *Pediatrics International*, **2015**, 57, 758-62	1.2	12
109	A pipeline for complete characterization of complex germline rearrangements from long DNA reads. *Genome Medicine*, **2020**, 12, 67	14.4	12
108	Dermatan 4-O-sulfotransferase 1-deficient Ehlers-Danlos syndrome complicated by a large subcutaneous hematoma on the back. *Journal of Dermatology*, **2016**, 43, 832-3	1.6	12
107	Delineation of musculocontractural Ehlers-Danlos Syndrome caused by dermatan sulfate epimerase deficiency. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1197	2.3	11
106	A unique case of de novo 5q33.3-q34 triplication with uniparental isodisomy of 5q34-qter. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1904-9	2.5	11
105	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. *American Journal of Human Genetics*, **2020**, 106, 13-25	11	11
104	Prenatal clinical manifestations in individuals with variants. *Journal of Medical Genetics*, **2021**, 58, 505-513	3.8	11
103	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. *Journal of Human Genetics*, **2018**, 63, 1223-1229	4.3	11
102	A severe pulmonary complication in a patient with COL4A1-related disorder: A case report. *European Journal of Medical Genetics*, **2017**, 60, 169-171	2.6	10
101	De Novo Truncating Variants in the Last Exon of SEMA6B Cause Progressive Myoclonic Epilepsy. *American Journal of Human Genetics*, **2020**, 106, 549-558	11	10
100	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies. *Brain and Development*, **2018**, 40, 406-409	2.2	10
99	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, 187-91	4.3	10
98	Phenotype-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	2.2	10
97	A female case of aromatic l-amino acid decarboxylase deficiency responsive to MAO-B inhibition. *Brain and Development*, **2016**, 38, 959-963	2.2	10
96	GRIN2D variants in three cases of developmental and epileptic encephalopathy. *Clinical Genetics*, **2018**, 94, 538-547	4	10
95	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. *Brain and Development*, **2018**, 40, 493-497	2.2	9
94	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. *Human Genome Variation*, **2015**, 2, 15024	1.8	9
93	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. *Journal of Human Genetics*, **2021**, 66, 499-507	4.3	9

92	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation. *Brain and Development*, **2018**, 40, 728-732	2.2	8
91	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. *Journal of Human Genetics*, **2019**, 64, 885-890	4.3	8
90	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	17.4	8
89	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	2.2	7
88	Cancer Management in Kabuki Syndrome: The First Case of Wilms Tumor and a Literature Review. *Journal of Pediatric Hematology/Oncology*, **2018**, 40, 391-394	1.2	7
87	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. *Journal of Human Genetics*, **2017**, 62, 997-1000	4.3	7
86	PCortical cerebellar atrophyPdwindles away in the era of next-generation sequencing. *Journal of Human Genetics*, **2014**, 59, 589-90	4.3	7
85	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. *Journal of Human Genetics*, **2021**, 66, 401-407	4.3	7
84	A recurrent homozygous variant in siblings with Lafora disease. *Human Genome Variation*, **2018**, 5, 16	1.8	6
83	Clinical and molecular features of 66 patients with musculocontractural Ehlers-Danlos syndrome caused by pathogenic variants in (mcEDS-). *Journal of Medical Genetics*, **2021**,	5.8	6
82	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. *Nature Communications*, **2021**, 12, 3750	17.4	6
81	Leaky splicing variant in sepiapterin reductase deficiency: Are milder cases escaping diagnosis?. *Neurology: Genetics*, **2019**, 5, e319	3.8	6
80	Rapid progression of a walking disability in a 5-year-old boy with a CLN6 mutation. *Brain and Development*, **2019**, 41, 726-730	2.2	5
79	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. *Journal of Human Genetics*, **2020**, 65, 751-757	4.3	5
78	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. *Neurology: Genetics*, **2020**, 6, e531	3.8	5
77	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. *Human Mutation*, **2020**, 41, 591-599	4.7	5
76	Clinical and genetic characteristics of patients with Doose syndrome. *Epilepsia Open*, **2020**, 5, 442-450	4	5
75	SOFT syndrome in a patient from Chile. *American Journal of Medical Genetics, Part A*, **2019**, 179, 338-340	2.5	5

74	Efficient detection of copy-number variations using exome data: Batch- and sex-based analyses. *Human Mutation*, **2021**, 42, 50-65	4.7	5
73	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. *Journal of Human Genetics*, **2019**, 64, 1005-1014	4.3	4
72	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). *Human Genome Variation*, **2015**, 2, 15012	1.8	4
71	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. *Human Genetics*, **2021**, 140, 1109-1120	6.3	4
70	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. *Human Mutation*, **2021**, 42, 66-76	4.7	4
69	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. *Science Advances*, **2021**, 7,	14.3	4
68	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	3.2	3
67	Early-onset epileptic encephalopathy and severe developmental delay in an association with de novo double mutations in and. *Epilepsia Open*, **2018**, 3, 81-85	4	3
66	A novel homozygous mutation of CLCN2 in a patient with characteristic brain MRI images - A first case of CLCN2-related leukoencephalopathy in Japan. *Brain and Development*, **2019**, 41, 101-105	2.2	3
65	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	7.7	3
64	De novo ATP1A3 variants cause polymicrogyria. *Science Advances*, **2021**, 7,	14.3	3
63	Complete sequencing of expanded SAMD12 repeats by long-read sequencing and Cas9-mediated enrichment. *Brain*, **2021**, 144, 1103-1117	11.2	3
62	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. *Brain and Development*, **2021**, 43, 505-514	2.2	3
61	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	4.3	3
60	Dystonia due to bilateral caudate hemorrhage associated with a COL4A1 mutation. *Parkinsonism and Related Disorders*, **2017**, 40, 80-82	3.6	2
59	Reply to "GGC Repeat Expansion of NOTCH2NLC is Rare in European Leukoencephalopathy". *Annals of Neurology*, **2020**, 88, 642-643	9.4	2
58	De novo mutations in epilepsy. *Developmental Medicine and Child Neurology*, **2011**, 53, 806-807	3.3	2
57	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy		2

56	Regulation of human development by ubiquitin chain editing of chromatin remodelers		2
55	The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. *Journal of Human Genetics*, **2021**, 66, 371-377	4.3	2
54	Clinical variations of epileptic syndrome associated with PACS2 variant. *Brain and Development*, **2021**, 43, 343-347	2.2	2
53	Expanding the phenotypic spectrum of TNFRSF11A-associated dysosteosclerosis: a case with intracranial extramedullary hematopoiesis. *Journal of Human Genetics*, **2021**, 66, 607-611	4.3	2
52	Clonazepam as an Effective Treatment for Epilepsy in a Female Patient with Mutation: Case Report. *Molecular Syndromology*, **2020**, 11, 232-237	1.5	1
51	Efficacy and Safety of Catheter Ablation for Persistent or Permanent Atrial Fibrillation. *Journal of Arrhythmia*, **2007**, 23, 229-235	1.5	1
50	A possible explanation for nocturnal hypertension in preeclamptics. *Clinical and Experimental Hypertension Part B, Hypertension in Pregnancy*, **1989**, 8, 495-506		1
49	Duplications in the G3 domain or switch II region in HRAS identified in patients with Costello syndrome. *Human Mutation*, **2021**,	4.7	1
48	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	8.6	1
47	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. *Journal of Human Genetics*, **2021**,	4.3	1
46	A method for complete characterization of complex germline rearrangements from long DNA reads		1
45	An atypical case of KMT2B -related dystonia manifesting asterixis and effect of deep brain stimulation of the globus pallidus. *Neurology and Clinical Neuroscience*, **2020**, 8, 36-38	0.3	1
44	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. *Journal of the Neurological Sciences*, **2020**, 416, 117047	3.2	1
43	GNAO1 organizes the cytoskeletal remodeling and firing of developing neurons. *FASEB Journal*, **2020**, 34, 16601-16621	0.9	1
42	De novo missense variants in are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. *Journal of Medical Genetics*, **2021**, 58, 712-716	5.8	1
41	Pathogenic MAST3 variants in the STK domain are associated with epilepsy		1
40	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. *Nature Communications*, **2021**, 12, 2046	17.4	1
39	Cerebrovascular diseases in two patients with entire NSD1 deletion. *Human Genome Variation*, **2021**, 8, 20	1.8	1

38	Progressive cerebral atrophies in three children with COL4A1 mutations. *Brain and Development*, **2021**, 43, 1033-1038	2.2	1
37	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. *Brain and Development*, **2021**, 43, 475-481	2.2	1
36	Association of early-onset epileptic encephalopathy with involuntary movements - Case series and literature review. *Epilepsy and Behavior Reports*, **2021**, 15, 100417	1.3	1
35	Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 631428	5.7	1
34	A novel LRP6 variant in a Japanese family with oligodontia. *Human Genome Variation*, **2021**, 8, 30	1.8	1
33	Intellectual disability and microcephaly associated with a novel CHAMP1 mutation. *Human Genome Variation*, **2021**, 8, 34	1.8	1
32	Biallelic null variants in ZNF142 cause global developmental delay with familial epilepsy and dysmorphic features. *Journal of Human Genetics*, **2021**,	4.3	1
31	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome.. *Brain*, **2022**,	11.2	1
30	Behçet's disease with a somatic UBA1 variant: Expanding spectrum of autoinflammatory phenotypes of VEXAS syndrome.. *Clinical Immunology*, **2022**, 108996	9	1
29	Legg-Calvé-Perthes disease in a patient with Bardet-Biedl syndrome: A case report of a novel mutation. *Clinical Case Reports (discontinued)*, **2020**, 8, 3110-3115	0.7	0
28	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant.. *Human Genetics*, **2022**, 141, 283	6.3	0
27	Cerebrospinal fluid abnormalities in developmental and epileptic encephalopathy with a variant. *Neurology: Genetics*, **2020**, 6, e527	3.8	0
26	COG1-congenital disorders of glycosylation: Milder presentation and review. *Clinical Genetics*, **2021**, 100, 318-323	4	0
25	De novo pathogenic DHX30 variants in two cases. *Clinical Genetics*, **2021**, 100, 350-351	4	0
24	Gait disturbance in a patient with de novo 1.0-kb SOX2 microdeletion. *Brain and Development*, **2021**,	2.2	0
23	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	3.7	0
22	Comprehensive Genetic Analysis of Non-syndromic Autism Spectrum Disorder in Clinical Settings. *Journal of Autism and Developmental Disorders*, **2021**, 51, 4655-4662	4.6	0
21	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	5.6	0

20	Clinical course of epilepsy and white matter abnormality linked to a novel DYRK1A variant. *Human Genome Variation*, **2021**, 8, 26	1.8	0
19	Expanding the KIF4A-associated phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3728-3739	2.5	0
18	Expanding the phenotypic spectrum of cardiospondylocarpofacial syndrome: From a detailed clinical and radiological observation of a boy with a novel missense variant in MAP3K7. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
17	A case of VEXAS syndrome with Sweet's disease and pulmonary involvement.. *Journal of Dermatology*, **2022**,	1.6	0
16	Hornerin deposits in neuronal intranuclear inclusion disease: direct identification of proteins with compositionally biased regions in inclusions.. *Acta Neuropathologica Communications*, **2022**, 10, 28	7.3	0
15	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	14.4	0
14	Clinical course of a Japanese patient with developmental delay linked to a small 6q16.1 deletion.. *Human Genome Variation*, **2022**, 9, 14	1.8	0
13	Does Atrial-Pacing from Different Intra-Atrial Sites for Atrial Fibrillation Effect Pulmonary Venous Pressure?. *Journal of Arrhythmia*, **2010**, 26, 176-180	1.5	
12	Efficacy of Additional Amiodarone Therapy in Patients with an Implantable Cardioverter-Defibrillator. *Journal of Arrhythmia*, **2010**, 26, 103-110	1.5	
11	Two males with sick sinus syndrome in a family with 0.6 kb deletions involving major domains in MECP2. *European Journal of Medical Genetics*, **2020**, 63, 103769	2.6	
10	A 2-year-old patient with a diffuse intrinsic pontine glioma and radiation-induced moyamoya syndrome. *Pediatric Blood and Cancer*, **2020**, 67, e28618	3	
9	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning.. *Med*, **2021**, 2, 611-632.e9	31.7	
8	A patient with a 6q22.1 deletion and a phenotype of non-progressive early-onset generalized epilepsy with tremor. *Epilepsy and Behavior Reports*, **2021**, 15, 100405	1.3	
7	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	4	
6	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	2.2	
5	Severe cardiac defect in Cornelia de Lange syndrome from a novel SMC1A variant.. *Pediatrics International*, **2022**, 64, e15031	1.2	
4	Long-term course of early onset developmental and epileptic encephalopathy associated with 2q24.3 microduplication. *Epilepsy and Behavior Reports*, **2022**, 100547	1.3	
3	Perampanel markedly improved clinical seizures in a patient with a Rett-like phenotype and 960-kb deletion on chromosome 9q34.11 including the STXBP1. *Clinical Case Reports (discontinued)*, **2022**, 10,	0.7	

2	Genetic and Imaging Characteristics of a Family With Neuronal Intranuclear Inclusion Disease.. *Journal of Clinical Neurology (Korea*, **2022**, 18, 358-360	1.7
1	Detecting the NOTCH2NLC Repeat Expansion in Neuronal Intranuclear Inclusion Disease. *Neuromethods*, **2022**, 121-138	0.4