Hirotomo Saitsu

List of Publications by Year in Descending Order

199 papers	4,632 citations	36 h-index	62 g-index
234 ext. papers	5,818 ext. citations	4.9 avg, IF	4.96 L-index

#	Paper	IF	Citations
199	A novel intronic PORCN variant creating an alternative splice acceptor site in a mother and her daughter with focal dermal hypoplasia.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
198	Retinitis pigmentosa with optic neuropathy and mutations: A case report.. *American Journal of Ophthalmology Case Reports*, **2022**, 25, 101298	1.3	1
197	An intronic GNAO1 variant leading to in-frame insertion cause movement disorder controlled by deep brain stimulation.. *Neurogenetics*, **2022**, 23, 129	3	1
196	Retrotransposition disrupting EBP in a girl and her mother with X-linked dominant chondrodysplasia punctata.. *Journal of Human Genetics*, **2022**,	4.3	1
195	Neurochemistry evaluated by MR spectroscopy in a patient with SPTAN1-related developmental and epileptic encephalopathy.. *Brain and Development*, **2022**,	2.2	0
194	Two novel heterozygous variants in ATP1A3 cause movement disorders.. *Human Genome Variation*, **2022**, 9, 7	1.8	
193	A new case of concurrent existence of PRRT2-associated paroxysmal movement disorders with c.649dup variant and 16p11.2 microdeletion syndrome.. *Brain and Development*, **2022**,	2.2	
192	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
191	A New Case With Cortical Malformation Caused by Biallelic Variants in LAMC3. *Neurology: Genetics*, **2022**, 8, e680	3.8	
190	Cognitive Impairment in a Complex Family With AAGGG and ACAGG Repeat Expansions in RFC1 Detected by ExpansionHunter Denovo. *Neurology: Genetics*, **2022**, 8, e682	3.8	
189	Compound heterozygous ADAMTS9 variants in Joubert syndrome-related disorders without renal manifestation. *Brain and Development*, **2021**,	2.2	1
188	Novel Variants Cause Progressive Leukodystrophy in Childhood: Case Report and Literature Review. *Child Neurology Open*, **2021**, 8, 2329048X211048613	1.3	1
187	Prenatal clinical manifestations in individuals with variants. *Journal of Medical Genetics*, **2021**, 58, 505-513	3.8	11
186	Genetic and phenotypic analysis of 101 patients with developmental delay or intellectual disability using whole-exome sequencing. *Clinical Genetics*, **2021**, 100, 40-50	4	3
185	De novo ATP1A3 variants cause polymicrogyria. *Science Advances*, **2021**, 7,	14.3	3
184	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia. *Clinical Epigenetics*, **2021**, 13, 73	7.7	1
183	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

182	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. *Brain and Development*, **2021**, 43, 505-514	2.2	3
181	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
180	ZNF445: a homozygous truncating variant in a patient with Temple syndrome and multilocus imprinting disturbance. *Clinical Epigenetics*, **2021**, 13, 119	7.7	3
179	Comprehensive genetic analysis confers high diagnostic yield in 16 Japanese patients with corpus callosum anomalies. *Journal of Human Genetics*, **2021**, 66, 1061-1068	4.3	1
178	Cerebrovascular diseases in two patients with entire NSD1 deletion. *Human Genome Variation*, **2021**, 8, 20	1.8	1
177	A boy with biallelic frameshift variants in TTC5 and brain malformation resembling tubulinopathies. *Journal of Human Genetics*, **2021**, 66, 1189-1192	4.3	0
176	Global developmental delay, systemic dysmorphism and epilepsy in a patient with a de novo U2AF2 variant. *Journal of Human Genetics*, **2021**, 66, 1185-1187	4.3	0
175	Identification of two novel de novo TUBB variants in cases with brain malformations: case reports and literature review. *Journal of Human Genetics*, **2021**, 66, 1193-1197	4.3	1
174	Progressive cerebral atrophies in three children with COL4A1 mutations. *Brain and Development*, **2021**, 43, 1033-1038	2.2	1
173	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
172	Congenital disorders of glycosylation type IIb with MOGS mutations cause early infantile epileptic encephalopathy, dysmorphic features, and hepatic dysfunction. *Brain and Development*, **2021**, 43, 402-410	2.2	1
171	Familial periodic paralysis associated with a rare KCNJ5 variant that supposed to have incomplete penetrance. *Brain and Development*, **2021**, 43, 470-474	2.2	2
170	Kagami-Ogata syndrome in a patient with 46,XX,t(2;14)(q11.2;q32.2)mat disrupting MEG3. *Journal of Human Genetics*, **2021**, 66, 439-443	4.3	
169	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
168	A novel method for isolating lymphatic endothelial cells from lymphatic malformations and detecting PIK3CA somatic mutation in these isolated cells. *Surgery Today*, **2021**, 51, 439-446	3	1
167	Association of early-onset epileptic encephalopathy with involuntary movements - Case series and literature review. *Epilepsy and Behavior Reports*, **2021**, 15, 100417	1.3	1
166	Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review. *Endocrine Journal*, **2021**, 68, 111-117	2.9	4
165	Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation. *Endocrine Journal*, **2021**, 68, 605-611	2.9	2

164	Biallelic CDK9 variants as a cause of a new multiple-malformation syndrome with retinal dystrophy mimicking the CHARGE syndrome. *Journal of Human Genetics*, **2021**, 66, 1021-1027	4.3	1
163	SCN8A-related developmental and epileptic encephalopathy with ictal asystole requiring cardiac pacemaker implantation. *Brain and Development*, **2021**, 43, 804-808	2.2	1
162	A novel de novo TMEM63A variant in a patient with severe hypomyelination and global developmental delay. *Brain and Development*, **2021**,	2.2	0
161	Novel ALG12 variants and hydronephrosis in siblings with impaired N-glycosylation. *Brain and Development*, **2021**, 43, 945-951	2.2	0
160	Leigh syndrome-like MRI changes in a patient with biallelic variants treated with ketogenic diet. *Molecular Genetics and Metabolism Reports*, **2021**, 29, 100800	1.8	2
159	TSC1 intragenic deletion transmitted from a mosaic father to two siblings with cardiac rhabdomyomas: Identification of two aberrant transcripts. *European Journal of Medical Genetics*, **2020**, 63, 104060	2.6	1
158	Identification of a deep intronic POLR3A variant causing inclusion of a pseudoexon derived from an Alu element in Pol III-related leukodystrophy. *Journal of Human Genetics*, **2020**, 65, 921-925	4.3	8
157	Intronic variant in IQGAP3 associated with hereditary neuropathy with proximal lower dominancy, urinary disturbance, and paroxysmal dry cough. *Journal of Human Genetics*, **2020**, 65, 717-725	4.3	
156	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
155	A recurrent TMEM106B mutation in hypomyelinating leukodystrophy: A rapid diagnostic assay. *Brain and Development*, **2020**, 42, 603-606	2.2	2
154	POLR3A variants in striatal involvement without diffuse hypomyelination. *Brain and Development*, **2020**, 42, 363-368	2.2	9
153	A de novo TOP2B variant associated with global developmental delay and autism spectrum disorder. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1145	2.3	6
152	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. *Journal of Human Genetics*, **2020**, 65, 727-734	4.3	5
151	A case of childhood glaucoma with a combined partial monosomy 6p25 and partial trisomy 18p11 due to an unbalanced translocation. *Ophthalmic Genetics*, **2020**, 41, 175-182	1.2	1
150	Long-term observation of a Japanese mucolipidosis IV patient with a novel homozygous p.F313del variant of MCOLN1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1500-1505	2.5	6
149	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant. *PLoS ONE*, **2020**, 15, e0231665	3.7	4
148	A case of CLCN2-related leukoencephalopathy with bright tree appearance during aseptic meningitis. *Brain and Development*, **2020**, 42, 462-467	2.2	0
147	A de novo GABRB2 variant associated with myoclonic status epilepticus and rhythmic high-amplitude delta with superimposed (poly) spikes (RHADS). *Epileptic Disorders*, **2020**, 22, 476-481	1.9	3

146	Fulminant myocarditis following recurrent generalized erythrokeratoderma in a child with a heterozygous GJA1 variant. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1933-1938	2.5	
145	Exome reports A de novo GNB2 variant associated with global developmental delay, intellectual disability, and dysmorphic features. *European Journal of Medical Genetics*, **2020**, 63, 103804	2.6	11
144	De novo ZBTB7A variant in a patient with macrocephaly, intellectual disability, and sleep apnea: implications for the phenotypic development in 19p13.3 microdeletions. *Journal of Human Genetics*, **2020**, 65, 181-186	4.3	2
143	Nanopore sequencing reveals a structural alteration of mirror-image duplicated genes in a genome-editing mouse line. *Congenital Anomalies (discontinued)*, **2020**, 60, 120-125	1.1	3
142	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. *European Journal of Medical Genetics*, **2020**, 63, 104061	2.6	2
141	variants dysregulate splicing and cause hypomyelinating leukodystrophy. *Neurology: Genetics*, **2020**, 6, e524	3.8	0
140	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. *Journal of the Neurological Sciences*, **2020**, 416, 117047	3.2	1
139	GNAO1 organizes the cytoskeletal remodeling and firing of developing neurons. *FASEB Journal*, **2020**, 34, 16601-16621	0.9	1
138	Nonsense-associated altered splicing of MAP3K1 in two siblings with 46,XY disorders of sex development. *Scientific Reports*, **2020**, 10, 17375	4.9	4
137	IGF2 Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	13
136	Life-threatening muscle complications of COL4A1-related disorder. *Brain and Development*, **2020**, 42, 93-97	2.2	2
135	Low-prevalence mosaicism of chromosome 18q distal deletion identified by exome-based copy number profiling in a child with cerebral hypomyelination. *Congenital Anomalies (discontinued)*, **2020**, 60, 94-96	1.1	2
134	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant **2020**, 15, e0231665		
133	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant **2020**, 15, e0231665		
132	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant **2020**, 15, e0231665		
131	CCNB2 and AURKA overexpression may cause atypical mitosis in Japanese cortisol-producing adrenocortical carcinoma with TP53 somatic variant **2020**, 15, e0231665		
130	A novel homozygous truncating variant of NECAP1 in early infantile epileptic encephalopathy: the second case report of EIEE21. *Journal of Human Genetics*, **2019**, 64, 347-350	4.3	3
129	De novo variants in cause intellectual disability, autism spectrum disorder, and epilepsy with myoclonic absences. *Epilepsia Open*, **2019**, 4, 476-481	4	13

128	A de novo variant in RAC3 causes severe global developmental delay and a middle interhemispheric variant of holoprosencephaly. *Journal of Human Genetics*, **2019**, 64, 1127-1132	4.3	8
127	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
126	A case of early-onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 645-649	2.5	3
125	A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e814	2.3	7
124	Ataxic phenotype with altered Ca3.1 channel property in a mouse model for spinocerebellar ataxia 42. *Neurobiology of Disease*, **2019**, 130, 104516	7.5	15
123	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. *Nature Communications*, **2019**, 10, 2506	17.4	22
122	Identification of novel compound heterozygous mutations in ACO2 in a patient with progressive cerebral and cerebellar atrophy. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00698	2.3	5
121	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. *Journal of Medical Genetics*, **2019**, 56, 396-407	5.8	20
120	Germline-Derived Gain-of-Function Variants of Gs-Coding Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 877-889	12.7	13
119	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. *Human Molecular Genetics*, **2019**, 28, 2319-2329	5.6	12
118	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases. *Brain and Development*, **2019**, 41, 474-479	2.2	3
117	De novo AFF3 variant in a patient with mesomelic dysplasia with foot malformation. *Journal of Human Genetics*, **2019**, 64, 1041-1044	4.3	2
116	Single-fiber electromyography-based diagnosis of CACNA1A mutation in children: A potential role of the electrodiagnosis in the era of whole exome sequencing. *Brain and Development*, **2019**, 41, 905-909	2.2	1
115	Comprehensive clinical and molecular studies in split-hand/foot malformation: identification of two plausible candidate genes (LRP6 and UBA2). *European Journal of Human Genetics*, **2019**, 27, 1845-1857	5.3	6
114	Reply to "Reduced CYFIP2 Stability by Arg87 Variants Causing Human Neurological Disorders". *Annals of Neurology*, **2019**, 86, 805-806	9.4	0
113	Quinidine therapy and therapeutic drug monitoring in four patients with KCNT1 mutations **2019**, 21, 48-54		7
112	Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan. *Journal of Human Genetics*, **2019**, 64, 145-152	4.3	3
111	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. *Genetics in Medicine*, **2019**, 21, 1629-1638	8.1	17

110	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
109	De novo truncating variants in PHF21A cause intellectual disability and craniofacial anomalies. *European Journal of Human Genetics*, **2019**, 27, 378-383	5.3	6
108	A male case with CDKL5-associated encephalopathy manifesting transient methylmalonic acidemia. *European Journal of Medical Genetics*, **2018**, 61, 451-454	2.6	7
107	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37
106	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations. *Japanese Journal of Ophthalmology*, **2018**, 62, 458-466	2.6	8
105	variants in and cause neurodevelopmental disorders. *Annals of Clinical and Translational Neurology*, **2018**, 5, 280-296	5.3	27
104	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
103	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. *Brain*, **2018**, 141, 1703-1718	11.2	44
102	Biallelic Variants in CNPY3, Encoding an Endoplasmic Reticulum Chaperone, Cause Early-Onset Epileptic Encephalopathy. *American Journal of Human Genetics*, **2018**, 102, 321-329	11	10
101	Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the AR gene. *Scientific Reports*, **2018**, 8, 2287	4.9	9
100	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. *Human Molecular Genetics*, **2018**, 27, 1421-1433	5.6	23
99	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. *Journal of Human Genetics*, **2018**, 63, 263-270	4.3	15
98	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. *Journal of Human Genetics*, **2018**, 63, 417-423	4.3	10
97	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. *Human Genetics*, **2018**, 137, 95-104	6.3	36
96	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation. *Brain and Development*, **2018**, 40, 728-732	2.2	8
95	A case of new PCDH12 gene variants presented as dyskinetic cerebral palsy with epilepsy. *Journal of Human Genetics*, **2018**, 63, 749-753	4.3	6
94	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
93	Epileptic apnea in a patient with inherited glycosylphosphatidylinositol anchor deficiency and PIGT mutations. *Brain and Development*, **2018**, 40, 53-57	2.2	17

92	Deletions of SCN2A and SCN3A genes in a patient with West syndrome and autistic spectrum disorder. *Seizure: the Journal of the British Epilepsy Association*, **2018**, 60, 91-93	3.2	6
91	A recurrent homozygous variant in siblings with Lafora disease. *Human Genome Variation*, **2018**, 5, 16	1.8	6
90	Recurrent SCN3A p.Ile875Thr variant in patients with polymicrogyria. *Annals of Neurology*, **2018**, 84, 159-161	9.4	8
89	Molecular Diagnosis of 34 Japanese Families with Leber Congenital Amaurosis Using Targeted Next Generation Sequencing. *Scientific Reports*, **2018**, 8, 8279	4.9	21
88	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene. *Brain and Development*, **2018**, 40, 819-823	2.2	6
87	FGFR1 disruption identified by whole genome sequencing in a male with a complex chromosomal rearrangement and hypogonadotropic hypogonadism. *American Journal of Medical Genetics, Part A*, **2018**, 176, 139-143	2.5	1
86	Biallelic COLGALT1 variants are associated with cerebral small vessel disease. *Annals of Neurology*, **2018**, 84, 843-853	9.4	20
85	variant identified by whole-exome sequencing in a Japanese family with atrial septal defect: Implications for male sex development. *Clinical Case Reports (discontinued)*, **2018**, 6, 2229-2233	0.7	1
84	mutations cause variable phenotypes of developmental and epileptic encephalopathy. *Epilepsia Open*, **2018**, 3, 495-502	4	21
83	De novo PHACTR1 mutations in West syndrome and their pathophysiological effects. *Brain*, **2018**, 141, 3098-3114	11.2	12
82	GRIN2D variants in three cases of developmental and epileptic encephalopathy. *Clinical Genetics*, **2018**, 94, 538-547	4	10
81	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. *Journal of Human Genetics*, **2018**, 63, 1223-1229	4.3	11
80	De novo variants in RHOBTB2, an atypical Rho GTPase gene, cause epileptic encephalopathy. *Human Mutation*, **2018**, 39, 1070-1075	4.7	14
79	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. *Cell Reports*, **2018**, 22, 734-747	10.6	86
78	A novel mutation in the proteolytic domain of LONP1 causes atypical CODAS syndrome. *Journal of Human Genetics*, **2017**, 62, 653-655	4.3	13
77	PARS2 and NARS2 mutations in infantile-onset neurodegenerative disorder. *Journal of Human Genetics*, **2017**, 62, 525-529	4.3	41
76	Biallelic mutations in the 3Pexonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464	36.3	43
75	Mutations in genes encoding polycomb repressive complex 2 subunits cause Weaver syndrome. *Human Mutation*, **2017**, 38, 637-648	4.7	50

74	Folate receptors and neural tube closure. *Congenital Anomalies (discontinued)*, **2017**, 57, 130-133	1.1	4
73	Neuroimaging findings in Joubert syndrome with C5orf42 gene mutations: A milder form of molar tooth sign and vermian hypoplasia. *Journal of the Neurological Sciences*, **2017**, 376, 7-12	3.2	11
72	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffin-Siris-like syndrome. *Journal of Human Genetics*, **2017**, 62, 741-746	4.3	31
71	Identification of novel SNORD118 mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. *Clinical Genetics*, **2017**, 92, 180-187	4	19
70	De novo IGF2 mutation on the paternal allele in a patient with Silver-Russell syndrome and ectrodactyly. *Human Mutation*, **2017**, 38, 953-958	4.7	23
69	Dystonia due to bilateral caudate hemorrhage associated with a COL4A1 mutation. *Parkinsonism and Related Disorders*, **2017**, 40, 80-82	3.6	2
68	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. *Scientific Reports*, **2017**, 7, 3552	4.9	35
67	A patient with Muenke syndrome manifesting migrating neonatal seizures. *Brain and Development*, **2017**, 39, 873-876	2.2	2
66	Biallelic Mutations in MYPN, Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. *American Journal of Human Genetics*, **2017**, 100, 169-178	11	57
65	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
64	Foxc2 knock-in mice mark stage-specific Foxc2-expressing cells during mouse organogenesis. *Congenital Anomalies (discontinued)*, **2017**, 57, 24-31	1.1	3
63	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
62	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*, **2017**, 140, 2322-2336	11.2	44
61	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. *Journal of Human Genetics*, **2017**, 62, 997-1000	4.3	7
60	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. *Human Mutation*, **2017**, 38, 1542-1554	4.7	19
59	Severe leukoencephalopathy with cortical involvement and peripheral neuropathy due to FOLR1 deficiency. *Brain and Development*, **2017**, 39, 266-270	2.2	15
58	A case of early onset epileptic encephalopathy with de novo mutation in SLC35A2: Clinical features and treatment for epilepsy. *Brain and Development*, **2017**, 39, 256-260	2.2	23
57	The first report of Japanese patients with asparagine synthetase deficiency. *Brain and Development* , **2017**, 39, 236-242	2.2	17

56	Quinidine therapy for West syndrome with KCNTI mutation: A case report. *Brain and Development*, **2017**, 39, 80-83	2.2	36
55	A Male Case with Propeller Protein-Associated Neurodegeneration (BPAN) with Somatic Mosaic Mutation in WDR45 **2016**, 6,		2
54	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
53	De novo missense mutations in NALCN cause developmental and intellectual impairment with hypotonia. *Journal of Human Genetics*, **2016**, 61, 451-5	4.3	22
52	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
51	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. *Journal of Human Genetics*, **2016**, 61, 381-7	4.3	30
50	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. *Journal of Human Genetics*, **2016**, 61, 527-31	4.3	24
49	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
48	Bortezomib-resistance is associated with increased levels of proteasome subunits and apoptosis-avoidance. *Oncotarget*, **2016**, 7, 77622-77634	3.3	25
47	Ineffective quinidine therapy in early onset epileptic encephalopathy with KCNT1 mutation. *Annals of Neurology*, **2016**, 79, 502-3	9.4	52
46	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. *Scientific Reports*, **2016**, 6, 30072	4.9	52
45	WDR45 mutations in three male patients with West syndrome. *Journal of Human Genetics*, **2016**, 61, 653-61	4.3	36
44	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. *Journal of Human Genetics*, **2016**, 61, 835-8	4.3	24
43	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. *European Journal of Paediatric Neurology*, **2016**, 20, 412-7	3.8	19
42	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
41	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
40	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 950-961	11	34
39	De novo DNM1 mutations in two cases of epileptic encephalopathy. *Epilepsia*, **2016**, 57, e18-23	6.4	17

38	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an Rexome-firstPapproach. *Journal of Human Genetics*, **2015**, 60, 175-82	4.3	41
37	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
36	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. *Journal of Human Genetics*, **2015**, 60, 631-5	4.3	14
35	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
34	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
33	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
32	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. *Human Genome Variation*, **2015**, 2, 15034	1.8	9
31	De novo KCNT1 mutations in early-onset epileptic encephalopathy. *Epilepsia*, **2015**, 56, e121-8	6.4	66
30	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
29	Electroclinical features of epileptic encephalopathy caused by SCN8A mutation. *Pediatrics International*, **2015**, 57, 758-62	1.2	12
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