Mitsuhiro Kato

List of Publications by Year
in descending order

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171	8,831	44069	51608
papers	citations	48 h-index	86 g-index

173	173	173	13386
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutation of ARX causes abnormal development of forebrain and testes in mice and X-linked lissencephaly with abnormal genitalia in humans. Nature Genetics, 2002, 32, 359-369.	21.4	647
2	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nature Genetics, 2008, 40, 782-788.	21.4	498
3	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	21.4	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.	21.4	396
5	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
6	Lissencephaly and the molecular basis of neuronal migration. Human Molecular Genetics, 2003, 12, 89R-96.	2.9	274
7	Clinical spectrum of early onset epileptic encephalopathies caused by <scp>KCNQ2</scp> mutation. Epilepsia, 2013, 54, 1282-1287.	5.1	195
8	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. Neurology, 2013, 81, 992-998.	1.1	188
9	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.	6.2	187
10	Somatic Mutations in the <scp>MTOR</scp> gene cause focal cortical dysplasia type <scp>II</scp>b. Annals of Neurology, 2015, 78, 375-386.	5.3	169
11	A Longer Polyalanine Expansion Mutation in the ARX Gene Causes Early Infantile Epileptic Encephalopathy with Suppression-Burst Pattern (Ohtahara Syndrome). American Journal of Human Genetics, 2007, 81, 361-366.	6.2	168
12	X-Linked Lissencephaly With Abnormal Genitalia as a Tangential Migration Disorder Causing Intractable Epilepsy: Proposal for a New Term, â€œInterneuronopathyâ€. Journal of Child Neurology, 2005, 20, 392-397.	1.4	143
13	Phenotypic Spectrum of COL4A1 Mutations: Porencephaly to Schizencephaly. Annals of Neurology, 2013, 73, 48-57.	5.3	143
14	Early onset epileptic encephalopathy caused by de novo <scp>SCN</scp>8A mutations. Epilepsia, 2014, 55, 994-1000.	5.1	142
15	STXBP1 mutations in early infantile epileptic encephalopathy with suppressionâ€burst pattern. Epilepsia, 2010, 51, 2397-2405.	5.1	133
16	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	6.4	132
17	Dominant-Negative Mutations in Î±-II Spectrin Cause West Syndrome with Severe Cerebral Hypomyelination, Spastic Quadriplegia, and Developmental Delay. American Journal of Human Genetics, 2010, 86, 881-891.	6.2	131
18	Gene therapy improves motor and mental function of aromatic l-amino acid decarboxylase deficiency. Brain, 2019, 142, 322-333.	7.6	116

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19	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	3.3	102
20	The somatic GNAQ mutation c.548G>A (p.R183Q) is consistently found in Sturgeâ€“Weber syndrome. Journal of Human Genetics, 2014, 59, 691-693.	2.3	100
21	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	5.1	95
22	PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. Neurology, 2014, 82, 1587-1596.	1.1	93
23	Clinical correlations of mutations affecting six components of the <scp>SWI</scp>/<scp>SNF</scp> complex: Detailed description of 21 patients and a review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 1221-1237.	1.2	91
24	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. Annals of Neurology, 2012, 72, 298-300.	5.3	88
25	De Novo Mutations inSLC35A2Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy. Human Mutation, 2013, 34, 1708-1714.	2.5	85
26	SPTAN1 encephalopathy: distinct phenotypes and genotypes. Journal of Human Genetics, 2015, 60, 167-173.	2.3	83
27	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
28	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
29	De Novo and Inherited Mutations in COL4A2, Encoding the Type IV Collagen Î±2 Chain Cause Porencephaly. American Journal of Human Genetics, 2012, 90, 86-90.	6.2	79
30	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	5.1	76
31	<scp>GRIN</scp>1 mutations cause encephalopathy with infantileâ€onset epilepsy, and hyperkinetic and stereotyped movement disorders. Epilepsia, 2015, 56, 841-848.	5.1	76
32	De novo GABRA1 mutations in Ohtahara and West syndromes. Epilepsia, 2016, 57, 566-573.	5.1	76
33	De novo KCNB1 mutations in infantile epilepsy inhibit repetitive neuronal firing. Scientific Reports, 2015, 5, 15199.	3.3	73
34	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	6.2	71
35	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	7.6	69
36	Acute cerebellar ataxia and consecutive cerebellitis produced by glutamate receptor Î´2 autoantibody. Brain and Development, 2007, 29, 254-256.	1.1	68

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37	Inflammatory changes in infantile-onset LMNA-associated myopathy. Neuromuscular Disorders, 2011, 21, 563-568.	0.6	67
38	CASK aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. Epilepsia, 2012, 53, 1441-1449.	5.1	66
39	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.	3.7	65
40	Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. Neurogenetics, 2014, 15, 193-200.	1.4	61
41	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	5.3	60
42	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. Human Genetics, 2018, 137, 95-104.	3.8	60
43	<scp>AADC</scp> deficiency from infancy to adulthood: Symptoms and developmental outcome in an international cohort of 63 patients. Journal of Inherited Metabolic Disease, 2020, 43, 1121-1130.	3.6	59
44	Cerebrospinal fluid levels of cytokines and soluble tumour necrosis factor receptor in acute disseminated encephalomyelitis. European Journal of Pediatrics, 2002, 161, 133-137.	2.7	58
45	BAC array CGH reveals genomic aberrations in idiopathic mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 205-211.	1.2	57
46	PARS2 and NARS2 mutations in infantile-onset neurodegenerative disorder. Journal of Human Genetics, 2017, 62, 525-529.	2.3	55
47	A new paradigm for West syndrome based on molecular and cell biology. Epilepsy Research, 2006, 70, 87-95.	1.6	54
48	High prevalence of genetic alterations in early-onset epileptic encephalopathies associated with infantile movement disorders. Brain and Development, 2016, 38, 285-292.	1.1	54
49	KIF1A mutation in a patient with progressive neurodegeneration. Journal of Human Genetics, 2014, 59, 639-641.	2.3	53
50	PCDH19-related female-limited epilepsy: Further details regarding early clinical features and therapeutic efficacy. Epilepsy Research, 2013, 106, 191-199.	1.6	52
51	Genotype-phenotype correlation in neuronal migration disorders and cortical dysplasias. Frontiers in Neuroscience, 2015, 9, 181.	2.8	52
52	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. American Journal of Human Genetics, 2016, 99, 950-961.	6.2	51
53	Loss of tuberin from cerebral tissues with tuberous sclerosis and astrocytoma. Annals of Neurology, 1996, 40, 941-944.	5.3	49
54	Mutation of the doublecortin gene in male patients with double cortex syndrome: Somatic mosaicism detected by hair root analysis. Annals of Neurology, 2001, 50, 547-551.	5.3	47

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55	Magnetoencephalography in Patients with Tuberous Sclerosis and Localization-related Epilepsy. Epilepsia, 2006, 47, 991-997.	5.1	47
56	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	1.8	46
57	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
58	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. Scientific Reports, 2017, 7, 3552.	3.3	46
59	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	12.8	46
60	A case of acute cerebellitis accompanied by autoantibodies against glutamate receptor Î´2. Brain and Development, 2007, 29, 224-226.	1.1	45
61	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. Journal of Human Genetics, 2014, 59, 581-583.	2.3	42
62	Somatic mutations in <scp>GLI</scp> 3 and <scp>OFD</scp> 1 involved in sonic hedgehog signaling cause hypothalamic hamartoma. Annals of Clinical and Translational Neurology, 2016, 3, 356-365.	3.7	42
63	Tuberin immunohistochemistry in brain, kidneys and heart with or without tuberous sclerosis. Acta Neuropathologica, 1997, 94, 525-531.	7.7	41
64	WDR45 mutations in three male patients with West syndrome. Journal of Human Genetics, 2016, 61, 653-661.	2.3	39
65	Molecular karyotyping in 17 patients and mutation screening in 41 patients with Kabuki syndrome. Journal of Human Genetics, 2009, 54, 304-309.	2.3	37
66	Mislocalization of syntaxinâ€1 and impaired neurite growth observed in a human <scp>iPSC</scp> model for <scp>STXBP</scp>1â€related epileptic encephalopathy. Epilepsia, 2016, 57, e81-6.	5.1	37
67	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	2.9	36
68	Early onset West syndrome with cerebral hypomyelination and reduced cerebral white matter. Brain and Development, 2008, 30, 349-355.	1.1	35
69	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.	1.2	31
70	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.	12.8	30
71	Novel splicing mutation in the ASXL3 gene causing Bainbridgeâ€“Ropers syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1863-1867.	1.2	28
72	New susceptibility variants to narcolepsy identified in HLA class II region. Human Molecular Genetics, 2015, 24, 891-898.	2.9	27

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73	Asymptomatic congenital cytomegalovirus infection with neurological sequelae: A retrospective study using umbilical cord. Brain and Development, 2016, 38, 819-826.	1.1	27
74	Novel MCA/ID syndrome with ASH1L mutation. American Journal of Medical Genetics, Part A, 2017, 173, 1644-1648.	1.2	27
75	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.	1.1	27
76	<scp>PLPBP</scp> mutations cause variable phenotypes of developmental and epileptic encephalopathy. Epilepsia Open, 2018, 3, 495-502.	2.4	27
77	Asymptomatic hereditary Alexander's disease caused by a novel mutation in GFAP. Journal of the Neurological Sciences, 2004, 225, 125-127.	0.6	26
78	De novo 5q14.3 translocation 121.5â€kb upstream of MEF2C in a patient with severe intellectual disability and earlyâ€onset epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2011, 155, 2879-2884.	1.2	26
79	Truncating mutation in NFIA causes brain malformation and urinary tract defects. Human Genome Variation, 2015, 2, 15007.	0.7	24
80	Pathogenic variants of DYNC2H1 , KIAA0556 , and PTPN11 associated with hypothalamic hamartoma. Neurology, 2019, 93, e237-e251.	1.1	24
81	Hypertrophy of the cerebral white matter in hemimegalencephaly. Pediatric Neurology, 1996, 14, 335-338.	2.1	23
82	Acute encephalopathy with refractory status epilepticus: Bilateral mesial temporal and claustral lesions, associated with a peripheral marker of oxidative DNA damage. Journal of the Neurological Sciences, 2006, 250, 159-161.	0.6	23
83	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. Epilepsia, 2014, 55, e22-6.	5.1	23
84	TUBA1A mutation can cause a hydranencephaly-like severe form of cortical dysgenesis. Scientific Reports, 2015, 5, 15165.	3.3	23
85	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. Journal of Human Genetics, 2020, 65, 727-734.	2.3	23
86	A case of TUBA1A mutation presenting with lissencephaly and Hirschsprung disease. Brain and Development, 2014, 36, 159-162.	1.1	22
87	In vitro characterization of neurite extension using induced pluripotent stem cells derived from lissencephaly patients with TUBA1A missense mutations. Molecular Brain, 2016, 9, 70.	2.6	22
88	A novel missense mutation in the HECT domain of NEDD4L identified in a girl with periventricular nodular heterotopia, polymicrogyria and cleft palate. Journal of Human Genetics, 2017, 62, 861-863.	2.3	22
89	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	3.2	22
90	A combination of genetic and biochemical analyses for the diagnosis of PI3K-AKT-mTOR pathway-associated megalencephaly. BMC Medical Genetics, 2017, 18, 4.	2.1	21

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91	De novo PHACTR1 mutations in West syndrome and their pathophysiological effects. Brain, 2018, 141, 3098-3114.	7.6	21
92	IMSindel: An accurate intermediate-size indel detection tool incorporating de novo assembly and gapped global-local alignment with split read analysis. Scientific Reports, 2018, 8, 5608.	3.3	20
93	Diffuse pachygyria with cerebellar hypoplasia: A milder form of microlissencephaly or a new genetic syndrome?. Annals of Neurology, 1999, 46, 660-663.	5.3	19
94	Costello Syndrome Showing Moyamoya-like Vasculopathy. Pediatric Neurology, 2005, 32, 361-363.	2.1	19
95	No detectable genomic aberrations by BAC array CGH in Kabuki make-up syndrome patients. American Journal of Medical Genetics, Part A, 2006, 140A, 291-293.	1.2	19
96	Megalencephaly and Polymicrogyria with Polydactyly Syndrome. Pediatric Neurology, 2007, 37, 148-151.	2.1	19
97	An association analysis of HLA-DQB1 with narcolepsy without cataplexy and idiopathic hypersomnia with/without long sleep time in a Japanese population. Human Genome Variation, 2015, 2, 15031.	0.7	19
98	Biallelic lossâ€ofâ€function UBA5 mutations in a patient with intractable West syndrome and profound failure to thrive. Epileptic Disorders, 2018, 20, 313-318.	1.3	18
99	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
100	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
101	Contrast Sensitivity of Patients With Severe Motor and Intellectual Disabilities and Cerebral Visual Impairment. Journal of Child Neurology, 2002, 17, 731-737.	1.4	17
102	Analysis of the hypothalamus in a case of X-linked lissencephaly with abnormal genitalia (XLAG). Brain and Development, 2009, 31, 456-460.	1.1	17
103	Compound heterozygosity in GPR56 with bilateral frontoparietal polymicrogyria. Brain and Development, 2014, 36, 528-531.	1.1	17
104	Biallelic Variants in CNPY3, Encoding an Endoplasmic Reticulum Chaperone, Cause Early-Onset Epileptic Encephalopathy. American Journal of Human Genetics, 2018, 102, 321-329.	6.2	17
105	Microcephaly, Cerebellar Atrophy, and Focal Segmental Glomerulosclerosis in Two Brothers: A Possible Mild Form of Galloway-Mowat Syndrome. Journal of Child Neurology, 2003, 18, 147-149.	1.4	16
106	Efficacy of long term weekly ACTH therapy for intractable epilepsy. Brain and Development, 2015, 37, 449-454.	1.1	16
107	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.	2.3	16
108	Developmental changes of heat shock protein 73 in human brain. Developmental Brain Research, 1995, 86, 180-186.	1.7	15

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109	Fluctuation of Computed Tomographic Findings in White Matter in Alexander's Disease. Journal of Child Neurology, 2002, 17, 227-230.	1.4	15
110	Clinical and molecular cytogenetic characterization of two patients with non-mutational aberrations of theFMR2 gene. American Journal of Medical Genetics, Part A, 2007, 143A, 687-693.	1.2	15
111	Compound heterozygous PMP22 deletion mutations causing severe Charcotâ€“Marieâ€“Tooth disease type 1. Journal of Human Genetics, 2010, 55, 771-773.	2.3	15
112	Abnormal glucose metabolism in aromatic l-amino acid decarboxylase deficiency. Brain and Development, 2010, 32, 506-510.	1.1	14
113	The incidence of hypoplasia of the corpus callosum in patients with dup (X)(q28) involving MECP2 is associated with the location of distal breakpoints. American Journal of Medical Genetics, Part A, 2012, 158A, 1292-1303.	1.2	14
114	Abnormal pupillary light reflex with chromatic pupillometry in <scp>G</scp> aucher disease. Annals of Clinical and Translational Neurology, 2014, 1, 135-140.	3.7	14
115	A combination of targeted enrichment methodologies for whole-exome sequencing reveals novel pathogenic mutations. Scientific Reports, 2015, 5, 9331.	3.3	14
116	Clinical manifestations and epilepsy treatment in Japanese patients with pathogenic CDKL5 variants. Brain and Development, 2021, 43, 505-514.	1.1	14
117	Sonic hedgehog signal peptide mutation in a patient with holoprosencephaly. Annals of Neurology, 2000, 47, 514-516.	5.3	13
118	A case of Baraitserâ€“Winter syndrome with unusual brain MRI findings: Pachygyria, subcortical-band heterotopia, and periventricular heterotopia. Brain and Development, 2010, 32, 502-505.	1.1	13
119	Role of a heterotrimeric Gâ€protein, Gi2, in the corticogenesis: possible involvement in periventricular nodular heterotopia and intellectual disability. Journal of Neurochemistry, 2017, 140, 82-95.	3.9	13
120	A recurrent homozygous NHLRC1 variant in siblings with Lafora disease. Human Genome Variation, 2018, 5, 16.	0.7	13
121	A novel de novo frameshift variant in SETD1B causes epilepsy. Journal of Human Genetics, 2019, 64, 821-827.	2.3	13
122	Sirolimus for epileptic seizures associated with focal cortical dysplasia type <scp>II</scp>. Annals of Clinical and Translational Neurology, 2022, 9, 181-192.	3.7	13
123	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.	8.2	13
124	Novel compound heterozygous variants in PLK4 identified in a patient with autosomal recessive microcephaly and chorioretinopathy. European Journal of Human Genetics, 2016, 24, 1702-1706.	2.8	12
125	MYCNde novo gain-of-function mutation in a patient with a novel megalencephaly syndrome. Journal of Medical Genetics, 2019, 56, 388-395.	3.2	12
126	Dopaminergic restoration of prefrontal cortico-putaminal network in gene therapy for aromatic <scp>l</scp>-amino acid decarboxylase deficiency. Brain Communications, 2021, 3, fcab078.	3.3	12

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127	A severe pulmonary complication in a patient with COL4A1 -related disorder: A case report. European Journal of Medical Genetics, 2017, 60, 169-171.	1.3	11
128	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
129	Brainstem lesion in Aicardi-GoutiÃ¨res syndrome. Pediatric Neurology, 1998, 19, 145-147.	2.1	10
130	Haploinsufficiency of STXBP1 and Ohtahara syndrome. Epilepsia, 2010, 51, 71-71.	5.1	10
131	Serum and CSF biomarkers in acute pediatric neurological disorders. Brain and Development, 2014, 36, 489-495.	1.1	10
132	Sudden death in a case of megalencephaly capillary malformation associated with a de novo mutation in AKT3. Child's Nervous System, 2015, 31, 465-471.	1.1	10
133	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome. Journal of Human Genetics, 2018, 63, 957-963.	2.3	10
134	A missense variant of SMC1A causes periodic pharmaco-resistant cluster seizures similar to PCDH19-related epilepsy. Epilepsy Research, 2019, 155, 106149.	1.6	10
135	Immunohistochemical and morphometrical development of the dorsal root ganglion as a neural crest derivative: Comparison with the fetal CNS. Early Human Development, 1994, 38, 81-90.	1.8	9
136	Progressive Sliding Hiatal Hernia as a Complication of Menkes' Syndrome. Journal of Child Neurology, 2002, 17, 401-402.	1.4	9
137	A Mutation in the Tubulin-Encoding TUBB3 Gene Causes Complex Cortical Malformations and Unilateral Hypohidrosis. Child Neurology Open, 2016, 3, 2329048X1666575.	1.1	9
138	Siblings with optic neuropathy and RTN4IP1 mutation. Journal of Human Genetics, 2017, 62, 927-929.	2.3	8
139	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene. Brain and Development, 2018, 40, 819-823.	1.1	8
140	High Expression on Kunitz-Type Protease Inhibitor-Containing Substances in the Cerebral Vessels of Patients with Down Syndrome.. Tohoku Journal of Experimental Medicine, 1994, 174, 181-187.	1.2	7
141	Choreoâ€“ballistic movements in a case carrying a missense mutation in syntaxin binding protein 1 gene. Movement Disorders, 2010, 25, 2265-2267.	3.9	7
142	A novel genetic syndrome with STARD9 mutation and abnormal spindle morphology. American Journal of Medical Genetics, Part A, 2017, 173, 2690-2696.	1.2	7
143	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.	2.9	7
144	Four pedigrees with aminoacyl-tRNA synthetase abnormalities. Neurological Sciences, 2022, 43, 2765-2774.	1.9	7

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145	The â€˜double cortexâ€™ syndrome Commentary to Hashimoto's paper (pp. 57-9). Brain and Development, 1993, 15, 83-84.	1.1	6
146	Respiratory syncytial virusâ€associated encephalopathy complicated by congenital myopathy. Pediatrics International, 2012, 54, 709-711.	0.5	6
147	Primary Microcephaly With Anterior Predominant Pachygyria Caused by Novel Compound Heterozygous Mutations in ASPM. Pediatric Neurology, 2015, 52, e7-e8.	2.1	6
148	Megalencephaly, polymicrogyria and ribbon-like band heterotopia: A new cortical malformation. Brain and Development, 2016, 38, 950-953.	1.1	6
149	Loss of Neurofibromin in the Leptomeningeal Astroglial Heterotopia of NF-1. Pediatric Neurology, 1998, 18, 227-230.	2.1	5
150	l -Thyroxine-responsive drop attacks in childhood benign hereditary chorea: A case report. Brain and Development, 2018, 40, 353-356.	1.1	5
151	Progressive subglottic stenosis in a child with <scp>P</scp>allisterâ€<scp>K</scp>illian syndrome. Congenital Anomalies (discontinued), 2018, 58, 102-104.	0.6	5
152	A variant at 9q34.11 is associated with HLA-DQB1*06:02Ânegative essential hypersomnia. Journal of Human Genetics, 2018, 63, 1259-1267.	2.3	5
153	Congenital Dysplastic Microcephaly and Hypoplasia of the Brainstem and Cerebellum With Diffuse Intracranial Calcification. Journal of Child Neurology, 2012, 27, 218-221.	1.4	4
154	Association of early-onset epileptic encephalopathy with involuntary movements â€“ Case series and literature review. Epilepsy and Behavior Reports, 2021, 15, 100417.	1.0	4
155	Comprehensive genetic analysis confers high diagnostic yield in 16 Japanese patients with corpus callosum anomalies. Journal of Human Genetics, 2021, 66, 1061-1068.	2.3	4
156	A tyrosine kinase-like molecule is localized in the nuclear membrane of neurons: Hippocampal behavior under stress*. Biology of the Cell, 1996, 88, 45-54.	2.0	3
157	Polymicrogyria with calcification in Pallister-Killian syndrome detected by microarray analysis. Brain and Development, 2021, 43, 448-453.	1.1	3
158	Novel <scp>DCX</scp> mutationâ€caused lissencephaly in a boy and very mild heterotopia in his mother. Pediatrics International, 2015, 57, 321-323.	0.5	2
159	Epileptic encephalopathy patients with SCN2A variant initiated by neonatal seizure. Pediatrics International, 2021, 63, 971-972.	0.5	2
160	A Single-Arm Open-Label Clinical Trial on the Efficacy and Safety of Sirolimus for Epileptic Seizures Associated with Focal Cortical Dysplasia Type II: A Study Protocol. Kurume Medical Journal, 2019, 66, 115-120.	0.1	2
161	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	3.8	2
162	Neural Crest and Central Nervous System Malformation. Congenital Anomalies (discontinued), 1993, 33, 327-335.	0.6	1

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163	Going BAC or oligo microarray to the well: A commentary on Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. Journal of Human Genetics, 2011, 56, 104-105.	2.3	1
164	Unusual ribbonâ€like periventricular heterotopia with congenital cataracts in a Japanese girl. American Journal of Medical Genetics, Part A, 2012, 158A, 674-677.	1.2	1
165	Cerebrospinal fluid abnormalities in developmental and epileptic encephalopathy with a de novo CDK19 variant. Neurology: Genetics, 2020, 6, e527.	1.9	1
166	A boy with biallelic frameshift variants in TTC5 and brain malformation resembling tubulinopathies. Journal of Human Genetics, 2021, 66, 1189-1192.	2.3	1
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