Yoshinori Tsurusaki

List of Publications by Year
in descending order

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167	7,833	46918	64668
papers	citations	47 h-index	79 g-index

170	170	170	15196
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	9.4	435
2	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
3	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
4	Clinical spectrum of early onset epileptic encephalopathies caused by <scp>KCNQ2</scp> mutation. Epilepsia, 2013, 54, 1282-1287.	2.6	195
5	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. Neurology, 2013, 81, 992-998.	1.5	188
6	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
7	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	2.6	186
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	KDM6A Point Mutations Cause Kabuki Syndrome. Human Mutation, 2013, 34, 108-110.	1.1	168
10	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	0.7	148
11	Phenotypic Spectrum of COL4A1 Mutations: Porencephaly to Schizencephaly. Annals of Neurology, 2013, 73, 48-57.	2.8	143
12	Early onset epileptic encephalopathy caused by de novo <scp>SCN</scp>8A mutations. Epilepsia, 2014, 55, 994-1000.	2.6	142
13	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. Human Mutation, 2010, 31, 966-974.	1.1	137
14	Mutations in POLR3A and POLR3B Encoding RNA Polymerase III Subunits Cause an Autosomal-Recessive Hypomyelinating Leukoencephalopathy. American Journal of Human Genetics, 2011, 89, 644-651.	2.6	137
15	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
16	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
17	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	2.6	112
18	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

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19	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. Neurogenetics, 2013, 14, 225-232.	0.7	104
20	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. American Journal of Human Genetics, 2011, 88, 30-41.	2.6	100
21	The somatic GNAQ mutation c.548G>A (p.R183Q) is consistently found in Sturgeâ€“Weber syndrome. Journal of Human Genetics, 2014, 59, 691-693.	1.1	100
22	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
23	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	2.6	95
24	PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. Neurology, 2014, 82, 1587-1596.	1.5	93
25	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.	0.7	91
26	Biallelic Mutations in Nuclear Pore Complex Subunit NUP107 Cause Early-Childhood-Onset Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 97, 555-566.	2.6	91
27	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. Journal of Human Genetics, 2016, 61, 199-206.	1.1	91
28	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. Annals of Neurology, 2012, 72, 298-300.	2.8	88
29	De Novo Mutations inSLC35A2Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy. Human Mutation, 2013, 34, 1708-1714.	1.1	85
30	Exome Sequencing Reveals a Homozygous SYT14 Mutation in Adult-Onset, Autosomal-Recessive Spinocerebellar Ataxia with Psychomotor Retardation. American Journal of Human Genetics, 2011, 89, 320-327.	2.6	79
31	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.	2.6	79
32	Mitochondrial Complex III Deficiency Caused by a HomozygousUQCRC2Mutation Presenting with Neonatal-Onset Recurrent Metabolic Decompensation. Human Mutation, 2013, 34, 446-452.	1.1	79
33	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76
34	<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
35	De novo GABRA1 mutations in Ohtahara and West syndromes. Epilepsia, 2016, 57, 566-573.	2.6	76
36	De novo KCNB1 mutations in infantile epilepsy inhibit repetitive neuronal firing. Scientific Reports, 2015, 5, 15199.	1.6	73

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37	Deletions and de novo mutations ofSOX11are associated with a neurodevelopmental disorder with features of Coffinâ€“Siris syndrome. Journal of Medical Genetics, 2016, 53, 152-162.	1.5	69
38	CASK aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. Epilepsia, 2012, 53, 1441-1449.	2.6	66
39	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance. Neurogenetics, 2012, 13, 327-332.	0.7	66
40	Biallelic Mutations in MYPN , Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. American Journal of Human Genetics, 2017, 100, 169-178.	2.6	66
41	Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. Neurogenetics, 2014, 15, 193-200.	0.7	61
42	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	2.8	60
43	Overexpression of regucalcin modulates tumor-related gene expression in cloned rat hepatoma H4-II-E cells. Journal of Cellular Biochemistry, 2003, 90, 619-626.	1.2	59
44	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
45	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. Neurogenetics, 2014, 15, 85-92.	0.7	57
46	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
47	Missense mutations in the DNA-binding/dimerization domain of NFIX cause Sotos-like features. Journal of Human Genetics, 2012, 57, 207-211.	1.1	53
48	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
49	Role of endogenous regucalcin in transgenic rats: Suppression of kidney cortex cytosolic protein phosphatase activity and enhancement of heart muscle microsomal Ca2+-ATPase activity. Journal of Cellular Biochemistry, 2002, 86, 520-529.	1.2	47
50	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. Journal of Human Genetics, 2014, 59, 649-654.	1.1	46
51	Role of endogenous regucalcin in nuclear regulation of regenerating rat liver: Suppression of the enhanced ribonucleic acid synthesis activity. Journal of Cellular Biochemistry, 2002, 87, 450-457.	1.2	45
52	Expanding the phenotypic spectrum of TUBB4A -associated hypomyelinating leukoencephalopathies. Neurology, 2014, 82, 2230-2237.	1.5	45
53	Suppressive role of endogenous regucalcin in the enhancement of deoxyribonucleic acid synthesis activity in the nucleus of regenerating rat liver. Journal of Cellular Biochemistry, 2002, 85, 516-522.	1.2	43
54	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffinâ€“Siris-like syndrome. Journal of Human Genetics, 2017, 62, 741-746.	1.1	43

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55	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. Journal of Human Genetics, 2014, 59, 581-583.	1.1	42
56	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
57	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.	1.7	42
58	Compound heterozygous BRAT1 mutations cause familial Ohtahara syndrome with hypertonia and microcephaly. Journal of Human Genetics, 2014, 59, 687-690.	1.1	38
59	<scp>PIGO</scp> mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels. Epilepsia, 2014, 55, e13-7.	2.6	38
60	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. Journal of Human Genetics, 2016, 61, 381-387.	1.1	38
61	Homozygous splicing mutation in NUP133 causes Gallowayâ€“Mowat syndrome. Annals of Neurology, 2018, 84, 814-828.	2.8	37
62	Rapid detection of a mutation causing X-linked leucoencephalopathy by exome sequencing. Journal of Medical Genetics, 2011, 48, 606-609.	1.5	36
63	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
64	Role of regucalcin in liver nuclear function: binding of regucalcin to nuclear protein or DNA and modulation of tumor-related gene expression. International Journal of Molecular Medicine, 2004, 14, 277-81.	1.8	35
65	The first Japanese case of leukodystrophy with ovarian failure arising from novel compound heterozygous AARS2 mutations. Journal of Human Genetics, 2016, 61, 899-902.	1.1	33
66	Enhancement of albumin expression in bone tissues with healing rat fractures. Journal of Cellular Biochemistry, 2003, 89, 356-363.	1.2	31
67	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
68	Performance Comparison of Bench-Top Next Generation Sequencers Using Microdroplet PCR-Based Enrichment for Targeted Sequencing in Patients with Autism Spectrum Disorder. PLoS ONE, 2013, 8, e74167.	1.1	31
69	A novel WTX mutation in a female patient with osteopathia striata with cranial sclerosis and hepatoblastoma. American Journal of Medical Genetics, Part A, 2014, 164, 998-1002.	0.7	31
70	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. Genetics in Medicine, 2019, 21, 1629-1638.	1.1	31
71	Numerous BAF complex genes are mutated in Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 257-261.	0.7	29
72	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29

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73	The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. Journal of Human Genetics, 2013, 58, 113-115.	1.1	28
74	Aortic aneurysm and craniosynostosis in a family with Cantu syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 231-236.	0.7	28
75	Role of endogenous regucalcin in bone metabolism: bone loss is induced in regucalcin transgenic rats. International Journal of Molecular Medicine, 2002, 10, 377-83.	1.8	28
76	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. Journal of Human Genetics, 2016, 61, 835-838.	1.1	27
77	De novo <scp>DNM1</scp> mutations in two cases of epileptic encephalopathy. Epilepsia, 2016, 57, e18-23.	2.6	27
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.	0.7	26
79	A novel homozygous YARS2 mutation causes severe myopathy, lactic acidosis, and sideroblastic anemia 2. Journal of Human Genetics, 2014, 59, 229-232.	1.1	26
80	A novel UBE2A mutation causes X-linked intellectual disability type Nascimento. Human Genome Variation, 2017, 4, 17019.	0.4	26
81	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. Human Genetics, 2014, 133, 225-234.	1.8	25
82	Suppressive effect of endogenous regucalcin on nitric oxide synthase activity in cloned rat hepatoma H4-II-E cells overexpressing regucalcin. Journal of Cellular Biochemistry, 2003, 89, 800-807.	1.2	24
83	Pathogenic variants of DYNC2H1 , KIAA0556 , and PTPN11 associated with hypothalamic hamartoma. Neurology, 2019, 93, e237-e251.	1.5	24
84	Potential role of regucalcin as a specific biochemical marker of chronic liver injury with carbon tetrachloride administration in rats. Molecular and Cellular Biochemistry, 2002, 241, 61-67.	1.4	23
85	Mutations in the glutaminyl-tRNA synthetase gene cause early-onset epileptic encephalopathy. Journal of Human Genetics, 2015, 60, 97-101.	1.1	23
86	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	1.5	22
87	De novo 19q13.42 duplications involving NLRP gene cluster in a patient with systemic-onset juvenile idiopathic arthritis. Journal of Human Genetics, 2011, 56, 343-347.	1.1	21
88	Wholeâ€exome sequencing identified a homozygous FNBP4 mutation in a family with a condition similar to microphthalmia with limb anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 1543-1546.	0.7	21
89	Refining the clinical phenotype of Okurâ€“Chung neurodevelopmental syndrome. Human Genome Variation, 2018, 5, 18011.	0.4	21
90	A novel gene (FAM20B encoding glycosaminoglycan xylosylkinase) for neonatal short limb dysplasia resembling Desbuquois dysplasia. Clinical Genetics, 2019, 95, 713-717.	1.0	21

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91	Update of the genotype and phenotype of <scp>KMT2D</scp> and <scp>KDM6A</scp> by genetic screening of 100 patients with clinically suspected Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2333-2344.	0.7	21
92	Role of regucalcin in liver nuclear function: Binding of regucalcin to nuclear protein or DNA and modulation of tumor-related gene expression. International Journal of Molecular Medicine, 2004, 14, 277.	1.8	20
93	Causative novel PNKP mutations and concomitant PCDH15 mutations in a patient with microcephaly with early-onset seizures and developmental delay syndrome and hearing loss. Journal of Human Genetics, 2014, 59, 471-474.	1.1	20
94	A female patient with Xâ€linked Ohdo syndrome of the Maatâ€Kievitâ€Brunner phenotype caused by a novel variant of MED12. Congenital Anomalies (discontinued), 2020, 60, 91-93.	0.3	20
95	Suppressive role of endogenous regucalcin in the enhancement of nitric oxide synthase activity in liver cytosol of normal and regucalcin transgenic rats. Journal of Cellular Biochemistry, 2003, 88, 1226-1234.	1.2	19
96	Growth Inhibition of Cultured Human Liver Carcinoma Cells by Ki-energy (Life-energy): Scientific Evidence for Ki-effects on Cancer Cells. Evidence-based Complementary and Alternative Medicine, 2005, 2, 387-393.	0.5	19
97	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
98	Rapid detection of gene mutations responsible for non-syndromic aortic aneurysm and dissection using two different methods: resequencing microarray technology and next-generation sequencing. Human Genetics, 2012, 131, 591-599.	1.8	18
99	SMARCE1, a rare cause of Coffinâ€“Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	0.7	18
100	Coffinâ€Siris syndrome and cardiac anomaly with a novel SOX11 mutation. Congenital Anomalies (discontinued), 2018, 58, 105-107.	0.3	18
101	A recurrent PJA1 variant in trigonocephaly and neurodevelopmental disorders. Annals of Clinical and Translational Neurology, 2020, 7, 1117-1131.	1.7	18
102	Novel FIG4 mutations in Yunisâ€“Varon syndrome. Journal of Human Genetics, 2013, 58, 822-824.	1.1	17
103	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. Journal of Human Genetics, 2015, 60, 631-635.	1.1	17
104	De novo deletion of 1q24.3â€q31.2 in a patient with severe growth retardation. American Journal of Medical Genetics, Part A, 2010, 152A, 1322-1325.	0.7	16
105	A family of oculofaciocardiodental syndrome (OFCD) with a novel BCOR mutation and genomic rearrangements involving NHS. Journal of Human Genetics, 2012, 57, 197-201.	1.1	16
106	A girl with early-onset epileptic encephalopathy associated with microdeletion involving CDKL5. Brain and Development, 2012, 34, 364-367.	0.6	16
107	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. Journal of Human Genetics, 2013, 58, 391-394.	1.1	16
108	A de novo CASK mutation in pontocerebellar hypoplasia type 3 with early myoclonic epilepsy and tetralogy of Fallot. Brain and Development, 2014, 36, 272-273.	0.6	16

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109	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioleiomyomatosis tissues by deep sequencing. Human Genetics, 2016, 135, 61-68.	1.8	16
110	Mandibulofacial dysostosis with microcephaly: A case presenting with seizures. Brain and Development, 2017, 39, 177-181.	0.6	16
111	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
112	Severe manifestations of handâ€footâ€genital syndrome associated with a novel HOXA13 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2398-2402.	0.7	15
113	Deep sequencing detects very-low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy. Neuromuscular Disorders, 2014, 24, 642-647.	0.3	15
114	Homozygous p.V116* mutation inC12orf65results in Leigh syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310084.	0.9	15
115	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
116	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. Journal of Human Genetics, 2018, 63, 417-423.	1.1	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	Novel COL4A1 mutation in a fetus with early prenatal onset of schizencephaly. Human Genome Variation, 2018, 5, 4.	0.4	14
119	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103610.	0.7	14
120	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
121	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
122	A de novo 1.4â€Mb deletion at 21q22.11 in a boy with developmental delay. American Journal of Medical Genetics, Part A, 2014, 164, 1021-1028.	0.7	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.	3.6	13
124	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. Human Genome Variation, 2015, 2, 15034.	0.4	12
125	Novel AMER1 frameshift mutation in a girl with osteopathia striata with cranial sclerosis. Congenital Anomalies (discontinued), 2018, 58, 145-146.	0.3	12
126	Duplication of the NPHP1 gene in patients with autism spectrum disorder and normal intellectual ability: a case series. Annals of General Psychiatry, 2014, 13, 22.	1.2	11

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127	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. Human Genome Variation, 2015, 2, 15024.	0.4	11
128	Familial schwannomatosis with a germline mutation of SMARCB1 in Japan. Brain Tumor Pathology, 2015, 32, 216-220.	1.1	11
129	Dual genetic diagnoses: Atypical handâ€footâ€genital syndrome and developmental delay due to de novo mutations in HOXA13 and NRXN1. American Journal of Medical Genetics, Part A, 2016, 170, 717-724.	0.7	11
130	A de novo deletion of 20q11.2â€“q12 in a boy presenting with abnormal hands and feet, retinal dysplasia, and intractable feeding difficulty. American Journal of Medical Genetics, Part A, 2011, 155, 409-414.	0.7	10
131	Mutations in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease. Brain and Development, 2015, 37, 960-966.	0.6	10
132	A novel <scp>SCARB</scp>2 mutation causing lateâ€onset progressive myoclonus epilepsy. Movement Disorders, 2013, 28, 552-553.	2.2	9
133	Identification of a Novel Homozygous SPG7 Mutation in a Japanese Patient with Spastic Ataxia: Making an Efficient Diagnosis Using Exome Sequencing for Autosomal Recessive Cerebellar Ataxia and Spastic Paraplegia. Internal Medicine, 2013, 52, 1629-1633.	0.3	9
134	Novel SYNGAP1 variant in a patient with intellectual disability and distinctive dysmorphisms. Congenital Anomalies (discontinued), 2018, 58, 188-190.	0.3	9
135	Nonsyndromic intellectual disability with novel heterozygous SCN2A mutation and epilepsy. Human Genome Variation, 2018, 5, 20.	0.4	9
136	Two unrelated girls with intellectual disability associated with a truncating mutation in the PPM1D penultimate exon. Brain and Development, 2019, 41, 538-541.	0.6	9
137	Role of endogenous regucalcin in the regulation of Ca2+-ATPase activity in rat liver nuclei. Journal of Cellular Biochemistry, 2000, 78, 541-549.	1.2	8
138	Suppressive Effect of Endogenous Regucalcin on Guanosine Triphosphatase Activity in Rat Liver Nucleus.. Biological and Pharmaceutical Bulletin, 2001, 24, 958-961.	0.6	8
139	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
140	A novel PITX2 mutation causing iris hypoplasia. Human Genome Variation, 2014, 1, 14005.	0.4	8
141	A Japanese girl with an early-infantile onset vanishing white matter disease resembling Cree leukoencephalopathy. Brain and Development, 2015, 37, 638-642.	0.6	8
142	Biallelic mutations of EGFR in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction. Human Genome Variation, 2018, 5, 11.	0.4	7
143	Novel CUL7 biallelic mutations alter the skeletal phenotype of 3M syndrome. Human Genome Variation, 2020, 7, 1.	0.4	7
144	Coâ€occurrence of 22q11 deletion syndrome and hdr syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2576-2581.	0.7	6

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145	White matter abnormalities in an adult patient with l-2-hydroxyglutaric aciduria. Brain and Development, 2016, 38, 142-144.	0.6	6
146	A severe form of Ellis-van Creveld syndrome caused by novel mutations in EVC2. Human Genome Variation, 2019, 6, 40.	0.4	6
147	Discordant phenotype caused by CASK mutation in siblings with NF1. Human Genome Variation, 2019, 6, 20.	0.4	6
148	Novel USP9X variants in two patients with X-linked intellectual disability. Human Genome Variation, 2019, 6, 49.	0.4	6
149	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. Brain and Development, 2020, 42, 289-292.	0.6	6
150	A Recurrent Variant in <i>POLR1B</i>, c.3007C>T; p.Arg1003Cys, Associated with Atresia of the External Canal and Microtia in Treacher Collins Syndrome Type 4. Molecular Syndromology, 2021, 12, 127-132.	0.3	6
151	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
152	Wholeâ€exome sequencing reveals the subclonal expression of NUP214â€ABL1 fusion gene in Tâ€cell acute lymphoblastic leukemia. Pediatric Blood and Cancer, 2020, 67, e28019.	0.8	5
153	Divergent variant patterns among 19 patients with<scp>Rubinsteinâ€Taybi</scp>syndrome uncovered by comprehensive genetic analysis including whole genome sequencing. Clinical Genetics, 2022, 101, 335-345.	1.0	5
154	Role of endogenous regucalcin in transgenic rats: suppression of protein tyrosine phosphatase and ribonucleic acid synthesis activities in liver nucleus. International Journal of Molecular Medicine, 2003, 12, 207-11.	1.8	5
155	POLR1C variants dysregulate splicing and cause hypomyelinating leukodystrophy. Neurology: Genetics, 2020, 6, e524.	0.9	4
156	An efficient genetic test flow for multiple congenital anomalies and intellectual disability. Pediatrics International, 2020, 62, 556-561.	0.2	4
157	Exome sequencing analysis of Japanese autism spectrum disorder case-control sample supports an increased burden of synaptic function-related genes. Translational Psychiatry, 2022, 12, .	2.4	4
158	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.	0.7	3
159	<scp>J</scp>apanese familial case of myoclonusâ€“dystonia syndrome with a splicing mutation in <scp>SGCE</scp>. Pediatrics International, 2015, 57, 324-326.	0.2	2
160	TMEM67 mutations found in a case of Joubert syndrome with renal hypodysplasia. CEN Case Reports, 2016, 5, 137-140.	0.5	2
161	Role of endogenous regucalcin in transgenic rats: Suppression of protein tyrosine phosphatase and ribonucleic acid synthesis activities in liver nucleus. International Journal of Molecular Medicine, 2003, 12, 207.	1.8	1
162	Hemoglobin beta Kanagawa [c.443A>C; p.(Ter148Serext*21)]: A novel Î²â€globin gene mutation causing dominantly inherited Î²â€thalassemia. Pediatric Blood and Cancer, 2019, 66, e27871.	0.8	1

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