

Yoshinori Tsurusaki

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

164
papers

6,024
citations

44
h-index

71
g-index

170
ext. papers

7,092
ext. citations

4.8
avg, IF

4.97
L-index

#	Paper	IF	Citations
164	Further delineation of SET-related intellectual disability syndrome.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	
163	Large-scale discovery of novel neurodevelopmental disorder-related genes through a unified analysis of single-nucleotide and copy number variants.. <i>Genome Medicine</i> , 2022 , 14, 40	14.4	0
162	Novel COL2A1 variants in Japanese patients with spondyloepiphyseal dysplasia congenita.. <i>Human Genome Variation</i> , 2022 , 9, 16	1.8	
161	Multiple alterations in glutamatergic transmission and dopamine D2 receptor splicing in induced pluripotent stem cell-derived neurons from patients with familial schizophrenia. <i>Translational Psychiatry</i> , 2021 , 11, 548	8.6	1
160	Prenatal clinical manifestations in individuals with variants. <i>Journal of Medical Genetics</i> , 2021 , 58, 505-513	3.8	11
159	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. <i>Human Mutation</i> , 2021 , 42, 66-76	4.7	4
158	A novel method for isolating lymphatic endothelial cells from lymphatic malformations and detecting PIK3CA somatic mutation in these isolated cells. <i>Surgery Today</i> , 2021 , 51, 439-446	3	1
157	A Recurrent Variant in , c.3007C>T; p.Arg1003Cys, Associated with Atresia of the External Canal and Microtia in Treacher Collins Syndrome Type 4. <i>Molecular Syndromology</i> , 2021 , 12, 127-132	1.5	0
156	Siblings with vascular Ehlers-Danlos syndrome inherited via maternal mosaicism. <i>Congenital Anomalies (discontinued)</i> , 2021 , 61, 101-102	1.1	0
155	A recurrent PJA1 variant in trigonocephaly and neurodevelopmental disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 1117-1131	5.3	5
154	Expanding the phenotype of COL4A1-related disorders-Four novel variants. <i>Brain and Development</i> , 2020 , 42, 639-645	2.2	0
153	Novel biallelic mutations alter the skeletal phenotype of 3M syndrome. <i>Human Genome Variation</i> , 2020 , 7, 1	1.8	4
152	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. <i>Brain and Development</i> , 2020 , 42, 289-292	2.2	3
151	An efficient genetic test flow for multiple congenital anomalies and intellectual disability. <i>Pediatrics International</i> , 2020 , 62, 556-561	1.2	2
150	Whole-exome sequencing reveals the subclonal expression of NUP214-ABL1 fusion gene in T-cell acute lymphoblastic leukemia. <i>Pediatric Blood and Cancer</i> , 2020 , 67, e28019	3	2
149	variants dysregulate splicing and cause hypomyelinating leukodystrophy. <i>Neurology: Genetics</i> , 2020 , 6, e524	3.8	0
148	Update of the genotype and phenotype of KMT2D and KDM6A by genetic screening of 100 patients with clinically suspected Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2333-2344	2.5	8

147	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103610	2.6	9
146	A female patient with X-linked Ohdo syndrome of the Maat-Kievit-Brunner phenotype caused by a novel variant of MED12. <i>Congenital Anomalies (discontinued)</i> , 2020 , 60, 91-93	1.1	12
145	A severe form of Ellis-van Creveld syndrome caused by novel mutations in. <i>Human Genome Variation</i> , 2019 , 6, 40	1.8	4
144	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. <i>Journal of Human Genetics</i> , 2019 , 64, 1173-1186	4.3	20
143	Discordant phenotype caused by mutation in siblings with. <i>Human Genome Variation</i> , 2019 , 6, 20	1.8	4
142	Hemoglobin beta Kanagawa [c.443A>C; p.(Ter148Serext*21)]: A novel β globin gene mutation causing dominantly inherited β thalassemia. <i>Pediatric Blood and Cancer</i> , 2019 , 66, e27871	3	0
141	A novel gene (FAM20B encoding glycosaminoglycan xylosylkinase) for neonatal short limb dysplasia resembling Desbuquois dysplasia. <i>Clinical Genetics</i> , 2019 , 95, 713-717	4	11
140	Two unrelated girls with intellectual disability associated with a truncating mutation in the PPM1D penultimate exon. <i>Brain and Development</i> , 2019 , 41, 538-541	2.2	7
139	Pathogenic variants of , , and associated with hypothalamic hamartoma. <i>Neurology</i> , 2019 , 93, e237-e251	6.5	8
138	Novel variants in two patients with X-linked intellectual disability. <i>Human Genome Variation</i> , 2019 , 6, 49	1.8	2
137	RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. <i>Genetics in Medicine</i> , 2019 , 21, 1629-1638	8.1	17
136	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. <i>Annals of Neurology</i> , 2018 , 83, 794-806	9.4	37
135	Novel SYNGAP1 variant in a patient with intellectual disability and distinctive dysmorphisms. <i>Congenital Anomalies (discontinued)</i> , 2018 , 58, 188-190	1.1	8
134	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. <i>Journal of Human Genetics</i> , 2018 , 63, 417-423	4.3	10
133	Novel AMER1 frameshift mutation in a girl with osteopathia striata with cranial sclerosis. <i>Congenital Anomalies (discontinued)</i> , 2018 , 58, 145-146	1.1	11
132	Nonsyndromic intellectual disability with novel heterozygous mutation and epilepsy. <i>Human Genome Variation</i> , 2018 , 5, 20	1.8	6
131	Novel mutation in a fetus with early prenatal onset of schizencephaly. <i>Human Genome Variation</i> , 2018 , 5, 4	1.8	12
130	Refining the clinical phenotype of Okur-Chung neurodevelopmental syndrome. <i>Human Genome Variation</i> , 2018 , 5, 18011	1.8	12

129	Coffin-Siris syndrome and cardiac anomaly with a novel SOX11 mutation. <i>Congenital Anomalies (discontinued)</i> , 2018 , 58, 105-107	1.1	10
128	Homozygous splicing mutation in NUP133 causes Galloway-Mowat syndrome. <i>Annals of Neurology</i> , 2018 , 84, 814-828	9.4	19
127	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. <i>Cell Reports</i> , 2018 , 22, 734-747	10.6	86
126	Biallelic mutations of in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction. <i>Human Genome Variation</i> , 2018 , 5, 11	1.8	3
125	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffin-Siris-like syndrome. <i>Journal of Human Genetics</i> , 2017 , 62, 741-746	4.3	31
124	A novel mutation causes X-linked intellectual disability type Nascimento. <i>Human Genome Variation</i> , 2017 , 4, 17019	1.8	20
123	Biallelic Mutations in MYPN, Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2017 , 100, 169-178	11	57
122	Mandibulofacial dysostosis with microcephaly: A case presenting with seizures. <i>Brain and Development</i> , 2017 , 39, 177-181	2.2	13
121	Human genetic variation database, a reference database of genetic variations in the Japanese population. <i>Journal of Human Genetics</i> , 2016 , 61, 547-53	4.3	212
120	De novo GABRA1 mutations in Ohtahara and West syndromes. <i>Epilepsia</i> , 2016 , 57, 566-73	6.4	50
119	TMEM67 mutations found in a case of Joubert syndrome with renal hypodysplasia. <i>CEN Case Reports</i> , 2016 , 5, 137-140	1	2
118	The first Japanese case of leukodystrophy with ovarian failure arising from novel compound heterozygous AARS2 mutations. <i>Journal of Human Genetics</i> , 2016 , 61, 899-902	4.3	28
117	Dual genetic diagnoses: Atypical hand-foot-genital syndrome and developmental delay due to de novo mutations in HOXA13 and NRXN1. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 717-24	2.5	6
116	Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. <i>European Journal of Human Genetics</i> , 2016 , 24, 129-34	5.3	72
115	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. <i>Journal of Human Genetics</i> , 2016 , 61, 381-7	4.3	30
114	White matter abnormalities in an adult patient with l-2-hydroxyglutaric aciduria. <i>Brain and Development</i> , 2016 , 38, 142-4	2.2	4
113	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioliomyomatosis tissues by deep sequencing. <i>Human Genetics</i> , 2016 , 135, 61-8	6.3	13
112	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. <i>Journal of Human Genetics</i> , 2016 , 61, 199-206	4.3	52

111	Homozygous p.V116* mutation in C12orf65 results in Leigh syndrome. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016 , 87, 212-6	5.5	12
110	SMARCE1, a rare cause of Coffin-Siris Syndrome: Clinical description of three additional cases. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 1967-73	2.5	15
109	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2662-70	2.5	13
108	Deletions and de novo mutations of SOX11 are associated with a neurodevelopmental disorder with features of Coffin-Siris syndrome. <i>Journal of Medical Genetics</i> , 2016 , 53, 152-62	5.8	41
107	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. <i>Journal of Human Genetics</i> , 2016 , 61, 835-8	4.3	24
106	Somatic mutations in GLI3 and OFD1 involved in sonic hedgehog signaling cause hypothalamic hamartoma. <i>Annals of Clinical and Translational Neurology</i> , 2016 , 3, 356-65	5.3	24
105	De novo DNM1 mutations in two cases of epileptic encephalopathy. <i>Epilepsia</i> , 2016 , 57, e18-23	6.4	17
104	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an @xome-first@approach. <i>Journal of Human Genetics</i> , 2015 , 60, 175-82	4.3	41
103	Familial schwannomatosis with a germline mutation of SMARCB1 in Japan. <i>Brain Tumor Pathology</i> , 2015 , 32, 216-20	3.2	9
102	A case of autism spectrum disorder arising from a de novo missense mutation in POGZ. <i>Journal of Human Genetics</i> , 2015 , 60, 277-9	4.3	33
101	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. <i>Journal of Human Genetics</i> , 2015 , 60, 631-5	4.3	14
100	Mutations in the glutamyl-tRNA synthetase gene cause early-onset epileptic encephalopathy. <i>Journal of Human Genetics</i> , 2015 , 60, 97-101	4.3	19
99	A Japanese girl with an early-infantile onset vanishing white matter disease resembling Cree leukoencephalopathy. <i>Brain and Development</i> , 2015 , 37, 638-42	2.2	6
98	Mutations in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease. <i>Brain and Development</i> , 2015 , 37, 960-6	2.2	6
97	Biallelic Mutations in Nuclear Pore Complex Subunit NUP107 Cause Early-Childhood-Onset Steroid-Resistant Nephrotic Syndrome. <i>American Journal of Human Genetics</i> , 2015 , 97, 555-66	11	68
96	DNA methylation and gene expression dynamics during spermatogonial stem cell differentiation in the early postnatal mouse testis. <i>BMC Genomics</i> , 2015 , 16, 624	4.5	84
95	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. <i>Journal of Human Genetics</i> , 2015 , 60, 739-42	4.3	37
94	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. <i>Epilepsia</i> , 2015 , 56, 841-8	6.4	56

93	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. <i>Human Genome Variation</i> , 2015 , 2, 15024	1.8	9
92	De novo KCNB1 mutations in infantile epilepsy inhibit repetitive neuronal firing. <i>Scientific Reports</i> , 2015 , 5, 15199	4.9	54
91	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. <i>Human Genome Variation</i> , 2015 , 2, 15034	1.8	9
90	De novo KCNT1 mutations in early-onset epileptic encephalopathy. <i>Epilepsia</i> , 2015 , 56, e121-8	6.4	66
89	Somatic Mutations in the MTOR gene cause focal cortical dysplasia type IIb. <i>Annals of Neurology</i> , 2015 , 78, 375-86	9.4	129
88	Japanese familial case of myoclonus-dystonia syndrome with a splicing mutation in SGCE. <i>Pediatrics International</i> , 2015 , 57, 324-6	1.2	1
87	A Japanese case of cerebellar ataxia, spastic paraparesis and deep sensory impairment associated with a novel homozygous TTC19 mutation. <i>Journal of Human Genetics</i> , 2015 , 60, 187-91	4.3	10
86	A de novo 1.4-Mb deletion at 21q22.11 in a boy with developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1021-8	2.5	13
85	Duplication of the NPHP1 gene in patients with autism spectrum disorder and normal intellectual ability: a case series. <i>Annals of General Psychiatry</i> , 2014 , 13, 22	3.4	7
84	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. <i>Journal of Human Genetics</i> , 2014 , 59, 649-54	4.3	40
83	Causative novel PNKP mutations and concomitant PCDH15 mutations in a patient with microcephaly with early-onset seizures and developmental delay syndrome and hearing loss. <i>Journal of Human Genetics</i> , 2014 , 59, 471-4	4.3	16
82	De novo SOX11 mutations cause Coffin-Siris syndrome. <i>Nature Communications</i> , 2014 , 5, 4011	17.4	84
81	Numerous BAF complex genes are mutated in Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014 , 166C, 257-61	3.1	21
80	Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. <i>Neurogenetics</i> , 2014 , 15, 193-200	3	54
79	Early onset epileptic encephalopathy caused by de novo SCN8A mutations. <i>Epilepsia</i> , 2014 , 55, 994-1000	6.4	122
78	A de novo CASK mutation in pontocerebellar hypoplasia type 3 with early myoclonic epilepsy and tetralogy of Fallot. <i>Brain and Development</i> , 2014 , 36, 272-3	2.2	11
77	Deep sequencing detects very-low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy. <i>Neuromuscular Disorders</i> , 2014 , 24, 642-7	2.9	11
76	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. <i>Journal of Human Genetics</i> , 2014 , 59, 292-5	4.3	44

75	A novel PITX2 mutation causing iris hypoplasia. <i>Human Genome Variation</i> , 2014 , 1, 14005	1.8	6
74	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. <i>Scientific Reports</i> , 2014 , 4, 7132	4.9	16
73	Cerebellar atrophy dwindles away in the era of next-generation sequencing. <i>Journal of Human Genetics</i> , 2014 , 59, 589-90	4.3	7
72	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. <i>Journal of Human Genetics</i> , 2014 , 59, 581-3	4.3	34
71	A novel homozygous YARS2 mutation causes severe myopathy, lactic acidosis, and sideroblastic anemia 2. <i>Journal of Human Genetics</i> , 2014 , 59, 229-32	4.3	23
70	Compound heterozygous BRAT1 mutations cause familial Ohtahara syndrome with hypertonia and microcephaly. <i>Journal of Human Genetics</i> , 2014 , 59, 687-90	4.3	30
69	Expanding the phenotypic spectrum of TUBB4A-associated hypomyelinating leukoencephalopathies. <i>Neurology</i> , 2014 , 82, 2230-7	6.5	38
68	PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. <i>Neurology</i> , 2014 , 82, 1587-96	6.5	80
67	Aortic aneurysm and craniosynostosis in a family with Cantu syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 231-6	2.5	27
66	A novel WTX mutation in a female patient with osteopathia striata with cranial sclerosis and hepatoblastoma. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 998-1002	2.5	19
65	Severe manifestations of hand-foot-genital syndrome associated with a novel HOXA13 mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2398-402	2.5	12
64	PIGO mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels. <i>Epilepsia</i> , 2014 , 55, e13-7	6.4	31
63	The somatic GNAQ mutation c.548G>A (p.R183Q) is consistently found in Sturge-Weber syndrome. <i>Journal of Human Genetics</i> , 2014 , 59, 691-3	4.3	70
62	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. <i>Human Genetics</i> , 2014 , 133, 225-34	6.3	15
61	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. <i>Neurogenetics</i> , 2014 , 15, 85-92	3	50
60	De Novo mutations in GNAO1, encoding a Gβ subunit of heterotrimeric G proteins, cause epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2013 , 93, 496-505	11	137
59	Mutations in KLHL40 are a frequent cause of severe autosomal-recessive nemaline myopathy. <i>American Journal of Human Genetics</i> , 2013 , 93, 6-18	11	142
58	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. <i>Neurogenetics</i> , 2013 , 14, 225-32	3	91

57	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. <i>Epilepsia</i> , 2013 , 54, 1262-9	6.4	64
56	De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy. <i>Human Mutation</i> , 2013 , 34, 1708-14	4.7	72
55	KDM6A point mutations cause Kabuki syndrome. <i>Human Mutation</i> , 2013 , 34, 108-10	4.7	129
54	Mutations in B3GALT6, which encodes a glycosaminoglycan linker region enzyme, cause a spectrum of skeletal and connective tissue disorders. <i>American Journal of Human Genetics</i> , 2013 , 92, 927-34	11	91
53	Clinical spectrum of early onset epileptic encephalopathies caused by KCNQ2 mutation. <i>Epilepsia</i> , 2013 , 54, 1282-7	6.4	143
52	A unique case of de novo 5q33.3-q34 triplication with uniparental isodisomy of 5q34-qter. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1904-9	2.5	11
51	Mitochondrial complex III deficiency caused by a homozygous UQCRC2 mutation presenting with neonatal-onset recurrent metabolic decompensation. <i>Human Mutation</i> , 2013 , 34, 446-52	4.7	68
50	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. <i>Nature Genetics</i> , 2013 , 45, 445-9, 449e1	36.3	330
49	Clinical correlations of mutations affecting six components of the SWI/SNF complex: detailed description of 21 patients and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1221-37	2.5	74
48	Phenotypic spectrum of COL4A1 mutations: porencephaly to schizencephaly. <i>Annals of Neurology</i> , 2013 , 73, 48-57	9.4	117
47	Whole-exome sequencing identified a homozygous FBNP4 mutation in a family with a condition similar to microphthalmia with limb anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1543-6	2.5	17
46	The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. <i>Journal of Human Genetics</i> , 2013 , 58, 113-5	4.3	27
45	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. <i>Neurology</i> , 2013 , 81, 992-8	6.5	158
44	Co-occurrence of 22q11 deletion syndrome and HDR syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2576-81	2.5	3
43	Novel FIG4 mutations in Yunis-Varon syndrome. <i>Journal of Human Genetics</i> , 2013 , 58, 822-4	4.3	15
42	MLL2 and KDM6A mutations in patients with Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2234-43	2.5	111
41	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. <i>Journal of Human Genetics</i> , 2013 , 58, 391-4	4.3	15
40	A novel SCARB2 mutation causing late-onset progressive myoclonus epilepsy. <i>Movement Disorders</i> , 2013 , 28, 552-3	7	7

39	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. <i>Internal Medicine</i> , 2013 , 52, 1629-33	1.1	9
38	Performance comparison of bench-top next generation sequencers using microdroplet PCR-based enrichment for targeted sequencing in patients with autism spectrum disorder. <i>PLoS ONE</i> , 2013 , 8, e74167	3.7	24
37	A girl with early-onset epileptic encephalopathy associated with microdeletion involving CDKL5. <i>Brain and Development</i> , 2012 , 34, 364-7	2.2	14
36	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slit-Robo Rho GTPase activating protein 2 (SRGAP2). <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 199-205	2.5	25
35	CASK aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. <i>Epilepsia</i> , 2012 , 53, 1441-9	6.4	51
34	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance. <i>Neurogenetics</i> , 2012 , 13, 327-32	3	55
33	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. <i>Annals of Neurology</i> , 2012 , 72, 298-300	9.4	74
32	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. <i>Nature Genetics</i> , 2012 , 44, 376-8	36.3	350
31	Missense mutations in the DNA-binding/dimerization domain of NFIX cause Sotos-like features. <i>Journal of Human Genetics</i> , 2012 , 57, 207-11	4.3	46
30	Rapid detection of gene mutations responsible for non-syndromic aortic aneurysm and dissection using two different methods: resequencing microarray technology and next-generation sequencing. <i>Human Genetics</i> , 2012 , 131, 591-9	6.3	18
29	De novo and inherited mutations in COL4A2, encoding the type IV collagen α chain cause porencephaly. <i>American Journal of Human Genetics</i> , 2012 , 90, 86-90	11	63
28	A family of oculofaciocardiodental syndrome (OFCD) with a novel BCOR mutation and genomic rearrangements involving NHS. <i>Journal of Human Genetics</i> , 2012 , 57, 197-201	4.3	13
27	De novo 19q13.42 duplications involving NLRP gene cluster in a patient with systemic-onset juvenile idiopathic arthritis. <i>Journal of Human Genetics</i> , 2011 , 56, 343-7	4.3	17
26	SMOC1 is essential for ocular and limb development in humans and mice. <i>American Journal of Human Genetics</i> , 2011 , 88, 30-41	11	82
25	Exome sequencing reveals a homozygous SYT14 mutation in adult-onset, autosomal-recessive spinocerebellar ataxia with psychomotor retardation. <i>American Journal of Human Genetics</i> , 2011 , 89, 320-7	11	60
24	Mutations in POLR3A and POLR3B encoding RNA Polymerase III subunits cause an autosomal-recessive hypomyelinating leukoencephalopathy. <i>American Journal of Human Genetics</i> , 2011 , 89, 644-51	11	112
23	A de novo deletion of 20q11.2-q12 in a boy presenting with abnormal hands and feet, retinal dysplasia, and intractable feeding difficulty. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 409-14	2.5	9
22	De novo 5q14.3 translocation 121.5-kb upstream of MEF2C in a patient with severe intellectual disability and early-onset epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2879-84	2.5	21

21	Rapid detection of a mutation causing X-linked leucoencephalopathy by exome sequencing. <i>Journal of Medical Genetics</i> , 2011 , 48, 606-9	5.8	34
20	Breakpoint determination of X;autosome balanced translocations in four patients with premature ovarian failure. <i>Journal of Human Genetics</i> , 2011 , 56, 156-60	4.3	17
19	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. <i>Human Mutation</i> , 2010 , 31, 966-74	4.7	119
18	De novo deletion of 1q24.3-q31.2 in a patient with severe growth retardation. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1322-5	2.5	15
17	Growth inhibition of cultured human liver carcinoma cells by Ki-energy (life-energy): scientific evidence for Ki-effects on cancer cells. <i>Evidence-based Complementary and Alternative Medicine</i> , 2005 , 2, 387-93	2.3	17
16	Suppressive effect of regucalcin on protein phosphatase activity in the heart cytosol of normal and regucalcin transgenic rats. <i>International Journal of Molecular Medicine</i> , 2004 , 13, 289	4.4	
15	Role of regucalcin in liver nuclear function: Binding of regucalcin to nuclear protein or DNA and modulation of tumor-related gene expression. <i>International Journal of Molecular Medicine</i> , 2004 , 14, 277	4.4	4
14	Role of regucalcin in liver nuclear function: binding of regucalcin to nuclear protein or DNA and modulation of tumor-related gene expression. <i>International Journal of Molecular Medicine</i> , 2004 , 14, 277-81	4.4	34
13	Role of endogenous regucalcin in transgenic rats: Suppression of protein tyrosine phosphatase and ribonucleic acid synthesis activities in liver nucleus. <i>International Journal of Molecular Medicine</i> , 2003 , 12, 207	4.4	
12	Suppressive role of endogenous regucalcin in the enhancement of nitric oxide synthase activity in liver cytosol of normal and regucalcin transgenic rats. <i>Journal of Cellular Biochemistry</i> , 2003 , 88, 1226-34	4.7	18
11	Enhancement of albumin expression in bone tissues with healing rat fractures. <i>Journal of Cellular Biochemistry</i> , 2003 , 89, 356-63	4.7	24
10	Suppressive effect of endogenous regucalcin on nitric oxide synthase activity in cloned rat hepatoma H4-II-E cells overexpressing regucalcin. <i>Journal of Cellular Biochemistry</i> , 2003 , 89, 800-7	4.7	24
9	Overexpression of regucalcin modulates tumor-related gene expression in cloned rat hepatoma H4-II-E cells. <i>Journal of Cellular Biochemistry</i> , 2003 , 90, 619-26	4.7	49
8	Role of endogenous regucalcin in transgenic rats: suppression of protein tyrosine phosphatase and ribonucleic acid synthesis activities in liver nucleus. <i>International Journal of Molecular Medicine</i> , 2003 , 12, 207-11	4.4	5
7	Suppressive role of endogenous regucalcin in the enhancement of deoxyribonucleic acid synthesis activity in the nucleus of regenerating rat liver. <i>Journal of Cellular Biochemistry</i> , 2002 , 85, 516-22	4.7	38
6	Role of endogenous regucalcin in transgenic rats: suppression of kidney cortex cytosolic protein phosphatase activity and enhancement of heart muscle microsomal Ca ²⁺ -ATPase activity. <i>Journal of Cellular Biochemistry</i> , 2002 , 86, 520-9	4.7	43
5	Role of endogenous regucalcin in nuclear regulation of regenerating rat liver: suppression of the enhanced ribonucleic acid synthesis activity. <i>Journal of Cellular Biochemistry</i> , 2002 , 87, 450-7	4.7	36
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