Yoshinori Tsurusaki

List of Publications by Citations

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	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. *Nature Genetics*, **2012**, 44, 376-8	36.3	350
163	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. *Nature Genetics*, **2013**, 45, 445-9, 449e1	36.3	330
162	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
161	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. *Neurology*, **2013**, 81, 992-8	6.5	158
160	Clinical spectrum of early onset epileptic encephalopathies caused by KCNQ2 mutation. *Epilepsia*, **2013**, 54, 1282-7	6.4	143
159	Mutations in KLHL40 are a frequent cause of severe autosomal-recessive nemaline myopathy. *American Journal of Human Genetics*, **2013**, 93, 6-18	11	142
158	De Novo mutations in GNAO1, encoding a Gα subunit of heterotrimeric G proteins, cause epileptic encephalopathy. *American Journal of Human Genetics*, **2013**, 93, 496-505	11	137
157	KDM6A point mutations cause Kabuki syndrome. *Human Mutation*, **2013**, 34, 108-10	4.7	129
156	Somatic Mutations in the MTOR gene cause focal cortical dysplasia type IIb. *Annals of Neurology*, **2015**, 78, 375-86	9.4	129
155	Early onset epileptic encephalopathy caused by de novo SCN8A mutations. *Epilepsia*, **2014**, 55, 994-1000	6.4	122
154	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. *Human Mutation*, **2010**, 31, 966-74	4.7	119
153	Phenotypic spectrum of COL4A1 mutations: porencephaly to schizencephaly. *Annals of Neurology*, **2013**, 73, 48-57	9.4	117
152	Mutations in POLR3A and POLR3B encoding RNA Polymerase III subunits cause an autosomal-recessive hypomyelinating leukoencephalopathy. *American Journal of Human Genetics*, **2011**, 89, 644-51	11	112
151	MLL2 and KDM6A mutations in patients with Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2234-43	2.5	111
150	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. *Neurogenetics*, **2013**, 14, 225-32	3	91
149	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, 927-34	11	91
148	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. *Cell Reports*, **2018**, 22, 734-747	10.6	86

147	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
146	De novo SOX11 mutations cause Coffin-Siris syndrome. *Nature Communications*, **2014**, 5, 4011	17.4	84
145	SMOC1 is essential for ocular and limb development in humans and mice. *American Journal of Human Genetics*, **2011**, 88, 30-41	11	82
144	PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. *Neurology*, **2014**, 82, 1587-96	6.5	80
143	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. *Annals of Neurology*, **2012**, 72, 298-300	9.4	74
142	Clinical correlations of mutations affecting six components of the SWI/SNF complex: detailed description of 21 patients and a review of the literature. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1221-37	2.5	74
141	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
140	De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy. *Human Mutation*, **2013**, 34, 1708-14	4.7	72
139	The somatic GNAQ mutation c.548G>A (p.R183Q) is consistently found in Sturge-Weber syndrome. *Journal of Human Genetics*, **2014**, 59, 691-3	4.3	70
138	Biallelic Mutations in Nuclear Pore Complex Subunit NUP107 Cause Early-Childhood-Onset Steroid-Resistant Nephrotic Syndrome. *American Journal of Human Genetics*, **2015**, 97, 555-66	11	68
137	Mitochondrial complex III deficiency caused by a homozygous UQCRC2 mutation presenting with neonatal-onset recurrent metabolic decompensation. *Human Mutation*, **2013**, 34, 446-52	4.7	68
136	De novo KCNT1 mutations in early-onset epileptic encephalopathy. *Epilepsia*, **2015**, 56, e121-8	6.4	66
135	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia*, **2013**, 54, 1262-9	6.4	64
134	De novo and inherited mutations in COL4A2, encoding the type IV collagen α2 chain cause porencephaly. *American Journal of Human Genetics*, **2012**, 90, 86-90	11	63
133	Exome sequencing reveals a homozygous SYT14 mutation in adult-onset, autosomal-recessive spinocerebellar ataxia with psychomotor retardation. *American Journal of Human Genetics*, **2011**, 89, 320-7	11	60
132	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
131	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
130	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance. *Neurogenetics*, **2012**, 13, 327-32	3	55

129	Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. *Neurogenetics*, **2014**, 15, 193-200	3	54
128	De novo KCNB1 mutations in infantile epilepsy inhibit repetitive neuronal firing. *Scientific Reports*, **2015**, 5, 15199	4.9	54
127	Whole-exome sequencing and neurite outgrowth analysis in autism spectrum disorder. *Journal of Human Genetics*, **2016**, 61, 199-206	4.3	52
126	CASK aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. *Epilepsia*, **2012**, 53, 1441-9	6.4	51
125	De novo GABRA1 mutations in Ohtahara and West syndromes. *Epilepsia*, **2016**, 57, 566-73	6.4	50
124	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. *Neurogenetics*, **2014**, 15, 85-92	3	50
123	Overexpression of regucalcin modulates tumor-related gene expression in cloned rat hepatoma H4-II-E cells. *Journal of Cellular Biochemistry*, **2003**, 90, 619-26	4.7	49
122	Missense mutations in the DNA-binding/dimerization domain of NFIX cause Sotos-like features. *Journal of Human Genetics*, **2012**, 57, 207-11	4.3	46
121	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. *Journal of Human Genetics*, **2014**, 59, 292-5	4.3	44
120	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, 520-9	4.7	43
119	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an @xome-first@pproach. *Journal of Human Genetics*, **2015**, 60, 175-82	4.3	41
118	Deletions and de novo mutations of SOX11 are associated with a neurodevelopmental disorder with features of Coffin-Siris syndrome. *Journal of Medical Genetics*, **2016**, 53, 152-62	5.8	41
117	Precise detection of chromosomal translocation or inversion breakpoints by whole-genome sequencing. *Journal of Human Genetics*, **2014**, 59, 649-54	4.3	40
116	Expanding the phenotypic spectrum of TUBB4A-associated hypomyelinating leukoencephalopathies. *Neurology*, **2014**, 82, 2230-7	6.5	38
115	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, 516-22	4.7	38
114	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
113	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37
112	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, 450-7	4.7	36

111	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. *Journal of Human Genetics*, **2014**, 59, 581-3	4.3	34
110	Rapid detection of a mutation causing X-linked leucoencephalopathy by exome sequencing. *Journal of Medical Genetics*, **2011**, 48, 606-9	5.8	34
109	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	4.4	34
108	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
107	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
106	PIGO mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels. *Epilepsia*, **2014**, 55, e13-7	6.4	31
105	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
104	Compound heterozygous BRAT1 mutations cause familial Ohtahara syndrome with hypertonia and microcephaly. *Journal of Human Genetics*, **2014**, 59, 687-90	4.3	30
103	The first Japanese case of leukodystrophy with ovarian failure arising from novel compound heterozygous AARS2 mutations. *Journal of Human Genetics*, **2016**, 61, 899-902	4.3	28
102	Role of endogenous regucalcin in bone metabolism: bone loss is induced in regucalcin transgenic rats. *International Journal of Molecular Medicine*, **2002**, 10, 377-83	4.4	28
101	Aortic aneurysm and craniosynostosis in a family with Cantu syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 231-6	2.5	27
100	The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. *Journal of Human Genetics*, **2013**, 58, 113-5	4.3	27
99	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slit-Robo Rho GTPase activating protein 2 (SRGAP2). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 199-205	2.5	25
98	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	3.7	24
97	Enhancement of albumin expression in bone tissues with healing rat fractures. *Journal of Cellular Biochemistry*, **2003**, 89, 356-63	4.7	24
96	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, 800-7	4.7	24
95	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. *Journal of Human Genetics*, **2016**, 61, 835-8	4.3	24
94	Somatic mutations in GLI3 and OFD1 involved in sonic hedgehog signaling cause hypothalamic hamartoma. *Annals of Clinical and Translational Neurology*, **2016**, 3, 356-65	5.3	24

93	A novel homozygous YARS2 mutation causes severe myopathy, lactic acidosis, and sideroblastic anemia 2. *Journal of Human Genetics*, **2014**, 59, 229-32	4.3	23
92	Numerous BAF complex genes are mutated in Coffin-Siris syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 257-61	3.1	21
91	De novo 5q14.3 translocation 121.5-kb upstream of MEF2C in a patient with severe intellectual disability and early-onset epileptic encephalopathy. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2879-84	2.5	21
90	A novel mutation causes X-linked intellectual disability type Nascimento. *Human Genome Variation*, **2017**, 4, 17019	1.8	20
89	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics* , **2019**, 64, 1173-1186	4.3	20
88	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, 61-7	4.2	20
87	Mutations in the glutaminyl-tRNA synthetase gene cause early-onset epileptic encephalopathy. *Journal of Human Genetics*, **2015**, 60, 97-101	4.3	19
86	A novel WTX mutation in a female patient with osteopathia striata with cranial sclerosis and hepatoblastoma. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 998-1002	2.5	19
85	Homozygous splicing mutation in NUP133 causes Galloway-Mowat syndrome. *Annals of Neurology*, **2018**, 84, 814-828	9.4	19
84	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, 591-9	6.3	18
83	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, 1226-34	4.7	18
82	Whole-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**, 161A, 1543-6	2.5	17
81	De novo 19q13.42 duplications involving NLRP gene cluster in a patient with systemic-onset juvenile idiopathic arthritis. *Journal of Human Genetics*, **2011**, 56, 343-7	4.3	17
80	Breakpoint determination of X;autosome balanced translocations in four patients with premature ovarian failure. *Journal of Human Genetics*, **2011**, 56, 156-60	4.3	17
79	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, 387-93	2.3	17
78	De novo DNM1 mutations in two cases of epileptic encephalopathy. *Epilepsia*, **2016**, 57, e18-23	6.4	17
77	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
76	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, 471-4	4.3	16

75	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. *Scientific Reports*, **2014**, 4, 7132	4.9	16
74	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. *Human Genetics*, **2014**, 133, 225-34	6.3	15
73	Novel FIG4 mutations in Yunis-Varon syndrome. *Journal of Human Genetics*, **2013**, 58, 822-4	4.3	15
72	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. *Journal of Human Genetics*, **2013**, 58, 391-4	4.3	15
71	De novo deletion of 1q24.3-q31.2 in a patient with severe growth retardation. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1322-5	2.5	15
70	SMARCE1, a rare cause of Coffin-Siris Syndrome: Clinical description of three additional cases. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1967-73	2.5	15
69	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. *Journal of Human Genetics*, **2015**, 60, 631-5	4.3	14
68	A girl with early-onset epileptic encephalopathy associated with microdeletion involving CDKL5. *Brain and Development*, **2012**, 34, 364-7	2.2	14
67	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioleiomyomatosis tissues by deep sequencing. *Human Genetics*, **2016**, 135, 61-8	6.3	13
66	A de novo 1.4-Mb deletion at 21q22.11 in a boy with developmental delay. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1021-8	2.5	13
65	Mandibulofacial dysostosis with microcephaly: A case presenting with seizures. *Brain and Development*, **2017**, 39, 177-181	2.2	13
64	A family of oculofaciocardiodental syndrome (OFCD) with a novel BCOR mutation and genomic rearrangements involving NHS. *Journal of Human Genetics*, **2012**, 57, 197-201	4.3	13
63	Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2662-70	2.5	13
62	Novel mutation in a fetus with early prenatal onset of schizencephaly. *Human Genome Variation*, **2018**, 5, 4	1.8	12
61	Refining the clinical phenotype of Okur-Chung neurodevelopmental syndrome. *Human Genome Variation*, **2018**, 5, 18011	1.8	12
60	Homozygous p.V116* mutation in C12orf65 results in Leigh syndrome. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 212-6	5.5	12
59	Severe manifestations of hand-foot-genital syndrome associated with a novel HOXA13 mutation. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2398-402	2.5	12
58	A female patient with X-linked Ohdo syndrome of the Maat-Kievit-Brunner phenotype caused by a novel variant of MED12. *Congenital Anomalies (discontinued)*, **2020**, 60, 91-93	1.1	12

57	A novel gene (FAM20B encoding glycosaminoglycan xylosylkinase) for neonatal short limb dysplasia resembling Desbuquois dysplasia. *Clinical Genetics*, **2019**, 95, 713-717	4	11
56	Novel AMER1 frameshift mutation in a girl with osteopathia striata with cranial sclerosis. *Congenital Anomalies (discontinued)*, **2018**, 58, 145-146	1.1	11
55	A de novo CASK mutation in pontocerebellar hypoplasia type 3 with early myoclonic epilepsy and tetralogy of Fallot. *Brain and Development*, **2014**, 36, 272-3	2.2	11
54	Deep sequencing detects very-low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy. *Neuromuscular Disorders*, **2014**, 24, 642-7	2.9	11
53	A unique case of de novo 5q33.3-q34 triplication with uniparental isodisomy of 5q34-qter. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1904-9	2.5	11
52	Prenatal clinical manifestations in individuals with variants. *Journal of Medical Genetics*, **2021**, 58, 505-513	3.8	11
51	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. *Journal of Human Genetics*, **2018**, 63, 417-423	4.3	10
50	A Japanese case of cerebellar ataxia, spastic paraparesis and deep sensory impairment associated with a novel homozygous TTC19 mutation. *Journal of Human Genetics*, **2015**, 60, 187-91	4.3	10
49	Coffin-Siris syndrome and cardiac anomaly with a novel SOX11 mutation. *Congenital Anomalies (discontinued)*, **2018**, 58, 105-107	1.1	10
48	Familial schwannomatosis with a germline mutation of SMARCB1 in Japan. *Brain Tumor Pathology*, **2015**, 32, 216-20	3.2	9
47	Novel rare variations of the oxytocin receptor (OXTR) gene in autism spectrum disorder individuals. *Human Genome Variation*, **2015**, 2, 15024	1.8	9
46	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. *Human Genome Variation*, **2015**, 2, 15034	1.8	9
45	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, 1629-33	1.1	9
44	A de novo deletion of 20q11.2-q12 in a boy presenting with abnormal hands and feet, retinal dysplasia, and intractable feeding difficulty. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 409-14	2.5	9
43	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. *European Journal of Medical Genetics*, **2020**, 63, 103610	2.6	9
42	Novel SYNGAP1 variant in a patient with intellectual disability and distinctive dysmorphisms. *Congenital Anomalies (discontinued)*, **2018**, 58, 188-190	1.1	8
41	Pathogenic variants of , , and associated with hypothalamic hamartoma. *Neurology*, **2019**, 93, e237-e251	6.5	8
40	Role of endogenous regucalcin in the regulation of Ca(2+)-ATPase activity in rat liver nuclei. *Journal of Cellular Biochemistry*, **2000**, 78, 541-9	4.7	8

39	Update of the genotype and phenotype of KMT2D and KDM6A by genetic screening of 100 patients with clinically suspected Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2333-2344	2.5	8
38	Two unrelated girls with intellectual disability associated with a truncating mutation in the PPM1D penultimate exon. *Brain and Development*, **2019**, 41, 538-541	2.2	7
37	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	3.4	7
36	Cortical cerebellar atrophy dwindles away in the era of next-generation sequencing. *Journal of Human Genetics*, **2014**, 59, 589-90	4.3	7
35	A novel SCARB2 mutation causing late-onset progressive myoclonus epilepsy. *Movement Disorders*, **2013**, 28, 552-3	7	7
34	A Japanese girl with an early-infantile onset vanishing white matter disease resembling Cree leukoencephalopathy. *Brain and Development*, **2015**, 37, 638-42	2.2	6
33	Mutations in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease. *Brain and Development*, **2015**, 37, 960-6	2.2	6
32	Dual genetic diagnoses: Atypical hand-foot-genital syndrome and developmental delay due to de novo mutations in HOXA13 and NRXN1. *American Journal of Medical Genetics, Part A*, **2016**, 170, 717-24	2.5	6
31	Nonsyndromic intellectual disability with novel heterozygous mutation and epilepsy. *Human Genome Variation*, **2018**, 5, 20	1.8	6
30	A novel PITX2 mutation causing iris hypoplasia. *Human Genome Variation*, **2014**, 1, 14005	1.8	6
29	A recurrent PJA1 variant in trigonocephaly and neurodevelopmental disorders. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1117-1131	5.3	5
28	Suppressive effect of endogenous regucalcin on guanosine triphosphatase activity in rat liver nucleus. *Biological and Pharmaceutical Bulletin*, **2001**, 24, 958-61	2.3	5
27	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	4.4	5
26	A severe form of Ellis-van Creveld syndrome caused by novel mutations in. *Human Genome Variation*, **2019**, 6, 40	1.8	4
25	Discordant phenotype caused by mutation in siblings with. *Human Genome Variation*, **2019**, 6, 20	1.8	4
24	Novel biallelic mutations alter the skeletal phenotype of 3M syndrome. *Human Genome Variation*, **2020**, 7, 1	1.8	4
23	White matter abnormalities in an adult patient with l-2-hydroxyglutaric aciduria. *Brain and Development*, **2016**, 38, 142-4	2.2	4
22	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	4.4	4

21	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
20	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. *Brain and Development*, **2020**, 42, 289-292	2.2	3
19	Co-occurrence of 22q11 deletion syndrome and HDR syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2576-81	2.5	3
18	Biallelic mutations of in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction. *Human Genome Variation*, **2018**, 5, 11	1.8	3
17	An efficient genetic test flow for multiple congenital anomalies and intellectual disability. *Pediatrics International*, **2020**, 62, 556-561	1.2	2
16	TMEM67 mutations found in a case of Joubert syndrome with renal hypodysplasia. *CEN Case Reports*, **2016**, 5, 137-140	1	2
15	Whole-exome sequencing reveals the subclonal expression of NUP214-ABL1 fusion gene in T-cell acute lymphoblastic leukemia. *Pediatric Blood and Cancer*, **2020**, 67, e28019	3	2
14	Novel variants in two patients with X-linked intellectual disability. *Human Genome Variation*, **2019**, 6, 49	1.8	2
13	Japanese familial case of myoclonus-dystonia syndrome with a splicing mutation in SGCE. *Pediatrics International*, **2015**, 57, 324-6	1.2	1
12	Multiple alterations in glutamatergic transmission and dopamine D2 receptor splicing in induced pluripotent stem cell-derived neurons from patients with familial schizophrenia. *Translational Psychiatry*, **2021**, 11, 548	8.6	1
11	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
10	Hemoglobin beta Kanagawa [c.443A>C; p.(Ter148Serext*21)]: A novel □globin gene mutation causing dominantly inherited □thalassemia. *Pediatric Blood and Cancer*, **2019**, 66, e27871	3	0
9	Expanding the phenotype of COL4A1-related disorders-Four novel variants. *Brain and Development*, **2020**, 42, 639-645	2.2	0
8	variants dysregulate splicing and cause hypomyelinating leukodystrophy. *Neurology: Genetics*, **2020**, 6, e524	3.8	0
7	A Recurrent Variant in , 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	1.5	0
6	Siblings with vascular Ehlers-Danlos syndrome inherited via maternal mosaicism. *Congenital Anomalies (discontinued)*, **2021**, 61, 101-102	1.1	0
5	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
4	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	4.4	

3	Suppressive effect of regucalcin on protein phosphatase activity in the heart cytosol of normal and regucalcin transgenic rats. *International Journal of Molecular Medicine*, **2004**, 13, 289	4.4
2	Further delineation of SET-related intellectual disability syndrome.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5
1	Novel COL2A1 variants in Japanese patients with spondyloepiphyseal dysplasia congenita.. *Human Genome Variation*, **2022**, 9, 16	1.8