Toshiyuki Yamamoto

List of Publications by Year
in descending order

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174 papers	2,623 citations	257101 24 h-index	329751 37 g-index
176 all docs	176 docs citations	176 times ranked	7040 citing authors

#	Article	IF	Citations
1	STXBP1 mutations cause not only Ohtahara syndrome but also West syndromeâ€"Result of Japanese cohort study. Epilepsia, 2010, 51, 2449-2452.	2.6	105
2	Reduced expression by SETBP1 haploinsufficiency causes developmental and expressive language delay indicating a phenotype distinct from Schinzel-Giedion syndrome. Journal of Medical Genetics, 2011, 48, 117-122.	1.5	80
3	A recurrent KCNT1 mutation in two sporadic cases with malignant migrating partial seizures in infancy. Gene, 2013, 531, 467-471.	1.0	80
4	NPC1 gene mutations in Japanese patients with Niemann-Pick disease typeÂC. Human Genetics, 1999, 105, 10-16.	1.8	78
5	Loss-of-function mutation of collybistin is responsible for X-linked mental retardation associated with epilepsy. Journal of Human Genetics, 2011, 56, 561-565.	1.1	72
6	CDKL5 alterations lead to early epileptic encephalopathy in both genders. Epilepsia, 2011, 52, 1835-1842.	2.6	62
7	Clinical manifestations of the deletion of Down syndrome critical region including DYRK1A and KCNJ6. American Journal of Medical Genetics, Part A, 2011, 155, 113-119.	0.7	52
8	MECP2 duplication syndrome in both genders. Brain and Development, 2013, 35, 411-419.	0.6	48
9	Epilepsy and neurological findings in 11 individuals with 1p36 deletion syndrome. Brain and Development, 2005, 27, 378-382.	0.6	45
10	A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination. , 2011, 155, 732-736.		43
11	Microarray analysis of 50 patients reveals the critical chromosomal regions responsible for 1p36 deletion syndrome-related complications. Brain and Development, 2015, 37, 515-526.	0.6	43
12	TULIP1 (RALGAPA1) haploinsufficiency with brain development delay. Genomics, 2009, 94, 414-422.	1.3	42
13	A familial 593-kb microdeletion of 16p11.2 associated with mental retardation and hemivertebrae. European Journal of Medical Genetics, 2009, 52, 433-435.	0.7	41
14	Genomic copy number variations at 17p13.3 and epileptogenesis. Epilepsy Research, 2010, 89, 303-309.	0.8	39
15	Ataxia-telangiectasia without immunodeficiency: Novel point mutations within and adjacent to the phosphatidylinositol 3-kinase-like domain. , 1998, 75, 141-144.		38
16	Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. Human Genome Variation, 2016, 3, 16025.	0.4	38
17	Genomic backgrounds of Japanese patients with undiagnosed neurodevelopmental disorders. Brain and Development, 2019, 41, 776-782.	0.6	36
18	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	0.9	35

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19	Lissencephaly with marked ventricular dilation, agenesis of corpus callosum, and cerebellar hypoplasia caused by TUBA1A mutation. Brain and Development, 2013, 35, 274-279.	0.6	34
20	PRRT2 mutation in Japanese children with benign infantile epilepsy. Brain and Development, 2013, 35, 641-646.	0.6	31
21	Narrowing of the responsible region for severe developmental delay and autistic behaviors in WAGR syndrome down to 1.6â€‰Mb including PAX6, WT1, and PRRG4. American Journal of Medical Genetics, Part A, 2014, 164, 634-638.	0.7	31
22	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	1.1	31
23	Clinical impacts of genomic copy number gains at Xq28. Human Genome Variation, 2014, 1, 14001.	0.4	30
24	GPC3 mutations in seven patients with Simpsonâ€“Golabiâ€“Behmel syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1703-1707.	0.7	29
25	An emerging phenotype of Xq22 microdeletions in females with severe intellectual disability, hypotonia and behavioral abnormalities. Journal of Human Genetics, 2014, 59, 300-306.	1.1	29
26	Comprehensive genetic analyses of PLP1 in patients with Pelizaeusâ€“Merzbacher disease applied by array-CGH and fiber-FISH analyses identified new mutations and variable sizes of duplications. Brain and Development, 2010, 32, 171-179.	0.6	28
27	Pelizaeus-Merzbacher disease caused by a duplication-inverted triplication-duplication in chromosomal segments including the PLP1 region. European Journal of Medical Genetics, 2012, 55, 400-403.	0.7	28
28	Jimpymsd mouse mutation and connatal Pelizaeus-Merzbacher disease. , 1998, 75, 439-440.		27
29	A functional analysis of GABARAP on 17p13.1 by knockdown zebrafish. Journal of Human Genetics, 2010, 55, 155-162.	1.1	27
30	Zebrafish gene knockdowns imply roles for human YWHAG in infantile spasms and cardiomegaly. Genesis, 2010, 48, 233-243.	0.8	26
31	Interstitial Duplication of 2q32.1â€“q33.3 in a Patient With Epilepsy, Developmental Delay, and Autistic Behavior. American Journal of Medical Genetics, Part A, 2013, 161, 1078-1084.	0.7	26
32	Severe pulmonary emphysema in a girl with interstitial deletion of 2q24.2q24.3 including ITGB6. American Journal of Medical Genetics, Part A, 2010, 152A, 1020-1025.	0.7	25
33	Submicroscopic deletion in 7q31 encompassing CADPS2 and TSPAN12 in a child with autism spectrum disorder and PHPV. American Journal of Medical Genetics, Part A, 2011, 155, 1568-1573.	0.7	24
34	Congenital anomaly of cervical vertebrae is a major complication of Rubinstein-Taybi syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 130-133.	0.7	23
35	Tattonâ€“Brownâ€“Rahman syndrome due to 2p23 microdeletion. American Journal of Medical Genetics, Part A, 2016, 170, 1339-1342.	0.7	23
36	MED13L haploinsufficiency syndrome: A de novo frameshift and recurrent intragenic deletions due to parental mosaicism. , 2017, 173, 1264-1269.		23

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37	De novo microdeletion of 5q14.3 excluding MEF2C in a patient with infantile spasms, microcephaly, and agenesis of the corpus callosum. American Journal of Medical Genetics, Part A, 2012, 158A, 2272-2276.	0.7	22
38	<scp>P</scp>elizaeusâ€“<scp>M</scp>erzbacher disease as a chromosomal disorder. Congenital Anomalies (discontinued), 2013, 53, 3-8.	0.3	22
39	Novel TSC1 and TSC2 mutations in Japanese patients with tuberous sclerosis complex. Brain and Development, 2002, 24, 227-230.	0.6	21
40	Clinical manifestations of Xq28 functional disomy involving MECP2 in one female and two male patients. American Journal of Medical Genetics, Part A, 2013, 161, 1779-1785.	0.7	21
41	Two concurrent chromosomal aberrations involving interstitial deletion in 1q24.2q25.2 and inverted duplication and deletion in 10q26 in a patient with stroke associated with antithrombin deficiency and a patent foramen ovale. American Journal of Medical Genetics, Part A, 2011, 155, 215-220.	0.7	20
42	Subtelomeric deletions of 1q43q44 and severe brain impairment associated with delayed myelination. Journal of Human Genetics, 2012, 57, 593-600.	1.1	20
43	Novel compound heterozygous mutations of POLR3A revealed by whole-exome sequencing in a patient with hypomyelination. Brain and Development, 2014, 36, 315-321.	0.6	20
44	Reduced PLP1 expression in induced pluripotent stem cells derived from a Pelizaeusâ€“Merzbacher disease patient with a partial PLP1 duplication. Journal of Human Genetics, 2012, 57, 580-586.	1.1	19
45	109â€‰kb deletion of chromosome 4p16.3 in a patient with mild phenotype of Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1465-1469.	0.7	19
46	Whole-exome sequencing identifies a de novo TUBA1A mutation in a patient with sporadic malformations of cortical development: a case report. BMC Research Notes, 2014, 7, 465.	0.6	19
47	A novel TUBB3 mutation in a sporadic patient with asymmetric cortical dysplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1076-1079.	0.7	19
48	A 7q31.33q32.1 microdeletion including LRRC4 and GRM8 is associated with severe intellectual disability and characteristics of autism. Human Genome Variation, 2017, 4, 17001.	0.4	19
49	Refractory neonatal epilepsy with a de novo duplication of chromosome 2q24.2q24.3. Epilepsia, 2011, 52, e66-e69.	2.6	18
50	A novel MED12 mutation associated with non-specific X-linked intellectual disability. Human Genome Variation, 2015, 2, 15018.	0.4	18
51	Leukoencephalopathy associated with 11q24 deletion involving the gene encoding hepatic and glial cell adhesion molecule in two patients. European Journal of Medical Genetics, 2015, 58, 492-496.	0.7	18
52	A novel <i>CASK</i> mutation identified in siblings exhibiting developmental disorders with/without microcephaly. Intractable and Rare Diseases Research, 2017, 6, 177-182.	0.3	18
53	A de novo 22q11.22q11.23 interchromosomal tandem duplication in a boy with developmental delay, hyperactivity, and epilepsy. American Journal of Medical Genetics, Part A, 2010, 152A, 2820-2826.	0.7	17
54	Somatic mosaic deletions involving SCN1A cause Dravet syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 657-662.	0.7	17

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55	Proximal interstitial 1p36 deletion syndrome: The most proximal 3.5-Mb microdeletion identified on a dysmorphic and mentally retarded patient with inv(3)(p14.1q26.2). Brain and Development, 2009, 31, 629-633.	0.6	16
56	An association of 19p13.2 microdeletions with Malan syndrome and Chiari malformation. American Journal of Medical Genetics, Part A, 2015, 167, 724-730.	0.7	16
57	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 1-5.	0.9	16
58	A novel KCNT1 mutation in a Japanese patient with epilepsy of infancy with migrating focal seizures. Human Genome Variation, 2014, 1, 14027.	0.4	15
59	Loss-of-function mutations of STXBP1 in patients with epileptic encephalopathy. Brain and Development, 2016, 38, 280-284.	0.6	15
60	A novel TUBB4A mutation G96R identified in a patient with hypomyelinating leukodystrophy onset beyond adolescence. Human Genome Variation, 2017, 4, 17035.	0.4	15
61	A cryptic microdeletion including MBD5 occurring within the breakpoint of a reciprocal translocation between chromosomes 2 and 5 in a patient with developmental delay and obesity. American Journal of Medical Genetics, Part A, 2013, 161, 850-855.	0.7	14
62	Novel SLC16A2 mutations in patients with Allan-Herndon-Dudley syndrome. Intractable and Rare Diseases Research, 2016, 5, 214-217.	0.3	14
63	A novel <i>PGK1</i> mutation associated with neurological dysfunction and the absence of episodes of hemolytic anemia or myoglobinuria. Intractable and Rare Diseases Research, 2017, 6, 132-136.	0.3	14
64	Congenital anemia reveals distinct targeting mechanisms for master transcription factor GATA1. Blood, 2022, 139, 2534-2546.	0.6	14
65	Clinical Course and Images of Four Familial Cases of Allan-Herndon-Dudley Syndrome With a Novel Monocarboxylate Transporter 8 Gene Mutation. Pediatric Neurology, 2014, 51, 414-416.	1.0	13
66	Characteristics of 2p15â€p16.1 microdeletion syndrome: Review and description of two additional patients. Congenital Anomalies (discontinued), 2015, 55, 125-132.	0.3	13
67	Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. PLoS ONE, 2015, 10, e0118946.	1.1	13
68	A de novo TUBB4A mutation in a patient with hypomyelination mimicking Pelizaeusâ€“Merzbacher disease. Brain and Development, 2015, 37, 281-285.	0.6	13
69	An episode of acute encephalopathy with biphasic seizures and late reduced diffusion followed by hemiplegia and intractable epilepsy observed in a patient with a novel frameshift mutation in HNRNPU. Brain and Development, 2018, 40, 813-818.	0.6	13
70	Application of induced pluripotent stem cells in epilepsy. Molecular and Cellular Neurosciences, 2020, 108, 103535.	1.0	13
71	A de novo intraâ€chromosomal tandem duplication at 22q13.1q13.31 including the Rubinsteinâ€“Taybi region but with no bipolar disorder. American Journal of Medical Genetics, Part A, 2009, 149A, 1359-1363.	0.7	12
72	A de novo 1.9â€Mb interstitial deletion of 3q13.2q13.31 in a girl with dysmorphic features, muscle hypotonia, and developmental delay. American Journal of Medical Genetics, Part A, 2009, 149A, 1818-1822.	0.7	12

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73	Microdeletions of 5.5â€‰Mb (4q13.2â€“q13.3) and 4.1â€‰Mb (7p15.3â€–p21.1) associated with a saethreâ€“chotzenâ€like phenotype, severe intellectual disability, and autism. American Journal of Medical Genetics, Part A, 2013, 161, 2078-2083.	0.7	12
74	A novel homozygous mutation of GJC2 derived from maternal uniparental disomy in a female patient with Pelizaeusâ€“Merzbacher-like disease. Journal of the Neurological Sciences, 2013, 330, 123-126.	0.3	12
75	Microdeletions of 3p21.31 characterized by developmental delay, distinctive features, elevated serum creatine kinase levels, and white matter involvement. American Journal of Medical Genetics, Part A, 2013, 161, 3049-3056.	0.7	12
76	CHCHD2 is down-regulated in neuronal cells differentiated from iPS cells derived from patients with lissencephaly. Genomics, 2015, 106, 196-203.	1.3	12
77	A 12p13 GRIN2B deletion is associated with developmental delay and macrocephaly. Human Genome Variation, 2016, 3, 16029.	0.4	12
78	A novel MLH1 mutation in a Japanese family with Lynch syndrome associated with small bowel cancer. Human Genome Variation, 2018, 5, 13.	0.4	12
79	The involvement of U-type dicentric chromosomes in the formation of terminal deletions with or without adjacent inverted duplications. Human Genetics, 2020, 139, 1417-1427.	1.8	12
80	SSCP analysis by RT-PCR for the prenatal diagnosis of Niemann-Pick disease type C. Prenatal Diagnosis, 2001, 21, 55-57.	1.1	11
81	An unmasked mutation of EIF2B2 due to submicroscopic deletion of 14q24.3 in a patient with vanishing white matter disease. American Journal of Medical Genetics, Part A, 2012, 158A, 1771-1777.	0.7	11
82	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. European Journal of Medical Genetics, 2014, 57, 163-168.	0.7	11
83	Identification of a rare homozygous <i>SZT2</i> variant due to uniparental disomy in a patient with a neurodevelopmental disorder. Intractable and Rare Diseases Research, 2018, 7, 245-250.	0.3	11
84	A 15q14 microdeletion involving MEIS2 identified in a patient with autism spectrum disorder. Human Genome Variation, 2017, 4, 17029.	0.4	11
85	NovelTSC2 mutation in a patient with pulmonary tuberous sclerosis: lack of loss of heterozygosity in a lung cyst. , 1999, 82, 368-370.		10
86	Neuropsychological profiles of patients with 2q37.3 deletion associated with developmental dyspraxia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 684-690.	1.1	10
87	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
88	7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. European Journal of Medical Genetics, 2016, 59, 502-506.	0.7	10
89	Clinical and genetic diagnosis of thirteen Japanese patients with hereditary spherocytosis. Human Genome Variation, 2022, 9, 1.	0.4	10
90	Tandem configurations of variably duplicated segments of 22q11.2 confirmed by fiber-FISH analysis. Journal of Human Genetics, 2011, 56, 810-812.	1.1	9

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91	Use of targeted nextâ€generation sequencing for molecular diagnosis of craniosynostosis: Identification of a novel de novo mutation of <scp>EFNB</scp>1. Congenital Anomalies (discontinued), 2016, 56, 91-93.	0.3	9
92	Elucidation of the pathogenic mechanism and potential treatment strategy for a female patient with spastic paraplegia derived from a single-nucleotide deletion in PLP1. Journal of Human Genetics, 2019, 64, 665-671.	1.1	9
93	MLC1 mutations in Japanese patients with megalencephalic leukoencephalopathy with subcortical cysts. Human Genome Variation, 2014, 1, 14019.	0.4	8
94	Recurrent occurrences of CDKL5 mutations in patients with epileptic encephalopathy. Human Genome Variation, 2015, 2, 15042.	0.4	8
95	Novel PLA2G6 mutations associated with an exonic deletion due to non-allelic homologous recombination in a patient with infantile neuroaxonal dystrophy. Human Genome Variation, 2015, 2, 15048.	0.4	8
96	Periventricular heterotopia and white matter abnormalities in a girl with mosaic ring chromosome 6. Molecular Cytogenetics, 2015, 8, 54.	0.4	8
97	Characteristics of patients with benign partial epilepsy in infancy without PRRT2 mutations. Epilepsy Research, 2015, 118, 10-13.	0.8	8
98	A de novo microdeletion in a patient with inner ear abnormalities suggests that the 10q26.13 region contains the responsible gene. Human Genome Variation, 2016, 3, 16008.	0.4	8
99	Analyses of breakpoint junctions of complex genomic rearrangements comprising multiple consecutive microdeletions by nanopore sequencing. Journal of Human Genetics, 2020, 65, 735-741.	1.1	8
100	Wholeâ€exome sequencing of a unique brain malformation with periventricular heterotopia, cingulate polymicrogyria and midbrain tectal hyperplasia. Neuropathology, 2013, 33, 553-560.	0.7	7
101	Megalencephalic leukoencephalopathy with subcortical cysts caused by compound heterozygous mutations in MLC1 , in patients with and without subcortical cysts in the brain. Journal of the Neurological Sciences, 2015, 351, 211-213.	0.3	7
102	Loss-of-function mutations and global rearrangements in GPC3 in patients with Simpsonâ€“Golabiâ€“Behmel syndrome. Human Genome Variation, 2016, 3, 16033.	0.4	7
103	Focal seizures and epileptic spasms in a child with Down syndrome from a family with a PRRT2 mutation. Brain and Development, 2016, 38, 597-600.	0.6	7
104	Clinical spectrum of individuals with de novo <scp>EBF3</scp> variants or deletions. American Journal of Medical Genetics, Part A, 2021, 185, 2913-2921.	0.7	7
105	Marfanoid hypermobility caused by an 862â€‰kb deletion of Xq22.3 in a patient with Sotos syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2293-2297.	0.7	6
106	Novel nucleotide mutation leading to a recurrent amino acid alteration in <scp>SH3BP2</scp> in a patient with cherubism. Congenital Anomalies (discontinued), 2013, 53, 166-169.	0.3	6
107	Pontine Malformation, Undecussated Pyramidal Tracts, and Regional Polymicrogyria: A New Syndrome. Pediatric Neurology, 2014, 50, 384-388.	1.0	6
108	SLC16A2 mutations in two Japanese patients with Allanâ€“Herndonâ€“Dudley syndrome. Human Genome Variation, 2014, 1, 14010.	0.4	6

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109	Pancreatic developmental defect evaluated by celiac artery angiography in a patient with MODY5. Human Genome Variation, 2016, 3, 16022.	0.4	6
110	White matter abnormalities in an adult patient with l-2-hydroxyglutaric aciduria. Brain and Development, 2016, 38, 142-144.	0.6	6
111	A novel DARS2 mutation in a Japanese patient with leukoencephalopathy with brainstem and spinal cord involvement but no lactate elevation. Human Genome Variation, 2017, 4, 17051.	0.4	6
112	Infantile spasms related to a 5q31.2-q31.3 microdeletion including PURA. Human Genome Variation, 2018, 5, 18007.	0.4	6
113	Interstitial deletion within 7q31.1q31.3 in a woman with mild intellectual disability and schizophrenia. Neuropsychiatric Disease and Treatment, 2018, Volume 14, 1773-1778.	1.0	6
114	Independent occurrence of de novo HSPD1 and HIP1 variants in brothers with different neurological disorders â€“ leukodystrophy and autism. Human Genome Variation, 2018, 5, 18.	0.4	6
115	Advantages of ddPCR in detection of <i>PLP1</i> duplications. Intractable and Rare Diseases Research, 2019, 8, 198-202.	0.3	6
116	Natural histories of patients with Wolfâ€Hirschhorn syndrome derived from variable chromosomal abnormalities. Congenital Anomalies (discontinued), 2019, 59, 169-173.	0.3	6
117	<scp>HECW2</scp>â€related disorder in four Japanese patients. American Journal of Medical Genetics, Part A, 2021, 185, 2895-2902.	0.7	6
118	Genomic Aberrations Associated with the Pathophysiological Mechanisms of Neurodevelopmental Disorders. Cells, 2021, 10, 2317.	1.8	6
119	Altered gene expression in umbilical cord mononuclear cells in preterm infants with periventricular leukomalacia. Early Human Development, 2010, 86, 665-667.	0.8	5
120	Coâ€occurrence of Praderâ€“Willi and Sotos syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 2103-2109.	0.7	5
121	Jacobsen syndrome due to an unbalanced translocation between 11q23 and 22q11.2 identified at age 40 years. American Journal of Medical Genetics, Part A, 2012, 158A, 220-223.	0.7	5
122	De novo triplication of 11q12.3 in a patient with developmental delay and distinctive facial features. Molecular Cytogenetics, 2013, 6, 15.	0.4	5
123	Mild developmental delay and obesity in two patients with mosaic 1p36 deletion syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 415-420.	0.7	5
124	A de novo microdeletion involving PAFAH1B (LIS1) related to lissencephaly phenotype. Data in Brief, 2015, 4, 488-491.	0.5	5
125	A 16q12.2q21 deletion identified in a patient with developmental delay, epilepsy, short stature, and distinctive features. Congenital Anomalies (discontinued), 2016, 56, 253-255.	0.3	5
126	Concurrent occurrence of an inherited 16p13.11 microduplication and a de novo 19p13.3 microdeletion involving MAP2K2 in a patient with developmental delay, distinctive facial features, and lambdoid synostosis. European Journal of Medical Genetics, 2016, 59, 559-563.	0.7	5

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127	Possible genes responsible for developmental delay observed in patients with rare 2q23q24 microdeletion syndrome: Literature review and description of an additional patient. Congenital Anomalies (discontinued), 2017, 57, 109-113.	0.3	5
128	An Xq22.1q22.2 nullisomy in a male patient with severe neurological impairment. American Journal of Medical Genetics, Part A, 2017, 173, 1124-1127.	0.7	5
129	Mutations in NSD1 and NFIX in Three Patients with Clinical Features of Sotos Syndrome and Malan Syndrome. Journal of Pediatric Genetics, 2017, 06, 234-237.	0.3	5
130	A case of Dravet syndrome with cortical myoclonus indicated by jerk-locked back-averaging of electroencephalogram data. Brain and Development, 2017, 39, 75-79.	0.6	5
131	Early-Onset Diabetes Mellitus in a Patient With a Chromosome 13q34qter Microdeletion Including IRS2. Journal of the Endocrine Society, 2018, 2, 1207-1213.	0.1	5
132	Primrose syndrome associated with unclassified immunodeficiency and a novel ZBTB20 mutation. American Journal of Medical Genetics, Part A, 2020, 182, 521-526.	0.7	5
133	Complex chromosomal rearrangements of human chromosome 21 in a patient manifesting clinical features partially overlapped with that of Down syndrome. Human Genetics, 2020, 139, 1555-1563.	1.8	5
134	A novel mutation (A246T) in exon 6 of the proteolipid protein gene associated with connatal Pelizaeus-Merzbacher disease. , 1999, 14, 182-182.		4
135	Growth patterns of patients with 1p36 deletion syndrome. Congenital Anomalies (discontinued), 2014, 54, 82-86.	0.3	4
136	Three Japanese patients with 3p13 microdeletions involving FOXP1. Brain and Development, 2019, 41, 257-262.	0.6	4
137	Establishment of a simple and rapid method to detect MECP2 duplications using digital polymerase chain reaction. Congenital Anomalies (discontinued), 2020, 60, 10-14.	0.3	4
138	Deep intronic deletion in intron 3 of PLP1 is associated with a severe phenotype of Pelizaeus-Merzbacher disease. Human Genome Variation, 2021, 8, 14.	0.4	4
139	Recurrent de novo pathogenic variant of WASF1 in a Japanese patient with neurodevelopmental disorder with absent language and variable seizures. Human Genome Variation, 2021, 8, 43.	0.4	4
140	Challenges in genetic counseling because of intraâ€familial phenotypic variation of oralâ€facialâ€digital syndrome type 1. Congenital Anomalies (discontinued), 2013, 53, 155-159.	0.3	3
141	Growth profiles of 34 patients with Wolf-Hirschhorn syndrome. Journal of Pediatric Genetics, 2015, 01, 033-037.	0.3	3
142	Comment on â€œDelayed myelination is not a constant feature of Allanâ€“Herndonâ€“Dudley syndrome: Report of a new case and review of the literatureâ€•by Azzolini S et al. Brain & Development 2014;36:716â€“720. Brain and Development, 2015, 37, 988-989.	0.6	3
143	A novel PLP1 mutation F240L identified in a patient with connatal type Pelizaeus-Merzbacher disease. Human Genome Variation, 2017, 4, 16044.	0.4	3
144	Aspartylglucosaminuria caused by a novel homozygous mutation in the AGA gene was identified by an exome-first approach in a patient from Japan. Brain and Development, 2017, 39, 422-425.	0.6	3

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145	A novel COL1A1 mutation in a family with osteogenesis imperfecta associated with phenotypic variabilities. Human Genome Variation, 2017, 4, 17007.	0.4	3
146	Familial 9q33q34 microduplication in siblings with developmental disorders and macrocephaly. European Journal of Medical Genetics, 2017, 60, 650-654.	0.7	3
147	The smallest de novo 20q11.2 microdeletion causing intellectual disability and dysmorphic features. Human Genome Variation, 2017, 4, 17050.	0.4	3
148	A 10q21.3q22.2 microdeletion identified in a patient with severe developmental delay and multiple congenital anomalies including congenital heart defects. Congenital Anomalies (discontinued), 2018, 58, 36-38.	0.3	3
149	Novel A178P mutation in SLC16A2 in a patient with Allanâ€Herndonâ€Dudley syndrome. Congenital Anomalies (discontinued), 2018, 58, 143-144.	0.3	3
150	Novel compound heterozygous EPG5 mutations consisted with a missense mutation and a microduplication in the exon 1 region identified in a Japanese patient with Vici syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2803-2807.	0.7	3
151	Phenotypic features of 1q41q42 microdeletion including WDR26 and FBXO28 are clinically recognizable: The first case from Japan. Brain and Development, 2019, 41, 452-455.	0.6	3
152	Epilepsy and Other Symptoms Associated with Chromosome 9q34.11 Microdeletion. Journal of Pediatric Epilepsy, 2015, 04, 023-029.	0.1	2
153	Abdominal paraganglioma in a young woman with 1p36 deletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 495-500.	0.7	2
154	De novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic West syndrome. Congenital Anomalies (discontinued), 2019, 59, 193-194.	0.3	2
155	Coffinâ€Siris syndrome with bilateral macular dysplasia caused by a novel exonic deletion in ARID1B. Congenital Anomalies (discontinued), 2020, 60, 189-193.	0.3	2
156	Molecular Profiles of Breast Cancer in a Single Institution. Anticancer Research, 2020, 40, 4567-4570.	0.5	2
157	Breakpoint junction analysis for complex genomic rearrangements with the caldera volcanoâ€like pattern. Human Mutation, 2020, 41, 2119-2127.	1.1	2
158	Novel COL4A1 mutations identified in infants with congenital hemolytic anemia in association with brain malformations. Human Genome Variation, 2020, 7, 42.	0.4	2
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