Shinji Saitoh

List of Publications by Year
in descending order

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237 papers	6,898 citations	87888 38 h-index	79698 73 g-index
241 all docs	241 docs citations	241 times ranked	8640 citing authors

#	Article	IF	Citations
1	Inherited microdeletions in the Angelman and Praderâ€“Willi syndromes define an imprinting centre on human chromosome 15. Nature Genetics, 1995, 9, 395-400.	21.4	589
2	Imprinting in Praderâ€“Willi and Angelman syndromes. Trends in Genetics, 1998, 14, 194-200.	6.7	422
3	Imprint switching on human chromosome 15 may involve alternative transcripts of the SNRPN gene. Nature Genetics, 1996, 14, 163-170.	21.4	250
4	A Novel Imprinted Gene, Encoding a RING Zinc-Finger Protein, and Overlapping Antisense Transcript in the Prader-Willi Syndrome Critical Region. Human Molecular Genetics, 1999, 8, 783-793.	2.9	220
5	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	6.2	208
6	An imprinted, mammalian bicistronic transcript encodes two independent proteins. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 5616-5621.	7.1	190
7	Molecular dissection of the Prader-Willi/Angelman syndrome region (15q11â€“13) by YAC cloning and FISH analysis. Human Molecular Genetics, 1992, 1, 417-425.	2.9	169
8	Minimal definition of the imprinting center and fixation of chromosome 15q11-q13 epigenotype by imprinting mutations.. Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 7811-7815.	7.1	168
9	A Longer Polyalanine Expansion Mutation in the ARX Gene Causes Early Infantile Epileptic Encephalopathy with Suppression-Burst Pattern (Ohtahara Syndrome). American Journal of Human Genetics, 2007, 81, 361-366.	6.2	168
10	Phenotypic Spectrum of COL4A1 Mutations: Porencephaly to Schizencephaly. Annals of Neurology, 2013, 73, 48-57.	5.3	143
11	A novel maternally expressed gene, ATP10C, encodes a putative aminophospholipid translocase associated with Angelman syndrome. Nature Genetics, 2001, 28, 19-20.	21.4	136
12	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	6.4	132
13	Dominant-Negative Mutations in Î±-II Spectrin Cause West Syndrome with Severe Cerebral Hypomyelination, Spastic Quadriplegia, and Developmental Delay. American Journal of Human Genetics, 2010, 86, 881-891.	6.2	131
14	Molecular and clinical study of 61 Angelman syndrome patients. American Journal of Medical Genetics Part A, 1994, 52, 158-163.	2.4	103
15	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	3.3	102
16	Familial Angelman syndrome caused by imprinted submicroscopic deletion encompassing GABAA receptor Î²3-subunit gene. Lancet, The, 1992, 339, 366-367.	13.7	96
17	Decreased Tonic Inhibition in Cerebellar Granule Cells Causes Motor Dysfunction in a Mouse Model of Angelman Syndrome. Science Translational Medicine, 2012, 4, 163ra157.	12.4	96
18	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients. Genetics in Medicine, 2017, 19, 1356-1366.	2.4	96

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19	Molecular Genetic Dissection and Neonatal/Infantile Intrahepatic Cholestasis Using Targeted Next-Generation Sequencing. Journal of Pediatrics, 2016, 171, 171-177.e4.	1.8	88
20	Clinical spectrum and molecular diagnosis of Angelman and Prader-Willi syndrome patients with an imprinting mutation. , 1997, 68, 195-206.		84
21	Parent-of-Origin Specific Histone Acetylation and Reactivation of a Key Imprinted Gene Locus in Prader-Willi Syndrome. American Journal of Human Genetics, 2000, 66, 1958-1962.	6.2	74
22	Breakage in the SNRPN locus in a balanced 46,XY,t(15;19) Prader-Willi syndrome patient. Human Molecular Genetics, 1996, 5, 517-524.	2.9	73
23	Application of Magnetoencephalography in Epilepsy Patients with Widespread Spike or Slow-wave Activity. Epilepsia, 2005, 46, 1264-1272.	5.1	72
24	Inflammatory changes in infantile-onset LMNA-associated myopathy. Neuromuscular Disorders, 2011, 21, 563-568.	0.6	67
25	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance. Neurogenetics, 2012, 13, 327-332.	1.4	66
26	Angelman Syndrome in Three Siblings: Characteristic Epileptic Seizures and EEG Abnormalities. Epilepsia, 1992, 33, 1078-1082.	5.1	64
27	Radial Glial Fibers Promote Neuronal Migration and Functional Recovery after Neonatal Brain Injury. Cell Stem Cell, 2018, 22, 128-137.e9.	11.1	63
28	Title is missing!. Nature Genetics, 2001, 28, 19-20.	21.4	57
29	A model system to study genomic imprinting of human genes. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 14857-14862.	7.1	55
30	Acute encephalopathy in children with Dravet syndrome. Epilepsia, 2012, 53, 79-86.	5.1	53
31	KIF1A mutation in a patient with progressive neurodegeneration. Journal of Human Genetics, 2014, 59, 639-641.	2.3	53
32	Wide clinical variability in a family with a CACNA1A T666m mutation: hemiplegic migraine, coma, and progressive ataxia. Pediatric Neurology, 2002, 26, 47-50.	2.1	52
33	Association of HLA-A*31:01 Screening With the Incidence of Carbamazepine-Induced Cutaneous Adverse Reactions in a Japanese Population. JAMA Neurology, 2018, 75, 842.	9.0	52
34	Mutations in <scp>COG2</scp> encoding a subunit of the conserved oligomeric golgi complex cause a congenital disorder of glycosylation. Clinical Genetics, 2015, 87, 455-460.	2.0	51
35	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	1.8	46
36	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. Scientific Reports, 2017, 7, 3552.	3.3	46

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37	Molecular genetic analysis of 30 families with Joubert syndrome. Clinical Genetics, 2016, 90, 526-535.	2.0	45
38	Parthenogenetic chimaerism/mosaicism with a Silver-Russell syndrome-like phenotype. Journal of Medical Genetics, 2010, 47, 782-785.	3.2	41
39	Phenotype variability and allelic heterogeneity in KMT2B-Associated disease. Parkinsonism and Related Disorders, 2018, 52, 55-61.	2.2	41
40	Clinical and neuroimaging findings in children with posterior reversible encephalopathy syndrome. European Journal of Paediatric Neurology, 2015, 19, 672-678.	1.6	40
41	Growth Factors Released from Gelatin Hydrogel Microspheres Increase New Neurons in the Adult Mouse Brain. Stem Cells International, 2012, 2012, 1-7.	2.5	38
42	Targeted nextâ€generation sequencing in the diagnosis of neurodevelopmental disorders. Clinical Genetics, 2015, 88, 288-292.	2.0	35
43	Clinical, Pathologic, and Genetic Features of Neonatal Dubin-Johnson Syndrome: A Multicenter Study in Japan. Journal of Pediatrics, 2018, 196, 161-167.e1.	1.8	35
44	Increased Protein Stability of CDKN1C Causes a Gain-of-Function Phenotype in Patients with IMAGe Syndrome. PLoS ONE, 2013, 8, e75137.	2.5	34
45	Enhancement of Neuroblast Migration into the Injured Cerebral Cortex Using Laminin-Containing Porous Sponge. Tissue Engineering - Part A, 2015, 21, 193-201.	3.1	33
46	Effects of long working hours and shift work during pregnancy on obstetric and perinatal outcomes: A large prospective cohort studyâ€"Japan Environment and Childrenâ€™s Study. Birth, 2020, 47, 67-79.	2.2	33
47	Epimutation (hypomethylation) affecting the chromosome 14q32.2 imprinted region in a girl with upd(14)mat-like phenotype. European Journal of Human Genetics, 2008, 16, 1019-1023.	2.8	32
48	West syndrome associated with mosaic duplication of FOXG1 in a patient with maternal uniparental disomy of chromosome 14. American Journal of Medical Genetics, Part A, 2011, 155, 2584-2588.	1.2	32
49	A lossâ€ofâ€function mutation in the SLC9A6 gene causes Xâ€linked mental retardation resembling Angelman syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 799-807.	1.7	32
50	Seizure characteristics of epilepsy in childhood after acute encephalopathy with biphasic seizures and late reduced diffusion. Epilepsia, 2015, 56, 1286-1293.	5.1	31
51	Good death for children with cancer: a qualitative study. Japanese Journal of Clinical Oncology, 2015, 45, 349-355.	1.3	29
52	De novo variants in SETD1B cause intellectual disability, autism spectrum disorder, and epilepsy with myoclonic absences. Epilepsia Open, 2019, 4, 476-481.	2.4	29
53	Novel splicing mutation in the ASXL3 gene causing Bainbridgeâ€"Ropers syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1863-1867.	1.2	28
54	Congenital goitrous hypothyroidism is caused by dysfunction of the iodide transporter SLC26A7. Communications Biology, 2019, 2, 270.	4.4	28

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55	Aberrant somatosensory-evoked responses imply GABAergic dysfunction in Angelman syndrome. NeuroImage, 2008, 39, 593-599.	4.2	27
56	Novel MCA/ID syndrome with ASH1L mutation. American Journal of Medical Genetics, Part A, 2017, 173, 1644-1648.	1.2	27
57	Biallelic VPS35L pathogenic variants cause 3C/Ritscher-Schinzel-like syndrome through dysfunction of retriever complex. Journal of Medical Genetics, 2020, 57, 245-253.	3.2	27
58	Dynamic Statistical Parametric Mapping for Analyzing the Magnetoencephalographic Epileptiform Activity in Patients With Epilepsy. Journal of Child Neurology, 2005, 20, 363-369.	1.4	26
59	Cumulative exposure assessment of neonicotinoids and an investigation into their intake-related factors in young children in Japan. Science of the Total Environment, 2021, 750, 141630.	8.0	26
60	Germline mosaicism of a novel mutation in lysosome-associated membrane protein-2 deficiency (Danon) Tj ETQq0 0 0 rgBT /Overlock 10	3.3	25
61	Clinical phenotype and candidate genes for the 5q31.3 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1891-1896.	1.2	25
62	Diclofenac enhances proinflammatory cytokine-induced phagocytosis of cultured microglia via nitric oxide production. Toxicology and Applied Pharmacology, 2013, 268, 99-105.	2.8	25
63	Adverse pregnancy and perinatal outcome in patients with recurrent pregnancy loss: Multiple imputation analyses with propensity score adjustment applied to a largeâ€scale birth cohort of the Japan Environment and Childrenâ€™s Study. American Journal of Reproductive Immunology, 2019, 81, e13072.	1.2	25
64	Truncating mutation in NFIA causes brain malformation and urinary tract defects. Human Genome Variation, 2015, 2, 15007.	0.7	24
65	Early phase 2 trial of TASâ€205 in patients with Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 181-190.	3.7	24
66	High uptake on 11C-methionine positron emission tomographic scan of basal ganglia germinoma with cerebral hemiatrophy. American Journal of Neuroradiology, 2003, 24, 1909-11.	2.4	24
67	Molecular genetic study of Japanese patients with X-linked ?-thalassemia/mental retardation syndrome (ATR-X). American Journal of Medical Genetics Part A, 2000, 94, 242-248.	2.4	23
68	TUBA1A mutation can cause a hydranencephaly-like severe form of cortical dysgenesis. Scientific Reports, 2015, 5, 15165.	3.3	23
69	CTCFdeletion syndrome: clinical features and epigenetic delineation. Journal of Medical Genetics, 2017, 54, 836-842.	3.2	23
70	Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. Journal of Human Genetics, 2011, 56, 110-124.	2.3	22
71	Diclofenac Enhances Proinflammatory Cytokine-Induced Aquaporin-4 Expression in Cultured Astrocyte. Cellular and Molecular Neurobiology, 2013, 33, 393-400.	3.3	22
72	Macitentan reverses early obstructive pulmonary vasculopathy in rats: early intervention in overcoming the survivin-mediated resistance to apoptosis. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2015, 308, L523-L538.	2.9	22

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73	A novel missense mutation in the HECT domain of NEDD4L identified in a girl with periventricular nodular heterotopia, polymicrogyria and cleft palate. Journal of Human Genetics, 2017, 62, 861-863.	2.3	22
74	Biallelic mutations in SZT2 cause a discernible clinical entity with epilepsy, developmental delay, macrocephaly and a dysmorphic corpus callosum. Brain and Development, 2018, 40, 134-139.	1.1	22
75	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	3.2	22
76	MERRF/MELAS overlap syndrome: a double pathogenic mutation in mitochondrial tRNA genes. Journal of Medical Genetics, 2010, 47, 659-664.	3.2	21
77	A combination of genetic and biochemical analyses for the diagnosis of PI3K-AKT-mTOR pathway-associated megalencephaly. BMC Medical Genetics, 2017, 18, 4.	2.1	21
78	Endometriosis and Recurrent Pregnancy Loss as New Risk Factors for Venous Thromboembolism during Pregnancy and Post-Partum: The JECS Birth Cohort. Thrombosis and Haemostasis, 2019, 119, 606-617.	3.4	21
79	Impact of School Closures due to COVID-19 on Children with Neurodevelopmental Disorders in Japan. Journal of Autism and Developmental Disorders, 2022, 52, 2149-2155.	2.7	21
80	Bilateral germinoma involving the basal ganglia and cerebral white matter. American Journal of Neuroradiology, 2005, 26, 1166-9.	2.4	21
81	IMSindel: An accurate intermediate-size indel detection tool incorporating de novo assembly and gapped global-local alignment with split read analysis. Scientific Reports, 2018, 8, 5608.	3.3	20
82	Cerebellar and brainstem involvement in familial juvenile nephronophthisis type I. Pediatric Neurology, 2003, 28, 142-144.	2.1	19
83	Non-skewed X-inactivation may cause mental retardation in a female carrier of X-linked Î±-thalassemia/mental retardation syndrome (ATR-X): X-inactivation study of nine female carriers of ATR-X. American Journal of Medical Genetics, Part A, 2005, 138A, 18-20.	1.2	19
84	Detection of lymphocytes and granulocytes expressing the mutant WASP message in carriers of Wiskottâ€Aldrich syndrome. British Journal of Haematology, 1999, 104, 893-900.	2.5	18
85	Mosaic paternally derived inv dup(15) may partially rescue the Praderâ€“Willi syndrome phenotype with uniparental disomy. Clinical Genetics, 2007, 72, 378-380.	2.0	18
86	Genetic analysis of two Japanese families with progressive external ophthalmoplegia and parkinsonism. Journal of Neurology, 2011, 258, 1327-1332.	3.6	18
87	Schaaf-Yang syndrome shows a Prader-Willi syndrome-like phenotype during infancy. Orphanet Journal of Rare Diseases, 2019, 14, 277.	2.7	18
88	Uniparental disomy and imprinting defects in Japanese patients with Angelman syndrome. Brain and Development, 2005, 27, 389-391.	1.1	17
89	The applications of time-frequency analyses to ictal magnetoencephalography in neocortical epilepsy. Epilepsy Research, 2010, 90, 199-206.	1.6	17
90	Adult Leigh Disease Without Failure to Thrive. Neurologist, 2011, 17, 222-227.	0.7	17

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91	Molecular genetic and clinical delineation of 22 patients with congenital hypogonadotropic hypogonadism. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1111-1118.	0.9	17
92	A novel truncating mutation in FLNA causes periventricular nodular heterotopia, Ehlers-Danlos-like collagenopathy and macrothrombocytopenia. Brain and Development, 2018, 40, 489-492.	1.1	17
93	Six yearsâ€™ accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. Journal of Human Genetics, 2022, 67, 505-513.	2.3	17
94	Possible involvement of the tip of temporal lobe in Landauâ€“Kleffner syndrome. Brain and Development, 2007, 29, 529-533.	1.1	16
95	Homoplasmy of a mitochondrial 3697G>A mutation causes Leigh syndrome. Journal of Human Genetics, 2014, 59, 405-407.	2.3	16
96	Magnetoencephalography localizing spike sources of atypical benign partial epilepsy. Brain and Development, 2014, 36, 21-27.	1.1	16
97	Downbeat positioning nystagmus is a common clinical feature despite variable phenotypes in an FHM1 family. Journal of Neurology, 2008, 255, 1541-1544.	3.6	15
98	[11C]Flumazenil Positron Emission Tomography Analyses of Brain Gamma-Aminobutyric Acid Type A Receptors in Angelman Syndrome. Journal of Pediatrics, 2008, 152, 546-549.e3.	1.8	15
99	Successful Alternative Treatment Containing Vindesine for Acute Lymphoblastic Leukemia With Charcot-Marie-Tooth Disease. Journal of Pediatric Hematology/Oncology, 2012, 34, 239-241.	0.6	15
100	Advantageous information provided by magnetoencephalography for patients with neocortical epilepsy. Brain and Development, 2015, 37, 237-242.	1.1	15
101	Exposure levels of organophosphate pesticides in Japanese diapered children: Contributions of exposure-related behaviors and mothersâ€™ considerations of food selection and preparation. Environment International, 2020, 134, 105294.	10.0	15
102	Association between Prenatal Exposure to Household Pesticides and Neonatal Weight and Length Growth in the Japan Environment and Childrenâ€™s Study. International Journal of Environmental Research and Public Health, 2020, 17, 4608.	2.6	15
103	Clinical and genetic investigation of 136 Japanese patients with congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 691-701.	0.9	15
104	A new detection method forATRX gene mutations using a mismatch-specific endonuclease. American Journal of Medical Genetics, Part A, 2006, 140A, 1519-1523.	1.2	14
105	Childhood-onset anti-MuSK antibody positive myasthenia gravis demonstrates a distinct clinical course. Brain and Development, 2012, 34, 784-786.	1.1	14
106	Inflammatory cytokine tumor necrosis factor Î± suppresses neuroprotective endogenous erythropoietin from astrocytes mediated by hypoxiaâ€inducible factorâ€2Î±. European Journal of Neuroscience, 2014, 40, 3620-3626.	2.6	14
107	A combination of targeted enrichment methodologies for whole-exome sequencing reveals novel pathogenic mutations. Scientific Reports, 2015, 5, 9331.	3.3	14
108	A novel P gene missense mutation in a Japanese patient with oculocutaneous albinism type II (OCA2). Journal of Dermatological Science, 2003, 31, 189-192.	1.9	13

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109	Visualization of the spatial positioning of the SNRPN, UBE3A, and GABRB3 genes in the normal human nucleus by three-color 3D fluorescence in situ hybridization. Chromosome Research, 2012, 20, 659-672.	2.2	13
110	Magnetoencephalographic analysis of paroxysmal fast activity in patients with epileptic spasms. Epilepsy Research, 2013, 104, 68-77.	1.6	13
111	Prenatal diagnosis of <scp>X</scp>â€linked recessive <scp>L</scp>enz microphthalmia syndrome. Journal of Obstetrics and Gynaecology Research, 2013, 39, 1545-1547.	1.3	13
112	Role of a heterotrimeric Gâ€protein, Gi2, in the corticogenesis: possible involvement in periventricular nodular heterotopia and intellectual disability. Journal of Neurochemistry, 2017, 140, 82-95.	3.9	13
113	Germline mosaicism of a novelUBE3A mutation in Angelman syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 187-189.	1.2	12
114	An infantileâ€“juvenile form of Alexander disease caused by a R79H mutation in GFAP. Brain and Development, 2006, 28, 131-133.	1.1	12
115	Direct correlation between the facial nerve nucleus and hemifacial seizures associated with a gangliocytoma of the floor of the fourth ventricle: A case report. Epilepsia, 2011, 52, e204-e206.	5.1	12
116	Novel compound heterozygous variants in PLK4 identified in a patient with autosomal recessive microcephaly and chorioretinopathy. European Journal of Human Genetics, 2016, 24, 1702-1706.	2.8	12
117	Distinctive facies, macrocephaly, and developmental delay are signs of a PTEN mutation in childhood. Brain and Development, 2018, 40, 678-684.	1.1	12
118	MYCNde novo gain-of-function mutation in a patient with a novel megalencephaly syndrome. Journal of Medical Genetics, 2019, 56, 388-395.	3.2	12
119	Angelman syndrome caused by an identical familial 1,487-kb deletion. American Journal of Medical Genetics, Part A, 2007, 143A, 98-101.	1.2	11
120	Probability curves focusing on symptom severity during an oral food challenge. Annals of Allergy, Asthma and Immunology, 2014, 112, 556-557.e2.	1.0	11
121	Development of tandem mass spectrometry-based creatinine measurement using dried blood spot for newborn mass screening. Pediatric Research, 2017, 82, 237-243.	2.3	11
122	Predominant area of brain lesions in neonates with herpes simplex encephalitis. Journal of Perinatology, 2017, 37, 1210-1214.	2.0	11
123	Constitutive activation of mTORC1 signaling induced by biallelic loss-of-function mutations in SZT2 underlies a discernible neurodevelopmental disease. PLoS ONE, 2019, 14, e0221482.	2.5	11
124	Transition from Leigh syndrome to MELAS syndrome in a patient with heteroplasmic MT-ND3 m.10158T>C. Brain and Development, 2019, 41, 803-807.	1.1	11
125	Clinical characteristics of Angelman syndrome patients with a non-IC-deleted imprinting mutation. Clinical Genetics, 1999, 55, 277-278.	2.0	10
126	A de novo direct duplication of 16q22.1â€‰â†’â€‰q23.1 in a boy with midface hypoplasia and mental retardation. American Journal of Medical Genetics, Part A, 2009, 149A, 2560-2563.	1.2	10

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127	MEG timeâ€“frequency analyses for pre- and post-surgical evaluation of patients with epileptic rhythmic fast activity. Epilepsy Research, 2010, 88, 100-107.	1.6	10
128	Handâ€“footâ€“genital syndrome with a 7p15 deletion: Clinically recognizable syndrome. Pediatrics International, 2012, 54, e22-5.	0.5	10
129	Episodic tremors representing cortical myoclonus are characteristic in Angelman syndrome due to UBE3A mutations. Brain and Development, 2015, 37, 216-222.	1.1	10
130	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome. Journal of Human Genetics, 2018, 63, 957-963.	2.3	10
131	Intrauterine growth and the maturation process of adrenal function. PeerJ, 2019, 7, e6368.	2.0	10
132	Endosomal Recycling Defects and Neurodevelopmental Disorders. Cells, 2022, 11, 148.	4.1	10
133	Phenotypic variability in a family with a mitochondrial DNA T8993C mutation. Pediatric Neurology, 1998, 19, 283-286.	2.1	9
134	Pitfall in the Diagnosis of Fructose-1,6-Bisphosphatase Deficiency: Difficulty in Detecting Glycerol-3-Phosphate with Solvent Extraction in Urinary GC/MS Analysis. Tohoku Journal of Experimental Medicine, 2015, 237, 235-239.	1.2	9
135	Concordance of DSMâ€5 and DSMâ€IVâ€TR classifications for autism spectrum disorder. Pediatrics International, 2015, 57, 1097-1100.	0.5	9
136	Combined genetic analyses can achieve efficient diagnostic yields for subjects with Alagille syndrome and incomplete Alagille syndrome. Acta Paediatrica, International Journal of Paediatrics, 2017, 106, 1817-1824.	1.5	9
137	A de novo p.Arg756Cys mutation in ATP1A3 causes a distinct phenotype with prolonged weakness and encephalopathy triggered by fever. Brain and Development, 2018, 40, 222-225.	1.1	9
138	Attitudes of school teachers toward epilepsy in Nagoya, Japan. Epilepsy and Behavior, 2020, 103, 106359.	1.7	9
139	Itpr1 regulates the formation of anterior eye segment tissues derived from neural crest cells. Development (Cambridge), 2021, 148, .	2.5	9
140	Comparison of Three Methods for Localizing Interictal Epileptiform Discharges With Magnetoencephalography. Journal of Clinical Neurophysiology, 2011, 28, 431-440.	1.7	9
141	A novel splicing mutation of the ATRX gene in ATR-X syndrome. Brain and Development, 2006, 28, 322-325.	1.1	8
142	Vaccineâ€associated paralytic poliomyelitis in a nonâ€immunocompromised infant. Pediatrics International, 2010, 52, 838-841.	0.5	8
143	Tissueâ€limited ring chromosome 18 mosaicism as a cause of Pittâ€“Hopkins syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2621-2623.	1.2	8
144	Effects of tolvaptan on congestive heart failure complicated with chylothorax in a neonate. Pediatrics International, 2015, 57, 1020-1022.	0.5	8

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145	Siblings with optic neuropathy and RTN4IP1 mutation. Journal of Human Genetics, 2017, 62, 927-929.	2.3	8
146	Delayed recognition of childhood arterial ischemic stroke. Pediatrics International, 2019, 61, 895-903.	0.5	8
147	A novel splicing mutation in SLC9A6 in a boy with Christianson syndrome. Human Genome Variation, 2019, 6, 15.	0.7	8
148	A novel missense variant in CUL3 shows altered binding ability to BTB-adaptor proteins leading to diverse phenotypes of CUL3-related disorders. Journal of Human Genetics, 2021, 66, 491-498.	2.3	8
149	Association of Maternal Total Cholesterol With SGA or LGA Birth at Term: the Japan Environment and Childrenâ€™s Study. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e118-e129.	3.6	8
150	Cortical reflex myoclonus associated with mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes (MELAS): A case report. Brain and Development, 1992, 14, 260-263.	1.1	7
151	Unmasking 15q12 deletion using microarrayâ€based comparative genomic hybridization in a mentally retarded boy with r(Y). American Journal of Medical Genetics Part A, 2004, 130A, 322-324.	2.4	7
152	SNP array screening of cryptic genomic imbalances in 450 Japanese subjects with intellectual disability and multiple congenital anomalies previously negative for large rearrangements. Journal of Human Genetics, 2016, 61, 335-343.	2.3	7
153	A novel genetic syndrome with STARD9 mutation and abnormal spindle morphology. American Journal of Medical Genetics, Part A, 2017, 173, 2690-2696.	1.2	7
154	Pharmacoresistant epileptic eyelid twitching in a child with a mutation in SYNGAP1. Epileptic Disorders, 2017, 19, 339-344.	1.3	7
155	Antiviral therapy for hepatitis B virus during second pregnancies. Journal of Obstetrics and Gynaecology Research, 2018, 44, 566-569.	1.3	7
156	Feeding-Induced Cortisol Response in Newborn Infants. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4450-4455.	3.6	7
157	Genome-wide methylation analysis in Silverâ€“Russell syndrome, Temple syndrome, and Praderâ€“Willi syndrome. Clinical Epigenetics, 2020, 12, 159.	4.1	7
158	Influence of Percutaneous Occlusion of Atrial Septal Defect on Left Atrial Function Evaluated Using 2D Speckle Tracking Echocardiography. International Heart Journal, 2020, 61, 83-88.	1.0	7
159	Relationship between delivery with anesthesia and postpartum depression: The Japan Environment and Childrenâ€™s Study (JECS). BMC Pregnancy and Childbirth, 2021, 21, 522.	2.4	7
160	Four pedigrees with aminoacyl-tRNA synthetase abnormalities. Neurological Sciences, 2022, 43, 2765-2774.	1.9	7
161	A common insertion/deletion polymorphism in the Praderâ€”Willi syndrome minimal critical region. Human Molecular Genetics, 1994, 3, 1912-1912.	2.9	6
162	Food sensitization in Japanese infants is associated with a common Filaggrin variant. Annals of Allergy, Asthma and Immunology, 2013, 110, 388-390.e1.	1.0	6

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163	Primary Microcephaly With Anterior Predominant Pachygyria Caused by Novel Compound Heterozygous Mutations in ASPM. Pediatric Neurology, 2015, 52, e7-e8.	2.1	6
164	Probability curves for predicting symptom severity during oral food challenge with milk. Annals of Allergy, Asthma and Immunology, 2015, 115, 251-253.	1.0	6
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