Shinji Saitoh

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/914765/publications.pdf

Version: 2024-02-01

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	House dust mite SLIT-tablet is well tolerated in pediatric patients with controlled asthma. Asian Pacific Journal of Allergy and Immunology, 2022, , .	0.4	1
2	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
3	Validation of actigraphy in hospitalised newborn infants using video polysomnography. Journal of Sleep Research, 2022, 31, e13437.	3.2	3
4	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
5	Four pedigrees with aminoacyl-tRNA synthetase abnormalities. Neurological Sciences, 2022, 43, 2765-2774.	1.9	7
6	Diagnostic rate of autism spectrum disorder in a highâ€survival cohort of children born very preterm: A crossâ€sectional study. International Journal of Developmental Neuroscience, 2022, 82, 188-195.	1.6	2
7	Endosomal Recycling Defects and Neurodevelopmental Disorders. Cells, 2022, 11, 148.	4.1	10
8	Hemorrhagic shock and encephalopathy syndrome in a patient with a de novo heterozygous variant in KIF1A. Brain and Development, 2022, 44, 249-253.	1.1	1
9	Impact of Ready-Meal Consumption during Pregnancy on Birth Outcomes: The Japan Environment and Childrenâ€™s Study. Nutrients, 2022, 14, 895.	4.1	3
10	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
11	Real-life Progression of the Use of a Genetic Panel in to Diagnose Neonatal Cholestasis. JPGN Reports, 2022, 3, e196.	0.4	2
12	Repeated maternal non-responsiveness to baby's crying during postpartum and infant neuropsychological development: The Japan Environment and Childrenâ€™s Study. Child Abuse and Neglect, 2022, 127, 105581.	2.6	1
13	Simultaneous quantification of pyrethroid metabolites in urine of non-toilet-trained children in Japan. Environmental Health and Preventive Medicine, 2022, 27, 25-25.	3.4	0
14	The iodide transporter Slc26a7 impacts thyroid function more strongly than Slc26a4 in mice. Scientific Reports, 2022, 12, .	3.3	3
15	Depression symptoms during pregnancy and postpartum in patients with recurrent pregnancy loss and infertility: The Japan environment and childrenâ€™s study. Journal of Reproductive Immunology, 2022, 152, 103659.	1.9	2
16	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	3.2	22
17	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
18	The identification of two pathogenic variants in a family with mild and severe forms of developmental delay. Journal of Human Genetics, 2021, 66, 445-448.	2.3	0

#	Article	IF	Citations
19	Phosphorylated proteome analysis of a novel germline ABL1 mutation causing an autosomal dominant syndrome with ventricular septal defect. International Journal of Cardiology, 2021, 326, 81-87.	1.7	2
20	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
21	Promoting sound development of preterm infants in the name of developmental neuroscience: Beyond advanced life support and neuroprotection. Pediatrics and Neonatology, 2021, 62, S10-S15.	0.9	3
22	Comprehensive Genetic Analysis of Non-syndromic Autism Spectrum Disorder in Clinical Settings. Journal of Autism and Developmental Disorders, 2021, 51, 4655-4662.	2.7	4
23	Homozygous ADCY5 mutation causes early-onset movement disorder with severe intellectual disability. Neurological Sciences, 2021, 42, 2975-2978.	1.9	3
24	Short-latency somatosensory-evoked potentials demonstrate cortical dysfunction in patients with Angelman syndrome. ENeurologicalSci, 2021, 22, 100298.	1.3	0
25	Temporal inversion of the acid-base equilibrium in newborns: an observational study. PeerJ, 2021, 9, e11240.	2.0	0
26	Variance in the pathophysiological impact of the hemizygosity of gamma-aminobutyric acid type A receptor subunit genes between Prader-Willi syndrome and Angelman syndrome. Brain and Development, 2021, 43, 521-527.	1.1	1
27	Peripheral nerves are involved in hypomyelinating leukodystrophy-3 caused by a homozygous AIMP1 variant. Brain and Development, 2021, 43, 590-595.	1.1	1
28	Airway gas temperature within endotracheal tube can be monitored using rapid response thermometer. Scientific Reports, 2021, 11, 9537.	3.3	2
29	Breakthrough HBV infection in a vaccinated child due to vaccine escape mutant. Acta Hepatologica Japonica, 2021, 62, 403-412.	0.1	0
30	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
31	Death review of children receiving medical care at home. Pediatric Research, 2021, , .	2.3	1
32	Evidence of both foetal inflammation and hypoxiaâ€“ischaemia is associated with meconium aspiration syndrome. Scientific Reports, 2021, 11, 16799.	3.3	1
33	Utility of breakpointâ€specific nested polymerase chain reaction for the diagnosis of Emanuel syndrome. Pediatrics International, 2021, 63, 1534-1536.	0.5	1
34	SCN8A-related developmental and epileptic encephalopathy with ictal asystole requiring cardiac pacemaker implantation. Brain and Development, 2021, 43, 804-808.	1.1	3
35	Itpr1 regulates the formation of anterior eye segment tissues derived from neural crest cells. Development (Cambridge), 2021, 148, .	2.5	9
36	A case of ATR-X syndrome with mitochondrial respiratory chain dysfunction. European Journal of Medical Genetics, 2021, 64, 104251.	1.3	1

#	Article	IF	Citations
37	Relationship between Physical Activity and Physical and Mental Health Status in Pregnant Women: A Prospective Cohort Study of the Japan Environment and Childrenâ€™s Study. International Journal of Environmental Research and Public Health, 2021, 18, 11373.	2.6	2
38	A Novel Î±-Spectrin Pathogenic Variant in Trans to Î±-Spectrin LELY Causing Neonatal Jaundice With Hemolytic Anemia From Hereditary Pyropoikilocytosis Coexisting With Gilbert Syndrome. Journal of Pediatric Hematology/Oncology, 2021, 43, e250-e254.	0.6	0
39	Acute encephalopathy with biphasic seizures and late reduced diffusion: Predictive EEG findings. Brain and Development, 2021, , .	1.1	5
40	Influence of mothersâ€™ nighttime responses on the sleepâ€“wake rhythm of 1-month-old infants. Scientific Reports, 2021, 11, 24363.	3.3	2
41	Splenial Lesions in Benign Convulsions With Gastroenteritis Associated With Rotavirus Infection. Pediatric Neurology, 2020, 109, 79-84.	2.1	6
42	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
43	Respiratory illness and acute flaccid myelitis in the Tokai district in 2018. Pediatrics International, 2020, 62, 337-340.	0.5	2
44	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
45	Attitudes of school teachers toward epilepsy in Nagoya, Japan. Epilepsy and Behavior, 2020, 103, 106359.	1.7	9
46	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
47	Successful treatment of adult-onset type II citrullinemia with a low-carbohydrate diet and l-arginine after DNA analysis produced a definitive diagnosis. Clinical Journal of Gastroenterology, 2020, 13, 823-833.	0.8	4
48	Novel compound heterozygous MCOLN1 mutations identified in a Japanese girl with severe developmental delay and thin corpus callosum. Brain and Development, 2020, 42, 298-301.	1.1	1
49	Reliability and validity of a Japanese version of the psychosocial assessment tool for families of children with cancer. Japanese Journal of Clinical Oncology, 2020, 50, 296-302.	1.3	3
50	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
51	Behavioral problems and family distress in tuberous sclerosis complex. Epilepsy and Behavior, 2020, 111, 107321.	1.7	4
52	Genome-wide methylation analysis in Silverâ€“Russell syndrome, Temple syndrome, and Praderâ€“Willi syndrome. Clinical Epigenetics, 2020, 12, 159.	4.1	7
53	A case of tricuspid atresia with Praderâ€Willi syndrome. Pediatrics International, 2020, 62, 1105-1106.	0.5	0
54	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes. PLoS ONE, 2020, 15, e0237814.	2.5	5

#	Article	IF	Citations
55	Transient cortical diffusion restriction in children immediately after prolonged febrile seizures. European Journal of Paediatric Neurology, 2020, 27, 30-36.	1.6	2
56	De novo 2q36.3q37.1 deletion encompassing TRIP12 and NPPC yields distinct phenotypes. Human Genome Variation, 2020, 7, 19.	0.7	1
57	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
58	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
59	Clinical and genetic investigation of 136 Japanese patients with congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 691-701.	0.9	15
60	Frequent epileptic apnoea in a patient with Pittâ€Hopkins syndrome. Epileptic Disorders, 2020, 22, 673-677.	1.3	2
61	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes. , 2020, 15, e0237814.		0
62	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes. , 2020, 15, e0237814.		0
63	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes. , 2020, 15, e0237814.		0
64	Two mouse models carrying truncating mutations in Magel2 show distinct phenotypes. , 2020, 15, e0237814.		0
65	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
66	Delayed recognition of childhood arterial ischemic stroke. Pediatrics International, 2019, 61, 895-903.	0.5	8
67	Peripartum depression and infant care, sleep and growth. Scientific Reports, 2019, 9, 10186.	3.3	6
68	Congenital goitrous hypothyroidism is caused by dysfunction of the iodide transporter SLC26A7. Communications Biology, 2019, 2, 270.	4.4	28
69	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
70	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
71	A novel CUL4B splice site variant in a young male exhibiting less pronounced features. Human Genome Variation, 2019, 6, 43.	0.7	4
72	A case of earlyâ€onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy. American Journal of Medical Genetics, Part A, 2019, 179, 645-649.	1.2	3

#	Article	IF	Citations
73	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
74	Visual function scale for identification of infants with low respiratory compliance. Pediatrics and Neonatology, 2019, 60, 611-616.	0.9	2
75	Chronological dynamic changes in cortico-subcortical imbalance of cerebral blood flow in a boy with CAPOS syndrome. Brain and Development, 2019, 41, 625-629.	1.1	0
76	A novel splicing mutation in SLC9A6 in a boy with Christianson syndrome. Human Genome Variation, 2019, 6, 15.	0.7	8
77	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
78	Cohort profile: Aichi regional sub-cohort of the Japan Environment and Childrenâ€™s Study (JECS-A). BMJ Open, 2019, 9, e028105.	1.9	6
79	Schaaf-Yang syndrome shows a Prader-Willi syndrome-like phenotype during infancy. Orphanet Journal of Rare Diseases, 2019, 14, 277.	2.7	18
80	Echovirus Type 7 Virus-Associated Hemophagocytic Syndrome in a Neonate Successfully Treated With Intravenous Immunoglobulin Therapy: A Case Report. Frontiers in Pediatrics, 2019, 7, 469.	1.9	5
81	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
82	Intrauterine growth and the maturation process of adrenal function. PeerJ, 2019, 7, e6368.	2.0	10
83	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
84	Phenotype variability and allelic heterogeneity in KMT2B-Associated disease. Parkinsonism and Related Disorders, 2018, 52, 55-61.	2.2	41
85	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
86	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
87	Radial Glial Fibers Promote Neuronal Migration and Functional Recovery after Neonatal Brain Injury. Cell Stem Cell, 2018, 22, 128-137.e9.	11.1	63
88	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
89	Regional Differences in Clinical Features of Kaposiform Hemangioendothelioma of the Intestinal Tract. Journal of Pediatric Hematology/Oncology, 2018, 40, 491-493.	0.6	2
90	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

#	Article	IF	Citations
91	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
92	Antiviral therapy for hepatitis B virus during second pregnancies. Journal of Obstetrics and Gynaecology Research, 2018, 44, 566-569.	1.3	7
93	Feeding-Induced Cortisol Response in Newborn Infants. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4450-4455.	3.6	7
94	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	6.4	132
95	Distinctive facies, macrocephaly, and developmental delay are signs of a PTEN mutation in childhood. Brain and Development, 2018, 40, 678-684.	1.1	12
96	Estimation of elevated intracranial pressure in infants with hydroce-phalus by using transcranial Doppler velocimetry with fontanel compression. Scientific Reports, 2018, 8, 11824.	3.3	3
97	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
98	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
99	Novel MCA/ID syndrome with ASH1L mutation. American Journal of Medical Genetics, Part A, 2017, 173, 1644-1648.	1.2	27
100	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
101	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
102	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. Scientific Reports, 2017, 7, 3552.	3.3	46
103	Probability curves for predicting symptom severity during an oral food challenge with wheat. Allergology International, 2017, 66, 627-628.	3.3	4
104	CTCFdeletion syndrome: clinical features and epigenetic delineation. Journal of Medical Genetics, 2017, 54, 836-842.	3.2	23
105	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
106	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
107	Predominant area of brain lesions in neonates with herpes simplex encephalitis. Journal of Perinatology, 2017, 37, 1210-1214.	2.0	11
108	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

#	Article	IF	Citations
109	Pharmacoresistant epileptic eyelid twitching in a child with a mutation in SYNGAP1. Epileptic Disorders, 2017, 19, 339-344.	1.3	7
110	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients. Genetics in Medicine, 2017, 19, 1356-1366.	2.4	96
111	Siblings with optic neuropathy and RTN4IP1 mutation. Journal of Human Genetics, 2017, 62, 927-929.	2.3	8
112	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
113	Effects of 4â€phenylbutyrate therapy in a preterm infant with cholestasis and liver fibrosis. Pediatrics International, 2016, 58, 506-509.	0.5	5
114	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
115	Single nucleotide polymorphisms in AGTR1, TFAP2B, and TRAF1 are not associated with the incidence of patent ductus arteriosus in Japanese preterm infants. Pediatrics International, 2016, 58, 461-466.	0.5	4
116	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	3.3	102
117	Treatment Strategy for Pediatric Paratesticular Rhabdomyosarcoma Based on Chimeric Gene Assessment. Urology, 2016, 95, 187-189.	1.0	1
118	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
119	Molecular genetic analysis of 30 families with Joubert syndrome. Clinical Genetics, 2016, 90, 526-535.	2.0	45
120	Effect of Japanese cedarâ€“specific sublingual immunotherapy on allergen-specific TH2 cell counts in blood. Annals of Allergy, Asthma and Immunology, 2016, 117, 72-78.e4.	1.0	6
121	Molecular Genetic Dissection and Neonatal/Infantile Intrahepatic Cholestasis Using Targeted Next-Generation Sequencing. Journal of Pediatrics, 2016, 171, 171-177.e4.	1.8	88
122	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
123	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
124	TUBA1A mutation can cause a hydranencephaly-like severe form of cortical dysgenesis. Scientific Reports, 2015, 5, 15165.	3.3	23
125	Truncating mutation in NFIA causes brain malformation and urinary tract defects. Human Genome Variation, 2015, 2, 15007.	0.7	24
126	A combination of targeted enrichment methodologies for whole-exome sequencing reveals novel pathogenic mutations. Scientific Reports, 2015, 5, 9331.	3.3	14

#	Article	IF	Citations
127	Seizure characteristics of epilepsy in childhood after acute encephalopathy with biphasic seizures and late reduced diffusion. Epilepsia, 2015, 56, 1286-1293.	5.1	31
128	Effects of tolvaptan on congestive heart failure complicated with chylothorax in a neonate. Pediatrics International, 2015, 57, 1020-1022.	0.5	8
129	Diffuse alveolar hemorrhage secondary to <scp>ANCA</scp>â€associated vasculitis in a patient with Down syndrome. Pediatrics International, 2015, 57, e45-7.	0.5	2
130	Concordance of DSMâ€5 and DSMâ€IVâ€TR classifications for autism spectrum disorder. Pediatrics International, 2015, 57, 1097-1100.	0.5	9
131	Good death for children with cancer: a qualitative study. Japanese Journal of Clinical Oncology, 2015, 45, 349-355.	1.3	29
132	Clinical and neuroimaging findings in children with posterior reversible encephalopathy syndrome. European Journal of Paediatric Neurology, 2015, 19, 672-678.	1.6	40
133	Primary Microcephaly With Anterior Predominant Pachygyria Caused by Novel Compound Heterozygous Mutations in ASPM. Pediatric Neurology, 2015, 52, e7-e8.	2.1	6
134	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
135	Clinical, Molecular, and Neurophysiological Features in Angelman Syndrome. Journal of Pediatric Epilepsy, 2015, 04, 017-022.	0.2	3
136	Novel <scp>DCX</scp> mutationâ€caused lissencephaly in a boy and very mild heterotopia in his mother. Pediatrics International, 2015, 57, 321-323.	0.5	2
137	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
138	Perinatal management of neonatal alloimmune thrombocytopenia associated with anti-group A antibody. Transfusion Medicine, 2015, 25, 42-46.	1.1	5
139	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
140	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
141	Advantageous information provided by magnetoencephalography for patients with neocortical epilepsy. Brain and Development, 2015, 37, 237-242.	1.1	15
142	Episodic tremors representing cortical myoclonus are characteristic in Angelman syndrome due to UBE3A mutations. Brain and Development, 2015, 37, 216-222.	1.1	10
143	Targeted nextâ€generation sequencing in the diagnosis of neurodevelopmental disorders. Clinical Genetics, 2015, 88, 288-292.	2.0	35
144	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

#	Article	IF	Citations
145	Homoplasmy of a mitochondrial 3697G>A mutation causes Leigh syndrome. Journal of Human Genetics, 2014, 59, 405-407.	2.3	16
146	Fulminant Encephalopathy with Marked Brain Edema and Bilateral Thalamic Lesions. Neuropediatrics, 2014, 45, 256-260.	0.6	2
147	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
148	Combination of Millerâ€“Dieker syndrome and VACTERL association causes extremely severe clinical presentation. European Journal of Pediatrics, 2014, 173, 1541-1544.	2.7	5
149	Thalamic Lesions in Acute Encephalopathy With Biphasic Seizures and Late Reduced Diffusion. Pediatric Neurology, 2014, 51, 701-705.	2.1	2
150	KIF1A mutation in a patient with progressive neurodegeneration. Journal of Human Genetics, 2014, 59, 639-641.	2.3	53
151	Magnetoencephalography localizing spike sources of atypical benign partial epilepsy. Brain and Development, 2014, 36, 21-27.	1.1	16
152	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
153	Effect of Japanese cedar specific immunotherapy on allergen-specific TH2 cells in peripheral blood. Annals of Allergy, Asthma and Immunology, 2013, 110, 380-385.e1.	1.0	4
154	Magnetoencephalographic analysis of paroxysmal fast activity in patients with epileptic spasms. Epilepsy Research, 2013, 104, 68-77.	1.6	13
155	Diclofenac enhances proinflammatory cytokine-induced phagocytosis of cultured microglia via nitric oxide production. Toxicology and Applied Pharmacology, 2013, 268, 99-105.	2.8	25
156	Diclofenac Enhances Proinflammatory Cytokine-Induced Aquaporin-4 Expression in Cultured Astrocyte. Cellular and Molecular Neurobiology, 2013, 33, 393-400.	3.3	22
157	Phenotypic Spectrum of COL4A1 Mutations: Porencephaly to Schizencephaly. Annals of Neurology, 2013, 73, 48-57.	5.3	143
158	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
159	Increased Protein Stability of CDKN1C Causes a Gain-of-Function Phenotype in Patients with IMAGe Syndrome. PLoS ONE, 2013, 8, e75137.	2.5	34
160	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
161	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
162	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

#	Article	IF	Citations
163	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
164	A DYNC1H1 mutation causes a dominant spinal muscular atrophy with lower extremity predominance. Neurogenetics, 2012, 13, 327-332.	1.4	66
165	Childhood-onset anti-MuSK antibody positive myasthenia gravis demonstrates a distinct clinical course. Brain and Development, 2012, 34, 784-786.	1.1	14
166	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
167	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
168	Acute encephalopathy in children with Dravet syndrome. Epilepsia, 2012, 53, 79-86.	5.1	53
169	Handâ€footâ€genital syndrome with a 7p15 deletion: Clinically recognizable syndrome. Pediatrics International, 2012, 54, e22-5.	0.5	10
170	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
171	Inflammatory changes in infantile-onset LMNA-associated myopathy. Neuromuscular Disorders, 2011, 21, 563-568.	0.6	67
172	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
173	Early-onset absence epilepsy at eight months of age. Epileptic Disorders, 2011, 13, 417-421.	1.3	1
174	Genetic analysis of two Japanese families with progressive external ophthalmoplegia and parkinsonism. Journal of Neurology, 2011, 258, 1327-1332.	3.6	18
175	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
176	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
177	Adult Leigh Disease Without Failure to Thrive. Neurologist, 2011, 17, 222-227.	0.7	17
178	Comparison of Three Methods for Localizing Interictal Epileptiform Discharges With Magnetoencephalography. Journal of Clinical Neurophysiology, 2011, 28, 431-440.	1.7	9
179	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
180	The applications of time-frequency analyses to ictal magnetoencephalography in neocortical epilepsy. Epilepsy Research, 2010, 90, 199-206.	1.6	17

#	Article	IF	Citations
181	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
182	Vaccineâ€associated paralytic poliomyelitis in a nonâ€immunocompromised infant. Pediatrics International, 2010, 52, 838-841.	0.5	8
183	Parthenogenetic chimaerism/mosaicism with a Silver-Russell syndrome-like phenotype. Journal of Medical Genetics, 2010, 47, 782-785.	3.2	41
184	MERRF/MELAS overlap syndrome: a double pathogenic mutation in mitochondrial tRNA genes. Journal of Medical Genetics, 2010, 47, 659-664.	3.2	21
185	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	1.8	46
186	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
187	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	6.2	208
188	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
189	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
190	[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
191	Aberrant somatosensory-evoked responses imply GABAergic dysfunction in Angelman syndrome. NeuroImage, 2008, 39, 593-599.	4.2	27
192	Long-term sequential magnetoencephalographic analyses for patients with atypical benign partial epilepsy in childhood. International Congress Series, 2007, 1300, 689-692.	0.2	2
193	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
194	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
195	DNA demethylation reactivation of imprinted genes in cell lines from patients with Prader-Willi syndrome and a mouse model. American Journal of Medical Genetics, Part A, 2007, 143A, 1386-1390.	1.2	2
196	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
197	Possible involvement of the tip of temporal lobe in Landauâ€“Kleffner syndrome. Brain and Development, 2007, 29, 529-533.	1.1	16
198	Non-chromosome 15 marker chromosome in a Prader-Willi syndrome patient with uniparental disomy. Pediatrics International, 2006, 48, 97-99.	0.5	1

#	Article	IF	Citations
199	An infantileâ€“juvenile form of Alexander disease caused by a R79H mutation in GFAP. Brain and Development, 2006, 28, 131-133.	1.1	12
200	A novel splicing mutation of the ATRX gene in ATR-X syndrome. Brain and Development, 2006, 28, 322-325.	1.1	8
201	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
202	Application of Magnetoencephalography in Epilepsy Patients with Widespread Spike or Slow-wave Activity. Epilepsia, 2005, 46, 1264-1272.	5.1	72
203	Uniparental disomy and imprinting defects in Japanese patients with Angelman syndrome. Brain and Development, 2005, 27, 389-391.	1.1	17
204	A woman with 46,XX,dup(16)(p13.11 p13.3) and the ATR-X phenotype. American Journal of Medical Genetics, Part A, 2005, 132A, 414-418.	1.2	3
205	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
206	Germline mosaicism of a novelUBE3A mutation in Angelman syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 187-189.	1.2	12
207	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
208	Bilateral germinoma involving the basal ganglia and cerebral white matter. American Journal of Neuroradiology, 2005, 26, 1166-9.	2.4	21
209	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
210	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
211	Cerebellar and brainstem involvement in familial juvenile nephronophthisis type I. Pediatric Neurology, 2003, 28, 142-144.	2.1	19
212	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
213	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
214	Germline mosaicism of a novel mutation in lysosome-associated membrane protein-2 deficiency (Danon) Tj ETQq0 0 0 rgBT /Overlock 10	5.3	25
215	A novel maternally expressed gene, ATP10C, encodes a putative aminophospholipid translocase associated with Angelman syndrome. Nature Genetics, 2001, 28, 19-20.	21.4	136
216	Title is missing!. Nature Genetics, 2001, 28, 19-20.	21.4	57

#	Article	IF	Citations
217	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
218	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
219	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
220	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
221	Clinical characteristics of Angelman syndrome patients with a non-IC-deleted imprinting mutation. Clinical Genetics, 1999, 55, 277-278.	2.0	10
222	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
223	Imprinting in Praderâ€“Willi and Angelman syndromes. Trends in Genetics, 1998, 14, 194-200.	6.7	422
224	Phenotypic variability in a family with a mitochondrial DNA T8993C mutation. Pediatric Neurology, 1998, 19, 283-286.	2.1	9
225	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
226	Clinical spectrum and molecular diagnosis of Angelman and Prader-Willi syndrome patients with an imprinting mutation. , 1997, 68, 195-206.		84
227	Clinical spectrum and molecular diagnosis of Angelman and Praderâ€Willi syndrome patients with an imprinting mutation. American Journal of Medical Genetics Part A, 1997, 68, 195-206.	2.4	2
228	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
229	Imprint switching on human chromosome 15 may involve alternative transcripts of the SNRPN gene. Nature Genetics, 1996, 14, 163-170.	21.4	250
230	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
231	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
232	A common insertion/deletion polymorphism in the Praderâ€”Willi syndrome minimal critical region. Human Molecular Genetics, 1994, 3, 1912-1912.	2.9	6
233	Molecular and clinical study of 61 Angelman syndrome patients. American Journal of Medical Genetics Part A, 1994, 52, 158-163.	2.4	103
234	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

#	Article	IF	Citations
235	Angelman Syndrome in Three Siblings: Characteristic Epileptic Seizures and EEG Abnormalities. Epilepsia, 1992, 33, 1078-1082.	5.1	64
236	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
237	Familial Angelman syndrome caused by imprinted submicroscopic deletion encompassing GABAA receptor Î²3-subunit gene. Lancet, The, 1992, 339, 366-367.	13.7	96