Tetsuya Okazaki

List of Publications by Year
in descending order

39	454	759233 12	752698 20
papers	citations	h-index	g-index

40	40	40	1040
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	3.3	77
2	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. American Journal of Human Genetics, 2018, 103, 1009-1021.	6.2	57
3	Staurosporine, a novel protein kinase inhibitor, enhances HL-60-cell differentiation induced by various compounds. Experimental Hematology, 1988, 16, 42-8.	0.4	34
4	Probing the Inhibitor versus Chaperone Properties of sp2-Iminosugars towards Human Î²-Glucocerebrosidase: A Picomolar Chaperone for Gaucher Disease. Molecules, 2018, 23, 927.	3.8	30
5	Characterization of SPATA5â€related encephalopathy in early childhood. Clinical Genetics, 2016, 90, 437-444.	2.0	20
6	Retrospective details of false-positive and false-negative results in non-invasive prenatal testing for fetal trisomies 21, 18 and 13. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 256, 75-81.	1.1	20
7	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
8	Clinical Diagnosis of Mendelian Disorders Using a Comprehensive Gene-Targeted Panel Test for Next-Generation Sequencing. Yonago Acta Medica, 2016, 59, 118-25.	0.7	17
9	Self-rating Barthel Index Compatible with the Original Barthel Index and the Functional Independence Measure Motor Score. Journal of UOEH, 1997, 19, 107-121.	0.6	15
10	Epileptic phenotype of FGFR3-related bilateral medial temporal lobe dysgenesis. Brain and Development, 2017, 39, 67-71.	1.1	15
11	Neurodevelopmental disorders in children with macrocephaly: A prevalence study and PTEN gene analysis. Brain and Development, 2018, 40, 36-41.	1.1	14
12	Evidence of intracellular and trans-acting differentiation-inducing activity in human promyelocytic leukemia HL-60 cells: Its possible involvement in process of cell differentiation from a commitment step to a phenotype-expression step. Journal of Cellular Physiology, 1988, 134, 261-268.	4.1	13
13	Late-onset epilepsy in children with acute febrile encephalopathy with prolonged convulsions: A clinical and encephalographic study. Brain and Development, 2013, 35, 531-539.	1.1	13
14	Persistent verbal and behavioral deficits after resection of the left supplementary motor area in epilepsy surgery. Brain and Development, 2014, 36, 74-79.	1.1	12
15	CD4+ T Cells Require Adhesion via LFA-1/ICAM-1 to Induce Target Apoptosis in TNF-independent Pathway. Cellular Immunology, 1994, 156, 135-145.	3.0	8
16	Use of high b value diffusion-weighted magnetic resonance imaging in acute encephalopathy/encephalitis during childhood. Brain and Development, 2018, 40, 116-125.	1.1	8
17	High expression of c-kit in K562YO cells due to the prolonged half-life of its mRNA: the effects of modification with serine/threonine kinase signals. Blood, 1995, 85, 1496-1503.	1.4	7
18	Pharmacoresistant epileptic eyelid twitching in a child with a mutation in SYNGAP1. Epileptic Disorders, 2017, 19, 339-344.	1.3	7

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19	Efficacy of longâ€term adrenocorticotropic hormone therapy for West syndrome: A retrospective multicenter case series. Epilepsia Open, 2021, 6, 402-412.	2.4	7
20	A Girl with Idiopathic Epilepsy Showing Forced Normalization after Levetiracetam Administration. Journal of Nippon Medical School, 2015, 82, 250-253.	0.9	6
21	Bilateral cerebellar cysts and cerebral white matter lesions with cortical dysgenesis: Expanding the phenotype of LAMB1 gene mutations. Clinical Genetics, 2018, 94, 391-392.	2.0	6
22	Successful treatment of migrating partial seizures in Wolfâ€“Hirschhorn syndrome with bromide. Brain and Development, 2016, 38, 658-662.	1.1	5
23	Comparison of Causative Variant Prioritization Tools Using Next-generation Sequencing Data in Japanese Patients with Mendelian Disorders. Yonago Acta Medica, 2019, 62, 244-252.	0.7	5
24	Evaluation of the Effects of Exercise and a Mild Hypocaloric Diet on Cardiovascular Risk Factors in Obese Subjects. Journal of UOEH, 2001, 23, 1-12.	0.6	5
25	Surface electromyogram and muscle ultrasonography for detection of muscle fasciculations in pediatric peripheral neuropathy. Brain and Development, 2017, 39, 617-620.	1.1	4
26	Three Japanese patients with 3p13 microdeletions involving FOXP1. Brain and Development, 2019, 41, 257-262.	1.1	4
27	Duchenne muscular dystrophyâ€“like phenotype in an LGMD2I patient with novel FKRP gene variants. Human Genome Variation, 2020, 7, 12.	0.7	4
28	Clinical Characteristics of Fragile X Syndrome Patients in Japan. Yonago Acta Medica, 2021, 64, 30-33.	0.7	4
29	Progressive cerebral atrophies in three children with COL4A1 mutations. Brain and Development, 2021, 43, 1033-1038.	1.1	4
30	Significance of Glutathione-Mediated Scavenger Potency in the Development of Seizure Susceptibility in the EL Mouse Brain. Journal of Pediatric Epilepsy, 2015, 04, 067-071.	0.2	2
31	Effect of Intrathecal Baclofen on Delayed-Onset Paroxysmal Dystonia due to Compression Injury Resulting From Congenital and Progressive Spinal Bone Deformities in Chondrodysplasia Punctata. Pediatric Neurology, 2016, 56, 80-85.e2.	2.1	2
32	Recurrent Erythema Nodosum in a Child with a <i>SHOC2</i> Gene Mutation. Yonago Acta Medica, 2019, 62, 159-162.	0.7	2
33	Evaluation of the clinical performance of noninvasive prenatal testing at a Japanese laboratory. Journal of Obstetrics and Gynaecology Research, 2021, 47, 3437-3446.	1.3	2
34	FKRP mutations cause congenital muscular dystrophy 1C and limb-girdle muscular dystrophy 2I in Asian patients. Journal of Clinical Neuroscience, 2021, 92, 215-221.	1.5	2
35	Clinical course of a Japanese patient with developmental delay linked to a small 6q16.1 deletion. Human Genome Variation, 2022, 9, 14.	0.7	2
36	Gait disturbance in a patient with de novo 1.0-kb SOX2 microdeletion. Brain and Development, 2022, 44, 68-72.	1.1	1

#	Article	IF	Citations
37	Clinical course of epilepsy and white matter abnormality linked to a novel DYRK1A variant. Human Genome Variation, 2021, 8, 26.	0.7	1
38	THE EFFECT OF ACETAZOLEAMIDE UPON THE CARDIOPULMONARY DYNAMICS AND ITS CLINICAL INDICATION. The Journal of the Japanese Society of Internal Medicine, 1959, 48, 288-297.	0.0	0
39	Near-infrared Spectroscopy-based Assessment of Ictal Cerebral Hemodynamics of a Frontal Area in Startle Epilepsy. Journal of the Japan Epilepsy Society, 2012, 29, 482-489.	0.2	0