Toshiki Takenouchi

List of Publications by Year
in descending order

70	741	687363 13	642732 23
papers	citations	h-index	g-index

72	72	72	1469
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Ketogenic Diet for KARS-Related Mitochondrial Dysfunction and Progressive Leukodystrophy. Neuropediatrics, 2022, 53, 065-068.	0.6	3
2	Vanishing basal ganglia in <scp>ATP1A3</scp>â€related polymicrogyria. American Journal of Medical Genetics, Part A, 2022, 188, 665-667.	1.2	0
3	Pro108Ser mutation of SARS-CoV-2 3CLpro reduces the enzyme activity and ameliorates the clinical severity of COVID-19. Scientific Reports, 2022, 12, 1299.	3.3	15
4	The p.<scp>Thr395Met</scp> missense variant of <scp>NFIA</scp> found in a patient with intellectual disability is a defective variant. American Journal of Medical Genetics, Part A, 2022, 188, 1184-1192.	1.2	5
5	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. Journal of Pediatrics, 2022, 244, 38-48.e1.	1.8	8
6	Neuropsychiatric systemic lupus erythematosus in a girl with neurocutaneous melanosis caused by a somatic mutation in NRAS. Rheumatology, 2022, , .	1.9	0
7	Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome. European Journal of Medical Genetics, 2022, , 104512.	1.3	0
8	Mutation of PTPN11 (Encoding SHP-2) Promotes MEK Activation and Malignant Progression in Neurofibromin-Deficient Cells in a Manner Sensitive to BRAP Mutation. Cancers, 2022, 14, 2377.	3.7	1
9	Precocious puberty in a case of Simpsonâ€“Golabiâ€“Behmel syndrome with a de novo 240-kb deletion including GPC3. Human Genome Variation, 2022, 9, .	0.7	0
10	Role of chimeric transcript formation in the pathogenesis of birth defects. Congenital Anomalies (discontinued), 2021, 61, 76-81.	0.6	11
11	Severe course with lethal hepatocellular injury and skeletal muscular dysgenesis in a neonate with infantile liver failure syndrome type 1 caused by novel <scp>LARS1</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 866-870.	1.2	4
12	Clinical utility of SARS-CoV-2 whole genome sequencing in deciphering source of infection. Journal of Hospital Infection, 2021, 107, 40-44.	2.9	19
13	Complex hereditary spastic paraplegia associated with episodic visual loss caused by ACO2 variants. Human Genome Variation, 2021, 8, 4.	0.7	3
14	Forkâ€shaped mandibular incisors as a novel phenotype of LRP5 â€associated disorder. American Journal of Medical Genetics, Part A, 2021, 185, 1544-1549.	1.2	1
15	Establishing intellectual disability as the key feature of patients with biallelic <scp>RNPC3</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 1836-1840.	1.2	3
16	Recurrent <scp>NFIA K125E</scp> substitution represents a lossâ€ofâ€function allele: Sensitive in vitro and in vivo assays forÂnontruncating alleles. American Journal of Medical Genetics, Part A, 2021, 185, 2084-2093.	1.2	9
17	A Japanese adult and two girls with NEDMIAL caused by de novo missense variants in DHX30. Human Genome Variation, 2021, 8, 24.	0.7	1
18	Decisive evidence of direct effect of ACTH treatment in West syndrome: A case report. Seizure: the Journal of the British Epilepsy Association, 2021, 91, 49-51.	2.0	1

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19	Identification of B.1.346 Lineage of SARS-CoV-2 in Japan: Genomic Evidence of Re-entry of Clade 20C. Keio Journal of Medicine, 2021, 70, 44-50.	1.1	4
20	Coloboma may be a shared feature in a spectrum of disorders caused by mutations in the <scp>WDR37â€PACS1â€PACS2</scp> axis. American Journal of Medical Genetics, Part A, 2021, 185, 884-888.	1.2	11
21	Biallelic loss of OTUD7A causes severe muscular hypotonia, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1182-1186.	1.2	14
22	Progressive cerebral and coronary aneurysms in the original two patients with Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 999-1003.	1.2	2
23	Hereditary spastic paraplegia masqueraded by congenital melanocytic nevus syndrome: Dual pathogenesis of germline non-mosaicism and somatic mosaicism. European Journal of Medical Genetics, 2020, 63, 103803.	1.3	2
24	Learning disability and myoclonic epilepsy associated with apparently synonymous but spliceâ€disrupting <scp>JMJD1C</scp> variant that led to 21â€‰bp deletion of the transcript. American Journal of Medical Genetics, Part A, 2020, 182, 3064-3067.	1.2	0
25	Hypercoagulopathy Associated With Uniparental Disomy of Chromosome 2. Journal of Pediatric Hematology/Oncology, 2020, 42, 370-371.	0.6	0
26	Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients. American Journal of Medical Genetics, Part A, 2020, 182, 2529-2532.	1.2	12
27	Protein elongation variant of <scp>PUF60</scp>: Milder phenotypic end of the Verheij syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2709-2714.	1.2	7
28	Consecutive medical exome analysis at a tertiary center: Diagnostic and healthâ€economic outcomes. American Journal of Medical Genetics, Part A, 2020, 182, 1601-1607.	1.2	10
29	Shortfall of exome analysis for diagnosis of <scp>Shwachmanâ€Diamond</scp> syndrome: Mismapping due to the pseudogene <scp>SBDSP1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 1631-1636.	1.2	11
30	Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Auâ€Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities. Molecular Genetics & Genomic Medicine, 2020, 8, e1364.	1.2	6
31	Novel ARX mutation identified in infantile spasm syndrome patient. Human Genome Variation, 2020, 7, 9.	0.7	2
32	Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. Journal of Pediatrics, 2020, 221, 251-254.	1.8	11
33	A paradoxical thrombogenic mutation in factor II at the target site of arthropod bleeding toxin. European Journal of Medical Genetics, 2019, 62, 93-95.	1.3	5
34	IFT172 as the 19th gene causative of oralâ€facialâ€digital syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2510-2513.	1.2	9
35	A case of autism spectrum disorder with cleft lip and palate carrying a mutation in exon 8 of AUTS2. Clinical Case Reports (discontinued), 2019, 7, 2059-2063.	0.5	5
36	Kosaki overgrowth syndrome: A newly identified entity caused by pathogenic variants in plateletâ€derived growth factor receptorâ€beta. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 650-657.	1.6	8

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37	De novo NSF mutations cause early infantile epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 2334-2339.	3.7	10
38	CNOT2 haploinsufficiency causes a neurodevelopmental disorder with characteristic facial features. American Journal of Medical Genetics, Part A, 2019, 179, 2506-2509.	1.2	9
39	Severe Noonan syndrome phenotype associated with a germline Q71R MRAS variant: a recurrent substitution in RAS homologs in various cancers. American Journal of Medical Genetics, Part A, 2019, 179, 1628-1630.	1.2	13
40	A novel missense PTEN mutation identified in a patient with macrocephaly and developmental delay. Human Genome Variation, 2019, 6, 25.	0.7	3
41	Noninvasive diagnosis of TRIT1â€related mitochondrial disorder by measuring i⁶A37 and ms²i⁶A37 modifications in tRNAs from blood and urine samples. American Journal of Medical Genetics, Part A, 2019, 179, 1609-1614.	1.2	6
42	Pathogenetic basis of Takenouchi-Kosaki syndrome: Electron microscopy study using platelets in patients and functional studies in a Caenorhabditis elegans model. Scientific Reports, 2019, 9, 4418.	3.3	16
43	SATB2â€associated syndrome in patients from Japan: Linguistic profiles. American Journal of Medical Genetics, Part A, 2019, 179, 896-899.	1.2	11
44	CNOT2 as the critical gene for phenotypes of 12q15 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 659-662.	1.2	8
45	Cover Image, Volume 179A, Number 6, June 2019. American Journal of Medical Genetics, Part A, 2019, 179, i.	1.2	0
46	Further evidence of a causal association between AGO1, a critical regulator of microRNA formation, and intellectual disability/autism spectrum disorder. European Journal of Medical Genetics, 2019, 62, 103537.	1.3	16
47	Schuursâ€Hoeijmakers syndrome in two patients from Japan. American Journal of Medical Genetics, Part A, 2019, 179, 341-343.	1.2	16
48	Tranilast inhibits the expression of genes related to epithelial-mesenchymal transition and angiogenesis in neurofibromin-deficient cells. Scientific Reports, 2018, 8, 6069.	3.3	16
49	Growth pattern of Rahman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 712-714.	1.2	14
50	Respiratory arrest at the onset of idiopathic childhood occipital epilepsy of Gastaut. Brain and Development, 2018, 40, 74-76.	1.1	2
51	Biallelic mutations in NALCN: Expanding the genotypic and phenotypic spectra of IHPRF1. American Journal of Medical Genetics, Part A, 2018, 176, 431-437.	1.2	16
52	Ablepharon and craniosynostosis in a patient with a localized TWIST1 basic domain substitution. American Journal of Medical Genetics, Part A, 2018, 176, 2777-2780.	1.2	7
53	Haploinsufficiency of NCOR1 associated with autism spectrum disorder, scoliosis, and abnormal palatogenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2466-2469.	1.2	5
54	Expanding Phenotype of Nephronophthisis-Related Ciliopathy: an Elderly Patient with Homozygous RPGRIP1L Mutation. Nephron, 2018, 140, 74-78.	1.8	2

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55	Cover Image, Volume 173A, Number 5, May 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
56	Diagnostic use of computational retrotransposon detection: Successful definition of pathogenetic mechanism in a ciliopathy phenotype. , 2017, 173, 1353-1357.		9
57	Expansion of the phenotype of Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2422-2427.	1.2	27
58	Further evidence of a mutation in CDC42 as a cause of a recognizable syndromic form of thrombocytopenia. American Journal of Medical Genetics, Part A, 2016, 170, 852-855.	1.2	49
59	Hirschsprung disease as a yet undescribed phenotype in a patient with ARID1B mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3249-3252.	1.2	20
60	Cover Image, Volume 170A, Number 12, December 2016. , 2016, 170, i-i.		0
61	Childhood SjÃ¶gren syndrome presenting as acute brainstem encephalitis. Brain and Development, 2016, 38, 158-162.	1.1	19
62	Macrothrombocytopenia and developmental delay with a de novo CDC42 mutation: Yet another locus for thrombocytopenia and developmental delay. American Journal of Medical Genetics, Part A, 2015, 167, 2822-2825.	1.2	80
63	Mosaic overgrowth with fibroadipose hyperplasia due to AKT1 mutation. American Journal of Medical Genetics, Part A, 2015, 167, 907-909.	1.2	1
64	Novel Overgrowth Syndrome Phenotype Due to Recurrent DeÂNovoÂPDGFRBÂMutation. Journal of Pediatrics, 2015, 166, 483-486.	1.8	89
65	Cilostazol strengthens the endothelial barrier of postcapillary venules from the rat mesentery in situ. Phlebology, 2014, 29, 594-599.	1.2	5
66	The Use of Next-Generation Sequencing in Molecular Diagnosis of Neurofibromatosis Type 1: A Validation Study. Genetic Testing and Molecular Biomarkers, 2014, 18, 722-735.	0.7	33
67	Therapeutic hypothermia achieves neuroprotection via a decrease in acetylcholine with a concurrent increase in carnitine in the neonatal hypoxiaâ€ischemia (877.2). FASEB Journal, 2014, 28, 877.2.	0.5	0
68	Hydrocephalus with Hirschsprung disease: Severe end of Xâ€linked hydrocephalus spectrum. American Journal of Medical Genetics, Part A, 2012, 158A, 812-815.	1.2	14
69	Congenital corneal staphyloma as a complication of Kabuki syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2000-2002.	1.2	14
70	Posterior cerebral artery dissection on a serial magnetic resonance angiography. Brain and Development, 2012, 34, 396-399.	1.1	1