Tomohiro Chiyonobu

List of Publications by Year
in descending order

34	514	840776 11	713466 21
papers	citations	h-index	g-index

35	35	35	1332
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Pleomorphic rhabdomyosarcoma in a young adult harboring a novel germline MSH2 variant. Human Genome Variation, 2022, 9, 8.	0.7	3
2	Thyroid hypogenesis is associated with a novel AKT3 germline variant that causes megalencephaly and cortical malformation. Human Genome Variation, 2022, 9, .	0.7	2
3	Clinical phenotypes of spinal muscular atrophy patients with hybrid SMN gene. Brain and Development, 2021, 43, 294-302.	1.1	18
4	Complex hereditary spastic paraplegia associated with episodic visual loss caused by ACO2 variants. Human Genome Variation, 2021, 8, 4.	0.7	3
5	Impaired neuronal activity and differential gene expression in STXBP1 encephalopathy patient iPSC-derived GABAergic neurons. Human Molecular Genetics, 2021, 30, 1337-1348.	2.9	11
6	Phenotypes of SMA patients retaining SMN1 with intragenic mutation. Brain and Development, 2021, 43, 745-758.	1.1	12
7	TUBB3 E410K Syndrome With Childhood-Onset Nonalcoholic Steatohepatitis. Journal of Clinical Endocrinology and Metabolism, 2021, , .	3.6	0
8	Lowâ€grade IVH in preterm infants causes cerebellar damage, motor, and cognitive impairment. Pediatrics International, 2021, 63, 1327-1333.	0.5	2
9	Early infantile-onset Leigh syndrome complicated with infantile spasms associated with the m.9185â€Tâ€>â€C variant in the MT-ATP6 gene: Expanding the clinical spectrum. Brain and Development, 2020, 42, 69-72.	1.1	8
10	Childhood-onset multifocal motor neuropathy with IgM antibodies to GM2 and GalNac-GD1a. Brain and Development, 2020, 42, 88-92.	1.1	3
11	Intrathecal nusinersen treatment after ventriculo-peritoneal shunt placement: A case report focusing on the neurofilament light chain in cerebrospinal fluid. Brain and Development, 2020, 42, 311-314.	1.1	12
12	Novel genetic link between the ATP-binding cassette subfamily A gene and hippo gene in Drosophila. Experimental Cell Research, 2020, 386, 111733.	2.6	5
13	Application of induced pluripotent stem cells in epilepsy. Molecular and Cellular Neurosciences, 2020, 108, 103535.	2.2	13
14	A novel Drosophila model for neurodevelopmental disorders associated with Shwachmanâ€“Diamond syndrome. Neuroscience Letters, 2020, 739, 135449.	2.1	8
15	Investigating Developmental and Epileptic Encephalopathy Using Drosophila melanogaster. International Journal of Molecular Sciences, 2020, 21, 6442.	4.1	14
16	Gender differences in occurrence of behavioral and emotional problems at the lower grades of elementary school: Association with developmental and behavioral characteristics at 5â€years. Brain and Development, 2019, 41, 760-768.	1.1	3
17	Plasma neurofilament light chain: A potential prognostic biomarker of dementia in adult Down syndrome patients. PLoS ONE, 2019, 14, e0211575.	2.5	24
18	Lumbosacral polyradiculopathy after intrathecal chemotherapy in pediatric acute lymphoblastic leukemia. International Journal of Hematology, 2018, 107, 499-501.	1.6	3

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19	A 5-Year Follow-Up of Triple-Seronegative Myasthenia Gravis Successfully Treated with Tacrolimus Therapy. Neuropediatrics, 2018, 49, 200-203.	0.6	3
20	Identification of novel <scp>BCL11A</scp> variants in patients with epileptic encephalopathy: Expanding the phenotypic spectrum. Clinical Genetics, 2018, 93, 368-373.	2.0	23
21	Detection of copy number variations in epilepsy using exome data. Clinical Genetics, 2018, 93, 577-587.	2.0	35
22	Cerebellar peduncle injury predicts motor impairments in preterm infants: A quantitative tractography study at term-equivalent age. Brain and Development, 2018, 40, 743-752.	1.1	4
23	Luteolin attenuates interleukin-6-mediated astrogliosis in human iPSC-derived neural aggregates: A candidate preventive substance for maternal immune activation-induced abnormalities. Neuroscience Letters, 2017, 653, 296-301.	2.1	21
24	Longitudinal change in white matter in preterm infants without magnetic resonance imaging abnormalities: Assessment of serial diffusion tensor imaging and their relationship to neurodevelopmental outcomes. Brain and Development, 2017, 39, 40-47.	1.1	10
25	Increased levels of plasma total tau in adult Down syndrome. PLoS ONE, 2017, 12, e0188802.	2.5	36
26	Mislocalization of syntaxinâ€1 and impaired neurite growth observed in a human <scp>iPSC</scp> model for <scp>STXBP</scp>1â€related epileptic encephalopathy. Epilepsia, 2016, 57, e81-6.	5.1	37
27	A Video Report of Brainâ€“Lungâ€“Thyroid Syndrome in a Japanese Female With a Novel Frameshift Mutation of the NKX2-1 Gene. Child Neurology Open, 2016, 3, 2329048X1666501.	1.1	2
28	Establishment of isogenic iPSCs from an individual with SCN1A mutation mosaicism as a model for investigating neurocognitive impairment in Dravet syndrome. Journal of Human Genetics, 2016, 61, 565-569.	2.3	20
29	Microarray analysis of 50 patients reveals the critical chromosomal regions responsible for 1p36 deletion syndrome-related complications. Brain and Development, 2015, 37, 515-526.	1.1	43
30	Serial investigation of PTPN11 mutation in nonhematopoietic tissues in a patient with juvenile myelomonocytic leukemia who was treated with unrelated cord blood transplantation. International Journal of Hematology, 2015, 102, 719-722.	1.6	2
31	Glycosylphosphatidylinositol (GPI) anchor deficiency caused by mutations inPIGWis associated with West syndrome and hyperphosphatasia with mental retardation syndrome. Journal of Medical Genetics, 2014, 51, 203-207.	3.2	93
32	Altered somatosensory barrel cortex refinement in the developing brain of Mecp2-null mice. Brain Research, 2013, 1537, 319-326.	2.2	7
33	Fatal varicella with multiâ€organ failure associated with lowâ€dose adrenocorticotropic hormone therapy. Pediatrics International, 2012, 54, 305-306.	0.5	3
34	Postnatal changes in serotonergic innervation to the hippocampus of methyl-CpG-binding protein 2-null mice. Neuroscience, 2010, 165, 1254-1260.	2.3	22