

Ryuta Tanaka

List of Publications by Year in descending order

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Version: 2024-02-01

17
papers

184
citations

1163117

8
h-index

1199594

12
g-index

18
all docs

18
docs citations

18
times ranked

390
citing authors

#	ARTICLE	IF	CITATIONS
1	Acute encephalopathy in children with tuberous sclerosis complex. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 5.	2.7	4
2	ATP6VOA1 encoding the α 1-subunit of the V0 domain of vacuolar H ⁺ -ATPases is essential for brain development in humans and mice. <i>Nature Communications</i> , 2021, 12, 2107.	12.8	30
3	Nusinersen improved respiratory function in spinal muscular atrophy type 2. <i>Pediatrics International</i> , 2021, 63, 973-974.	0.5	2
4	Variants in <i>KIF2A</i> cause broad clinical presentation; the computational structural analysis of a novel variant in a patient with a cortical dysplasia, complex, with other brain malformations 3. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1113-1119.	1.2	5
5	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 71, 1-5.	2.0	16
6	Schuurs-Hoeijmakers syndrome in two patients from Japan. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 341-343.	1.2	16
7	Hospital-based care utilization of children with medical complexity in Japan. <i>Pediatrics International</i> , 2018, 60, 626-633.	0.5	6
8	Long-term accumulation of diphenylarsinic acid in the central nervous system of cynomolgus monkeys. <i>Archives of Toxicology</i> , 2017, 91, 2799-2812.	4.2	12
9	Identification of novel <i>SNORD118</i> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. <i>Clinical Genetics</i> , 2017, 92, 180-187.	2.0	28
10	Cyclic vomiting syndrome after acute autonomic and sensory neuropathy. <i>Pediatrics International</i> , 2017, 59, 503-505.	0.5	3
11	A novel <i>BBS10</i> mutation identified in a patient with Bardet-Biedl syndrome with a violent emotional outbreak. <i>Human Genome Variation</i> , 2017, 4, 17033.	0.7	4
12	Preaxial polydactyly in an individual with Wiedemann-Steiner syndrome caused by a novel nonsense mutation in <i>KMT2A</i> . , 2017, 173, 2821-2825.		11
13	Monozygotic twins with de novo <i>ZIC2</i> gene mutations discordant for the type of holoprosencephaly. <i>Neurology</i> , 2016, 86, 1456-1458.	1.1	5
14	Novel compound heterozygous <i>LIAS</i> mutations cause glycine encephalopathy. <i>Journal of Human Genetics</i> , 2015, 60, 631-635.	2.3	17
15	Neurochemistry in shiverer mouse depicted on MR spectroscopy. <i>Journal of Magnetic Resonance Imaging</i> , 2014, 39, 1550-1557.	3.4	10
16	Abnormal brain MRI signal in 18q-syndrome not due to dysmyelination. <i>Brain and Development</i> , 2012, 34, 234-237.	1.1	15
17	Ethosuximide completely suppressed epileptic negative myoclonus in childhood localization-related epilepsy. <i>Epilepsy and Seizure</i> , 2010, 3, 1-9.	0.2	0