Ryuta Tanaka

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3179993/publications.pdf

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17	184	1163117	1199594
papers	citations	8	12
		h-index	g-index

18	18	18	390
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. Nature Communications, 2021, 12, 2107.	12.8	30
2	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	2.0	28
3	Novel compound heterozygous LIAS mutations cause glycine encephalopathy. Journal of Human Genetics, 2015, 60, 631-635.	2.3	17
4	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 1-5.	2.0	16
5	Schuursâ€Hoeijmakers syndrome in two patients from Japan. American Journal of Medical Genetics, Part A, 2019, 179, 341-343.	1.2	16
6	Abnormal brain MRI signal in 18q-syndrome not due to dysmyelination. Brain and Development, 2012, 34, 234-237.	1.1	15
7	Long-term accumulation of diphenylarsinic acid in the central nervous system of cynomolgus monkeys. Archives of Toxicology, 2017, 91, 2799-2812.	4.2	12
8	Preaxial polydactyly in an individual with Wiedemann-Steiner syndrome caused by a novel nonsense mutation in KMT2A. , 2017, 173, 2821-2825.		11
9	Neurochemistry in shiverer mouse depicted on MR spectroscopy. Journal of Magnetic Resonance Imaging, 2014, 39, 1550-1557.	3.4	10
10	Hospitalâ€based care utilization of children with medical complexity inÂJapan. Pediatrics International, 2018, 60, 626-633.	0.5	6
11	Monozygotic twins with de novo ZIC2 gene mutations discordant for the type of holoprosencephaly. Neurology, 2016, 86, 1456-1458.	1.1	5
12	Variants in <scp>KIF2A</scp> 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. American Journal of Medical Genetics, Part A, 2021, 185, 1113-1119.	1.2	5
13	A novel BBS10 mutation identified in a patient with Bardetâ€“Biedl syndrome with a violent emotional outbreak. Human Genome Variation, 2017, 4, 17033.	0.7	4
14	Acute encephalopathy in children with tuberous sclerosis complex. Orphanet Journal of Rare Diseases, 2021, 16, 5.	2.7	4
15	Cyclic vomiting syndrome after acute autonomic and sensory neuropathy. Pediatrics International, 2017, 59, 503-505.	0.5	3
16	Nusinersen improved respiratory function in spinal muscular atrophy type 2. Pediatrics International, 2021, 63, 973-974.	0.5	2
17	Ethosuximide completely suppressed epileptic negative myoclonus in childhood localization-related epilepsy. Epilepsy and Seizure, 2010, 3, 1-9.	0.2	0