Brendan C Lanpher

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6653955/publications.pdf

Version: 2024-02-01

17	507	933447	940533
papers	citations	10	16
		h-index	g-index

17	17	17	1205
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	3.2	146
2	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
3	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
4	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	6.2	39
5	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Reports, 2017, 40, 63-69.	1.5	27
6	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
7	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
8	The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. Molecular Genetics and Metabolism Reports, 2017, 13, 46-51.	1.1	17
9	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
10	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
11	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333.		11
12	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	1.2	10
13	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931.	2.3	7
14	Diagnosis of Attenuated Mucopolysaccharidosis VI: Clinical, Biochemical, and Genetic Pitfalls. Pediatrics, 2018, 142, e20180658.	2.1	6
15	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219.	2.1	6
16	A novel missense variant and multiexon deletion causing a delayed presentation of xeroderma pigmentosum, group C. Journal of Physical Education and Sports Management, 2020, 6, a005165.	1.2	1
17	Bilateral subdural hematomas and retinal hemorrhages mimicking nonaccidental trauma in a patient with Dâ€2â€hydroxyglutaric aciduria. JIMD Reports, 2021, 58, 21-28.	1.5	1