Laura E Schultz-Rogers

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5852381/publications.pdf

Version: 2024-02-01

16	460	933447 10	996975 15
papers	citations	h-index	g-index

17	17	17	1181
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
2	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
3	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
4	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
5	Clinical and Biochemical Phenotypes in a Family With ENPP1 Mutations. Journal of Bone and Mineral Research, 2020, 35, 662-670.	2.8	33
6	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	2.5	27
7	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
8	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	4.1	22
9	Proteinâ€elongating mutations inMYH11are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. Human Mutation, 2020, 41, 973-982.	2.5	18
10	SPECC1L regulates palate development downstream of IRF6. Human Molecular Genetics, 2020, 29, 845-858.	2.9	18
11	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
12	Haploinsufficiency as a disease mechanism in GNB1â€associated neurodevelopmental disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1477.	1.2	12
13	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219.	2.1	6
14	Three rare disease diagnoses in one patient through exome sequencing. Journal of Physical Education and Sports Management, 2019, 5, a004390.	1.2	5
15	Novel biallelic variants in MSTO1 associated with mitochondrial myopathy. Journal of Physical Education and Sports Management, 2019, 5, a004309.	1.2	5
16	Spectrum of Hematological Malignancies in 130 Patients with Germline Predisposition Syndromes - Mayo Clinic Germline Predisposition Study. Blood, 2020, 136, 34-35.	1.4	0