Laura E Schultz-Rogers

List of Publications by Year
in descending order

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	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
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	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
4	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
5	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	4.1	22
6	Clinical and Biochemical Phenotypes in a Family With ENPP1 Mutations. Journal of Bone and Mineral Research, 2020, 35, 662-670.	2.8	33
7	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219.	2.1	6
8	Haploinsufficiency as a disease mechanism in GNB1â€associated neurodevelopmental disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1477.	1.2	12
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	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
12	Spectrum of Hematological Malignancies in 130 Patients with Germline Predisposition Syndromes - Mayo Clinic Germline Predisposition Study. Blood, 2020, 136, 34-35.	1.4	0
13	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
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	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	2.5	27
16	Novel biallelic variants in MSTO1 associated with mitochondrial myopathy. Journal of Physical Education and Sports Management, 2019, 5, a004309.	1.2	5