

Pilar Cacheiro

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5427886/publications.pdf>

Version: 2024-02-01

9
papers

709
citations

1163117
8
h-index

1474206
9
g-index

12
all docs

12
docs citations

12
times ranked

1061
citing authors

#	ARTICLE	IF	CITATIONS
1	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. <i>Nature Genetics</i> , 2021, 53, 1006-1021.	21.4	44
2	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care – Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	27.0	352
3	Human and mouse essentiality screens as a resource for disease gene discovery. <i>Nature Communications</i> , 2020, 11, 655.	12.8	64
4	New models for human disease from the International Mouse Phenotyping Consortium. <i>Mammalian Genome</i> , 2019, 30, 143-150.	2.2	57
5	The International Mouse Phenotyping Consortium (IMPC): a functional catalogue of the mammalian genome that informs conservation. <i>Conservation Genetics</i> , 2018, 19, 995-1005.	1.5	82
6	Synaptotagmin XI in Parkinson's disease: New evidence from an association study in Spain and Mexico. <i>Journal of the Neurological Sciences</i> , 2016, 362, 321-325.	0.6	17
7	A replication study of a GWAS finding in migraine does not identify association in a Spanish case-control sample. <i>Cephalalgia</i> , 2012, 32, 1076-1080.	3.9	11
8	Databases for neurogenetics: Introduction, overview, and challenges. <i>Human Mutation</i> , 2012, 33, 1311-1314.	2.5	3
9	“Costa da Morte” ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. <i>Brain</i> , 2012, 135, 1423-1435.	7.6	78