

Constance Smith-Hicks

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10650512/publications.pdf>

Version: 2024-02-01

10
papers

250
citations

1478505

6
h-index

1372567

10
g-index

10
all docs

10
docs citations

10
times ranked

607
citing authors

#	ARTICLE	IF	CITATIONS
1	Delineating the genotypic and phenotypic spectrum of <i>HECW2</i> -related neurodevelopmental disorders. <i>Journal of Medical Genetics</i> , 2022, 59, 669-677.	3.2	5
2	mTORC1 functional assay reveals <i>SZT2</i> loss-of-function variants and a founder in-frame deletion. <i>Brain</i> , 2022, 145, 1939-1948.	7.6	1
3	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2021, 23, 1912-1921.	2.4	5
4	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. <i>American Journal of Human Genetics</i> , 2021, 108, 2006-2016.	6.2	11
5	Heterozygous de novo variants in <i>CSNK1G1</i> are associated with syndromic developmental delay and autism spectrum disorder. <i>Clinical Genetics</i> , 2020, 98, 571-576.	2.0	10
6	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 640-657.	6.2	31
7	Rett Syndrome and CDKL5 Deficiency Disorder: From Bench to Clinic. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5098.	4.1	30
8	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. <i>American Journal of Human Genetics</i> , 2019, 104, 203-212.	6.2	44
9	Monogenic disorders that mimic the phenotype of Rett syndrome. <i>Neurogenetics</i> , 2018, 19, 41-47.	1.4	41
10	SRF binding to SRE 6.9 in the Arc promoter is essential for LTD in cultured Purkinje cells. <i>Nature Neuroscience</i> , 2010, 13, 1082-1089.	14.8	72