

Miranda Splitt

List of Publications by Year in descending order

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8
papers

706
citations

1307594
7
h-index

1588992
8
g-index

8
all docs

8
docs citations

8
times ranked

2184
citing authors

#	ARTICLE	IF	CITATIONS
1	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	27.8	343
2	Quantifying the contribution of recessive coding variation to developmental disorders. <i>Science</i> , 2018, 362, 1161-1164.	12.6	158
3	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	2.8	108
4	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. <i>Nature Communications</i> , 2021, 12, 833.	12.8	41
5	De novo variants in CNOT3 cause a variable neurodevelopmental disorder. <i>European Journal of Human Genetics</i> , 2019, 27, 1677-1682.	2.8	19
6	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. <i>European Journal of Human Genetics</i> , 2018, 26, 1611-1622.	2.8	18
7	Phenotype of CNTNAP1: a study of patients demonstrating a specific severe congenital hypomyelinating neuropathy with survival beyond infancy. <i>European Journal of Human Genetics</i> , 2018, 26, 796-807.	2.8	13
8	SET de novo frameshift variants associated with developmental delay and intellectual disabilities. <i>European Journal of Human Genetics</i> , 2018, 26, 1306-1311.	2.8	6