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