Benjamin CognÃ©

List of Publications by Year
in descending order

11 papers	176 citations	1307594 7 h-index	1281871 11 g-index
13 all docs	13 docs citations	13 times ranked	329 citing authors

#	Article	IF	Citations
1	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
2	A dominant vimentin variant causes a rare syndrome with premature aging. European Journal of Human Genetics, 2020, 28, 1218-1230.	2.8	23
3	SETD1B-associated neurodevelopmental disorder. Journal of Medical Genetics, 2021, 58, 196-204.	3.2	22
4	THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 587-600.	6.2	19
5	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
6	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
7	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. European Journal of Human Genetics, 2017, 25, 150-152.	2.8	13
8	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
9	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	12.8	6
10	Patients with KCNH1-related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome. Journal of Medical Genetics, 2022, 59, 505-510.	3.2	5
11	Lossâ€ofâ€function variants in ARHGEF9 are associated with an Xâ€linked intellectual disability dominant disorder. Human Mutation, 2021, 42, 498-505.	2.5	1