Lydie Burglen

List of Publications by Year
in descending order

12	495	1040056 9	1281871 11
papers	citations	h-index	g-index

13	13	13	1240
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recessive PRDM13 mutations cause fatal perinatal brainstem dysfunction with cerebellar hypoplasia and disrupt Purkinje cell differentiation. American Journal of Human Genetics, 2022, 109, 909-927.	6.2	10
2	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
3	Childhoodâ€onset progressive dystonia associated with pathogenic truncating variants in CHD8. Annals of Clinical and Translational Neurology, 2021, 8, 1986-1990.	3.7	5
4	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
5	MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087.	12.8	28
6	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
7	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	5.3	52
8	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
9	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
10	Validation of a clinical practice-based algorithm for the diagnosis of autosomal recessive cerebellar ataxias based on NGS identified cases. Journal of Neurology, 2016, 263, 1314-1322.	3.6	15
11	GRID2 mutations span from congenital to mild adult-onset cerebellar ataxia. Neurology, 2015, 84, 1751-1759.	1.1	70
12	Phenotypic variability in ARCA2 and identification of a core ataxic phenotype with slow progression. Orphanet Journal of Rare Diseases, 2013, 8, 173.	2.7	63