Amy Pizzino

List of Publications by Year
in descending order

19	796	687363 13	794594 19
papers	citations	h-index	g-index

19	19	19	1585
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Early-Onset Vascular Leukoencephalopathy Caused by Bi-Allelic NOTCH3 Variants. Neuropediatrics, 2022, 53, 115-121.	0.6	4
2	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
3	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
4	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. Pediatric Neurology, 2021, 121, 11-19.	2.1	2
5	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	3.7	26
6	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
7	Janus Kinase Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 986-989.	27.0	109
8	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2020, 182, 1906-1912.	1.2	22
9	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	2.1	16
10	Randomized Clinical Trial of <scp>Firstâ€Line</scp> Genome Sequencing in Pediatric White Matter Disorders. Annals of Neurology, 2020, 88, 264-273.	5.3	17
11	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	6.2	92
12	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	2.1	9
13	Mutations in SZT2 result in earlyâ€onset epileptic encephalopathy and leukoencephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 1443-1448.	1.2	15
14	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
15	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	1.1	42
16	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Lossâ€”Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	2.1	12
17	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	5.3	116
18	Consensus statement on preventive and symptomatic care of leukodystrophy patients. Molecular Genetics and Metabolism, 2015, 114, 516-526.	1.1	29

#	Article	IF	Citations
19	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170