

Amy Pizzino

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

796
citations

687363

13
h-index

794594

19
g-index

19
all docs

19
docs citations

19
times ranked

1585
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical spectrum of 4H leukodystrophy caused by <i>POLR3A</i> and <i>POLR3B</i> mutations. <i>Neurology</i> , 2014, 83, 1898-1905.	1.1	170
2	Whole exome sequencing in patients with white matter abnormalities. <i>Annals of Neurology</i> , 2016, 79, 1031-1037.	5.3	116
3	Janus Kinase Inhibition in the Aicardi-Goutières Syndrome. <i>New England Journal of Medicine</i> , 2020, 383, 986-989.	27.0	109
4	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. <i>American Journal of Human Genetics</i> , 2019, 104, 925-935.	6.2	92
5	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020, 6, .	10.3	43
6	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 18-32.	1.1	42
7	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in <i>AIFM1</i> . <i>Neurogenetics</i> , 2017, 18, 185-194.	1.4	38
8	Consensus statement on preventive and symptomatic care of leukodystrophy patients. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 516-526.	1.1	29
9	Genome sequencing in persistently unsolved white matter disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 144-152.	3.7	26
10	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e660-e674.	3.6	26
11	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of next-generation sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1906-1912.	1.2	22
12	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. <i>Annals of Neurology</i> , 2020, 88, 264-273.	5.3	17
13	Phenotypic and Imaging Spectrum Associated With <i>WDR45</i> . <i>Pediatric Neurology</i> , 2020, 109, 56-62.	2.1	16
14	Mutations in <i>SZT2</i> result in early-onset epileptic encephalopathy and leukoencephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1443-1448.	1.2	15
15	<i>RMND1</i> -Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss—Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. <i>Pediatric Neurology</i> , 2017, 66, 59-62.	2.1	12
16	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. <i>Pediatric Neurology</i> , 2018, 84, 21-26.	2.1	9
17	Expanded phenotype of <i>AARS1</i> -related white matter disease. <i>Genetics in Medicine</i> , 2021, 23, 2352-2359.	2.4	8
18	Early-Onset Vascular Leukoencephalopathy Caused by Bi-Allelic <i>NOTCH3</i> Variants. <i>Neuropediatrics</i> , 2022, 53, 115-121.	0.6	4

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19	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. <i>Pediatric Neurology</i> , 2021, 121, 11-19.	2.1	2