

Grace M Hobson

List of Publications by Year in descending order

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Version: 2024-02-01

17
papers

606
citations

759233

12
h-index

996975

15
g-index

18
all docs

18
docs citations

18
times ranked

948
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. <i>Human Mutation</i> , 2020, 41, 150-168.	2.5	15
3	Inside Back Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, ii.	2.5	0
4	Distinct patterns of complex rearrangements and a mutational signature of microhomeology are frequently observed in PLP1 copy number gain structural variants. <i>Genome Medicine</i> , 2019, 11, 80.	8.2	24
5	Morpholino Antisense Oligomers as a Potential Therapeutic Option for the Correction of Alternative Splicing in PMD, SPG2, and HEMS. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 12, 420-432.	5.1	13
6	Modeling the Mutational and Phenotypic Landscapes of Pelizaeus-Merzbacher Disease with Human iPSC-Derived Oligodendrocytes. <i>American Journal of Human Genetics</i> , 2017, 100, 617-634.	6.2	52
7	Altered <i>PLP1</i> splicing causes hypomyelination of early myelinating structures. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 648-661.	3.7	27
8	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. <i>PLoS Genetics</i> , 2015, 11, e1005050.	3.5	57
9	<i>Plp1</i> gene duplication inhibits airway responsiveness and induces lung inflammation. <i>Pulmonary Pharmacology and Therapeutics</i> , 2015, 30, 22-31.	2.6	0
10	PMD patient mutations reveal a long-distance intronic interaction that regulates PLP1/DM20 alternative splicing. <i>Human Molecular Genetics</i> , 2014, 23, 5464-5478.	2.9	32
11	CJC2 promoter mutations causing Pelizaeus-Merzbacher-like disease. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 393-398.	1.1	19
12	Spastic paraplegia type 2 associated with axonal neuropathy and apparent <i>PLP1</i> position effect. <i>Annals of Neurology</i> , 2006, 59, 398-403.	5.3	83
13	Three or more copies of the proteolipid protein gene PLP1 cause severe Pelizaeus-Merzbacher disease. <i>Brain</i> , 2005, 128, 743-751.	7.6	91
14	Heterogeneous Duplications in Patients with Pelizaeus-Merzbacher Disease Suggest a Mechanism of Coupled Homologous and Nonhomologous Recombination. <i>American Journal of Human Genetics</i> , 2005, 77, 966-987.	6.2	93
15	A new polymorphism in the proteolipid protein (PLP1) gene and its use for carrier detection of PLP1 gene duplication in Pelizaeus-Merzbacher disease. <i>Human Mutation</i> , 2001, 17, 152-152.	2.5	28
16	Developmental Expression of Creatine Kinase Isoenzymes in Chicken Growth Cartilage. <i>Journal of Bone and Mineral Research</i> , 1999, 14, 747-756.	2.8	8
17	Glucocorticoids Decrease Interleukin-6 Levels and Induce Mineralization of Cultured Osteogenic Cells from Children with Fibrous Dysplasia. <i>Journal of Bone and Mineral Research</i> , 1999, 14, 1104-1114.	2.8	38