Martin Engvall

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9852493/publications.pdf

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20	476	933447 10	794594 19
papers	citations	h-index	g-index

22	22	22	939
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotypic spectrum and clinical course of single large-scale mitochondrial DNA deletion disease in the paediatric population: a multicentre study. Journal of Medical Genetics, 2023, 60, 65-73.	3.2	2
2	DNAJC30 defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. Brain, 2022, 145, 1624-1631.	7.6	21
3	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. Journal of Pediatrics, 2021, 228, 240-251.e2.	1.8	6
4	Case Report: A Novel Mutation in the Mitochondrial MT-ND5 Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). Frontiers in Neurology, 2021, 12, 652590.	2.4	4
5	Novel Mutation m.10372A>G in MT-ND3 Causing Sensorimotor Axonal Polyneuropathy. Neurology: Genetics, 2021, 7, e566.	1.9	3
6	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
7	Simplifying the clinical classification of polymerase gamma (POLG) disease based on age of onset; studies using a cohort of 155 cases. Journal of Inherited Metabolic Disease, 2020, 43, 726-736.	3.6	33
8	The impact of gender, puberty, and pregnancy in patients with POLG disease. Annals of Clinical and Translational Neurology, 2020, 7, 2019-2025.	3.7	7
9	Expanded Screening of One Million Swedish Babies with R4S and CLIR for Post-Analytical Evaluation of Data. International Journal of Neonatal Screening, 2020, 6, 42.	3.2	13
10	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	12.8	11
11	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6
12	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	6.2	41
13	POLG-Associated Ataxia Presenting as a Fragile X Tremor/Ataxia Phenocopy Syndrome. Cerebellum, 2016, 15, 632-635.	2.5	1
14	Epidemiology and Screening for Pompe Disease in Sweden. Journal of Neuromuscular Diseases, 2015, 2, S38-S38.	2.6	0
15	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	3.2	94
16	Epidemiology and Screening for Pompe Disease in Sweden. Journal of Neuromuscular Diseases, 2015, 2, S38.	2.6	0
17	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	2.8	54
18	Î²-Ureidopropionase deficiency: phenotype, genotype and protein structural consequences in 16 patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 73-74.	0.0	1

#	Article	IF	Citations
19	Substrate reduction therapy with miglustat for type 1 Gaucher disease: A retrospective analysis from a single institution. Upsala Journal of Medical Sciences, 2012, 117, 28-34.	0.9	26
20	ÃŸ-Ureidopropionase deficiency: Phenotype, genotype and protein structural consequences in 16 patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1096-1108.	3.8	27