

Martin Engvall

List of Publications by Year in descending order

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

Version: 2024-02-01

20
papers

476
citations

933447

10
h-index

794594

19
g-index

22
all docs

22
docs citations

22
times ranked

939
citing authors

#	ARTICLE	IF	CITATIONS
1	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
2	Rescue of primary ubiquinone deficiency due to a novel <i>COQ7</i> defect using 2,4-dihydroxybenzoic acid. <i>Journal of Medical Genetics</i> , 2015, 52, 779-783.	3.2	94
3	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. <i>BMC Genomics</i> , 2014, 15, 1090.	2.8	54
4	Allelic Expression Imbalance Promoting a Mutant <i>PEX6</i> Allele Causes Zellweger Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 965-976.	6.2	41
5	Simplifying the clinical classification of polymerase gamma (<i>POLG</i>) disease based on age of onset; studies using a cohort of 155 cases. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 726-736.	3.6	33
6	Î²-Ureidopropionase deficiency: Phenotype, genotype and protein structural consequences in 16 patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1096-1108.	3.8	27
7	Substrate reduction therapy with miglustat for type 1 Gaucher disease: A retrospective analysis from a single institution. <i>Upsala Journal of Medical Sciences</i> , 2012, 117, 28-34.	0.9	26
8	<i>DNAJC30</i> defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. <i>Brain</i> , 2022, 145, 1624-1631.	7.6	21
9	Expanded Screening of One Million Swedish Babies with R4S and CLIR for Post-Analytical Evaluation of Data. <i>International Journal of Neonatal Screening</i> , 2020, 6, 42.	3.2	13
10	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. <i>Nature Communications</i> , 2019, 10, 1396.	12.8	11
11	The impact of gender, puberty, and pregnancy in patients with <i>POLG</i> disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2019-2025.	3.7	7
12	Elevated cerebrospinal fluid protein in <i>POLG</i> -related epilepsy: Diagnostic and prognostic implications. <i>Epilepsia</i> , 2018, 59, 1595-1602.	5.1	6
13	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. <i>Journal of Pediatrics</i> , 2021, 228, 240-251.e2.	1.8	6
14	Case Report: A Novel Mutation in the Mitochondrial <i>MT-ND5</i> Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). <i>Frontiers in Neurology</i> , 2021, 12, 652590.	2.4	4
15	Novel Mutation m.10372A>G in <i>MT-ND3</i> Causing Sensorimotor Axonal Polyneuropathy. <i>Neurology: Genetics</i> , 2021, 7, e566.	1.9	3
16	Phenotypic spectrum and clinical course of single large-scale mitochondrial DNA deletion disease in the paediatric population: a multicentre study. <i>Journal of Medical Genetics</i> , 2023, 60, 65-73.	3.2	2
17	Î²-Ureidopropionase deficiency: phenotype, genotype and protein structural consequences in 16 patients. <i>Tijdschrift Voor Kindergeneeskunde</i> , 2013, 81, 73-74.	0.0	1
18	<i>POLG</i> -Associated Ataxia Presenting as a Fragile X Tremor/Ataxia Phenocopy Syndrome. <i>Cerebellum</i> , 2016, 15, 632-635.	2.5	1

#	ARTICLE	IF	CITATIONS
19	Epidemiology and Screening for Pompe Disease in Sweden. Journal of Neuromuscular Diseases, 2015, 2, S38-S38.	2.6	0
20	Epidemiology and Screening for Pompe Disease in Sweden. Journal of Neuromuscular Diseases, 2015, 2, S38.	2.6	0