

Magnhild Rasmussen

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

562
citations

759233

12
h-index

642732

23
g-index

26
all docs

26
docs citations

26
times ranked

1119
citing authors

#	ARTICLE	IF	CITATIONS
1	Novel mutations in the <i>HADHB</i> gene causing a mild phenotype of mitochondrial trifunctional protein (MTP) deficiency. <i>JIMD Reports</i> , 2022, 63, 193-198.	1.5	1
2	Priority setting at the clinical level: the case of nusinersen and the Norwegian national expert group. <i>BMC Medical Ethics</i> , 2021, 22, 54.	2.4	2
3	Simplifying the clinical classification of polymerase gamma (POLG) 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
4	Mental health and health related quality of life in mitochondrial POLG disease. <i>Mitochondrion</i> , 2020, 55, 95-99.	3.4	4
5	The impact of gender, puberty, and pregnancy in patients with POLG disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2019-2025.	3.7	7
6	Parent-child communication and timing of interventions are challenges in the Duchenne muscular dystrophy care. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2019, 108, 535-540.	1.5	4
7	Molecular and Clinical Characteristics of a National Cohort of Paediatric Duchenne Muscular Dystrophy Patients in Norway. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 349-359.	2.6	11
8	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 241-258.	2.6	32
9	Fever-related ataxia: a case report of CAPOS syndrome. <i>Cerebellum and Ataxias</i> , 2019, 6, 2.	1.9	4
10	The role of delayed bone age in the evaluation of stature and bone health in glucocorticoid treated patients with Duchenne muscular dystrophy. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2019, 2019, 4.	1.6	10
11	Elevated cerebrospinal fluid protein in <i>POLG</i> -related epilepsy: Diagnostic and prognostic implications. <i>Epilepsia</i> , 2018, 59, 1595-1602.	5.1	6
12	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017, 48, 166-184.	0.6	62
13	Juvenile myasthenia gravis in Norway: Clinical characteristics, treatment, and long-term outcome in a nationwide population-based cohort. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 707-714.	1.6	30
14	The presence of anaemia negatively influences survival in patients with POLG disease. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 861-866.	3.6	8
15	Juvenile myasthenia gravis in Norway: A nationwide epidemiological study. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 312-317.	1.6	26
16	A novel mutation in FBXL4 in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. <i>European Journal of Medical Genetics</i> , 2016, 59, 342-346.	1.3	16
17	Clinical and molecular characteristics in three families with biallelic mutations in IGHMBP2. <i>Neuromuscular Disorders</i> , 2016, 26, 570-575.	0.6	17
18	Loss-of-function mutations in <i>SCN4A</i> cause severe foetal hypokinesia or "classical" congenital myopathy. <i>Brain</i> , 2016, 139, 674-691.	7.6	100

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19	Succinateâ€CoA ligase deficiency due to mutations in <i>SUCLA2</i> and <i>SUCLG1</i>: phenotype and genotype correlations in 71 patients. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 243-252.	3.6	79
20	Neuromuscular disorders in children in South-Eastern Norway. <i>Journal of Pediatric Neurology</i> , 2015, 10, 095-100.	0.2	2
21	A de novo Mutation in the SCN4A Gene Causing Sodium Channel Myotonia. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 181-184.	2.6	4
22	Clinical and muscle biopsy findings in Norwegian paediatric patients with limb girdle muscular dystrophy 2I. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2014, 103, 553-558.	1.5	12
23	Prevalence, mutation spectrum and phenotypic variability in Norwegian patients with Limb Girdle Muscular Dystrophy 2I. <i>Neuromuscular Disorders</i> , 2011, 21, 41-46.	0.6	59
24	The activity of acyl CoA: retinol acyltransferase in the rat: variation with vitamin A status. <i>British Journal of Nutrition</i> , 1984, 51, 245.	2.3	24
25	Microsomal Esterification of Retinol in Human Liver. <i>Acta Medica Scandinavica</i> , 1984, 216, 403-407.	0.0	9