Magnhild Rasmussen

List of Publications by Year
in descending order

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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	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	7.6	100
2	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	3.6	79
3	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
4	Prevalence, mutation spectrum and phenotypic variability in Norwegian patients with Limb Girdle Muscular Dystrophy 2I. Neuromuscular Disorders, 2011, 21, 41-46.	0.6	59
5	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
6	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258.	2.6	32
7	Juvenile myasthenia gravis in Norway: Clinical characteristics, treatment, and long-term outcome in a nationwide population-based cohort. European Journal of Paediatric Neurology, 2017, 21, 707-714.	1.6	30
8	Juvenile myasthenia gravis in Norway: A nationwide epidemiological study. European Journal of Paediatric Neurology, 2017, 21, 312-317.	1.6	26
9	The activity of acyl CoA: retinol acyltransferase in the rat: variation with vitamin A status. British Journal of Nutrition, 1984, 51, 245.	2.3	24
10	Clinical and molecular characteristics in three families with biallelic mutations in IGHMBP2. Neuromuscular Disorders, 2016, 26, 570-575.	0.6	17
11	A novel mutation in FBXL4 in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. European Journal of Medical Genetics, 2016, 59, 342-346.	1.3	16
12	Clinical and muscle biopsy findings in Norwegian paediatric patients with limb girdle muscular dystrophy 2I. Acta Paediatrica, International Journal of Paediatrics, 2014, 103, 553-558.	1.5	12
13	Molecular and Clinical Characteristics of a National Cohort of Paediatric Duchenne Muscular Dystrophy Patients in Norway. Journal of Neuromuscular Diseases, 2019, 6, 349-359.	2.6	11
14	The role of delayed bone age in the evaluation of stature and bone health in glucocorticoid treated patients with Duchenne muscular dystrophy. International Journal of Pediatric Endocrinology (Springer), 2019, 2019, 4.	1.6	10
15	Microsomal Esterification of Retinol in Human Liver. Acta Medica Scandinavica, 1984, 216, 403-407.	0.0	9
16	The presence of anaemia negatively influences survival in patients with POLG disease. Journal of Inherited Metabolic Disease, 2017, 40, 861-866.	3.6	8
17	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
18	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6

#	Article	IF	Citations
19	A de novo Mutation in the SCN4A Gene Causing Sodium Channel Myotonia. Journal of Neuromuscular Diseases, 2015, 2, 181-184.	2.6	4
20	Parent-child communication and timing of interventions are challenges in the Duchenne muscular dystrophy care. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 535-540.	1.5	4
21	Fever-related ataxia: a case report of CAPOS syndrome. Cerebellum and Ataxias, 2019, 6, 2.	1.9	4
22	Mental health and health related quality of life in mitochondrial POLG disease. Mitochondrion, 2020, 55, 95-99.	3.4	4
23	Neuromuscular disorders in children in South-Eastern Norway. Journal of Pediatric Neurology, 2015, 10, 095-100.	0.2	2
24	Priority setting at the clinical level: the case of nusinersen and the Norwegian national expert group. BMC Medical Ethics, 2021, 22, 54.	2.4	2
25	Novel mutations in the <scp> HADHB </scp> gene causing a mild phenotype of mitochondrial trifunctional protein (<scp>MTP</scp>) deficiency. JIMD Reports, 2022, 63, 193-198.	1.5	1