Elsebet Ã˜stergaard

List of Publications by Year
in descending order

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39	1,171	471509	395702
papers	citations	17 h-index	33 g-index

39	39	39	2022
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	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	2.9	12
3	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
4	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
5	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159.	2.5	3
6	Excellent response to asfotase alfa treatment in an adolescent patient with hypophosphatasia. JIMD Reports, 2021, 59, 10-15.	1.5	2
7	A novel homozygous variant in C1QBP causes severe IUGR , edema, and cardiomyopathy in two fetuses. JIMD Reports, 2021, 59, 20-25.	1.5	3
8	NANS-CDG: Delineation of the Genetic, Biochemical, and Clinical Spectrum. Frontiers in Neurology, 2021, 12, 668640.	2.4	11
9	A novel homoplasmic mt-tRNAGlu m.14701C>T variant presenting with a partially reversible infantile respiratory chain deficiency. European Journal of Medical Genetics, 2021, 64, 104306.	1.3	0
10	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
11	SUCLA2 mutations cause global protein succinylation contributing to the pathomechanism of a hereditary mitochondrial disease. Nature Communications, 2020, 11, 5927.	12.8	35
12	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
13	Mitochondrial dysfunction induced by variation in the non-coding genome â€“ A proposed workflow to improve diagnostics. Mitochondrion, 2020, 53, 255-259.	3.4	1
14	Genotype and phenotype classification of 29 patients affected by Krabbe disease. JIMD Reports, 2019, 46, 35-45.	1.5	6
15	Phenotype-genotype correlations in Leigh syndrome: new insights from a multicentre study of 96 patients. Journal of Medical Genetics, 2018, 55, 21-27.	3.2	54
16	A recurrent de novo CUX2 missense variant associated with intellectual disability, seizures, and autism spectrum disorder. European Journal of Human Genetics, 2018, 26, 1388-1391.	2.8	17
17	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
18	A Faroese founder variant in TBCD causes early onset, progressive encephalopathy with a homogenous clinical course. European Journal of Human Genetics, 2018, 26, 1512-1520.	2.8	13

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19	Correspondence to: â€œHeterozygous mutation in the X chromosomal NDUFA1 gene in a girl with complex I deficiencyâ€•and â€œA novel NDUFA1 mutation leads to progressive mitochondrial complex I-specific neurodegenerative diseaseâ€• Molecular Genetics and Metabolism Reports, 2017, 13, 30.	1.1	1
20	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	3.6	27
21	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
22	Two transgenic mouse models for Î²-subunit components of succinate-CoA ligase yielding pleiotropic metabolic alterations. Biochemical Journal, 2016, 473, 3463-3485.	3.7	26
23	Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling. JIMD Reports, 2016, 33, 69-77.	1.5	17
24	Hearing impairment and renal failure associated with RMND1 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 142-147.	1.2	13
25	Hypomyelinating Leukodystrophy due to HSPD1 Mutations: A New Patient. Neuropediatrics, 2016, 47, 332-335.	0.6	15
26	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
27	An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. Human Molecular Genetics, 2015, 24, 4103-4113.	2.9	22
28	Neonatal mitochondrial hepatoencephalopathy caused by novel GFM1 mutations. Molecular Genetics and Metabolism Reports, 2015, 3, 5-10.	1.1	14
29	Mutations inCOA3cause isolated complex IV deficiency associated with neuropathy, exercise intolerance, obesity, and short stature. Journal of Medical Genetics, 2015, 52, 203-207.	3.2	49
30	CLPB Variants Associated with Autosomal-Recessive Mitochondrial Disorder with Cataract, Neutropenia, Epilepsy, and Methylglutaconic Aciduria. American Journal of Human Genetics, 2015, 96, 258-265.	6.2	58
31	Exercise Intolerance and Myoglobinuria Associated with a Novel Maternally Inherited MT-ND1 Mutation. JIMD Reports, 2015, 25, 65-70.	1.5	4
32	A mitochondrial tRNAMet mutation causing developmental delay, exercise intolerance and limb girdle phenotype with onset in early childhood. European Journal of Paediatric Neurology, 2015, 19, 69-71.	1.6	8
33	Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. American Journal of Medical Genetics, Part A, 2014, 164, 1143-1150.	1.2	27
34	A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet Journal of Rare Diseases, 2014, 9, 52.	2.7	182
35	Novel Mutations in the PC Gene in Patients with Type B Pyruvate Carboxylase Deficiency. JIMD Reports, 2012, 9, 1-5.	1.5	9
36	A novel RNASEH2B splice site mutation responsible for Aicardiâ€“Goutieres syndrome in the Faroe Islands. Acta Paediatrica, International Journal of Paediatrics, 2012, 101, e509-13.	1.5	5

#	Article	IF	Citations
37	Respiratory chain complex I deficiency due to NDUFA12 mutations as a new cause of Leigh syndrome. Journal of Medical Genetics, 2011, 48, 737-740.	3.2	54
38	A novel MERTK deletion is a common founder mutation in the Faroe Islands and is responsible for a high proportion of retinitis pigmentosa cases. Molecular Vision, 2011, 17, 1485-92.	1.1	55
39	Mitochondrial encephalomyopathy with elevated methylmalonic acid is caused by SUCLA2 mutations. Brain, 2007, 130, 853-861.	7.6	162