Pirjo Isohanni

List of Publications by Year
in descending order

39 papers	1,852 citations	361413 20 h-index	315739 38 g-index
39 all docs	39 docs citations	39 times ranked	3773 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	Cost-effectiveness of whole-exome sequencing in progressive neurological disorders of children. European Journal of Paediatric Neurology, 2022, 36, 30-36.	1.6	12
3	Duplication/triplication mosaicism of EBF3 and expansion of the EBF3 neurodevelopmental disorder phenotype. European Journal of Paediatric Neurology, 2022, 37, 1-7.	1.6	5
4	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159.	2.5	3
5	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 469-480.	3.6	34
6	Vegan diet in young children remodels metabolism and challenges the statuses of essential nutrients. EMBO Molecular Medicine, 2021, 13, e13492.	6.9	43
7	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
8	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
9	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
10	De novo SPTAN1mutation in axonal sensorimotor neuropathy and developmental disorder. Brain, 2020, 143, e104-e104.	7.6	8
11	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
12	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	1.9	6
13	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	3.7	11
14	Reply to â€˜Letter to Editor by Finsterer J and Zarrouk-Mahjoub S: Phenotypic manifestations of the m.8969G>A variantâ€™. Neurogenetics, 2018, 19, 133-134.	1.4	0
15	Defective mitochondrial ATPase due to rare mtDNA m.8969G>A mutationâ€”causing lactic acidosis, intellectual disability, and poor growth. Neurogenetics, 2018, 19, 49-53.	1.4	7
16	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
17	A complex genomic locus drives mt DNA replicase POLG expression to its diseaseâ€related nervous system regions. EMBO Molecular Medicine, 2018, 10, 13-21.	6.9	8
18	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73

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19	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
20	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	3.2	59
21	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6
22	Absence of Hikeshi, a nuclear transporter for heat-shock protein HSP70, causes infantile hypomyelinating leukoencephalopathy. European Journal of Human Genetics, 2017, 25, 366-370.	2.8	11
23	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
24	Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865.	6.2	49
25	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
26	Defective mitochondrial RNA processing due to PNPT1 variants causes Leigh syndrome. Human Molecular Genetics, 2017, 26, 3352-3361.	2.9	41
27	ATPase-deficient mitochondrial inner membrane protein ATAD3A disturbs mitochondrial dynamics in dominant hereditary spastic paraplegia. Human Molecular Genetics, 2017, 26, 1432-1443.	2.9	63
28	Splicing Defect in Mitochondrial Seryl-tRNA Synthetase Gene Causes Progressive Spastic Paresis Instead of HUPRA Syndrome. Human Mutation, 2016, 37, 884-888.	2.5	23
29	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyÂpatients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
30	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
31	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
32	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
33	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. Cell Metabolism, 2016, 23, 635-648.	16.2	222
34	Dominant transmission of de novo KIF1A motor domain variant underlying pure spastic paraplegia. European Journal of Human Genetics, 2015, 23, 1427-1430.	2.8	44
35	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology, 2015, 85, 306-315.	1.1	52
36	Mitochondrial encephalomyopathy and retinoblastoma explained by compound heterozygosity of SUCLA2 point mutation and 13q14 deletion. European Journal of Human Genetics, 2015, 23, 325-330.	2.8	20

#	Article	IF	Citations
37	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology, 2014, 83, 743-751.	1.1	31
38	Fatal neonatal lactic acidosis caused by a novel de novo mitochondrial G7453A tRNA-Serine (UCN) mutation. Pediatric Research, 2012, 72, 90-94.	2.3	17
39	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurology, The, 2011, 10, 806-818.	10.2	352