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	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
2	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. Cell Metabolism, 2016, 23, 635-648.	16.2	222
3	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
4	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
5	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
6	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
7	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
8	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
9	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
10	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyÂpatients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
11	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
12	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology, 2015, 85, 306-315.	1.1	52
13	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
14	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
15	Vegan diet in young children remodels metabolism and challenges the statuses of essential nutrients. EMBO Molecular Medicine, 2021, 13, e13492.	6.9	43
16	Defective mitochondrial RNA processing due to PNPT1 variants causes Leigh syndrome. Human Molecular Genetics, 2017, 26, 3352-3361.	2.9	41
17	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
18	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

#	Article	IF	Citations
19	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology, 2014, 83, 743-751.	1.1	31
20	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
21	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
22	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
23	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
24	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
25	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
26	Cost-effectiveness of whole-exome sequencing in progressive neurological disorders of children. European Journal of Paediatric Neurology, 2022, 36, 30-36.	1.6	12
27	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
28	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	3.7	11
29	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
30	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
31	De novo SPTAN1mutation in axonal sensorimotor neuropathy and developmental disorder. Brain, 2020, 143, e104-e104.	7.6	8
32	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
33	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
34	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6
35	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	1.9	6
36	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

#	Article	IF	Citations
37	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159.	2.5	3
38	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
39	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