Johanna Uusimaa

List of Publications by Year in descending order

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25 papers	816 citations	623734 14 h-index	610901 24 g-index
25 all docs	25 docs citations	25 times ranked	1972 citing authors

#	Article	IF	Citations
1	Cytosolic phosphoenolpyruvate carboxykinase deficiency: Expanding the clinical phenotype and novel laboratory findings. Journal of Inherited Metabolic Disease, 2022, 45, 223-234.	3.6	7
2	Starting a DBS service for children: Itâ€™s not the latitude but the attitude - Establishment of the paediatric DBS centre in Northern Finland. European Journal of Paediatric Neurology, 2022, 36, 107-114.	1.6	0
3	Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159.	2.5	3
4	Nhlrc2 is crucial during mouse gastrulation. Genesis, 2022, 60, e23470.	1.6	5
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	Loss of DIAPH1 causes SCBMS, combined immunodeficiency, and mitochondrial dysfunction. Journal of Allergy and Clinical Immunology, 2021, 148, 599-611.	2.9	23
7	Variant in NHLRC2 leads to increased hnRNP C2 in developing neurons and the hippocampus of a mouse model of FINCAÂdisease. Molecular Medicine, 2020, 26, 123.	4.4	5
8	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	7.8	26
9	Novel variants and phenotypes widen the phenotypic spectrum of GABRG2-related disorders. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 99-104.	2.0	16
10	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). Genetics in Medicine, 2019, 21, 2355-2363.	2.4	19
11	NHLRC2 variants identified in patients with fibrosis, neurodegeneration, and cerebral angiomatosis (FINCA): characterisation of a novel cerebropulmonary disease. Acta Neuropathologica, 2018, 135, 727-742.	7.7	21
12	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
13	Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. Blood, 2017, 129, 2266-2279.	1.4	152
14	Novel homozygous PCK1 mutation causing cytosolic phosphoenolpyruvate carboxykinase deficiency presenting as childhood hypoglycemia, an abnormal pattern of urine metabolites and liver dysfunction. Molecular Genetics and Metabolism, 2017, 120, 337-341.	1.1	29
15	Riboflavin-Responsive Multiple Acyl-CoA Dehydrogenase Deficiency Associated with Hepatoencephalomyopathy and White Matter Signal Abnormalities on Brain MRI. Neuropediatrics, 2017, 48, 194-198.	0.6	9
16	Ataxia-pancytopenia syndrome with SAMD9L mutations. Neurology: Genetics, 2017, 3, e183.	1.9	24
17	A novel MTTT mutation m.15933Gâ€‰>â€‰A revealed in analysis of mitochondrial DNA in patients with suspected mitochondrial disease. BMC Medical Genetics, 2017, 18, 14.	2.1	2
18	Case report: a novel frameshift mutation in the mitochondrial cytochrome c oxidase II gene causing mitochondrial disorder. BMC Neurology, 2017, 17, 96.	1.8	6

#	Article	IF	Citations
19	Evaluating clinical mitochondrial respiratory chain enzymes from biopsy specimens presenting skewed probability distribution of activity data. Mitochondrion, 2016, 29, 53-58.	3.4	3
20	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
21	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
22	Sodium valproate induces mitochondrial respiration dysfunction in HepG2 in vitro cell model. Toxicology, 2015, 331, 47-56.	4.2	71
23	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. European Journal of Human Genetics, 2014, 22, 184-191.	2.8	52
24	SIRT5 is under the control of PGCâ€1Î± and AMPK and is involved in regulation of mitochondrial energy metabolism. FASEB Journal, 2014, 28, 3225-3237.	0.5	105
25	Nodularia spumigena extract induces upregulation of mitochondrial respiratory chain complexes in spinach (Spinacia oleracea L.). Acta Physiologiae Plantarum, 2013, 35, 969-974.	2.1	6