# Johanna Uusimaa

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5647280/publications.pdf

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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 | Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. Blood, 2017, 129, 2266-2279. | 1.4 | 152 |
| 2 | 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 |
| 3 | 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 |
| 4 | Sodium valproate induces mitochondrial respiration dysfunction in HepG2 in vitro cell model. Toxicology, 2015, 331, 47-56. | 4.2 | 71 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | 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 |
| 10 | Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364. | 7.8 | 26 |
| 11 | Ataxia-pancytopenia syndrome with SAMD9L mutations. Neurology: Genetics, 2017, 3, e183. | 1.9 | 24 |
| 12 | Loss of DIAPH1 causes SCBMS, combined immunodeficiency, and mitochondrial dysfunction. Journal of Allergy and Clinical Immunology, 2021, 148, 599-611. | 2.9 | 23 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Cytosolic phosphoenolpyruvate carboxykinase deficiency: Expanding the clinical phenotype and novel laboratory findings. Journal of Inherited Metabolic Disease, 2022, 45, 223-234. | 3.6 | 7 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | Nhlrc2 is crucial during mouse gastrulation. Genesis, 2022, 60, e23470. | 1.6 | 5 |
| 22 | Evaluating clinical mitochondrial respiratory chain enzymes from biopsy specimens presenting skewed probability distribution of activity data. Mitochondrion, 2016, 29, 53-58. | 3.4 | 3 |
| 23 | Renal Phenotype in Mitochondrial Diseases: A Multicenter Study. Kidney Diseases (Basel, Switzerland), 2022, 8, 148-159. | 2.5 | 3 |
| 24 | 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 |
| 25 | 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 |