

Hannaleena Kokkonen

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8480939/publications.pdf>

Version: 2024-02-01

13
papers

343
citations

1307594

7
h-index

1125743

13
g-index

13
all docs

13
docs citations

13
times ranked

857
citing authors

#	ARTICLE	IF	CITATIONS
1	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. <i>Human Genetics</i> , 2021, 140, 1011-1029.	3.8	23
2	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. <i>Molecular Genetics & Genomic Medicine</i> , 2021, , e1703.	1.2	3
3	Loss of DIAPH1 causes SCBMS, combined immunodeficiency, and mitochondrial dysfunction. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 599-611.	2.9	23
4	<i>COL4A1</i> and <i>COL4A2</i> Duplication Causes Cerebral Small Vessel Disease With Recurrent Early Onset Ischemic Strokes. <i>Stroke</i> , 2021, 52, e624-e625.	2.0	9
5	Homozygous <i>TAF1C</i> variants are associated with a novel childhood-onset neurological phenotype. <i>Clinical Genetics</i> , 2020, 98, 493-498.	2.0	2
6	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). <i>Genetics in Medicine</i> , 2019, 21, 2355-2363.	2.4	19
7	<i>NHLRC2</i> variants identified in patients with fibrosis, neurodegeneration, and cerebral angiomas (FINCA): characterisation of a novel cerebropulmonary disease. <i>Acta Neuropathologica</i> , 2018, 135, 727-742.	7.7	21
8	Gain-of-function <i>SAMD9L</i> mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. <i>Blood</i> , 2017, 129, 2266-2279.	1.4	152
9	Biallelic Mutations in <i>PDE10A</i> Lead to Loss of Striatal <i>PDE10A</i> and a Hyperkinetic Movement Disorder with Onset in Infancy. <i>American Journal of Human Genetics</i> , 2016, 98, 735-743.	6.2	65
10	Infantile spasms and 15q11.2q13.1 chromosome duplication in two successive generations. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 164-167.	1.6	4
11	Combined First-Trimester Screening in Northern Finland: Experiences of the First Ten Years. <i>Clinical Medicine Insights Reproductive Health</i> , 2014, 8, CMRH.S14958.	3.9	4
12	An unexpected recurrence of Angelman syndrome suggestive of maternal germ-line mosaicism of del(15)(q11q13) in a Finnish family. <i>Human Genetics</i> , 2000, 107, 83-85.	3.8	3
13	An unexpected recurrence of Angelman syndrome suggestive of maternal germ-line mosaicism of del(15)(q11q13) in a Finnish family. <i>Human Genetics</i> , 2000, 107, 83-85.	3.8	15