Hannaleena Kokkonen

List of Publications by Year
in descending order

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

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13	343	1307594 7	1125743 13
papers	citations	h-index	g-index

13	13	13	857
all docs	docs citations	times ranked	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	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
3	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029.	3.8	23
4	Loss of DIAPH1 causes SCBMS, combined immunodeficiency, and mitochondrial dysfunction. Journal of Allergy and Clinical Immunology, 2021, 148, 599-611.	2.9	23
5	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
6	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
7	An unexpected recurrence of Angelman syndrome suggestive of maternal germ-line mosaicism of del(15)(q11q13) in a Finnish family. Human Genetics, 2000, 107, 83-85.	3.8	15
8	COL4A1 and COL4A2 Duplication Causes Cerebral Small Vessel Disease With Recurrent Early Onset Ischemic Strokes. Stroke, 2021, 52, e624-e625.	2.0	9
9	Combined First-Trimester Screening in Northern Finland: Experiences of the First Ten Years. Clinical Medicine Insights Reproductive Health, 2014, 8, CMRH.S14958.	3.9	4
10	Infantile spasms and 15q11.2q13.1 chromosome duplication in two successive generations. European Journal of Paediatric Neurology, 2016, 20, 164-167.	1.6	4
11	An unexpected recurrence of Angelman syndrome suggestive of maternal germ-line mosaicism of del(15)(q11q13) in a Finnish family. Human Genetics, 2000, 107, 83-85.	3.8	3
12	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. Molecular Genetics & Genomic Medicine, 2021, , e1703.	1.2	3
13	Homozygous <scp>TAF1C</scp> variants are associated with a novel childhoodâ€onset neurological phenotype. Clinical Genetics, 2020, 98, 493-498.	2.0	2