

Anneke Ja Kievit

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

366
citations

1163117

8
h-index

1281871

11
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13
all docs

13
docs citations

13
times ranked

789
citing authors

#	ARTICLE	IF	CITATIONS
1	Early onset X-linked female limited high myopia in three multigenerational families caused by novel mutations in the <i>ARR3</i> gene. <i>Human Mutation</i> , 2022, 43, 380-388.	2.5	11
2	Experience in Genetic Counseling for GBA1 Variants in Parkinson's Disease. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 33-36.	1.5	5
3	A new alpha-synuclein missense variant (Thr72Met) in two Turkish families with Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2021, 89, 63-72.	2.2	11
4	Modelling the cascade of biomarker changes in progranulin-related frontotemporal dementia. <i>Alzheimer's and Dementia</i> , 2020, 16, e040934.	0.8	0
5	Lessons learned from 40 novel <i>PIGA</i> patients and a review of the literature. <i>Epilepsia</i> , 2020, 61, 1142-1155.	5.1	32
6	Deciphering the natural history of SCA7 in children. <i>European Journal of Neurology</i> , 2020, 27, 2267-2276.	3.3	12
7	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 241-258.	2.6	32
8	Cysteinyl-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. <i>American Journal of Human Genetics</i> , 2019, 104, 520-529.	6.2	31
9	The Effect of Predictive Testing in Adult-Onset Neurodegenerative Diseases on Social and Personal Life. <i>Journal of Genetic Counseling</i> , 2018, 27, 947-954.	1.6	10
10	Mutations in <i>TMEM230</i> are not a common cause of Parkinson's disease. <i>Movement Disorders</i> , 2017, 32, 302-304.	3.9	14
11	<i>DNAJC6</i> Mutations Associated with Early-Onset Parkinson's Disease. <i>Annals of Neurology</i> , 2016, 79, 244-256.	5.3	148
12	Paroxysmal exercise-induced dystonia within the phenotypic spectrum of <i>ECHS1</i> deficiency. <i>Movement Disorders</i> , 2016, 31, 1041-1048.	3.9	58