Jenny Lord

List of Publications by Year in descending order

35 papers	5,421 citations	279778 23 h-index	377849 34 g-index
47 all docs	47 docs citations	47 times ranked	10825 citing authors

#	Article	IF	Citations
1	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	21.4	1,962
2	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783
3	Prenatal exome sequencing analysis in fetal structural anomalies detected by ultrasonography (PAGE): a cohort study. Lancet, The, 2019, 393, 747-757.	13.7	443
4	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
5	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
6	Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in Medicine, 2018, 20, 1216-1223.	2.4	255
7	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
8	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
9	The epigenetic landscape of Alzheimer's disease. Nature Neuroscience, 2014, 17, 1138-1140.	14.8	101
10	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	2.4	99
11	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
12	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.2	65
13	Investigating the role of rare coding variability in Mendelian dementia genes (APP , PSEN1 , PSEN2 , GRN) Tj ETQq1 1 0.784314 rgBT	3.1	53
14	Molecular autopsy by trio exome sequencing (ES) and postmortem examination in fetuses and neonates with prenatally identified structural anomalies. Genetics in Medicine, 2019, 21, 1065-1073.	2.4	47
15	A potential endophenotype for Alzheimer's disease: cerebrospinal fluid clusterin. Neurobiology of Aging, 2016, 37, 208.e1-208.e9.	3.1	44
16	Splicing in the Diagnosis of Rare Disease: Advances and Challenges. Frontiers in Genetics, 2021, 12, 689892.	2.3	41
17	Caspase-8, association with Alzheimerâ€™s Disease and functional analysis of rare variants. PLoS ONE, 2017, 12, e0185777.	2.5	38
18	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. Neurobiology of Aging, 2016, 46, 235.e1-235.e9.	3.1	37

#	Article	IF	Citations
19	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	3.3	37
20	Influence of Coding Variability in APP-AÎ² Metabolism Genes in Sporadic Alzheimerâ€™s Disease. PLoS ONE, 2016, 11, e0150079.	2.5	34
21	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. Neurobiology of Aging, 2018, 66, 179.e17-179.e29.	3.1	32
22	Identification of rare variants in AlzheimerÃ¢â‚¬â„¢s disease. Frontiers in Genetics, 2014, 5, 369.	2.3	30
23	Exome sequencing identifies 2 novel presenilin 1 mutations (p.L166V and p.S230R) in British early-onset Alzheimer's disease. Neurobiology of Aging, 2014, 35, 2422.e13-2422.e16.	3.1	28
24	Genetic studies of plasma analytes identify novel potential biomarkers for several complex traits. Scientific Reports, 2016, 6, .	3.3	25
25	Splicing in the pathogenesis, diagnosis and treatment of ciliopathies. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2019, 1862, 194433.	1.9	25
26	Fetal central nervous system anomalies: When should we offer exome sequencing?. Prenatal Diagnosis, 2022, 42, 736-743.	2.3	16
27	Whole genome sequencing in the diagnosis of primary ciliary dyskinesia. BMC Medical Genomics, 2021, 14, 234.	1.5	15
28	CI-SpliceAIâ€”Improving machine learning predictions of disease causing splicing variants using curated alternative splice sites. PLoS ONE, 2022, 17, e0269159.	2.5	15
29	A CRISPR and high-content imaging assay compliant with ACMG/AMP guidelines for clinical variant interpretation in ciliopathies. Human Genetics, 2021, 140, 593-607.	3.8	6
30	Janus-faced EPHB4-associated disorders: novel pathogenic variants and unreported intrafamilial overlapping phenotypes. Genetics in Medicine, 2021, 23, 1315-1324.	2.4	6
31	Prenatal Exome Sequencing Analysis in Fetal Structural Anomalies Detected by Ultrasonography (PAGE): A Cohort Study. Obstetrical and Gynecological Survey, 2019, 74, 394-396.	0.4	2
32	Next generation sequencing of CLU, PICALM and CR1: pitfalls and potential solutions. International Journal of Molecular Epidemiology and Genetics, 2012, 3, 262-75.	0.4	2
33	P1-055: Exome-sequencing in a large dataset of late-onset families with Alzheimer's disease. , 2015, 11, P359-P359.		1
34	O4-05-06: A potential endophenotype for Alzheimer's disease: Cerebrospinal fluid clusterin. , 2015, 11, P280-P280.		0
35	Primary ciliary dyskinesia and non-CF bronchiectasis in the 100,000 Genomes Project. , 2020, , .		0