Elisa Majounie

List of Publications by Year
in descending order

28 papers	10,504 citations	257450 24 h-index	526287 27 g-index

28 all docs	28 docs citations	28 times ranked	13600 citing authors

#	Article	IF	Citations
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
2	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	27.0	2,385
3	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
4	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
5	Common polygenic variation enhances risk prediction for Alzheimerâ€™s disease. Brain, 2015, 138, 3673-3684.	7.6	359
6	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	6.2	333
7	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
8	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain, 2012, 135, 784-793.	7.6	182
9	Repeat Expansion inC9ORF72in Alzheimer's Disease. New England Journal of Medicine, 2012, 366, 283-284.	27.0	156
10	Extensive genetics of ALS. Neurology, 2012, 79, 1983-1989.	1.1	145
11	Characterization of the somatic mutational spectrum of the neurofibromatosis type 1 (NF1) gene in neurofibromatosis patients with benign and malignant tumors. Human Mutation, 2004, 23, 134-146.	2.5	97
12	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	3.1	96
13	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	8.8	96
14	A molecular analysis of individuals with neurofibromatosis type 1 (NF1) and optic pathway gliomas (OPGs), and an assessment of genotype-phenotype correlations. Journal of Medical Genetics, 2011, 48, 256-260.	3.2	90
15	Familial frontotemporal dementia with amyotrophic lateral sclerosis and a shared haplotype on chromosomeÂ9p. Journal of Neurology, 2011, 258, 647-655.	3.6	76
16	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20.	3.1	76
17	Tdp-43 cryptic exons are highly variable between cell types. Molecular Neurodegeneration, 2017, 12, 13.	10.8	63
18	ALS/FTD phenotype in two Sardinian families carrying bothC9ORF72andTARDBPmutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 730-733.	1.9	57

#	Article	IF	Citations
19	Somatic loss of wild typeNF1 allele in neurofibromas: Comparison ofNF1 microdeletion and non-microdeletion patients. Genes Chromosomes and Cancer, 2006, 45, 893-904.	2.8	56
20	The Correlation between Inflammatory Biomarkers and Polygenic Risk Score inÂAlzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2017, 56, 25-36.	2.6	51
21	Variation in tau isoform expression in different brain regions and disease states. Neurobiology of Aging, 2013, 34, 1922.e7-1922.e12.	3.1	49
22	Transcriptomic profiling of the human brain reveals that altered synaptic gene expression is associated with chronological aging. Scientific Reports, 2017, 7, 16890.	3.3	47
23	Large C9orf72 repeat expansions are not a common cause of Parkinson's disease. Neurobiology of Aging, 2012, 33, 2527.e1-2527.e2.	3.1	40
24	Multiple system atrophy is not caused by C9orf72 hexanucleotide repeat expansions. Neurobiology of Aging, 2015, 36, 1223.e1-1223.e2.	3.1	25
25	Three different pathological lesions in the NF1 gene originating de novo in a family with neurofibromatosis type 1. Human Genetics, 2003, 112, 12-17.	3.8	24
26	Familial Lund frontotemporal dementia caused by C9ORF72 hexanucleotide expansion. Neurobiology of Aging, 2012, 33, 1850.e13-1850.e16.	3.1	14
27	Juvenile onset Parkinsonism with â€œpure nigralâ€•degeneration and POLG1 mutation. Parkinsonism and Related Disorders, 2016, 30, 83-85.	2.2	9
28	P2â€•112: NEXT GENERATION EXOME SEQUENCING IN A LARGE SAMPLE OF ALZHEIMER'S PATIENTS. Alzheimer's and Dementia, 2018, 14, P712.	0.8	0