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	P2â€112: NEXT GENERATION EXOME SEQUENCING IN A LARGE SAMPLE OF ALZHEIMER'S PATIENTS. Alzheimer's and Dementia, 2018, 14, P712.	0.8	0
2	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	8.8	96
3	Tdp-43 cryptic exons are highly variable between cell types. Molecular Neurodegeneration, 2017, 12, 13.	10.8	63
4	The Correlation between Inflammatory Biomarkers and Polygenic Risk Score inÂAlzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2017, 56, 25-36.	2.6	51
5	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
6	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
7	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
8	Juvenile onset Parkinsonism with â€œpure nigralâ€•degeneration and POLG1 mutation. Parkinsonism and Related Disorders, 2016, 30, 83-85.	2.2	9
9	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
10	Multiple system atrophy is not caused by C9orf72 hexanucleotide repeat expansions. Neurobiology of Aging, 2015, 36, 1223.e1-1223.e2.	3.1	25
11	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	3.1	96
12	Common polygenic variation enhances risk prediction for Alzheimerâ€™s disease. Brain, 2015, 138, 3673-3684.	7.6	359
13	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	27.0	2,385
14	Variation in tau isoform expression in different brain regions and disease states. Neurobiology of Aging, 2013, 34, 1922.e7-1922.e12.	3.1	49
15	Repeat Expansion inC9ORF72in Alzheimer's Disease. New England Journal of Medicine, 2012, 366, 283-284.	27.0	156
16	Extensive genetics of ALS. Neurology, 2012, 79, 1983-1989.	1.1	145
17	ALS/FTD phenotype in two Sardinian families carrying bothC9ORF72andTARDBPmutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 730-733.	1.9	57
18	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20.	3.1	76

#	Article	IF	Citations
19	Familial Lund frontotemporal dementia caused by C9ORF72 hexanucleotide expansion. Neurobiology of Aging, 2012, 33, 1850.e13-1850.e16.	3.1	14
20	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
21	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
22	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
23	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
24	Familial frontotemporal dementia with amyotrophic lateral sclerosis and a shared haplotype on chromosomeÂ9p. Journal of Neurology, 2011, 258, 647-655.	3.6	76
25	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
26	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
27	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
28	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