Ian P Blair

List of Publications by Year
in descending order

97 papers	11,691 citations	116194 36 h-index	38517 99 g-index
109 all docs	109 docs citations	109 times ranked	14409 citing authors

#	Article	IF	Citations
1	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	6.0	2,295
2	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	6.0	2,268
3	DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4). American Journal of Human Genetics, 2004, 74, 1128-1135.	2.6	717
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
5	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
6	C9ORF72, implicated in amytrophic lateral sclerosis and frontotemporal dementia, regulates endosomal trafficking. Human Molecular Genetics, 2014, 23, 3579-3595.	1.4	410
7	A yeast functional screen predicts new candidate ALS disease genes. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 20881-20890.	3.3	365
8	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
9	Life events, first depression onset and the serotonin transporter gene. British Journal of Psychiatry, 2006, 188, 210-215.	1.7	262
10	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 2899-2911.	1.4	246
11	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
12	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
13	FUS mutations in amyotrophic lateral sclerosis: clinical, pathological, neurophysiological and genetic analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 639-645.	0.9	205
14	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
15	Sigma nonopioid intracellular receptor 1 mutations cause frontotemporal lobar degenerationâ€“motor neuron disease. Annals of Neurology, 2010, 68, 639-649.	2.8	168
16	The gene for hereditary sensory neuropathy type I (HSNâ€“I) maps to chromosome 9q22.1â€“q22.3. Nature Genetics, 1996, 13, 101-104.	9.4	130
17	Exome sequencing to identify de novo mutations in sporadic ALS trios. Nature Neuroscience, 2013, 16, 851-855.	7.1	129
18	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. American Journal of Human Genetics, 2013, 93, 900-905.	2.6	123

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19	UBQLN2/ubiquilin 2 mutation and pathology in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 2527.e3-2527.e10.	1.5	114
20	Distinct partitioning of ALS associated TDP-43, FUS and SOD1 mutants into cellular inclusions. Scientific Reports, 2015, 5, 13416.	1.6	109
21	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. Nature Communications, 2017, 8, 611.	5.8	93
22	Ataxin-2 interacts with FUS and intermediate-length polyglutamine expansions enhance FUS-related pathology in amyotrophic lateral sclerosis. Human Molecular Genetics, 2013, 22, 717-728.	1.4	90
23	Pathophysiological insights into ALS with C9ORF72 expansions. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 931-935.	0.9	89
24	Mutant FUS induces endoplasmic reticulum stress in amyotrophic lateral sclerosis and interacts with protein disulfide-isomerase. Neurobiology of Aging, 2012, 33, 2855-2868.	1.5	88
25	Defects in optineurin- and myosin VI-mediated cellular trafficking in amyotrophic lateral sclerosis. Human Molecular Genetics, 2015, 24, 3830-3846.	1.4	71
26	TDP-43 mutations causing amyotrophic lateral sclerosis are associated with altered expression of RNA-binding protein hnRNP K and affect the Nrf2 antioxidant pathway. Human Molecular Genetics, 2017, 26, 1732-1746.	1.4	62
27	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	3.7	62
28	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	3.8	61
29	The genotypeâ€“phenotype landscape of familial amyotrophic lateral sclerosis in Australia. Clinical Genetics, 2017, 92, 259-266.	1.0	58
30	Impaired NHEJ repair in amyotrophic lateral sclerosis is associated with TDP-43 mutations. Molecular Neurodegeneration, 2020, 15, 51.	4.4	54
31	TDP-43: A DNA and RNA binding protein with roles in neurodegenerative diseases. International Journal of Biochemistry and Cell Biology, 2010, 42, 1606-1609.	1.2	53
32	Ubiquilin 2: A component of the ubiquitinâ€“proteasome system with an emerging role in neurodegeneration. International Journal of Biochemistry and Cell Biology, 2014, 50, 123-126.	1.2	53
33	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	3.8	51
34	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	3.8	49
35	Phosphorylation of hnRNP K by cyclin-dependent kinase 2 controls cytosolic accumulation of TDP-43. Human Molecular Genetics, 2015, 24, 1655-1669.	1.4	48
36	Evidence for polygenic and oligogenic basis of Australian sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2021, 58, 87-95.	1.5	48

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37	Expression of ALS/FTD-linked mutant CCNF in zebrafish leads to increased cell death in the spinal cord and an aberrant motor phenotype. Human Molecular Genetics, 2017, 26, 2616-2626.	1.4	44
38	Pathogenic mutation in the ALS/FTD gene, CCNF, causes elevated Lys48-linked ubiquitylation and defective autophagy. Cellular and Molecular Life Sciences, 2018, 75, 335-354.	2.4	44
39	Genome-wide Meta-analysis Finds the ACSL5-ZDHHC6 Locus Is Associated with ALS and Links Weight Loss to the Disease Genetics. Cell Reports, 2020, 33, 108323.	2.9	41
40	Neuronal intranuclear inclusion disease is genetically heterogeneous. Annals of Clinical and Translational Neurology, 2020, 7, 1716-1725.	1.7	38
41	Monozygotic twins and triplets discordant for amyotrophic lateral sclerosis display differential methylation and gene expression. Scientific Reports, 2019, 9, 8254.	1.6	36
42	Novel TBK1 truncating mutation in a familial amyotrophic lateral sclerosis patient of Chinese origin. Neurobiology of Aging, 2015, 36, 3334.e1-3334.e5.	1.5	35
43	Search for genes involved in Joubert syndrome: Evidence that one or more major loci are yet to be identified and exclusion of candidate genesEN1,EN2,FGF8, andBARHL1. American Journal of Medical Genetics Part A, 2002, 107, 190-196.	2.4	34
44	Fused in sarcoma/translocated in liposarcoma: A multifunctional DNA/RNA binding protein. International Journal of Biochemistry and Cell Biology, 2010, 42, 1408-1411.	1.2	30
45	Evaluation of Skin Fibroblasts from Amyotrophic Lateral Sclerosis Patients for the Rapid Study of Pathological Features. Neurotoxicity Research, 2015, 28, 138-146.	1.3	30
46	Casein kinase II phosphorylation of cyclin F at serine 621 regulates the Lys48-ubiquitylation E3 ligase activity of the SCF (cyclin F) complex. Open Biology, 2017, 7, 170058.	1.5	29
47	Ubiquitin Homeostasis Is Disrupted in TDP-43 and FUS Cell Models of ALS. IScience, 2020, 23, 101700.	1.9	28
48	Genetic and Pathological Assessment of hnRNPA1, hnRNPA2/B1, and hnRNPA3 in Familial and Sporadic Amyotrophic Lateral Sclerosis. Neurodegenerative Diseases, 2017, 17, 304-312.	0.8	27
49	A Simple Differentiation Protocol for Generation of Induced Pluripotent Stem Cell-Derived Basal Forebrain-Like Cholinergic Neurons for Alzheimerâ€™s Disease and Frontotemporal Dementia Disease Modeling. Cells, 2020, 9, 2018.	1.8	27
50	Predictive genetic testing for amyotrophic lateral sclerosis and frontotemporal dementia: genetic counselling considerations. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 475-485.	1.1	26
51	Cyclin F: A component of an E3 ubiquitin ligase complex with roles in neurodegeneration and cancer. International Journal of Biochemistry and Cell Biology, 2017, 89, 216-220.	1.2	26
52	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. Npj Genomic Medicine, 2020, 5, 10.	1.7	25
53	Altered SOD1 maturation and post-translational modification in amyotrophic lateral sclerosis spinal cord. Brain, 2022, 145, 3108-3130.	3.7	25
54	Frontotemporal dementiaâ€“amyotrophic lateral sclerosis syndrome locus on chromosome 16p12.1â€“q12.2: genetic, clinical and neuropathological analysis. Acta Neuropathologica, 2013, 125, 523-533.	3.9	24

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55	A novel amyotrophic lateral sclerosis mutation in OPTN induces ER stress and Golgi fragmentation in vitro. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 126-133.	1.1	24
56	ERp57 is protective against mutant SOD1-induced cellular pathology in amyotrophic lateral sclerosis. Human Molecular Genetics, 2018, 27, 1311-1331.	1.4	24
57	Amyotrophic lateral sclerosis-linked UBQLN2 mutants inhibit endoplasmic reticulum to Golgi transport, leading to Golgi fragmentation and ER stress. Cellular and Molecular Life Sciences, 2020, 77, 3859-3873.	2.4	24
58	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456.	2.9	24
59	A YAC-Based Transcript Map of Human Chromosome 9q22.1â€“q22.3 Encompassing the Loci for Hereditary Sensory Neuropathy Type I and Multiple Self-Healing Squamous Epithelioma. Genomics, 1998, 51, 277-281.	1.3	23
60	A novel locus for distal motor neuron degeneration maps to chromosome 7q34-q36. Human Genetics, 2007, 121, 559-564.	1.8	23
61	Accumulation of dysfunctional SOD1 protein in Parkinsonâ€™s disease is not associated with mutations in the SOD1 gene. Acta Neuropathologica, 2018, 135, 155-156.	3.9	23
62	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.1	22
63	The metastability of the proteome of spinal motor neurons underlies their selective vulnerability in ALS. Neuroscience Letters, 2019, 704, 89-94.	1.0	22
64	Identity by descent analysis identifies founder events and links SOD1 familial and sporadic ALS cases. Npj Genomic Medicine, 2020, 5, 32.	1.7	20
65	Mutant Human FUS Is Ubiquitously Mislocalized and Generates Persistent Stress Granules in Primary Cultured Transgenic Zebrafish Cells. PLoS ONE, 2014, 9, e90572.	1.1	19
66	Association between the serotonin 2A receptor gene and bipolar affective disorder in an Australian cohort. Psychiatric Genetics, 2009, 19, 244-252.	0.6	18
67	Mutation analysis of VCP in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 1488.e15-1488.e16.	1.5	17
68	Mutation analysis and immunopathological studies of PFN1 in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2235.e7-2235.e10.	1.5	16
69	The C9orf72 hexanucleotide repeat expansion presents a challenge for testing laboratories and genetic counseling. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 310-316.	1.1	16
70	Motor Neuron Abnormalities Correlate with Impaired Movement in Zebrafish that Express Mutant Superoxide Dismutase 1. Zebrafish, 2019, 16, 8-14.	0.5	16
71	Sensitive Detection of Motor Neuron Disease Derived Exosomal miRNA Using Electrocatalytic Activity of Goldâ€Loaded Superparamagnetic Ferric Oxide Nanocubes. ChemElectroChem, 2020, 7, 3459-3467.	1.7	16
72	ALS/FTD-causing mutation in cyclin F causes the dysregulation of SFPQ. Human Molecular Genetics, 2021, 30, 971-984.	1.4	16

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73	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. European Journal of Human Genetics, 2022, 30, 532-539.	1.4	16
74	Mutation analysis of the optineurin gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 210.e9-210.e10.	1.5	13
75	Mutation analysis of MATR3 in Australian familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1602.e1-1602.e2.	1.5	13
76	A novelTARDBPinsertion/deletion mutation in the flail arm variant of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 465-470.	2.3	12
77	Unbiased Label-Free Quantitative Proteomics of Cells Expressing Amyotrophic Lateral Sclerosis (ALS) Mutations in CCNF Reveals Activation of the Apoptosis Pathway: A Workflow to Screen Pathogenic Gene Mutations. Frontiers in Molecular Neuroscience, 2021, 14, 627740.	1.4	12
78	Splicing factor proline and glutamine rich intron retention, reduced expression and aggregate formation are pathological features of amyotrophic lateral sclerosis. Neuropathology and Applied Neurobiology, 2021, 47, 990-1003.	1.8	11
79	TDP-43 is a ubiquitylation substrate of the SCFcyclin F complex. Neurobiology of Disease, 2022, 167, 105673.	2.1	11
80	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy typeÂI by mutation analysis. Human Genetics, 2000, 106, 594-596.	1.8	8
81	Neuronal cell culture from transgenic zebrafish models of neurodegenerative disease. Biology Open, 2018, 7, .	0.6	8
82	Genetic and immunopathological analysis of CHCHD10 in Australian amyotrophic lateral sclerosis and frontotemporal dementia and transgenic TDP-43 mice. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 162-171.	0.9	8
83	Genetic Analysis of Tryptophan Metabolism Genes in Sporadic Amyotrophic Lateral Sclerosis. Frontiers in Immunology, 2021, 12, 701550.	2.2	8
84	Search for genes involved in Joubert syndrome: evidence that one or more major loci are yet to be identified and exclusion of candidate genes EN1, EN2, FGF8, and BARHL1. American Journal of Medical Genetics Part A, 2002, 107, 190-6.	2.4	8
85	Generation and characterization of a human induced pluripotent stem cell line UOWi005-A from dermal fibroblasts derived from a CCNF familial amyotrophic lateral sclerosis patient using mRNA reprogramming. Stem Cell Research, 2019, 40, 101530.	0.3	6
86	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 101, 297.e9-297.e11.	1.5	6
87	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy type I by mutation analysis. Human Genetics, 2000, 106, 594-596.	1.8	5
88	Genome screen of 15 Australian bipolar affective disorder pedigrees supports previously identified loci for bipolar susceptibility genes. Psychiatric Genetics, 2008, 18, 156-161.	0.6	5
89	Association analysis of transcripts from the bipolar susceptibility locus on chromosome 4q35, exclusion of a pathogenic role for eight positional candidate genes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 134B, 56-59.	1.1	4
90	The mRNA-based reprogramming of fibroblasts from a SOD1E101G familial amyotrophic lateral sclerosis patient to induced pluripotent stem cell line UOWi007. Stem Cell Research, 2020, 42, 101701.	0.3	4

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91	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
92	Simultaneous Isolation of High-Quality RNA and DNA From Postmortem Human Central Nervous System Tissues for Omics Studies. Journal of Neuropathology and Experimental Neurology, 2022, 81, 135-145.	0.9	3
93	Identification, characterization, and association analysis of novel genes from the bipolar disorder susceptibility locus on chromosome 4q35. Psychiatric Genetics, 2005, 15, 199-204.	0.6	2
94	Theme 13 Clinical management and support. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 327-347.	1.1	2
95	Postnatal Development of Spasticity Following Transgene Insertion in the Mouse Î²IV Spectrin Gene (SPTBN4). Journal of Neuromuscular Diseases, 2017, 4, 159-164.	1.1	1
96	Theme 3 In vitro experimental models. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 135-159.	1.1	1
97	NEK1 and STMN2 short tandem repeat lengths are not associated with Australian amyotrophic lateral sclerosis risk. Neurobiology of Aging, 2022, , .	1.5	0