

Ian P Blair

List of Publications by Year in descending order

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97
papers

11,691
citations

101543

36
h-index

33894

99
g-index

109
all docs

109
docs citations

109
times ranked

13113
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. <i>Science</i> , 2009, 323, 1208-1211.	12.6	2,295
2	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. <i>Science</i> , 2008, 319, 1668-1672.	12.6	2,268
3	DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4). <i>American Journal of Human Genetics</i> , 2004, 74, 1128-1135.	6.2	717
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
5	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	21.4	494
6	C9ORF72, implicated in amyotrophic lateral sclerosis and frontotemporal dementia, regulates endosomal trafficking. <i>Human Molecular Genetics</i> , 2014, 23, 3579-3595.	2.9	410
7	A yeast functional screen predicts new candidate ALS disease genes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 20881-20890.	7.1	365
8	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	8.1	308
9	Life events, first depression onset and the serotonin transporter gene. <i>British Journal of Psychiatry</i> , 2006, 188, 210-215.	2.8	262
10	Evaluating the role of the FUS/TLS-related gene EWSR1 in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2012, 21, 2899-2911.	2.9	246
11	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	21.4	223
12	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	21.4	218
13	FUS mutations in amyotrophic lateral sclerosis: clinical, pathological, neurophysiological and genetic analysis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 639-645.	1.9	205
14	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	12.8	174
15	Sigma nonopioid intracellular receptor 1 mutations cause frontotemporal lobar degeneration—motor neuron disease. <i>Annals of Neurology</i> , 2010, 68, 639-649.	5.3	168
16	The gene for hereditary sensory neuropathy type I (HSN—1) maps to chromosome 9q22.1—q22.3. <i>Nature Genetics</i> , 1996, 13, 101-104.	21.4	130
17	Exome sequencing to identify de novo mutations in sporadic ALS trios. <i>Nature Neuroscience</i> , 2013, 16, 851-855.	14.8	129
18	ERBB4 Mutations that Disrupt the Neuregulin-ErbB4 Pathway Cause Amyotrophic Lateral Sclerosis Type 19. <i>American Journal of Human Genetics</i> , 2013, 93, 900-905.	6.2	123

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19	UBQLN2/ubiquilin 2 mutation and pathology in familial amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2012, 33, 2527.e3-2527.e10.	3.1	114
20	Distinct partitioning of ALS associated TDP-43, FUS and SOD1 mutants into cellular inclusions. <i>Scientific Reports</i> , 2015, 5, 13416.	3.3	109
21	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. <i>Nature Communications</i> , 2017, 8, 611.	12.8	93
22	Ataxin-2 interacts with FUS and intermediate-length polyglutamine expansions enhance FUS-related pathology in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2013, 22, 717-728.	2.9	90
23	Pathophysiological insights into ALS with C9ORF72 expansions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 931-935.	1.9	89
24	Mutant FUS induces endoplasmic reticulum stress in amyotrophic lateral sclerosis and interacts with protein disulfide-isomerase. <i>Neurobiology of Aging</i> , 2012, 33, 2855-2868.	3.1	88
25	Defects in optineurin- and myosin VI-mediated cellular trafficking in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2015, 24, 3830-3846.	2.9	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. <i>Human Molecular Genetics</i> , 2017, 26, 1732-1746.	2.9	62
27	CYLD is a causative gene for frontotemporal dementia “ amyotrophic lateral sclerosis. <i>Brain</i> , 2020, 143, 783-799.	7.6	62
28	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. <i>Neuron</i> , 2017, 94, 322-336.e5.	8.1	61
29	The genotype“phenotype landscape of familial amyotrophic lateral sclerosis in Australia. <i>Clinical Genetics</i> , 2017, 92, 259-266.	2.0	58
30	Impaired NHEJ repair in amyotrophic lateral sclerosis is associated with TDP-43 mutations. <i>Molecular Neurodegeneration</i> , 2020, 15, 51.	10.8	54
31	TDP-43: A DNA and RNA binding protein with roles in neurodegenerative diseases. <i>International Journal of Biochemistry and Cell Biology</i> , 2010, 42, 1606-1609.	2.8	53
32	Ubiquilin 2: A component of the ubiquitin“proteasome system with an emerging role in neurodegeneration. <i>International Journal of Biochemistry and Cell Biology</i> , 2014, 50, 123-126.	2.8	53
33	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. <i>Neuron</i> , 2022, 110, 992-1008.e11.	8.1	51
34	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. <i>Genome Biology</i> , 2021, 22, 90.	8.8	49
35	Phosphorylation of hnRNP K by cyclin-dependent kinase 2 controls cytosolic accumulation of TDP-43. <i>Human Molecular Genetics</i> , 2015, 24, 1655-1669.	2.9	48
36	Evidence for polygenic and oligogenic basis of Australian sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2021, 58, 87-95.	3.2	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. <i>Human Molecular Genetics</i> , 2017, 26, 2616-2626.	2.9	44
38	Pathogenic mutation in the ALS/FTD gene, CCNF, causes elevated Lys48-linked ubiquitylation and defective autophagy. <i>Cellular and Molecular Life Sciences</i> , 2018, 75, 335-354.	5.4	44
39	Genome-wide Meta-analysis Finds the ACSL5-ZDHHC6 Locus Is Associated with ALS and Links Weight Loss to the Disease Genetics. <i>Cell Reports</i> , 2020, 33, 108323.	6.4	41
40	Neuronal intranuclear inclusion disease is genetically heterogeneous. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1716-1725.	3.7	38
41	Monozygotic twins and triplets discordant for amyotrophic lateral sclerosis display differential methylation and gene expression. <i>Scientific Reports</i> , 2019, 9, 8254.	3.3	36
42	Novel TBK1 truncating mutation in a familial amyotrophic lateral sclerosis patient of Chinese origin. <i>Neurobiology of Aging</i> , 2015, 36, 3334.e1-3334.e5.	3.1	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 genes EN1, EN2, FGF8, and BARHL1. <i>American Journal of Medical Genetics Part A</i> , 2002, 107, 190-196.	2.4	34
44	Fused in sarcoma/translocated in liposarcoma: A multifunctional DNA/RNA binding protein. <i>International Journal of Biochemistry and Cell Biology</i> , 2010, 42, 1408-1411.	2.8	30
45	Evaluation of Skin Fibroblasts from Amyotrophic Lateral Sclerosis Patients for the Rapid Study of Pathological Features. <i>Neurotoxicity Research</i> , 2015, 28, 138-146.	2.7	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. <i>Open Biology</i> , 2017, 7, 170058.	3.6	29
47	Ubiquitin Homeostasis Is Disrupted in TDP-43 and FUS Cell Models of ALS. <i>iScience</i> , 2020, 23, 101700.	4.1	28
48	Genetic and Pathological Assessment of hnRNPA1, hnRNPA2/B1, and hnRNPA3 in Familial and Sporadic Amyotrophic Lateral Sclerosis. <i>Neurodegenerative Diseases</i> , 2017, 17, 304-312.	1.4	27
49	A Simple Differentiation Protocol for Generation of Induced Pluripotent Stem Cell-Derived Basal Forebrain-Like Cholinergic Neurons for Alzheimer's Disease and Frontotemporal Dementia Disease Modeling. <i>Cells</i> , 2020, 9, 2018.	4.1	27
50	Predictive genetic testing for amyotrophic lateral sclerosis and frontotemporal dementia: genetic counselling considerations. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 475-485.	1.7	26
51	Cyclin F: A component of an E3 ubiquitin ligase complex with roles in neurodegeneration and cancer. <i>International Journal of Biochemistry and Cell Biology</i> , 2017, 89, 216-220.	2.8	26
52	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , 2020, 5, 10.	3.8	25
53	Altered SOD1 maturation and post-translational modification in amyotrophic lateral sclerosis spinal cord. <i>Brain</i> , 2022, 145, 3108-3130.	7.6	25
54	Frontotemporal dementia-amyotrophic lateral sclerosis syndrome locus on chromosome 16p12.1-q12.2: genetic, clinical and neuropathological analysis. <i>Acta Neuropathologica</i> , 2013, 125, 523-533.	7.7	24

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55	A novel amyotrophic lateral sclerosis mutation in <i>OPTN</i> induces ER stress and Golgi fragmentation in vitro. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 126-133.	1.7	24
56	ERp57 is protective against mutant SOD1-induced cellular pathology in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2018, 27, 1311-1331.	2.9	24
57	Amyotrophic lateral sclerosis-linked UBQLN2 mutants inhibit endoplasmic reticulum to Golgi transport, leading to Golgi fragmentation and ER stress. <i>Cellular and Molecular Life Sciences</i> , 2020, 77, 3859-3873.	5.4	24
58	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. <i>Cell Reports</i> , 2020, 33, 108456.	6.4	24
59	A YAC-Based Transcript Map of Human Chromosome 9q22.1–q22.3 Encompassing the Loci for Hereditary Sensory Neuropathy Type I and Multiple Self-Healing Squamous Epithelioma. <i>Genomics</i> , 1998, 51, 277-281.	2.9	23
60	A novel locus for distal motor neuron degeneration maps to chromosome 7q34-q36. <i>Human Genetics</i> , 2007, 121, 559-564.	3.8	23
61	Accumulation of dysfunctional SOD1 protein in Parkinson's disease is not associated with mutations in the SOD1 gene. <i>Acta Neuropathologica</i> , 2018, 135, 155-156.	7.7	23
62	Reconsidering the causality of TIA1 mutations in ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2018, 19, 1-3.	1.7	22
63	The metastability of the proteome of spinal motor neurons underlies their selective vulnerability in ALS. <i>Neuroscience Letters</i> , 2019, 704, 89-94.	2.1	22
64	Identity by descent analysis identifies founder events and links SOD1 familial and sporadic ALS cases. <i>Npj Genomic Medicine</i> , 2020, 5, 32.	3.8	20
65	Mutant Human FUS Is Ubiquitously Mislocalized and Generates Persistent Stress Granules in Primary Cultured Transgenic Zebrafish Cells. <i>PLoS ONE</i> , 2014, 9, e90572.	2.5	19
66	Association between the serotonin 2A receptor gene and bipolar affective disorder in an Australian cohort. <i>Psychiatric Genetics</i> , 2009, 19, 244-252.	1.1	18
67	Mutation analysis of VCP in familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2012, 33, 1488.e15-1488.e16.	3.1	17
68	Mutation analysis and immunopathological studies of PFN1 in familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2013, 34, 2235.e7-2235.e10.	3.1	16
69	The <i>C9orf72</i> hexanucleotide repeat expansion presents a challenge for testing laboratories and genetic counseling. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 310-316.	1.7	16
70	Motor Neuron Abnormalities Correlate with Impaired Movement in Zebrafish that Express Mutant Superoxide Dismutase 1. <i>Zebrafish</i> , 2019, 16, 8-14.	1.1	16
71	Sensitive Detection of Motor Neuron Disease Derived Exosomal miRNA Using Electrocatalytic Activity of Gold-Loaded Superparamagnetic Ferric Oxide Nanocubes. <i>ChemElectroChem</i> , 2020, 7, 3459-3467.	3.4	16
72	ALS/FTD-causing mutation in cyclin F causes the dysregulation of SFPQ. <i>Human Molecular Genetics</i> , 2021, 30, 971-984.	2.9	16

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73	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. <i>European Journal of Human Genetics</i> , 2022, 30, 532-539.	2.8	16
74	Mutation analysis of the optineurin gene in familial amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2012, 33, 210.e9-210.e10.	3.1	13
75	Mutation analysis of MATR3 in Australian familial amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2015, 36, 1602.e1-1602.e2.	3.1	13
76	A novel <i>TARDBP</i> insertion/deletion mutation in the flail arm variant of amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 465-470.	2.1	12
77	Unbiased Label-Free Quantitative Proteomics of Cells Expressing Amyotrophic Lateral Sclerosis (ALS) Mutations in CCFN Reveals Activation of the Apoptosis Pathway: A Workflow to Screen Pathogenic Gene Mutations. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 627740.	2.9	12
78	Splicing factor proline and glutamine rich intron retention, reduced expression and aggregate formation are pathological features of amyotrophic lateral sclerosis. <i>Neuropathology and Applied Neurobiology</i> , 2021, 47, 990-1003.	3.2	11
79	TDP-43 is a ubiquitylation substrate of the SCF _{cyclin F} complex. <i>Neurobiology of Disease</i> , 2022, 167, 105673.	4.4	11
80	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy type I by mutation analysis. <i>Human Genetics</i> , 2000, 106, 594-596.	3.8	8
81	Neuronal cell culture from transgenic zebrafish models of neurodegenerative disease. <i>Biology Open</i> , 2018, 7, .	1.2	8
82	Genetic and immunopathological analysis of CHCHD10 in Australian amyotrophic lateral sclerosis and frontotemporal dementia and transgenic TDP-43 mice. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 162-171.	1.9	8
83	Genetic Analysis of Tryptophan Metabolism Genes in Sporadic Amyotrophic Lateral Sclerosis. <i>Frontiers in Immunology</i> , 2021, 12, 701550.	4.8	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. <i>American Journal of Medical Genetics Part A</i> , 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 CCFN familial amyotrophic lateral sclerosis patient using mRNA reprogramming. <i>Stem Cell Research</i> , 2019, 40, 101530.	0.7	6
86	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2021, 101, 297.e9-297.e11.	3.1	6
87	Exclusion of NFIL3 as the gene causing hereditary sensory neuropathy type I by mutation analysis. <i>Human Genetics</i> , 2000, 106, 594-596.	3.8	5
88	Genome screen of 15 Australian bipolar affective disorder pedigrees supports previously identified loci for bipolar susceptibility genes. <i>Psychiatric Genetics</i> , 2008, 18, 156-161.	1.1	5
89	Association analysis of transcripts from the bipolar susceptibility locus on chromosome 4q35, exclusion of a pathogenic role for eight positional candidate genes. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2005, 134B, 56-59.	1.7	4
90	The mRNA-based reprogramming of fibroblasts from a SOD1E101G familial amyotrophic lateral sclerosis patient to induced pluripotent stem cell line UOWi007. <i>Stem Cell Research</i> , 2020, 42, 101701.	0.7	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.	1.7	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.	1.1	2
94	Theme 13 Clinical management and support. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 327-347.	1.7	2
95	Postnatal Development of Spasticity Following Transgene Insertion in the Mouse β IV Spectrin Gene (SPTBN4). Journal of Neuromuscular Diseases, 2017, 4, 159-164.	2.6	1
96	Theme 3 In vitro experimental models. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 135-159.	1.7	1
97	NEK1 and STMN2 short tandem repeat lengths are not associated with Australian amyotrophic lateral sclerosis risk. Neurobiology of Aging, 2022, , .	3.1	0