Elizabeth M C Fisher

List of Publications by Year
in descending order

225 papers	22,179 citations	12330 69 h-index	10158 140 g-index
303 all docs	303 docs citations	303 times ranked	25234 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	The sex-determining region of the human Y chromosome encodes a finger protein. Cell, 1987, 51, 1091-1104.	28.9	881
3	Genealogies of mouse inbred strains. Nature Genetics, 2000, 24, 23-25.	21.4	769
4	Mutations in the endosomal ESCRTIII-complex subunit CHMP2B in frontotemporal dementia. Nature Genetics, 2005, 37, 806-808.	21.4	752
5	Behavioral and functional analysis of mouse phenotype: SHIRPA, a proposed protocol for comprehensive phenotype assessment. Mammalian Genome, 1997, 8, 711-713.	2.2	721
6	A systematic, genome-wide, phenotype-driven mutagenesis programme for gene function studies in the mouse. Nature Genetics, 2000, 25, 440-443.	21.4	657
7	Mutations in Dynein Link Motor Neuron Degeneration to Defects in Retrograde Transport. Science, 2003, 300, 808-812.	12.6	652
8	C9orf72 repeat expansions cause neurodegeneration in Drosophila through arginine-rich proteins. Science, 2014, 345, 1192-1194.	12.6	632
9	Functional multivesicular bodies are required for autophagic clearance of protein aggregates associated with neurodegenerative disease. Journal of Cell Biology, 2007, 179, 485-500.	5.2	559
10	Mutation of Celsr1 Disrupts Planar Polarity of Inner Ear Hair Cells and Causes Severe Neural Tube Defects in the Mouse. Current Biology, 2003, 13, 1129-1133.	3.9	552
11	Homologous ribosomal protein genes on the human X and Y chromosomes: Escape from X inactivation and possible implications for turner syndrome. Cell, 1990, 63, 1205-1218.	28.9	414
12	A genetic cause of Alzheimer disease: mechanistic insights from Down syndrome. Nature Reviews Neuroscience, 2015, 16, 564-574.	10.2	404
13	An Aneuploid Mouse Strain Carrying Human Chromosome 21 with Down Syndrome Phenotypes. Science, 2005, 309, 2033-2037.	12.6	390
14	A Genome-Wide Investigation of SNPs and CNVs in Schizophrenia. PLoS Genetics, 2009, 5, e1000373.	3.5	383
15	Balancing Selection at the Prion Protein Gene Consistent with Prehistoric Kurulike Epidemics. Science, 2003, 300, 640-643.	12.6	347
16	Molecular mapping of alzheimerâ€type dementia in Down's syndrome. Annals of Neurology, 1998, 43, 380-383.	5.3	334
17	The origins and uses of mouse outbred stocks. Nature Genetics, 2005, 37, 1181-1186.	21.4	316
18	Genetic Analysis of the Cytoplasmic Dynein Subunit Families. PLoS Genetics, 2006, 2, e1.	3.5	276

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19	C9orf72 hexanucleotide repeat associated with amyotrophic lateral sclerosis and frontotemporal dementia forms RNA G-quadruplexes. Scientific Reports, 2012, 2, 1016.	3.3	275
20	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	3.5	269
21	Species-Specific Transcription in Mice Carrying Human Chromosome 21. Science, 2008, 322, 434-438.	12.6	260
22	Is SOD1 loss of function involved in amyotrophic lateral sclerosis?. Brain, 2013, 136, 2342-2358.	7.6	237
23	A mutation in dynein rescues axonal transport defects and extends the life span of ALS mice. Journal of Cell Biology, 2005, 169, 561-567.	5.2	223
24	FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. Acta Neuropathologica, 2010, 120, 33-41.	7.7	222
25	Evidence that a locus for familial psoriasis maps to chromosome 4q. Nature Genetics, 1996, 14, 231-233.	21.4	203
26	Down syndrome--recent progress and future prospects. Human Molecular Genetics, 2009, 18, R75-R83.	2.9	199
27	Human haploinsufficiency â€" one for sorrow, two for joy. Nature Genetics, 1994, 7, 5-7.	21.4	193
28	TDP-43 loss and ALS-risk SNPs drive mis-splicing and depletion of UNC13A. Nature, 2022, 603, 131-137.	27.8	188
29	A point mutation in TRPC3 causes abnormal Purkinje cell development and cerebellar ataxia in moonwalker mice. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 6706-6711.	7.1	187
30	Application of neurite orientation dispersion and density imaging (NODDI) to a tau pathology model of Alzheimer's disease. NeuroImage, 2016, 125, 739-744.	4.2	179
31	Turner syndrome: the case of the missing sex chromosome. Trends in Genetics, 1993, 9, 90-93.	6.7	176
32	Identification of multiple quantitative trait loci linked to prion disease incubation period in mice. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 6279-6283.	7.1	176
33	Cytoplasmic dynein nomenclature. Journal of Cell Biology, 2005, 171, 411-413.	5.2	171
34	SHIRPA, a protocol for behavioral assessment: validation for longitudinal study of neurological dysfunction in mice. Neuroscience Letters, 2001, 306, 89-92.	2.1	169
35	Disruption of endocytic trafficking in frontotemporal dementia with CHMP2B mutations. Human Molecular Genetics, 2010, 19, 2228-2238.	2.9	163
36	Species-specific pace of development is associated with differences in protein stability. Science, 2020, 369, .	12.6	163

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37	Paralogy Mapping: Identification of a Region in the Human MHC Triplicated onto Human Chromosomes 1 and 9 Allows the Prediction and Isolation of NovelPBXandNOTCHLoci. Genomics, 1996, 35, 101-108.	2.9	161
38	The importance of understanding individual differences in Down syndrome. F1000Research, 2016, 5, 389.	1.6	151
39	Rodent models in Down syndrome research: impact and future opportunities. DMM Disease Models and Mechanisms, 2017, 10, 1165-1186.	2.4	149
40	DYRK1A-Dosage Imbalance Perturbs NRSF/REST Levels, Deregulating Pluripotency and Embryonic Stem Cell Fate in Down Syndrome. American Journal of Human Genetics, 2008, 83, 388-400.	6.2	139
41	Rodent models of amyotrophic lateral sclerosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1421-1436.	3.8	137
42	CHMP2B C-truncating mutations in frontotemporal lobar degeneration are associated with an aberrant endosomal phenotype in vitro. Human Molecular Genetics, 2008, 17, 313-322.	2.9	131
43	Mice with endogenous <scp>TDP</scp> â€43 mutations exhibit gain of splicing function and characteristics of amyotrophic lateral sclerosis. EMBO Journal, 2018, 37, .	7.8	129
44	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. Acta Neuropathologica, 2013, 126, 401-409.	7.7	126
45	Tumour angiogenesis is reduced in the Tc1 mouse model of Downâ€™s syndrome. Nature, 2010, 465, 813-817.	27.8	122
46	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	8.2	117
47	SOD1 and TDP-43 animal models of amyotrophic lateral sclerosis: recent advances in understanding disease toward the development of clinical treatments. Mammalian Genome, 2011, 22, 420-448.	2.2	113
48	SOD1 Function and Its Implications for Amyotrophic Lateral Sclerosis Pathology. Neuroscientist, 2015, 21, 519-529.	3.5	113
49	Superoxide Dismutase 1 and tgSOD1G93A Mouse Spinal Cord Seed Fibrils, Suggesting a Propagative Cell Death Mechanism in Amyotrophic Lateral Sclerosis. PLoS ONE, 2010, 5, e10627.	2.5	113
50	Cytoplasmic dynein heavy chain: the servant of many masters. Trends in Neurosciences, 2013, 36, 641-651.	8.6	111
51	Association of Dementia With Mortality Among Adults With Down Syndrome Older Than 35 Years. JAMA Neurology, 2019, 76, 152.	9.0	110
52	Implementation of a large-scale ENU mutagenesis program: towards increasing the mouse mutant resource. Mammalian Genome, 2000, 11, 500-506.	2.2	109
53	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
54	Down syndrome: searching for the genetic culprits. DMM Disease Models and Mechanisms, 2011, 4, 586-595.	2.4	106

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55	Progressive neuronal inclusion formation and axonal degeneration in CHMP2B mutant transgenic mice. Brain, 2012, 135, 819-832.	7.6	97
56	Trisomy of human chromosome 21 enhances amyloid-Î² deposition independently of an extra copy ofAPP. Brain, 2018, 141, 2457-2474.	7.6	96
57	Sporadicâ€"but Not Variantâ€"Creutzfeldt-Jakob Disease Is Associated with Polymorphisms Upstream of PRNP Exon 1. American Journal of Human Genetics, 2001, 69, 1225-1235.	6.2	95
58	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	7.6	95
59	Preservation of long-term memory and synaptic plasticity despite short-term impairments in the Tc1 mouse model of Down syndrome. Learning and Memory, 2008, 15, 492-500.	1.3	94
60	Additional deletion in sex-determining region of human Y chromosome resolves paradox of X,t(Y;22) female. Nature, 1990, 346, 279-281.	27.8	93
61	Massively Parallel Sequencing Reveals the Complex Structure of an Irradiated Human Chromosome on a Mouse Background in the Tc1 Model of Down Syndrome. PLoS ONE, 2013, 8, e60482.	2.5	93
62	An ENU-induced mutation in mouse glycyl-tRNA synthetase (GARS) causes peripheral sensory and motor phenotypes creating a model of Charcot-Marie-Tooth type 2D peripheral neuropathy. DMM Disease Models and Mechanisms, 2009, 2, 359-373.	2.4	91
63	The Frequency and Position of Alu Repeats in cDNAs, as Determined by Database Searching. Genomics, 1995, 27, 544-548.	2.9	86
64	A comprehensive assessment of the SOD1G93A low-copy transgenic mouse, which models human amyotrophic lateral sclerosis. DMM Disease Models and Mechanisms, 2011, 4, 686-700.	2.4	86
65	Frontotemporal Dementia Caused by CHMP2B Mutations. Current Alzheimer Research, 2011, 8, 246-251.	1.4	85
66	Impairments in motor coordination without major changes in cerebellar plasticity in the Tc1 mouse model of Down syndrome. Human Molecular Genetics, 2009, 18, 1449-1463.	2.9	80
67	Genomically humanized mice: technologies and promises. Nature Reviews Genetics, 2012, 13, 14-20.	16.3	80
68	Humanising the mouse genome piece by piece. Nature Communications, 2019, 10, 1845.	12.8	78
69	Genetic dissection of Down syndrome-associated congenital heart defects using a new mouse mapping panel. ELife, 2016, 5, .	6.0	77
70	Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology, 2014, 82, 2077-2084.	1.1	76
71	The mapping of a cDNA from the human X-linked Duchenne muscular dystrophy gene to the mouse X chromosome. Nature, 1987, 328, 166-168.	27.8	71
72	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	14.5	70

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73	Identification and mapping of a novel human gene, HRMT1L1, homologous to the rat protein arginine N-methyltransferase 1 (PRMT1) gene. Mammalian Genome, 1997, 8, 526-529.	2.2	68
74	Quiet mutations in inbred strains of mice. Trends in Molecular Medicine, 2007, 13, 512-519.	6.7	68
75	Down syndrome genetics: unravelling a multifactorial disorder. Human Molecular Genetics, 1996, 5, 1411-1416.	2.9	66
76	Transgenic and physiological mouse models give insights into different aspects of amyotrophic lateral sclerosis. DMM Disease Models and Mechanisms, 2019, 12, .	2.4	65
77	Perturbed hematopoiesis in the Tc1 mouse model of Down syndrome. Blood, 2010, 115, 2928-2937.	1.4	64
78	TDP-43 is a culprit in human neurodegeneration, and not just an innocent bystander. Mammalian Genome, 2008, 19, 299-305.	2.2	63
79	Modification of Superoxide Dismutase 1 (SOD1) Properties by a GFP Tag â€“ Implications for Research into Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2010, 5, e9541.	2.5	63
80	Microdissection and microcloning of the mouse X chromosome.. Proceedings of the National Academy of Sciences of the United States of America, 1985, 82, 5846-5849.	7.1	62
81	An improved protocol for the analysis of SOD1 gene mutations, and a new mutation in exon 4. Human Molecular Genetics, 1995, 4, 1101-1104.	2.9	62
82	Novel phenotypes identified by plasma biochemical screening in the mouse. Mammalian Genome, 2002, 13, 595-602.	2.2	62
83	A Novel C-Terminal Binding Protein (CTBP2) Is Closely Related toCTBP1,an Adenovirus E1A-Binding Protein, and Maps to Human Chromosome 21q21.3. Genomics, 1998, 47, 294-299.	2.9	61
84	Automatic Structural Parcellation of Mouse Brain MRI Using Multi-Atlas Label Fusion. PLoS ONE, 2014, 9, e86576.	2.5	60
85	The Grb2 binding domain of mSos1 is not required for downstream signal transduction. Nature Genetics, 1995, 10, 294-300.	21.4	59
86	Examination of the human prion protein-like gene Doppel for genetic susceptibility to sporadic and variant Creutzfeldtâ€“Jakob disease. Neuroscience Letters, 2000, 290, 117-120.	2.1	59
87	Identification of genetic loci affecting mouse-adapted bovine spongiform encephalopathy incubation time in mice. Neurogenetics, 2002, 4, 77-81.	1.4	58
88	Identification of two new Pmp22 mouse mutants using large-scale mutagenesis and a novel rapid mapping strategy. Human Molecular Genetics, 2000, 9, 1865-1871.	2.9	56
89	A Motor-Driven Mechanism for Cell-Length Sensing. Cell Reports, 2012, 1, 608-616.	6.4	55
90	Altered regulation of tau phosphorylation in a mouse model of down syndrome aging. Neurobiology of Aging, 2012, 33, 828.e31-828.e44.	3.1	54

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91	Transchromosomal Mouse Embryonic Stem Cell Lines and Chimeric Mice That Contain Freely Segregating Segments of Human Chromosome 21. Human Molecular Genetics, 1999, 8, 923-933.	2.9	53
92	Microcell-mediated chromosome transfer (MMCT): small cells with huge potential. Mammalian Genome, 2003, 14, 583-592.	2.2	52
93	A novel SOD1-ALS mutation separates central and peripheral effects of mutant SOD1 toxicity. Human Molecular Genetics, 2015, 24, 1883-1897.	2.9	52
94	Down's syndrome-like cardiac developmental defects in embryos of the transchromosomic Tc1 mouse. Cardiovascular Research, 2010, 88, 287-295.	3.8	51
95	Mouse models of neurodegeneration: Know your question, know your mouse. Science Translational Medicine, 2019, 11, .	12.4	51
96	Mice Carrying ALS Mutant TDP-43, but Not Mutant FUS, Display InÂVivo Defects in Axonal Transport of Signaling Endosomes. Cell Reports, 2020, 30, 3655-3662.e2.	6.4	51
97	Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the C9orf72 expansion. Neurobiology of Aging, 2015, 36, 546.e1-546.e7.	3.1	48
98	Mouse autosomal trisomy: twoâ€™s company, threeâ€™s a crowd. Trends in Genetics, 1999, 15, 241-247.	6.7	47
99	Molecular Genetic Characterisation of Frontotemporal Dementia on Chromosome 3. Dementia and Geriatric Cognitive Disorders, 1999, 10, 93-101.	1.5	44
100	Mutations in the Gabrb1 gene promote alcohol consumption through increased tonic inhibition. Nature Communications, 2013, 4, 2816.	12.8	44
101	Protein profiles in Tc1 mice implicate novel pathway perturbations in the Down syndrome brain. Human Molecular Genetics, 2013, 22, 1709-1724.	2.9	43
102	Imaging the accumulation and suppression of tau pathology using multiparametric MRI. Neurobiology of Aging, 2016, 39, 184-194.	3.1	42
103	Evidence for evolutionary divergence of activity-dependent gene expression in developing neurons. ELife, 2016, 5, .	6.0	42
104	Mouse models for neurological disease. Lancet Neurology, The, 2002, 1, 215-224.	10.2	41
105	The phagocytic capacity of neurones. European Journal of Neuroscience, 2007, 25, 2947-2955.	2.6	41
106	Dissecting Alzheimer disease in Down syndrome using mouse models. Frontiers in Behavioral Neuroscience, 2015, 9, 268.	2.0	41
107	Alterations to Dendritic Spine Morphology, but Not Dendrite Patterning, of Cortical Projection Neurons in Tc1 and Ts1Rhr Mouse Models of Down Syndrome. PLoS ONE, 2013, 8, e78561.	2.5	39
108	A Syntenic Cross Species Aneuploidy Genetic Screen Links RCAN1 Expression to Î²-Cell Mitochondrial Dysfunction in Type 2 Diabetes. PLoS Genetics, 2016, 12, e1006033.	3.5	39

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109	Comparison of In Vivo and Ex Vivo MRI for the Detection of Structural Abnormalities in a Mouse Model of Tauopathy. Frontiers in Neuroinformatics, 2017, 11, 20.	2.5	37
110	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	3.1	36
111	FUS-ALS mutants alter FMRP phase separation equilibrium and impair protein translation. Science Advances, 2021, 7, .	10.3	36
112	Mice, the Motor System, and Human Motor Neuron Pathology. Mammalian Genome, 2000, 11, 1041-1052.	2.2	35
113	Mouse Cytoplasmic Dynein Intermediate Chains: Identification of New Isoforms, Alternative Splicing and Tissue Distribution of Transcripts. PLoS ONE, 2010, 5, e11682.	2.5	35
114	Novel Mouse Model of Autosomal Semidominant Adult Hypophosphatasia Has a Splice Site Mutation in the Tissue Nonspecific Alkaline Phosphatase Gene Akp2. Journal of Bone and Mineral Research, 2007, 22, 1397-1407.	2.8	34
115	The telomeric part of the human chromosome 21 from Cstb to Prmt2 is not necessary for the locomotor and short-term memory deficits observed in the Tc1 mouse model of Down syndrome. Behavioural Brain Research, 2011, 217, 271-281.	2.2	34
116	Large-scale pathways-based association study in amyotrophic lateral sclerosis. Brain, 2007, 130, 2292-2301.	7.6	32
117	Hippocampal circuit dysfunction in the Tc1 mouse model of Down syndrome. Nature Neuroscience, 2015, 18, 1291-1298.	14.8	32
118	Altered Hippocampal-Prefrontal Neural Dynamics in Mouse Models of Down Syndrome. Cell Reports, 2020, 30, 1152-1163.e4.	6.4	32
119	Intracerebral haemorrhage in Down syndrome: protected or predisposed?. F1000Research, 2016, 5, 876.	1.6	30
120	Analysis of motor dysfunction in Down Syndrome reveals motor neuron degeneration. PLoS Genetics, 2018, 14, e1007383.	3.5	29
121	Structural correlates of active-staining following magnetic resonance microscopy in the mouse brain. NeuroImage, 2011, 56, 974-983.	4.2	28
122	Identification, Expression, and Chromosomal Localization of Ubiquitin Conjugating Enzyme 7 (UBE2G2), a Human Homologue of theSaccharomyces cerevisiae Ubc7Gene. Genomics, 1998, 51, 128-131.	2.9	27
123	Cytoplasmic dynein could be key to understanding neurodegeneration. Genome Biology, 2008, 9, 214.	9.6	27
124	Neurodegenerative Mutation in Cytoplasmic Dynein Alters Its Organization and Dynein-Dynactin and Dynein-Kinesin Interactions*. Journal of Biological Chemistry, 2010, 285, 39922-39934.	3.4	27
125	No association with common Caucasian genotypes in exons 8, 13 and 14 of the human cytoplasmic dynein heavy chain gene (DNCHC1) and familial motor neuron disorders. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 150-157.	1.2	26
126	Identification and characterization of a novel mouse prion gene allele. Mammalian Genome, 2004, 15, 383-389.	2.2	26

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127	A landmark-free morphometrics pipeline for high-resolution phenotyping: application to a mouse model of Down syndrome. Development (Cambridge), 2021, 148, .	2.5	26
128	Quantitative Proteomics Characterization of a Mouse Embryonic Stem Cell Model of Down Syndrome. Molecular and Cellular Proteomics, 2009, 8, 585-595.	3.8	25
129	Increased Cerebral Vascular Reactivity in the Tau Expressing rTg4510 Mouse: Evidence against the Role of Tau Pathology to Impair Vascular Health in Alzheimer's Disease. Journal of Cerebral Blood Flow and Metabolism, 2015, 35, 359-362.	4.3	25
130	Mapping the Gene That Encodes Phosphatidylinositol-Specific Phospholipase C-Î³2 in the Human and the Mouse. Genomics, 1994, 23, 504-507.	2.9	23
131	Mighty mice. Nature, 2000, 404, 815-815.	27.8	23
132	The Legs at odd angles (Loa) Mutation in Cytoplasmic Dynein Ameliorates Mitochondrial Function in SOD1G93A Mouse Model for Motor Neuron Disease. Journal of Biological Chemistry, 2010, 285, 18627-18639.	3.4	23
133	Behavioral and Other Phenotypes in a Cytoplasmic Dynein Light Intermediate Chain 1 Mutant Mouse. Journal of Neuroscience, 2011, 31, 5483-5494.	3.6	23
134	Uses for humanised mouse models in precision medicine for neurodegenerative disease. Mammalian Genome, 2019, 30, 173-191.	2.2	22
135	Overexpression of the Hspa13 (Stch) gene reduces prion disease incubation time in mice. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 13722-13727.	7.1	21
136	DYNC1H1 mutation alters transport kinetics and ERK1/2-cFos signalling in a mouse model of distal spinal muscular atrophy. Brain, 2014, 137, 1883-1893.	7.6	21
137	ALS-related FUS mutations alter axon growth in motoneurons and affect HuD/ELAVL4 and FMRP activity. Communications Biology, 2021, 4, 1025.	4.4	21
138	Mapping GRB2, a Signal Transduction Gene in the Human and the Mouse. Genomics, 1994, 22, 313-318.	2.9	20
139	Paradigms for the identification of new genes in motor neuron degeneration. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 249-257.	1.2	20
140	Downregulated Wnt/Î²-catenin signalling in the Down syndrome hippocampus. Scientific Reports, 2019, 9, 7322.	3.3	20
141	Human sex-chromosome-specific repeats within a region of pseudoautosomal/Yq homology. Genomics, 1990, 7, 625-628.	2.9	19
142	The SOD1 transgene in the G93A mouse model of amyotrophic lateral sclerosis lies on distal mouse chromosome 12. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2005, 6, 111-114.	2.1	19
143	Cognitive impairment in the preclinical stage of dementia in FTD-3CHMP2Bmutation carriers: a longitudinal prospective study. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 170-176.	1.9	19
144	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. Human Molecular Genetics, 2016, 25, 291-307.	2.9	19

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145	Gene expression dysregulation domains are not a specific feature of Down syndrome. Nature Communications, 2019, 10, 2489.	12.8	19
146	Interaction of sexual dimorphism and gene dosage imbalance in skeletal deficits associated with Down syndrome. Bone, 2020, 136, 115367.	2.9	19
147	ENU Mutagenesis Reveals a Novel Phenotype of Reduced Limb Strength in Mice Lacking Fibrillin 2. PLoS ONE, 2010, 5, e9137.	2.5	19
148	Fully-Automated Î¼MRI Morphometric Phenotyping of the Tc1 Mouse Model of Down Syndrome. PLoS ONE, 2016, 11, e0162974.	2.5	19
149	Mapping TNNC1, the Gene That Encodes Cardiac Troponin I in the Human and the Mouse. Genomics, 1995, 30, 620-622.	2.9	18
150	Ain't misbehavin' - it's genetic!. Nature Genetics, 1996, 12, 115-116.	21.4	18
151	A Nonsense Mutation in Mouse Tardbp Affects TDP43 Alternative Splicing Activity and Causes Limb-Clasping and Body Tone Defects. PLoS ONE, 2014, 9, e85962.	2.5	18
152	Tc1 mouse model of trisomy-21 dissociates properties of short- and long-term recognition memory. Neurobiology of Learning and Memory, 2016, 130, 118-128.	1.9	18
153	Human glial cell line-derived neurotrophic factor (GDNF) maps to chromosome 5. Human Genetics, 1995, 96, 671-673.	3.8	17
154	Phenotypic analysis â€" making the most of your mouse. Trends in Genetics, 1997, 13, 254-256.	6.7	17
155	Towards a mutant map of the mouse ? new models of neurological, behavioural, deafness, bone, renal and blood disorders. Genetica, 2004, 122, 47-49.	1.1	17
156	An additional human chromosome 21 causes suppression of neural fate of pluripotent mouse embryonic stem cells in a teratoma model. BMC Developmental Biology, 2007, 7, 131.	2.1	17
157	Presymptomatic Generalized Brain Atrophy in Frontotemporal Dementia Caused by CHMP2B Mutation. Dementia and Geriatric Cognitive Disorders, 2009, 27, 182-186.	1.5	17
158	Profilin1 E117G is a moderate risk factor for amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 506-508.	1.9	17
159	Maternal iron deficiency perturbs embryonic cardiovascular development in mice. Nature Communications, 2021, 12, 3447.	12.8	17
160	Comprehensive phenotypic analysis of the Dp1Tyb mouse strain reveals a broad range of Down syndrome-related phenotypes. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	17
161	The DAD1 protein, whose defect causes apoptotic cell death, maps to human chromosome 14. Genomics, 1995, 26, 433-435.	2.9	16
162	5. The Contribution of the Mouse to Advances in Human Genetics. Advances in Genetics, 1997, 35, 155-205.	1.8	16

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163	Mouse models as a tool for understanding neurodegenerative diseases. Current Opinion in Neurology, 2003, 16, 451-458.	3.6	16
164	New approaches for modelling sporadic genetic disease in the mouse. DMM Disease Models and Mechanisms, 2009, 2, 446-453.	2.4	16
165	Sequencing analysis of the spinal bulbar muscular atrophy CAG expansion reveals absence of repeat interruptions. Neurobiology of Aging, 2014, 35, 443.e1-443.e3.	3.1	16
166	New techniques to understand chromosome dosage: mouse models of aneuploidy. Human Molecular Genetics, 2006, 15, R103-R109.	2.9	15
167	The integration site of the APP transgene in the J20 mouse model of Alzheimerâ€™s disease. Wellcome Open Research, 2017, 2, 84.	1.8	15
168	DYNLRB1 is essential for dynein mediated transport and neuronal survival. Neurobiology of Disease, 2020, 140, 104816.	4.4	15
169	The integration site of the APP transgene in the J20 mouse model of Alzheimerâ€™s disease. Wellcome Open Research, 2017, 2, 84.	1.8	15
170	Mutant Glycyl-tRNA Synthetase (Gars) Ameliorates SOD1G93A Motor Neuron Degeneration Phenotype but Has Little Affect on Loa Dynein Heavy Chain Mutant Mice. PLoS ONE, 2009, 4, e6218.	2.5	15
171	Characterisation and expression analysis of the WDR9 gene, located in the Down critical region-2 of the human chromosome 21. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2002, 1577, 377-383.	2.4	14
172	Prion disease incubation time is not affected in mice heterozygous for a dynein mutation. Biochemical and Biophysical Research Communications, 2004, 326, 18-22.	2.1	14
173	Sequencing analysis of the ITPR1 gene in a pure autosomal dominant spinocerebellar ataxia series. Movement Disorders, 2010, 25, 771-773.	3.9	14
174	Mouse Models of Aneuploidy. Scientific World Journal, The, 2012, 2012, 1-6.	2.1	14
175	Aging rather than aneuploidy affects monoamine neurotransmitters in brain regions of Down syndrome mouse models. Neurobiology of Disease, 2017, 105, 235-244.	4.4	14
176	Mouse models of aneuploidy to understand chromosome disorders. Mammalian Genome, 2022, 33, 157-168.	2.2	14
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