Elizabeth M C Fisher

List of Publications by Citations

224	17,278	64	128
papers	citations	h-index	g-index
303	20,032	9.7	6.01
ext. papers	ext. citations	avg, IF	L-index

#	Paper	IF	Citations
224	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A, tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
223	The sex-determining region of the human Y chromosome encodes a finger protein. *Cell*, **1987**, 51, 1091-1104	56.2	801
222	Genealogies of mouse inbred strains. *Nature Genetics*, **2000**, 24, 23-5	36.3	676
221	Mutations in the endosomal ESCRTIII-complex subunit CHMP2B in frontotemporal dementia. *Nature Genetics*, **2005**, 37, 806-8	36.3	648
220	Behavioral and functional analysis of mouse phenotype: SHIRPA, a proposed protocol for comprehensive phenotype assessment. *Mammalian Genome*, **1997**, 8, 711-3	3.2	633
219	A systematic, genome-wide, phenotype-driven mutagenesis programme for gene function studies in the mouse. *Nature Genetics*, **2000**, 25, 440-3	36.3	582
218	Mutations in dynein link motor neuron degeneration to defects in retrograde transport. *Science*, **2003**, 300, 808-12	33.3	577
217	Functional multivesicular bodies are required for autophagic clearance of protein aggregates associated with neurodegenerative disease. *Journal of Cell Biology*, **2007**, 179, 485-500	7.3	500
216	Mutation of Celsr1 disrupts planar polarity of inner ear hair cells and causes severe neural tube defects in the mouse. *Current Biology*, **2003**, 13, 1129-33	6.3	484
215	C9orf72 repeat expansions cause neurodegeneration in Drosophila through arginine-rich proteins. *Science*, **2014**, 345, 1192-1194	33.3	454
214	Homologous ribosomal protein genes on the human X and Y chromosomes: escape from X inactivation and possible implications for Turner syndrome. *Cell*, **1990**, 63, 1205-18	56.2	374
213	A genome-wide investigation of SNPs and CNVs in schizophrenia. *PLoS Genetics*, **2009**, 5, e1000373	6	357
212	An aneuploid mouse strain carrying human chromosome 21 with Down syndrome phenotypes. *Science*, **2005**, 309, 2033-7	33.3	324
211	A genetic cause of Alzheimer disease: mechanistic insights from Down syndrome. *Nature Reviews Neuroscience*, **2015**, 16, 564-74	13.5	301
210	Balancing selection at the prion protein gene consistent with prehistoric kurulike epidemics. *Science*, **2003**, 300, 640-3	33.3	287
209	Molecular mapping of Alzheimer-type dementia in Down's syndrome. *Annals of Neurology*, **1998**, 43, 380-3	9.4	275
208	The origins and uses of mouse outbred stocks. *Nature Genetics*, **2005**, 37, 1181-6	36.3	273

207	C9orf72 hexanucleotide repeat associated with amyotrophic lateral sclerosis and frontotemporal dementia forms RNA G-quadruplexes. *Scientific Reports*, **2012**, 2, 1016	4.9	234
206	Species-specific transcription in mice carrying human chromosome 21. *Science*, **2008**, 322, 434-8	33.3	231
205	Genetic analysis of the cytoplasmic dynein subunit families. *PLoS Genetics*, **2006**, 2, e1	6	231
204	Deletion at ITPR1 underlies ataxia in mice and spinocerebellar ataxia 15 in humans. *PLoS Genetics*, **2007**, 3, e108	6	221
203	FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. *Acta Neuropathologica*, **2010**, 120, 33-41	14.3	198
202	A mutation in dynein rescues axonal transport defects and extends the life span of ALS mice. *Journal of Cell Biology*, **2005**, 169, 561-7	7.3	198
201	Evidence that a locus for familial psoriasis maps to chromosome 4q. *Nature Genetics*, **1996**, 14, 231-3	36.3	182
200	Is SOD1 loss of function involved in amyotrophic lateral sclerosis?. *Brain*, **2013**, 136, 2342-58	11.2	178
199	Down syndrome--recent progress and future prospects. *Human Molecular Genetics*, **2009**, 18, R75-83	5.6	168
198	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, 6279-83	11.5	163
197	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, 6706-11	11.5	160
196	Turner syndrome: the case of the missing sex chromosome. *Trends in Genetics*, **1993**, 9, 90-3	8.5	157
195	Cytoplasmic dynein nomenclature. *Journal of Cell Biology*, **2005**, 171, 411-3	7.3	154
194	SHIRPA, a protocol for behavioral assessment: validation for longitudinal study of neurological dysfunction in mice. *Neuroscience Letters*, **2001**, 306, 89-92	3.3	154
193	Paralogy mapping: identification of a region in the human MHC triplicated onto human chromosomes 1 and 9 allows the prediction and isolation of novel PBX and NOTCH loci. *Genomics*, **1996**, 35, 101-8	4.3	144
192	Disruption of endocytic trafficking in frontotemporal dementia with CHMP2B mutations. *Human Molecular Genetics*, **2010**, 19, 2228-38	5.6	140
191	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	11	123
190	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. *Acta Neuropathologica*, **2013**, 126, 401-9	14.3	119

189	Rodent models of amyotrophic lateral sclerosis. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2013**, 1832, 1421-36	6.9	119
188	Application of neurite orientation dispersion and density imaging (NODDI) to a tau pathology model of Alzheimer's disease. *NeuroImage*, **2016**, 125, 739-744	7.9	118
187	CHMP2B C-truncating mutations in frontotemporal lobar degeneration are associated with an aberrant endosomal phenotype in vitro. *Human Molecular Genetics*, **2008**, 17, 313-22	5.6	112
186	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, 420-48	3.2	102
185	Tumour angiogenesis is reduced in the Tc1 mouse model of Down's syndrome. *Nature*, **2010**, 465, 813-7	50.4	101
184	Superoxide dismutase 1 and tgSOD1 mouse spinal cord seed fibrils, suggesting a propagative cell death mechanism in amyotrophic lateral sclerosis. *PLoS ONE*, **2010**, 5, e10627	3.7	100
183	Implementation of a large-scale ENU mutagenesis program: towards increasing the mouse mutant resource. *Mammalian Genome*, **2000**, 11, 500-6	3.2	99
182	The importance of understanding individual differences in Down syndrome. *F1000Research*, **2016**, 5,	3.6	98
181	Cytoplasmic dynein heavy chain: the servant of many masters. *Trends in Neurosciences*, **2013**, 36, 641-51	13.3	91
180	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2009**, 18, 1524-32	5.6	91
179	Rodent models in Down syndrome research: impact and future opportunities. *DMM Disease Models and Mechanisms*, **2017**, 10, 1165-1186	4.1	87
178	Sporadic--but not variant--Creutzfeldt-Jakob disease is associated with polymorphisms upstream of PRNP exon 1. *American Journal of Human Genetics*, **2001**, 69, 1225-35	11	86
177	Mice with endogenous TDP-43 mutations exhibit gain of splicing function and characteristics of amyotrophic lateral sclerosis. *EMBO Journal*, **2018**, 37,	13	84
176	Additional deletion in sex-determining region of human Y chromosome resolves paradox of X,t(Y;22) female. *Nature*, **1990**, 346, 279-81	50.4	83
175	The frequency and position of Alu repeats in cDNAs, as determined by database searching. *Genomics*, **1995**, 27, 544-8	4.3	82
174	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	2.8	81
173	Progressive neuronal inclusion formation and axonal degeneration in CHMP2B mutant transgenic mice. *Brain*, **2012**, 135, 819-32	11.2	80
172	Association of Dementia With Mortality Among Adults With Down Syndrome Older Than 35 Years. *JAMA Neurology*, **2019**, 76, 152-160	17.2	76

171	Down syndrome: searching for the genetic culprits. *DMM Disease Models and Mechanisms*, **2011**, 4, 586-95	4.1	74
170	Frontotemporal dementia caused by CHMP2B mutations. *Current Alzheimer Research*, **2011**, 8, 246-51	3	73
169	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in 'FUSDelta14' knockin mice. *Brain*, **2017**, 140, 2797-2805	11.2	70
168	SOD1 Function and Its Implications for Amyotrophic Lateral Sclerosis Pathology: New and Renascent Themes. *Neuroscientist*, **2015**, 21, 519-29	7.6	69
167	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	3.7	69
166	Genomically humanized mice: technologies and promises. *Nature Reviews Genetics*, **2011**, 13, 14-20	30.1	68
165	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	4.1	68
164	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, 359-73	4.1	67
163	Impairments in motor coordination without major changes in cerebellar plasticity in the Tc1 mouse model of Down syndrome. *Human Molecular Genetics*, **2009**, 18, 1449-63	5.6	65
162	Trisomy of human chromosome 21 enhances amyloid-β deposition independently of an extra copy of APP. *Brain*, **2018**, 141, 2457-2474	11.2	64
161	Identification and mapping of a novel human gene, HRMT1L1, homologous to the rat protein arginine N-methyltransferase 1 (PRMT1) gene. *Mammalian Genome*, **1997**, 8, 526-9	3.2	62
160	TDP-43 is a culprit in human neurodegeneration, and not just an innocent bystander. *Mammalian Genome*, **2008**, 19, 299-305	3.2	61
159	The mapping of a cDNA from the human X-linked Duchenne muscular dystrophy gene to the mouse X chromosome. *Nature*, **1987**, 328, 166-8	50.4	60
158	Quiet mutations in inbred strains of mice. *Trends in Molecular Medicine*, **2007**, 13, 512-9	11.5	59
157	The Grb2 binding domain of mSos1 is not required for downstream signal transduction. *Nature Genetics*, **1995**, 10, 294-300	36.3	58
156	An improved protocol for the analysis of SOD1 gene mutations, and a new mutation in exon 4. *Human Molecular Genetics*, **1995**, 4, 1101-4	5.6	57
155	A novel C-terminal binding protein (CTBP2) is closely related to CTBP1, an adenovirus E1A-binding protein, and maps to human chromosome 21q21.3. *Genomics*, **1998**, 47, 294-9	4.3	56
154	Down syndrome genetics: unravelling a multifactorial disorder. *Human Molecular Genetics*, **1996**, 5 Spec No, 1411-6	5.6	56

153	Species-specific pace of development is associated with differences in protein stability. *Science*, **2020**, 369,	33.3	56
152	Correlation of clinical and molecular features in spinal bulbar muscular atrophy. *Neurology*, **2014**, 82, 2077-84	6.5	55
151	Identification of genetic loci affecting mouse-adapted bovine spongiform encephalopathy incubation time in mice. *Neurogenetics*, **2002**, 4, 77-81	3	55
150	Novel phenotypes identified by plasma biochemical screening in the mouse. *Mammalian Genome*, **2002**, 13, 595-602	3.2	53
149	Microdissection and microcloning of the mouse X chromosome. *Proceedings of the National Academy of Sciences of the United States of America*, **1985**, 82, 5846-9	11.5	53
148	Modification of superoxide dismutase 1 (SOD1) properties by a GFP tag--implications for research into amyotrophic lateral sclerosis (ALS). *PLoS ONE*, **2010**, 5, e9541	3.7	52
147	Transchromosomal mouse embryonic stem cell lines and chimeric mice that contain freely segregating segments of human chromosome 21. *Human Molecular Genetics*, **1999**, 8, 923-33	5.6	49
146	Genetic dissection of Down syndrome-associated congenital heart defects using a new mouse mapping panel. *ELife*, **2016**, 5,	8.9	48
145	Down's syndrome-like cardiac developmental defects in embryos of the transchromosomic Tc1 mouse. *Cardiovascular Research*, **2010**, 88, 287-95	9.9	47
144	Identification of two new Pmp22 mouse mutants using large-scale mutagenesis and a novel rapid mapping strategy. *Human Molecular Genetics*, **2000**, 9, 1865-71	5.6	47
143	Examination of the human prion protein-like gene doppel for genetic susceptibility to sporadic and variant Creutzfeldt-Jakob disease. *Neuroscience Letters*, **2000**, 290, 117-20	3.3	47
142	Perturbed hematopoiesis in the Tc1 mouse model of Down syndrome. *Blood*, **2010**, 115, 2928-37	2.2	46
141	Microcell-mediated chromosome transfer (MMCT): small cells with huge potential. *Mammalian Genome*, **2003**, 14, 583-92	3.2	44
140	Mouse autosomal trisomy: two's company, three's a crowd. *Trends in Genetics*, **1999**, 15, 241-7	8.5	42
139	Transgenic and physiological mouse models give insights into different aspects of amyotrophic lateral sclerosis. *DMM Disease Models and Mechanisms*, **2019**, 12,	4.1	41
138	Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the C9orf72 expansion. *Neurobiology of Aging*, **2015**, 36, 546.e1-7	5.6	41
137	Automatic structural parcellation of mouse brain MRI using multi-atlas label fusion. *PLoS ONE*, **2014**, 9, e86576	3.7	41
136	A motor-driven mechanism for cell-length sensing. *Cell Reports*, **2012**, 1, 608-16	10.6	41

135	Altered regulation of tau phosphorylation in a mouse model of down syndrome aging. *Neurobiology of Aging*, **2012**, 33, 828.e31-44	5.6	39
134	Humanising the mouse genome piece by piece. *Nature Communications*, **2019**, 10, 1845	17.4	37
133	Mutations in the Gabrb1 gene promote alcohol consumption through increased tonic inhibition. *Nature Communications*, **2013**, 4, 2816	17.4	37
132	A novel SOD1-ALS mutation separates central and peripheral effects of mutant SOD1 toxicity. *Human Molecular Genetics*, **2015**, 24, 1883-97	5.6	37
131	Protein profiles in Tc1 mice implicate novel pathway perturbations in the Down syndrome brain. *Human Molecular Genetics*, **2013**, 22, 1709-24	5.6	36
130	Imaging the accumulation and suppression of tau pathology using multiparametric MRI. *Neurobiology of Aging*, **2016**, 39, 184-94	5.6	35
129	Mouse cytoplasmic dynein intermediate chains: identification of new isoforms, alternative splicing and tissue distribution of transcripts. *PLoS ONE*, **2010**, 5, e11682	3.7	34
128	Mouse models for neurological disease. *Lancet Neurology, The*, **2002**, 1, 215-24	24.1	34
127	Molecular genetic characterisation of frontotemporal dementia on chromosome 3. *Dementia and Geriatric Cognitive Disorders*, **1999**, 10 Suppl 1, 93-101	2.6	34
126	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. *Journal of Clinical Investigation*, **2020**, 130, 6080-6092	15.9	34
125	Mice, the motor system, and human motor neuron pathology. *Mammalian Genome*, **2000**, 11, 1041-52	3.2	33
124	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	3.7	31
123	The phagocytic capacity of neurones. *European Journal of Neuroscience*, **2007**, 25, 2947-55	3.5	30
122	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, 1397-407	6.3	30
121	A Syntenic Cross Species Aneuploidy Genetic Screen Links RCAN1 Expression to β Cell Mitochondrial Dysfunction in Type 2 Diabetes. *PLoS Genetics*, **2016**, 12, e1006033	6	30
120	Dissecting Alzheimer disease in Down syndrome using mouse models. *Frontiers in Behavioral Neuroscience*, **2015**, 9, 268	3.5	29
119	Large-scale pathways-based association study in amyotrophic lateral sclerosis. *Brain*, **2007**, 130, 2292-301	11.2	29
118	Mouse models of neurodegeneration: Know your question, know your mouse. *Science Translational Medicine*, **2019**, 11,	17.5	27

117	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, 271-81	3.4	27
116	Identification, expression, and chromosomal localization of ubiquitin conjugating enzyme 7 (UBE2G2), a human homologue of the Saccharomyces cerevisiae ubc7 gene. *Genomics*, **1998**, 51, 128-31	4.3	27
115	Hippocampal circuit dysfunction in the Tc1 mouse model of Down syndrome. *Nature Neuroscience*, **2015**, 18, 1291-1298	25.5	26
114	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. *Neurobiology of Aging*, **2014**, 35, 1491-8	5.6	25
113	Structural correlates of active-staining following magnetic resonance microscopy in the mouse brain. *NeuroImage*, **2011**, 56, 974-83	7.9	25
112	Cytoplasmic dynein could be key to understanding neurodegeneration. *Genome Biology*, **2008**, 9, 214	18.3	25
111	Intracerebral haemorrhage in Down syndrome: protected or predisposed?. *F1000Research*, **2016**, 5,	3.6	25
110	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. *Nucleic Acids Research*, **2020**, 48, 6889-6905	20.1	24
109	Neurodegenerative mutation in cytoplasmic dynein alters its organization and dynein-dynactin and dynein-kinesin interactions. *Journal of Biological Chemistry*, **2010**, 285, 39922-34	5.4	23
108	Comparison of and MRI for the Detection of Structural Abnormalities in a Mouse Model of Tauopathy. *Frontiers in Neuroinformatics*, **2017**, 11, 20	3.9	22
107	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, 150-7		22
106	Identification and characterization of a novel mouse prion gene allele. *Mammalian Genome*, **2004**, 15, 383-9	3.2	22
105	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, 359-62	7.3	21
104	Mice Carrying ALS Mutant TDP-43, but Not Mutant FUS, Display In Vivo Defects in Axonal Transport of Signaling Endosomes. *Cell Reports*, **2020**, 30, 3655-3662.e2	10.6	21
103	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, 18627-39	5.4	20
102	Evidence for evolutionary divergence of activity-dependent gene expression in developing neurons. *ELife*, **2016**, 5,	8.9	20
101	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, 111-4		19
100	Quantitative proteomics characterization of a mouse embryonic stem cell model of Down syndrome. *Molecular and Cellular Proteomics*, **2009**, 8, 585-95	7.6	18

99	Mighty mice. *Nature*, **2000**, 404, 815	50.4	17
98	Human sex-chromosome-specific repeats within a region of pseudoautosomal/Yq homology. *Genomics*, **1990**, 7, 625-8	4.3	17
97	ENU mutagenesis reveals a novel phenotype of reduced limb strength in mice lacking fibrillin 2. *PLoS ONE*, **2010**, 5, e9137	3.7	17
96	Profilin1 E117G is a moderate risk factor for amyotrophic lateral sclerosis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 506-8	5.5	16
95	Behavioral and other phenotypes in a cytoplasmic Dynein light intermediate chain 1 mutant mouse. *Journal of Neuroscience*, **2011**, 31, 5483-94	6.6	16
94	Towards a mutant map of the mouse--new models of neurological, behavioural, deafness, bone, renal and blood disorders. *Genetica*, **2004**, 122, 47-9	1.5	16
93	The DAD1 protein, whose defect causes apoptotic cell death, maps to human chromosome 14. *Genomics*, **1995**, 26, 433-5	4.3	16
92	Mapping GRB2, a signal transduction gene in the human and the mouse. *Genomics*, **1994**, 22, 313-8	4.3	16
91	Mapping the gene that encodes phosphatidylinositol-specific phospholipase C-gamma 2 in the human and the mouse. *Genomics*, **1994**, 23, 504-7	4.3	16
90	Tc1 mouse model of trisomy-21 dissociates properties of short- and long-term recognition memory. *Neurobiology of Learning and Memory*, **2016**, 130, 118-28	3.1	15
89	DYNC1H1 mutation alters transport kinetics and ERK1/2-cFos signalling in a mouse model of distal spinal muscular atrophy. *Brain*, **2014**, 137, 1883-93	11.2	15
88	Cognitive impairment in the preclinical stage of dementia in FTD-3 CHMP2B mutation carriers: a longitudinal prospective study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 170-6	5.5	15
87	A nonsense mutation in mouse Tardbp affects TDP43 alternative splicing activity and causes limb-clasping and body tone defects. *PLoS ONE*, **2014**, 9, e85962	3.7	15
86	Presymptomatic generalized brain atrophy in frontotemporal dementia caused by CHMP2B mutation. *Dementia and Geriatric Cognitive Disorders*, **2009**, 27, 182-6	2.6	15
85	Phenotypic analysis--making the most of your mouse. *Trends in Genetics*, **1997**, 13, 254-6	8.5	15
84	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, 249-57		15
83	Mutant glycyl-tRNA synthetase (Gars) ameliorates SOD1(G93A) motor neuron degeneration phenotype but has little affect on Loa dynein heavy chain mutant mice. *PLoS ONE*, **2009**, 4, e6218	3.7	15
82	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, 13722-7	11.5	14

81	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	3.1	14
80	New techniques to understand chromosome dosage: mouse models of aneuploidy. *Human Molecular Genetics*, **2006**, 15 Spec No 2, R103-9	5.6	14
79	Human glial cell line-derived neurotrophic factor (GDNF) maps to chromosome 5. *Human Genetics*, **1995**, 96, 671-3	6.3	14
78	TDP-43 loss and ALS-risk SNPs drive mis-splicing and depletion of UNC13A.. *Nature*, **2022**, 603, 131-137	50.4	14
77	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. *Human Molecular Genetics*, **2016**, 25, 291-307	5.6	13
76	New approaches for modelling sporadic genetic disease in the mouse. *DMM Disease Models and Mechanisms*, **2009**, 2, 446-53	4.1	13
75	Sequencing analysis of the ITPR1 gene in a pure autosomal dominant spinocerebellar ataxia series. *Movement Disorders*, **2010**, 25, 771-3	7	13
74	Mapping TNNC1, the gene that encodes cardiac troponin I in the human and the mouse. *Genomics*, **1995**, 30, 620-2	4.3	13
73	Sequencing analysis of the spinal bulbar muscular atrophy CAG expansion reveals absence of repeat interruptions. *Neurobiology of Aging*, **2014**, 35, 443.e1-3	5.6	12
72	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, 377-83		12
71	Prion disease incubation time is not affected in mice heterozygous for a dynein mutation. *Biochemical and Biophysical Research Communications*, **2005**, 326, 18-22	3.4	12
70	A novel phenotype for the dynein heavy chain mutation Loa: altered dendritic morphology, organelle density, and reduced numbers of trigeminal motoneurons. *Journal of Comparative Neurology*, **2012**, 520, 2757-73	3.4	11
69	The contribution of the mouse to advances in human genetics. *Advances in Genetics*, **1997**, 35, 155-205	3.3	11
68	The integration site of the APP transgene in the J20 mouse model of Alzheimer's disease. *Wellcome Open Research*, **2017**, 2, 84	4.8	11
67	Aging rather than aneuploidy affects monoamine neurotransmitters in brain regions of Down syndrome mouse models. *Neurobiology of Disease*, **2017**, 105, 235-244	7.5	10
66	The integration site of the transgene in the J20 mouse model of Alzheimer's disease. *Wellcome Open Research*, **2017**, 2, 84	4.8	10
65	Fully-Automated MRI Morphometric Phenotyping of the Tc1 Mouse Model of Down Syndrome. *PLoS ONE*, **2016**, 11, e0162974	3.7	10
64	Analysis of motor dysfunction in Down Syndrome reveals motor neuron degeneration. *PLoS Genetics*, **2018**, 14, e1007383	6	10

63	Downregulated Wnt/Ecatenin signalling in the Down syndrome hippocampus. *Scientific Reports*, **2019**, 9, 7322	4.9	9
62	Mouse models of aneuploidy. *Scientific World Journal, The*, **2012**, 2012, 214078	2.2	9
61	Analysis of European case-control studies suggests that common inherited variation in mitochondrial DNA is not involved in susceptibility to amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 341-6		9
60	Mouse models as a tool for understanding neurodegenerative diseases. *Current Opinion in Neurology*, **2003**, 16, 451-8	7.1	9
59	Four PCR-based polymorphisms in the pseudoautosomal region of the human X and Y chromosomes. *Human Molecular Genetics*, **1993**, 2, 1978	5.6	9
58	Gene expression dysregulation domains are not a specific feature of Down syndrome. *Nature Communications*, **2019**, 10, 2489	17.4	8
57	Altered Hippocampal-Prefrontal Neural Dynamics in Mouse Models of Down Syndrome. *Cell Reports*, **2020**, 30, 1152-1163.e4	10.6	8
56	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. *Molecular BioSystems*, **2013**, 9, 1736-42		8
55	Down syndrome and the molecular pathogenesis resulting from trisomy of human chromosome 21. *Journal of Biomedical Research*, **2010**, 24, 87-99	1.5	8
54	High resolution physical mapping and identification of transcribed sequences in the Down syndrome region-2. *Biochemical and Biophysical Research Communications*, **1998**, 243, 572-8	3.4	8
53	CHCHD10 Pro34Ser is not a highly penetrant pathogenic variant for amyotrophic lateral sclerosis and frontotemporal dementia. *Brain*, **2016**, 139, e9	11.2	7
52	Uses for humanised mouse models in precision medicine for neurodegenerative disease. *Mammalian Genome*, **2019**, 30, 173-191	3.2	7
51	No association of DYNC1H1 with sporadic ALS in a case-control study of a northern European derived population: a tagging SNP approach. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2006**, 7, 46-56		7
50	Frontotemporal dementia linked to chromosome 3. *Dementia and Geriatric Cognitive Disorders*, **2004**, 17, 274-6	2.6	7
49	Common ALS/FTD risk variants in UNC13A exacerbate its cryptic splicing and loss upon TDP-43 mislocalization		7
48	Interaction of sexual dimorphism and gene dosage imbalance in skeletal deficits associated with Down syndrome. *Bone*, **2020**, 136, 115367	4.7	6
47	An integrated genetic, radiation hybrid, physical and transcription map of a region of distal mouse chromosome 12, including an imprinted locus and the 'Legs at odd angles' (Loa) mutation. *Gene*, **2002**, 283, 71-82	3.8	6
46	Genomic organization and mapping of the mouse P26s4 ATPase gene: a member of the remarkably conserved AAA gene family. *Genomics*, **1996**, 31, 115-8	4.3	6

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