Huda Y Zoghbi

List of Publications by Year
in descending order

366	60,963	872 117	1009 236
papers	citations	h-index	g-index

409	409	409	39988
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. Nature Genetics, 1999, 23, 185-188.	21.4	4,459
2	Towards a proteome-scale map of the human proteinâ€“protein interaction network. Nature, 2005, 437, 1173-1178.	27.8	2,676
3	Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1. Nature Genetics, 1993, 4, 221-226.	21.4	1,673
4	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the Î±1A-voltage-dependent calcium channel. Nature Genetics, 1997, 15, 62-69.	21.4	1,606
5	MeCP2, a Key Contributor to Neurological Disease, Activates and Represses Transcription. Science, 2008, 320, 1224-1229.	12.6	1,582
6	Trinucleotide Repeat Disorders. Annual Review of Neuroscience, 2007, 30, 575-621.	10.7	1,289
7	Glutamine Repeats and Neurodegeneration. Annual Review of Neuroscience, 2000, 23, 217-247.	10.7	1,243
8	The Story of Rett Syndrome: From Clinic to Neurobiology. Neuron, 2007, 56, 422-437.	8.1	1,097
9	Math1: An Essential Gene for the Generation of Inner Ear Hair Cells. Science, 1999, 284, 1837-1841.	12.6	1,042
10	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269.	27.8	1,042
11	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	27.8	985
12	Ataxin-1 Nuclear Localization and Aggregation. Cell, 1998, 95, 41-53.	28.9	965
13	Requirement of Math1 for Secretory Cell Lineage Commitment in the Mouse Intestine. Science, 2001, 294, 2155-2158.	12.6	808
14	Chaperone suppression of aggregation and altered subcellular proteasome localization imply protein misfolding in SCA1. Nature Genetics, 1998, 19, 148-154.	21.4	802
15	Mice with Truncated MeCP2 Recapitulate Many Rett Syndrome Features and Display Hyperacetylation of Histone H3. Neuron, 2002, 35, 243-254.	8.1	723
16	Diseases of Unstable Repeat Expansion: Mechanisms and Common Principles. Nature Reviews Genetics, 2005, 6, 743-755.	16.3	716
17	A Proteinâ€“Protein Interaction Network for Human Inherited Ataxias and Disorders of Purkinje Cell Degeneration. Cell, 2006, 125, 801-814.	28.9	714
18	Synaptic Dysfunction in Neurodevelopmental Disorders Associated with Autism and Intellectual Disabilities. Cold Spring Harbor Perspectives in Biology, 2012, 4, a009886-a009886.	5.5	650

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19	Identification of genes that modify ataxin-1-induced neurodegeneration. Nature, 2000, 408, 101-106.	27.8	648
20	Math1 is essential for genesis of cerebellar granule neurons. Nature, 1997, 390, 169-172.	27.8	636
21	Postnatal Neurodevelopmental Disorders: Meeting at the Synapse?. Science, 2003, 302, 826-830.	12.6	621
22	SCA1 transgenic mice: A model for neurodegeneration caused by an expanded CAG trinucleotide repeat. Cell, 1995, 82, 937-948.	28.9	567
23	Mild overexpression of MeCP2 causes a progressive neurological disorder in mice. Human Molecular Genetics, 2004, 13, 2679-2689.	2.9	540
24	Ataxin-1 with an expanded glutamine tract alters nuclear matrix-associated structures. Nature, 1997, 389, 971-974.	27.8	531
25	DJ-1 Is a Redox-Dependent Molecular Chaperone That Inhibits Î±-Synuclein Aggregate Formation. PLoS Biology, 2004, 2, e362.	5.6	529
26	Math1 Expression Redefines the Rhombic Lip Derivatives and Reveals Novel Lineages within the Brainstem and Cerebellum. Neuron, 2005, 48, 31-43.	8.1	519
27	Large expansion of the ATTCT pentanucleotide repeat in spinocerebellar ataxia type 10. Nature Genetics, 2000, 26, 191-194.	21.4	505
28	Learning and Memory and Synaptic Plasticity Are Impaired in a Mouse Model of Rett Syndrome. Journal of Neuroscience, 2006, 26, 319-327.	3.6	493
29	Evidence for a mechanism predisposing to intergenerational CAG repeat instability in spinocerebellar ataxia type I. Nature Genetics, 1993, 5, 254-258.	21.4	489
30	Mutation of the E6-AP Ubiquitin Ligase Reduces Nuclear Inclusion Frequency While Accelerating Polyglutamine-Induced Pathology in SCA1 Mice. Neuron, 1999, 24, 879-892.	8.1	482
31	Rett Syndrome and Beyond: Recurrent Spontaneous and Familial MECP2 Mutations at CpG Hotspots. American Journal of Human Genetics, 1999, 65, 1520-1529.	6.2	463
32	Insight into Rett syndrome: MeCP2 levels display tissue- and cell-specific differences and correlate with neuronal maturation. Human Molecular Genetics, 2002, 11, 115-124.	2.9	461
33	MeCP2 Controls Excitatory Synaptic Strength by Regulating Glutamatergic Synapse Number. Neuron, 2007, 56, 58-65.	8.1	439
34	Genetic regulation of cerebellar development. Nature Reviews Neuroscience, 2001, 2, 484-491.	10.2	427
35	Regulation of RNA splicing by the methylation-dependent transcriptional repressor methyl-CpG binding protein 2. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 17551-17558.	7.1	413
36	Interaction of Akt-Phosphorylated Ataxin-1 with 14-3-3 Mediates Neurodegeneration in Spinocerebellar Ataxia Type 1. Cell, 2003, 113, 457-468.	28.9	402

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37	The role of Math1 in inner ear development: Uncoupling the establishment of the sensory primordium from hair cell fate determination. Development (Cambridge), 2002, 129, 2495-2505.	2.5	396
38	Failure of neuronal homeostasis results in common neuropsychiatric phenotypes. Nature, 2008, 455, 912-918.	27.8	367
39	Polyglutamine expansion down-regulates specific neuronal genes before pathologic changes in SCA1. Nature Neuroscience, 2000, 3, 157-163.	14.8	341
40	Sensitivity to Oxidative Stress in DJ-1-Deficient Dopamine Neurons: An ES- Derived Cell Model of Primary Parkinsonism. PLoS Biology, 2004, 2, e327.	5.6	338
41	SHANK3 overexpression causes manic-like behaviour with unique pharmacogenetic properties. Nature, 2013, 503, 72-77.	27.8	323
42	A Long CAG Repeat in the Mouse Sca1 Locus Replicates SCA1 Features and Reveals the Impact of Protein Solubility on Selective Neurodegeneration. Neuron, 2002, 34, 905-919.	8.1	320
43	Abnormalities of social interactions and home-cage behavior in a mouse model of Rett syndrome. Human Molecular Genetics, 2005, 14, 205-220.	2.9	318
44	Influence of mutation type and X chromosome inactivation on Rett syndrome phenotypes. Annals of Neurology, 2000, 47, 670-679.	5.3	314
45	Serine 776 of Ataxin-1 Is Critical for Polyglutamine-Induced Disease in SCA1 Transgenic Mice. Neuron, 2003, 38, 375-387.	8.1	303
46	Expression analysis of the ataxinâ€“1 protein in tissues from normal and spinocerebellar ataxia type 1 individuals. Nature Genetics, 1995, 10, 94-98.	21.4	291
47	Opposing effects of polyglutamine expansion on native protein complexes contribute to SCA1. Nature, 2008, 452, 713-718.	27.8	287
48	ATAXIN-1 Interacts with the Repressor Capicua in Its Native Complex to Cause SCA1 Neuropathology. Cell, 2006, 127, 1335-1347.	28.9	284
49	Proprioceptor Pathway Development Is Dependent on MATH1. Neuron, 2001, 30, 411-422.	8.1	280
50	Autism and other neuropsychiatric symptoms are prevalent in individuals with MeCP2 duplication syndrome. Annals of Neurology, 2009, 66, 771-782.	5.3	271
51	Gfi1 functions downstream of Math1 to control intestinal secretory cell subtype allocation and differentiation. Genes and Development, 2005, 19, 2412-2417.	5.9	267
52	The role of Math1 in inner ear development: Uncoupling the establishment of the sensory primordium from hair cell fate determination. Development (Cambridge), 2002, 129, 2495-505.	2.5	265
53	Purkinje Cell Expression of a Mutant Allele ofSCA1in Transgenic Mice Leads to Disparate Effects on Motor Behaviors, Followed by a Progressive Cerebellar Dysfunction and Histological Alterations. Journal of Neuroscience, 1997, 17, 7385-7395.	3.6	261
54	MeCP2 dysfunction in Rett syndrome and related disorders. Current Opinion in Genetics and Development, 2006, 16, 276-281.	3.3	258

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55	Intestine-Specific Ablation of Mouse atonal homolog 1 (Math1) Reveals a Role in Cellular Homeostasis. Gastroenterology, 2007, 132, 2478-2488.	1.3	258
56	Recovery from Polyglutamine-Induced Neurodegeneration in Conditional SCA1 Transgenic Mice. Journal of Neuroscience, 2004, 24, 8853-8861.	3.6	257
57	MeCP2 binds to non-CG methylated DNA as neurons mature, influencing transcription and the timing of onset for Rett syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 5509-5514.	7.1	256
58	Merkel Cells Are Essential for Light-Touch Responses. Science, 2009, 324, 1580-1582.	12.6	253
59	The cerebellar leucine-rich acidic nuclear protein interacts with ataxin-1. Nature, 1997, 389, 974-978.	27.8	246
60	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. Genetics in Medicine, 2006, 8, 784-792.	2.4	245
61	Loss of MeCP2 in aminergic neurons causes cell-autonomous defects in neurotransmitter synthesis and specific behavioral abnormalities. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21966-21971.	7.1	240
62	The zinc finger transcription factorGfi1, implicated in lymphomagenesis, is required for inner ear hair cell differentiation and survival. Development (Cambridge), 2003, 130, 221-232.	2.5	233
63	Deletion of Mecp2 in Sim1-Expressing Neurons Reveals a Critical Role for MeCP2 in Feeding Behavior, Aggression, and the Response to Stress. Neuron, 2008, 59, 947-958.	8.1	230
64	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. Human Molecular Genetics, 2009, 18, 2431-2442.	2.9	228
65	Adult Neural Function Requires MeCP2. Science, 2011, 333, 186-186.	12.6	227
66	Enhanced anxiety and stress-induced corticosterone release are associated with increased Crh expression in a mouse model of Rett syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 18267-18272.	7.1	225
67	RORÎ±-Mediated Purkinje Cell Development Determines Disease Severity in Adult SCA1 Mice. Cell, 2006, 127, 697-708.	28.9	210
68	Pathogenic Mechanisms of a Polyglutamine-mediated Neurodegenerative Disease, Spinocerebellar Ataxia Type 1. Journal of Biological Chemistry, 2009, 284, 7425-7429.	3.4	206
69	Identification of a putative Î³-aminobutyric acid (GABA) receptor subunit rho2 cDNA and colocalization of the genes encoding rho2 (GABRR2) and rho1 (GABRR1) to human chromosome 6q14â€“q21 and mouse chromosome 4. Genomics, 1992, 12, 801-806.	2.9	204
70	SCA7 Knockin Mice Model Human SCA7 and Reveal Gradual Accumulation of Mutant Ataxin-7 in Neurons and Abnormalities in Short-Term Plasticity. Neuron, 2003, 37, 383-401.	8.1	201
71	Protein Interactome Reveals Converging Molecular Pathways Among Autism Disorders. Science Translational Medicine, 2011, 3, 86ra49.	12.4	201
72	Mice Lacking Ataxin-1 Display Learning Deficits and Decreased Hippocampal Paired-Pulse Facilitation. Journal of Neuroscience, 1998, 18, 5508-5516.	3.6	197

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73	Neuronal dysfunction in a polyglutamine disease model occurs in the absence of ubiquitinâ€“proteasome system impairment and inversely correlates with the degree of nuclear inclusion formation. Human Molecular Genetics, 2005, 14, 679-691.	2.9	196
74	miR-19, miR-101 and miR-130 co-regulate ATXN1 levels to potentially modulate SCA1 pathogenesis. Nature Neuroscience, 2008, 11, 1137-1139.	14.8	194
75	Organization, inducible-expression and chromosome localization of the human HMG-I(Y) nonhistone protein gene. Nucleic Acids Research, 1993, 21, 4259-4267.	14.5	191
76	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
77	The AXH Domain of Ataxin-1 Mediates Neurodegeneration through Its Interaction with Gfi-1/Senseless Proteins. Cell, 2005, 122, 633-644.	28.9	189
78	Lessons learned from studying syndromic autism spectrum disorders. Nature Neuroscience, 2016, 19, 1408-1417.	14.8	185
79	Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics, 2011, 43, 1074-1081.	21.4	184
80	Preclinical research in Rett syndrome: setting the foundation for translational success. DMM Disease Models and Mechanisms, 2012, 5, 733-745.	2.4	183
81	Rett Syndrome and MeCP2: Linking Epigenetics and Neuronal Function. American Journal of Human Genetics, 2002, 71, 1259-1272.	6.2	181
82	Rett syndrome: Methyl-CpG-binding protein 2 mutations and phenotype-genotype correlations. American Journal of Medical Genetics Part A, 2000, 97, 147-152.	2.4	180
83	Gametic and somatic tissueâ€“specific heterogeneity of the expanded SCA1 CAG repeat in spinocerebellar ataxia type 1. Nature Genetics, 1995, 10, 344-350.	21.4	179
84	Epigenetics and Human Disease. Cold Spring Harbor Perspectives in Biology, 2016, 8, a019497.	5.5	177
85	CHIP Protects from the Neurotoxicity of Expanded and Wild-type Ataxin-1 and Promotes Their Ubiquitination and Degradation. Journal of Biological Chemistry, 2006, 281, 26714-26724.	3.4	176
86	A partial loss of function allele of Methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. Human Molecular Genetics, 2008, 17, 1718-1727.	2.9	173
87	MECP2 disorders: from the clinic to mice and back. Journal of Clinical Investigation, 2015, 125, 2914-2923.	8.2	172
88	The E-protein Tcf4 interacts with Math1 to regulate differentiation of a specific subset of neuronal progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 15382-15387.	7.1	169
89	Gene profiling links SCA1 pathophysiology to glutamate signaling in Purkinje cells of transgenic mice. Human Molecular Genetics, 2004, 13, 2535-2543.	2.9	168
90	Diagnostic Testing for Rett Syndrome by DHPLC and Direct Sequencing Analysis of the MECP2 Gene: Identification of Several Novel Mutations and Polymorphisms. American Journal of Human Genetics, 2000, 67, 1428-1436.	6.2	167

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91	Reversal of phenotypes in MECP2 duplication mice using genetic rescue or antisense oligonucleotides. Nature, 2015, 528, 123-126.	27.8	166
92	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	2.9	165
93	The Future of Psychiatric Research: Genomes and Neural Circuits. Science, 2010, 327, 1580-1581.	12.6	164
94	Deletion of Atoh1 Disrupts Sonic Hedgehog Signaling in the Developing Cerebellum and Prevents Medulloblastoma. Science, 2009, 326, 1424-1427.	12.6	163
95	Female Mecp2+/â˜’ mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. Human Molecular Genetics, 2013, 22, 96-109.	2.9	158
96	atonal Regulates Neurite Arborization but Does Not Act as a Proneural Gene in the Drosophila Brain. Neuron, 2000, 25, 549-561.	8.1	156
97	Spinocerebellar ataxia type 6 knockin mice develop a progressive neuronal dysfunction with age-dependent accumulation of mutant Ca _V 2.1 channels. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 11987-11992.	7.1	156
98	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. Human Molecular Genetics, 2011, 20, 3366-3375.	2.9	149
99	Lithium Therapy Improves Neurological Function and Hippocampal Dendritic Arborization in a Spinocerebellar Ataxia Type 1 Mouse Model. PLoS Medicine, 2007, 4, e182.	8.4	147
100	Math1 Is Essential for the Development of Hindbrain Neurons Critical for Perinatal Breathing. Neuron, 2009, 64, 341-354.	8.1	146
101	Crh and Oprm1 mediate anxiety-related behavior and social approach in a mouse model of MECP2 duplication syndrome. Nature Genetics, 2012, 44, 206-211.	21.4	146
102	Forniceal deep brain stimulation rescues hippocampal memory in Rett syndrome mice. Nature, 2015, 526, 430-434.	27.8	146
103	Exercise and Genetic Rescue of SCA1 via the Transcriptional Repressor Capicua. Science, 2011, 334, 690-693.	12.6	144
104	Loss of MeCP2 in Parvalbumin-and Somatostatin-Expressing Neurons in Mice Leads to Distinct Rett Syndrome-like Phenotypes. Neuron, 2015, 88, 651-658.	8.1	144
105	Glutamine-Expanded Ataxin-7 Alters TFTC/STAGA Recruitment and Chromatin Structure Leading to Photoreceptor Dysfunction. PLoS Biology, 2006, 4, e67.	5.6	143
106	Reduction of Biogenic Amine Levels in the Rett Syndrome. New England Journal of Medicine, 1985, 313, 921-924.	27.0	142
107	In vivo Atoh1 targetome reveals how a proneural transcription factor regulates cerebellar development. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 3288-3293.	7.1	141
108	Interruptions in the Triplet Repeats of SCA1 and FRAXA Reduce the Propensity and Complexity of Slipped Strand DNA (S-DNA) Formationâ€. Biochemistry, 1998, 37, 2701-2708.	2.5	139

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109	Pumilio1 Haploinsufficiency Leads to SCA1-like Neurodegeneration by Increasing Wild-Type Ataxin1 Levels. Cell, 2015, 160, 1087-1098.	28.9	139
110	SCA1-like Disease in Mice Expressing Wild-Type Ataxin-1 with a Serine to Aspartic Acid Replacement at Residue 776. Neuron, 2010, 67, 929-935.	8.1	137
111	Retinal degeneration characterizes a spinocerebellar ataxia mapping to chromosome 3p. Nature Genetics, 1995, 10, 89-93.	21.4	136
112	Characterization of the Transcriptome of Nascent Hair Cells and Identification of Direct Targets of the Atoh1 Transcription Factor. Journal of Neuroscience, 2015, 35, 5870-5883.	3.6	136
113	Rett Syndrome: A Prototypical Neurodevelopmental Disorder. Neuroscientist, 2004, 10, 118-128.	3.5	135
114	Solving the Autism Puzzle a Few Pieces at a Time. Neuron, 2011, 70, 806-808.	8.1	134
115	Generation of a Mouse Model for Arginase II Deficiency by Targeted Disruption of the Arginase II Gene. Molecular and Cellular Biology, 2001, 21, 811-813.	2.3	128
116	Spinocerebellar ataxia: Variable age of onset and linkage to human leukocyte antigen in a large kindred. Annals of Neurology, 1988, 23, 580-584.	5.3	126
117	Spinocerebellar ataxia type 1. Seminars in Cell Biology, 1995, 6, 29-35.	3.4	125
118	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	6.2	125
119	Rett's syndrome: Characterization of respiratory patterns and sleep. Annals of Neurology, 1987, 21, 377-382.	5.3	124
120	Excitatory neurons of the proprioceptive, interoceptive, and arousal hindbrain networks share a developmental requirement for Math1. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 22462-22467.	7.1	124
121	Dendritic Arborization and Spine Dynamics Are Abnormal in the Mouse Model ofMECP2Duplication Syndrome. Journal of Neuroscience, 2013, 33, 19518-19533.	3.6	123
122	Molecular profiling predicts meningioma recurrence and reveals loss of DREAM complex repression in aggressive tumors. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21715-21726.	7.1	122
123	Molecular genetics of Rett syndrome and clinical spectrum of MECP2 mutations. Current Opinion in Neurology, 2001, 14, 171-176.	3.6	120
124	X-Chromosome Inactivation Patterns Are Unbalanced and Affect the Phenotypic Outcome in a Mouse Model of Rett Syndrome. American Journal of Human Genetics, 2004, 74, 511-520.	6.2	120
125	Reduction of Nuak1 Decreases Tau and Reverses Phenotypes in a Tauopathy Mouse Model. Neuron, 2016, 92, 407-418.	8.1	120
126	Patterns of X chromosome inactivation in the rett syndrome. Brain and Development, 1990, 12, 131-135.	1.1	119

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127	RASâ€“MAPKâ€“MSK1 pathway modulates ataxin 1 protein levels and toxicity in SCA1. Nature, 2013, 498, 325-331.	27.8	119
128	Partial Loss of Ataxin-1 Function Contributes to Transcriptional Dysregulation in Spinocerebellar Ataxia Type 1 Pathogenesis. PLoS Genetics, 2010, 6, e1001021.	3.5	113
129	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
130	Atoh1-Lineal Neurons Are Required for Hearing and for the Survival of Neurons in the Spiral Ganglion and Brainstem Accessory Auditory Nuclei. Journal of Neuroscience, 2009, 29, 11123-11133.	3.6	111
131	Mouse and fly models of neurodegeneration. Trends in Genetics, 2002, 18, 463-471.	6.7	110
132	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	6.2	110
133	The CAG/Polyglutamine Tract Diseases: Gene Products and Molecular Pathogenesis. Brain Pathology, 1997, 7, 927-942.	4.1	109
134	A New Rett Syndrome Family Consistent with X-Linked Inheritance Expands the X Chromosome Exclusion Map. American Journal of Human Genetics, 1997, 61, 634-641.	6.2	107
135	Molecular and clinical correlations in spinocerebellar ataxia type 3 and Machado-Joseph disease. Annals of Neurology, 1995, 38, 68-72.	5.3	106
136	Antisense oligonucleotideâ€“mediated ataxin-1 reduction prolongs survival in SCA1 mice and reveals disease-associated transcriptome profiles. JCI Insight, 2018, 3, .	5.0	106
137	Impaired Conditioned Fear and Enhanced Long-Term Potentiation inFmr2 Knock-Out Mice. Journal of Neuroscience, 2002, 22, 2753-2763.	3.6	105
138	Increased Expression of Î±_{1A}Ca²⁺Channel Currents Arising from Expanded Trinucleotide Repeats in Spinocerebellar Ataxia Type 6. Journal of Neuroscience, 2001, 21, 9185-9193.	3.6	104
139	Drosophila atonal Fully Rescues the Phenotype of Math1 Null Mice. Current Biology, 2002, 12, 1611-1616.	3.9	104
140	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	28.9	103
141	Polyglutamine diseases: protein cleavage and aggregation. Current Opinion in Neurobiology, 1999, 9, 566-570.	4.2	102
142	Inactivation of hnRNP K by Expanded Intronic AUUCU Repeat Induces Apoptosis Via Translocation of PKCÎ´ to Mitochondria in Spinocerebellar Ataxia 10. PLoS Genetics, 2010, 6, e1000984.	3.5	102
143	Advances in understanding of Rett syndrome and MECP2 duplication syndrome: prospects for future therapies. Lancet Neurology, The, 2020, 19, 689-698.	10.2	102
144	A high resolution deletion map of human chromosome Xp22. Nature Genetics, 1993, 4, 272-279.	21.4	101

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145	Microphthalmia with linear skin defects (MLS) syndrome: Clinical, cytogenetic, and molecular characterization. American Journal of Medical Genetics Part A, 1994, 49, 229-234.	2.4	100
146	Mapping of the gene for a novel spinocerebellar ataxia with pure cerebellar signs and epilepsy. Annals of Neurology, 1999, 45, 407-411.	5.3	100
147	Increased Trinucleotide Repeat Instability with Advanced Maternal Age. Human Molecular Genetics, 1997, 6, 2135-2139.	2.9	97
148	TRIM28 regulates the nuclear accumulation and toxicity of both alpha-synuclein and tau. ELife, 2016, 5, .	6.0	97
149	Human-specific regulation of MeCP2 levels in fetal brains by microRNA miR-483-5p. Genes and Development, 2013, 27, 485-490.	5.9	95
150	Cerebrospinal fluid biogenic amines and biopterin in rett syndrome. Annals of Neurology, 1989, 25, 56-60.	5.3	92
151	Parkinson's Disease Genetics and Pathophysiology. Annual Review of Neuroscience, 2021, 44, 87-108.	10.7	92
152	ATXN1 Protein Family and CIC Regulate Extracellular Matrix Remodeling and Lung Alveolarization. Developmental Cell, 2011, 21, 746-757.	7.0	89
153	Restoration of Mecp2 expression in GABAergic neurons is sufficient to rescue multiple disease features in a mouse model of Rett syndrome. ELife, 2016, 5, .	6.0	89
154	Aberrant myofibril assembly in tropomodulin1 null mice leads to aborted heart development and embryonic lethality. Journal of Cell Biology, 2003, 163, 1033-1044.	5.2	88
155	Loss and Gain of MeCP2 Cause Similar Hippocampal Circuit Dysfunction that Is Rescued by Deep Brain Stimulation in a Rett Syndrome Mouse Model. Neuron, 2016, 91, 739-747.	8.1	88
156	SUMOylation of the Polyglutamine Repeat Protein, Ataxin-1, Is Dependent on a Functional Nuclear Localization Signal. Journal of Biological Chemistry, 2005, 280, 21942-21948.	3.4	87
157	Cerebellar Transcriptome Profiles of ATXN1 Transgenic Mice Reveal SCA1 Disease Progression and Protection Pathways. Neuron, 2016, 89, 1194-1207.	8.1	86
158	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. ELife, 2016, 5, .	6.0	86
159	The insulin-like growth factor pathway is altered in spinocerebellar ataxia type 1 and type 7. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1291-1296.	7.1	85
160	Rapid identification of yeast artificial chromosome clones by matrix pooling and crude lysate PCR. Nucleic Acids Research, 1990, 18, 7191-7191.	14.5	84
161	Altered Trafficking of Membrane Proteins in Purkinje Cells of SCA1 Transgenic Mice. American Journal of Pathology, 2001, 159, 905-913.	3.8	83
162	dAtaxin-2 Mediates Expanded Ataxin-1-Induced Neurodegeneration in a Drosophila Model of SCA1. PLoS Genetics, 2007, 3, e234.	3.5	83

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163	Sequencing, Expression Analysis, and Mapping of Three Unique Human Tropomodulin Genes and Their Mouse Orthologs. Genomics, 2000, 63, 97-107.	2.9	81
164	Reversing Neurodegeneration:A Promise Unfolds. Cell, 2000, 101, 1-4.	28.9	81
165	Modelling brain diseases in mice: the challenges of design and analysis. Nature Reviews Genetics, 2003, 4, 296-307.	16.3	81
166	Molecular Cloning of the cDNA Encoding a Human Renal Sodium Phosphate Transport Protein and Its Assignment to Chromosome 6p21.3-p23. Genomics, 1993, 18, 355-359.	2.9	79
167	ATXN1-CIC Complex Is the Primary Driver of Cerebellar Pathology in Spinocerebellar Ataxia Type 1 through a Gain-of-Function Mechanism. Neuron, 2018, 97, 1235-1243.e5.	8.1	79
168	Expanding Our Understanding of Polyglutamine Diseases through Mouse Models. Neuron, 1999, 24, 499-502.	8.1	78
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