Huda Y Zoghbi

List of Publications by Citations

349 papers	51,590 citations	111 h-index	223 g-index
409 ext. papers	57,110 ext. citations	15 avg, IF	7.49 L-index

#	Paper	IF	Citations
349	Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics*, **1999**, 23, 185-8	36.3	3780
348	Towards a proteome-scale map of the human protein-protein interaction network. *Nature*, **2005**, 437, 1173-8	50.4	2287
347	Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1. *Nature Genetics*, **1993**, 4, 221-6	36.3	1453
346	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the alpha 1A-voltage-dependent calcium channel. *Nature Genetics*, **1997**, 15, 62-9	36.3	1440
345	MeCP2, a key contributor to neurological disease, activates and represses transcription. *Science*, **2008**, 320, 1224-9	33.3	1360
344	Trinucleotide repeat disorders. *Annual Review of Neuroscience*, **2007**, 30, 575-621	17	1120
343	Glutamine repeats and neurodegeneration. *Annual Review of Neuroscience*, **2000**, 23, 217-47	17	1110
342	The story of Rett syndrome: from clinic to neurobiology. *Neuron*, **2007**, 56, 422-37	13.9	936
341	Ataxin-1 nuclear localization and aggregation: role in polyglutamine-induced disease in SCA1 transgenic mice. *Cell*, **1998**, 95, 41-53	56.2	898
340	Math1: an essential gene for the generation of inner ear hair cells. *Science*, **1999**, 284, 1837-41	33.3	884
339	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. *Nature*, **2010**, 468, 263-9	50.4	849
338	The DNA sequence of the human X chromosome. *Nature*, **2005**, 434, 325-37	50.4	822
337	Chaperone suppression of aggregation and altered subcellular proteasome localization imply protein misfolding in SCA1. *Nature Genetics*, **1998**, 19, 148-54	36.3	766
336	Requirement of Math1 for secretory cell lineage commitment in the mouse intestine. *Science*, **2001**, 294, 2155-8	33.3	722
335	Mice with truncated MeCP2 recapitulate many Rett syndrome features and display hyperacetylation of histone H3. *Neuron*, **2002**, 35, 243-54	13.9	645
334	A protein-protein interaction network for human inherited ataxias and disorders of Purkinje cell degeneration. *Cell*, **2006**, 125, 801-14	56.2	637
333	Diseases of unstable repeat expansion: mechanisms and common principles. *Nature Reviews Genetics*, **2005**, 6, 743-55	30.1	613

332	Math1 is essential for genesis of cerebellar granule neurons. *Nature*, **1997**, 390, 169-72	50.4	564
331	Identification of genes that modify ataxin-1-induced neurodegeneration. *Nature*, **2000**, 408, 101-6	50.4	560
330	Postnatal neurodevelopmental disorders: meeting at the synapse?. *Science*, **2003**, 302, 826-30	33.3	532
329	SCA1 transgenic mice: a model for neurodegeneration caused by an expanded CAG trinucleotide repeat. *Cell*, **1995**, 82, 937-48	56.2	505
328	Ataxin-1 with an expanded glutamine tract alters nuclear matrix-associated structures. *Nature*, **1997**, 389, 971-4	50.4	498
327	Synaptic dysfunction in neurodevelopmental disorders associated with autism and intellectual disabilities. *Cold Spring Harbor Perspectives in Biology*, **2012**, 4,	10.2	488
326	Mild overexpression of MeCP2 causes a progressive neurological disorder in mice. *Human Molecular Genetics*, **2004**, 13, 2679-89	5.6	478
325	DJ-1 is a redox-dependent molecular chaperone that inhibits alpha-synuclein aggregate formation. *PLoS Biology*, **2004**, 2, e362	9.7	458
324	Large expansion of the ATTCT pentanucleotide repeat in spinocerebellar ataxia type 10. *Nature Genetics*, **2000**, 26, 191-4	36.3	450
323	Mutation of the E6-AP ubiquitin ligase reduces nuclear inclusion frequency while accelerating polyglutamine-induced pathology in SCA1 mice. *Neuron*, **1999**, 24, 879-92	13.9	449
322	Math1 expression redefines the rhombic lip derivatives and reveals novel lineages within the brainstem and cerebellum. *Neuron*, **2005**, 48, 31-43	13.9	440
321	Evidence for a mechanism predisposing to intergenerational CAG repeat instability in spinocerebellar ataxia type I. *Nature Genetics*, **1993**, 5, 254-8	36.3	436
320	Learning and memory and synaptic plasticity are impaired in a mouse model of Rett syndrome. *Journal of Neuroscience*, **2006**, 26, 319-27	6.6	417
319	Rett syndrome and beyond: recurrent spontaneous and familial MECP2 mutations at CpG hotspots. *American Journal of Human Genetics*, **1999**, 65, 1520-9	11	417
318	Insight into Rett syndrome: MeCP2 levels display tissue- and cell-specific differences and correlate with neuronal maturation. *Human Molecular Genetics*, **2002**, 11, 115-24	5.6	397
317	Genetic regulation of cerebellar development. *Nature Reviews Neuroscience*, **2001**, 2, 484-91	13.5	378
316	MeCP2 controls excitatory synaptic strength by regulating glutamatergic synapse number. *Neuron*, **2007**, 56, 58-65	13.9	373
315	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, 17551-8	11.5	356

314	Interaction of Akt-phosphorylated ataxin-1 with 14-3-3 mediates neurodegeneration in spinocerebellar ataxia type 1. *Cell*, **2003**, 113, 457-68	56.2	344
313	Sensitivity to oxidative stress in DJ-1-deficient dopamine neurons: an ES- derived cell model of primary Parkinsonism. *PLoS Biology*, **2004**, 2, e327	9.7	304
312	Failure of neuronal homeostasis results in common neuropsychiatric phenotypes. *Nature*, **2008**, 455, 912-8	50.4	301
311	Polyglutamine expansion down-regulates specific neuronal genes before pathologic changes in SCA1. *Nature Neuroscience*, **2000**, 3, 157-63	25.5	299
310	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	6.6	295
309	Abnormalities of social interactions and home-cage behavior in a mouse model of Rett syndrome. *Human Molecular Genetics*, **2005**, 14, 205-20	5.6	284
308	Influence of mutation type and X chromosome inactivation on Rett syndrome phenotypes. *Annals of Neurology*, **2000**, 47, 670-679	9.4	283
307	Serine 776 of ataxin-1 is critical for polyglutamine-induced disease in SCA1 transgenic mice. *Neuron*, **2003**, 38, 375-87	13.9	275
306	Expression analysis of the ataxin-1 protein in tissues from normal and spinocerebellar ataxia type 1 individuals. *Nature Genetics*, **1995**, 10, 94-8	36.3	265
305	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	6.6	251
304	Opposing effects of polyglutamine expansion on native protein complexes contribute to SCA1. *Nature*, **2008**, 452, 713-8	50.4	250
303	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, 905-19	13.9	250
302	Proprioceptor pathway development is dependent on Math1. *Neuron*, **2001**, 30, 411-22	13.9	250
301	SHANK3 overexpression causes manic-like behaviour with unique pharmacogenetic properties. *Nature*, **2013**, 503, 72-7	50.4	249
300	ATAXIN-1 interacts with the repressor Capicua in its native complex to cause SCA1 neuropathology. *Cell*, **2006**, 127, 1335-47	56.2	242
299	Autism and other neuropsychiatric symptoms are prevalent in individuals with MeCP2 duplication syndrome. *Annals of Neurology*, **2009**, 66, 771-82	9.4	237
298	Gfi1 functions downstream of Math1 to control intestinal secretory cell subtype allocation and differentiation. *Genes and Development*, **2005**, 19, 2412-7	12.6	237
297	Purkinje cell expression of a mutant allele of SCA1 in transgenic mice leads to disparate effects on motor behaviors, followed by a progressive cerebellar dysfunction and histological alterations. *Journal of Neuroscience*, **1997**, 17, 7385-95	6.6	233

296	Recovery from polyglutamine-induced neurodegeneration in conditional SCA1 transgenic mice. *Journal of Neuroscience*, **2004**, 24, 8853-61	6.6	229
295	The cerebellar leucine-rich acidic nuclear protein interacts with ataxin-1. *Nature*, **1997**, 389, 974-8	50.4	227
294	MeCP2 dysfunction in Rett syndrome and related disorders. *Current Opinion in Genetics and Development*, **2006**, 16, 276-81	4.9	227
293	Intestine-specific ablation of mouse atonal homolog 1 (Math1) reveals a role in cellular homeostasis. *Gastroenterology*, **2007**, 132, 2478-88	13.3	225
292	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. *Genetics in Medicine*, **2006**, 8, 784-92	8.1	225
291	Merkel cells are essential for light-touch responses. *Science*, **2009**, 324, 1580-2	33.3	205
290	The zinc finger transcription factor Gfi1, implicated in lymphomagenesis, is required for inner ear hair cell differentiation and survival. *Development (Cambridge)*, **2003**, 130, 221-32	6.6	203
289	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, 947-58	13.9	202
288	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, 21966-71	11.5	198
287	Adult neural function requires MeCP2. *Science*, **2011**, 333, 186	33.3	195
286	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, 18267-72	11.5	192
285	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, 5509-14	11.5	190
284	Neuronal dysfunction in a polyglutamine disease model occurs in the absence of ubiquitin-proteasome system impairment and inversely correlates with the degree of nuclear inclusion formation. *Human Molecular Genetics*, **2005**, 14, 679-91	5.6	187
283	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. *Human Molecular Genetics*, **2009**, 18, 2431-42	5.6	185
282	Identification of a putative gamma-aminobutyric acid (GABA) receptor subunit rho2 cDNA and colocalization of the genes encoding rho2 (GABRR2) and rho1 (GABRR1) to human chromosome 6q14-q21 and mouse chromosome 4. *Genomics*, **1992**, 12, 801-6	4.3	183
281	miR-19, miR-101 and miR-130 co-regulate ATXN1 levels to potentially modulate SCA1 pathogenesis. *Nature Neuroscience*, **2008**, 11, 1137-9	25.5	182
280	Mice lacking ataxin-1 display learning deficits and decreased hippocampal paired-pulse facilitation. *Journal of Neuroscience*, **1998**, 18, 5508-16	6.6	181
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278	Organization, inducible-expression and chromosome localization of the human HMG-I(Y) nonhistone protein gene. *Nucleic Acids Research*, **1993**, 21, 4259-67	20.1	178
277	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	13.9	173
276	Pathogenic mechanisms of a polyglutamine-mediated neurodegenerative disease, spinocerebellar ataxia type 1. *Journal of Biological Chemistry*, **2009**, 284, 7425-9	5.4	165
275	The AXH domain of Ataxin-1 mediates neurodegeneration through its interaction with Gfi-1/Senseless proteins. *Cell*, **2005**, 122, 633-44	56.2	164
274	Gametic and somatic tissue-specific heterogeneity of the expanded SCA1 CAG repeat in spinocerebellar ataxia type 1. *Nature Genetics*, **1995**, 10, 344-50	36.3	163
273	A partial loss of function allele of methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. *Human Molecular Genetics*, **2008**, 17, 1718-27	5.6	157
272	CHIP protects from the neurotoxicity of expanded and wild-type ataxin-1 and promotes their ubiquitination and degradation. *Journal of Biological Chemistry*, **2006**, 281, 26714-24	5.4	156
271	Rett syndrome and MeCP2: linking epigenetics and neuronal function. *American Journal of Human Genetics*, **2002**, 71, 1259-72	11	156
270	Rett syndrome: methyl-CpG-binding protein 2 mutations and phenotype-genotype correlations. *American Journal of Medical Genetics Part A*, **2000**, 97, 147-52		156
269	Protein interactome reveals converging molecular pathways among autism disorders. *Science Translational Medicine*, **2011**, 3, 86ra49	17.5	155
268	Preclinical research in Rett syndrome: setting the foundation for translational success. *DMM Disease Models and Mechanisms*, **2012**, 5, 733-45	4.1	154
267	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, 1428-36	11	154
266	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, 15382-7	11.5	153
265	Gene profiling links SCA1 pathophysiology to glutamate signaling in Purkinje cells of transgenic mice. *Human Molecular Genetics*, **2004**, 13, 2535-43	5.6	152
264	Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. *Nature Genetics*, **2011**, 43, 1074-81	36.3	147
263	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. *Human Molecular Genetics*, **2009**, 18, 2188-203	5.6	143
262	MECP2 disorders: from the clinic to mice and back. *Journal of Clinical Investigation*, **2015**, 125, 2914-23	15.9	143
261	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway. *Genetics in Medicine*, **2014**, 16, 751-8	8.1	138

260	Deletion of Atoh1 disrupts Sonic Hedgehog signaling in the developing cerebellum and prevents medulloblastoma. *Science*, **2009**, 326, 1424-7	33.3	138
259	A mutant form of MeCP2 protein associated with human Rett syndrome cannot be displaced from methylated DNA by notch in Xenopus embryos. *Molecular Cell*, **2003**, 12, 425-35	17.6	138
258	Medicine. The future of psychiatric research: genomes and neural circuits. *Science*, **2010**, 327, 1580-1	33.3	137
257	atonal regulates neurite arborization but does not act as a proneural gene in the Drosophila brain. *Neuron*, **2000**, 25, 549-61	13.9	135
256	Math1 is essential for the development of hindbrain neurons critical for perinatal breathing. *Neuron* , **2009**, 64, 341-54	13.9	132
255	Reduction of biogenic amine levels in the Rett syndrome. *New England Journal of Medicine*, **1985**, 313, 921-4	59.2	131
254	Spinocerebellar ataxia type 6 knockin mice develop a progressive neuronal dysfunction with age-dependent accumulation of mutant CaV2.1 channels. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 11987-92	11.5	130
253	Interruptions in the triplet repeats of SCA1 and FRAXA reduce the propensity and complexity of slipped strand DNA (S-DNA) formation. *Biochemistry*, **1998**, 37, 2701-8	3.2	130
252	Lithium therapy improves neurological function and hippocampal dendritic arborization in a spinocerebellar ataxia type 1 mouse model. *PLoS Medicine*, **2007**, 4, e182	11.6	129
251	Glutamine-expanded ataxin-7 alters TFTC/STAGA recruitment and chromatin structure leading to photoreceptor dysfunction. *PLoS Biology*, **2006**, 4, e67	9.7	125
250	Retinal degeneration characterizes a spinocerebellar ataxia mapping to chromosome 3p. *Nature Genetics*, **1995**, 10, 89-93	36.3	125
249	SCA1-like disease in mice expressing wild-type ataxin-1 with a serine to aspartic acid replacement at residue 776. *Neuron*, **2010**, 67, 929-35	13.9	120
248	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, 3288-93	11.5	120
247	Forniceal deep brain stimulation rescues hippocampal memory in Rett syndrome mice. *Nature*, **2015**, 526, 430-4	50.4	119
246	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. *Human Molecular Genetics*, **2011**, 20, 3366-75	5.6	118
245	Female Mecp2(+/-) mice display robust behavioral deficits on two different genetic backgrounds providing a framework for pre-clinical studies. *Human Molecular Genetics*, **2013**, 22, 96-109	5.6	117
244	Exercise and genetic rescue of SCA1 via the transcriptional repressor Capicua. *Science*, **2011**, 334, 690-3	33.3	117
243	Generation of a mouse model for arginase II deficiency by targeted disruption of the arginase II gene. *Molecular and Cellular Biology*, **2001**, 21, 811-3	4.8	117

242	Lessons learned from studying syndromic autism spectrum disorders. *Nature Neuroscience*, **2016**, 19, 1408-1417	25.5	116
241	Solving the autism puzzle a few pieces at a time. *Neuron*, **2011**, 70, 806-8	13.9	115
240	Rett syndrome: a prototypical neurodevelopmental disorder. *Neuroscientist*, **2004**, 10, 118-28	7.6	114
239	Reversal of phenotypes in MECP2 duplication mice using genetic rescue or antisense oligonucleotides. *Nature*, **2015**, 528, 123-6	50.4	113
238	Epigenetics and Human Disease. *Cold Spring Harbor Perspectives in Biology*, **2016**, 8, a019497	10.2	111
237	Rett syndrome: characterization of respiratory patterns and sleep. *Annals of Neurology*, **1987**, 21, 377-82	9.4	110
236	Crh and Oprm1 mediate anxiety-related behavior and social approach in a mouse model of MECP2 duplication syndrome. *Nature Genetics*, **2012**, 44, 206-11	36.3	109
235	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, 511-20	11	109
234	Patterns of X chromosome inactivation in the Rett syndrome. *Brain and Development*, **1990**, 12, 131-5	2.2	107
233	Dendritic arborization and spine dynamics are abnormal in the mouse model of MECP2 duplication syndrome. *Journal of Neuroscience*, **2013**, 33, 19518-33	6.6	104
232	Loss of MeCP2 in Parvalbumin-and Somatostatin-Expressing Neurons in Mice Leads to Distinct Rett Syndrome-like Phenotypes. *Neuron*, **2015**, 88, 651-8	13.9	103
231	Spinocerebellar ataxia: variable age of onset and linkage to human leukocyte antigen in a large kindred. *Annals of Neurology*, **1988**, 23, 580-4	9.4	102
230	RAS-MAPK-MSK1 pathway modulates ataxin 1 protein levels and toxicity in SCA1. *Nature*, **2013**, 498, 325-331	50.4	101
229	The CAG/polyglutamine tract diseases: gene products and molecular pathogenesis. *Brain Pathology* , **1997**, 7, 927-42	6	101
228	Pumilio1 haploinsufficiency leads to SCA1-like neurodegeneration by increasing wild-type Ataxin1 levels. *Cell*, **2015**, 160, 1087-98	56.2	100
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225	Increased expression of alpha 1A Ca2+ channel currents arising from expanded trinucleotide repeats in spinocerebellar ataxia type 6. *Journal of Neuroscience*, **2001**, 21, 9185-93	6.6	98

224	Molecular genetics of Rett syndrome and clinical spectrum of MECP2 mutations. *Current Opinion in Neurology*, **2001**, 14, 171-6	7.1	98
223	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, 11123-33	6.6	97
222	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, 22462-7	11.5	96
221	Partial loss of ataxin-1 function contributes to transcriptional dysregulation in spinocerebellar ataxia type 1 pathogenesis. *PLoS Genetics*, **2010**, 6, e1001021	6	95
220	Polyglutamine diseases: protein cleavage and aggregation. *Current Opinion in Neurobiology*, **1999**, 9, 566-70	7.6	94
219	Molecular and clinical correlations in spinocerebellar ataxia type 3 and Machado-Joseph disease. *Annals of Neurology*, **1995**, 38, 68-72	9.4	94
218	A high resolution deletion map of human chromosome Xp22. *Nature Genetics*, **1993**, 4, 272-9	36.3	94
217	Impaired conditioned fear and enhanced long-term potentiation in Fmr2 knock-out mice. *Journal of Neuroscience*, **2002**, 22, 2753-63	6.6	93
216	Mapping of the gene for a novel spinocerebellar ataxia with pure cerebellar signs and epilepsy. *Annals of Neurology*, **1999**, 45, 407-11	9.4	92
215	Microphthalmia with linear skin defects (MLS) syndrome: clinical, cytogenetic, and molecular characterization. *American Journal of Medical Genetics Part A*, **1994**, 49, 229-34		92
214	Inactivation of hnRNP K by expanded intronic AUUCU repeat induces apoptosis via translocation of PKCdelta to mitochondria in spinocerebellar ataxia 10. *PLoS Genetics*, **2010**, 6, e1000984	6	91
213	Characterization of the transcriptome of nascent hair cells and identification of direct targets of the Atoh1 transcription factor. *Journal of Neuroscience*, **2015**, 35, 5870-83	6.6	88
212	Spinocerebellar ataxia type 1. *Seminars in Cell Biology*, **1995**, 6, 29-35		88
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208	Altered trafficking of membrane proteins in purkinje cells of SCA1 transgenic mice. *American Journal of Pathology*, **2001**, 159, 905-13	5.8	80
207	Human-specific regulation of MeCP2 levels in fetal brains by microRNA miR-483-5p. *Genes and Development*, **2013**, 27, 485-90	12.6	78

206	14-3-3 binding to Ataxin-1 (ATXN1) regulates its dephosphorylation at Ser-776 and transport to the nucleus.. *Journal of Biological Chemistry*, **2013**, 288, 6590	5.4	78
205	SUMOylation of the polyglutamine repeat protein, ataxin-1, is dependent on a functional nuclear localization signal. *Journal of Biological Chemistry*, **2005**, 280, 21942-8	5.4	77
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201	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, 1291-6	11.5	74
200	dAtaxin-2 mediates expanded Ataxin-1-induced neurodegeneration in a Drosophila model of SCA1. *PLoS Genetics*, **2007**, 3, e234	6	74
199	Reduction of Nuak1 Decreases Tau and Reverses Phenotypes in a Tauopathy Mouse Model. *Neuron*, **2016**, 92, 407-418	13.9	74
198	ATXN1 protein family and CIC regulate extracellular matrix remodeling and lung alveolarization. *Developmental Cell*, **2011**, 21, 746-57	10.2	73
197	Rapid identification of yeast artificial chromosome clones by matrix pooling and crude lysate PCR. *Nucleic Acids Research*, **1990**, 18, 7191-2	20.1	73
196	Disruption of the ATXN1-CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. *Nature Genetics*, **2017**, 49, 527-536	36.3	71
195	Molecular cloning of the cDNA encoding a human renal sodium phosphate transport protein and its assignment to chromosome 6p21.3-p23. *Genomics*, **1993**, 18, 355-9	4.3	71
194	Expanding our understanding of polyglutamine diseases through mouse models. *Neuron*, **1999**, 24, 499-502	13.9	70
193	Restoration of Mecp2 expression in GABAergic neurons is sufficient to rescue multiple disease features in a mouse model of Rett syndrome. *ELife*, **2016**, 5,	8.9	69
192	Modelling brain diseases in mice: the challenges of design and analysis. *Nature Reviews Genetics*, **2003**, 4, 296-307	30.1	68
191	Discordance of muscular dystrophy in monozygotic female twins: evidence supporting asymmetric splitting of the inner cell mass in a manifesting carrier of Duchenne dystrophy. *American Journal of Medical Genetics Part A*, **1991**, 40, 354-64		68
190	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	13.9	68
189	Sequencing, expression analysis, and mapping of three unique human tropomodulin genes and their mouse orthologs. *Genomics*, **2000**, 63, 97-107	4.3	66

188	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. *Cell*, **2018**, 172, 924-936.e11	56.2	65
187	Manipulations of MeCP2 in glutamatergic neurons highlight their contributions to Rett and other neurological disorders. *ELife*, **2016**, 5,	8.9	65
186	Duplication of Atxn1l suppresses SCA1 neuropathology by decreasing incorporation of polyglutamine-expanded ataxin-1 into native complexes. *Nature Genetics*, **2007**, 39, 373-9	36.3	64
185	The expansion of the CAG repeat in ataxin-2 is a frequent cause of autosomal dominant spinocerebellar ataxia. *Neurology*, **1997**, 49, 1009-13	6.5	63
184	Enhanced SUMOylation in polyglutamine diseases. *Biochemical and Biophysical Research Communications*, **2002**, 293, 307-13	3.4	63
183	Rett syndrome: controlled study of an oral opiate antagonist, naltrexone. *Annals of Neurology*, **1994**, 35, 464-70	9.4	62
182	Cerebellar Transcriptome Profiles of ATXN1 Transgenic Mice Reveal SCA1 Disease Progression and Protection Pathways. *Neuron*, **2016**, 89, 1194-1207	13.9	60
181	Cell-specific expression of wild-type MeCP2 in mouse models of Rett syndrome yields insight about pathogenesis. *Human Molecular Genetics*, **2007**, 16, 2315-25	5.6	60
180	Somatic and germline instability of the ATTCT repeat in spinocerebellar ataxia type 10. *American Journal of Human Genetics*, **2004**, 74, 1216-24	11	60
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178	Mice lacking Tropomodulin-2 show enhanced long-term potentiation, hyperactivity, and deficits in learning and memory. *Molecular and Cellular Neurosciences*, **2003**, 23, 1-12	4.8	58
177	Genetic modifiers of MeCP2 function in Drosophila. *PLoS Genetics*, **2008**, 4, e1000179	6	57
176	Dissection of the cellular and molecular events that position cerebellar Purkinje cells: a study of the math1 null-mutant mouse. *Journal of Neuroscience*, **2002**, 22, 8110-6	6.6	57
175	Rett-causing mutations reveal two domains critical for MeCP2 function and for toxicity in MECP2 duplication syndrome mice. *ELife*, **2014**, 3,	8.9	57
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