Cheryl A Shoubridge

List of Publications by Year
in descending order

57 papers	3,070 citations	236925 25 h-index	168389 53 g-index
65 all docs	65 docs citations	65 times ranked	4716 citing authors

#	Article	IF	Citations
1	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	21.4	528
2	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	21.4	397
3	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. Nature Genetics, 2007, 39, 1127-1133.	21.4	228
4	The genetic landscape of intellectual disability arising from chromosome X. Trends in Genetics, 2009, 25, 308-316.	6.7	190
5	ARX spectrum disorders: making inroads into the molecular pathology. Human Mutation, 2010, 31, 889-900.	2.5	151
6	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. Nature Genetics, 2010, 42, 486-488.	21.4	134
7	Mutations of the UPF3B gene, which encodes a protein widely expressed in neurons, are associated with nonspecific mental retardation with or without autism. Molecular Psychiatry, 2010, 15, 767-776.	7.9	113
8	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. Molecular Psychiatry, 2012, 17, 1103-1115.	7.9	97
9	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
10	Large Scale Gene Expression Meta-Analysis Reveals Tissue-Specific, Sex-Biased Gene Expression in Humans. Frontiers in Genetics, 2016, 7, 183.	2.3	91
11	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
12	Aristaless-related homeobox gene, the gene responsible for West syndrome and related disorders, is a Groucho/transducin-like enhancer of split dependent transcriptional repressor. Neuroscience, 2007, 146, 236-247.	2.3	62
13	Degradation of IGF-I in the adult rat gastrointestinal tract is limited by a specific antiserum or the dietary protein casein. Journal of Endocrinology, 1995, 146, 215-225.	2.6	60
14	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. Neurology, 2021, 96, e1770-e1782.	1.1	53
15	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
16	Protein and gene expression analysis of Phf6, the gene mutated in the BÃ¶rjesonâ€“Forssmanâ€“Lehmann Syndrome of intellectual disability and obesity. Gene Expression Patterns, 2007, 7, 858-871.	0.8	45
17	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. European Journal of Human Genetics, 2016, 24, 373-380.	2.8	43
18	Molecular pathology of expanded polyalanine tract mutations in the Aristaless-related homeobox gene. Genomics, 2007, 90, 59-71.	2.9	42

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19	Polyalanine expansions drive a shift into Î±-helical clusters without amyloid-fibril formation. Nature Structural and Molecular Biology, 2015, 22, 1008-1015.	8.2	42
20	A Regulatory Path Associated with X-Linked Intellectual Disability and Epilepsy Links KDM5C to the Polyalanine Expansions in ARX. American Journal of Human Genetics, 2013, 92, 114-125.	6.2	39
21	IQSEC2mutation update and review of the female-specific phenotype spectrum including intellectual disability and epilepsy. Human Mutation, 2019, 40, 5-24.	2.5	36
22	Mutations in the nuclear localization sequence of the Aristaless related homeobox; sequestration of mutant ARX with IPO13 disrupts normal subcellular distribution of the transcription factor and retards cell division. PathoGenetics, 2010, 3, 1.	5.7	33
23	Enhancement of intestinal growth and repair by growth factors. Current Opinion in Pharmacology, 2001, 1, 568-574.	3.5	32
24	Subtle functional defects in the Arf-specific guanine nucleotide exchange factor IQSEC2 cause non-syndromic X-linked intellectual disability. Small GTPases, 2010, 1, 98-103.	1.6	31
25	Incorrect dosage of IQSEC2, a known intellectual disability and epilepsy gene, disrupts dendritic spine morphogenesis. Translational Psychiatry, 2017, 7, e1110-e1110.	4.8	27
26	Placental transcriptome co-expression analysis reveals conserved regulatory programs across gestation. BMC Genomics, 2017, 18, 10.	2.8	26
27	Screening and cell-based assessment of mutations in the Aristaless-related homeobox (ARX) gene. Clinical Genetics, 2011, 80, 510-522.	2.0	23
28	Unraveling the pathogenesis of ARX polyalanine tract variants using a clinical and molecular interfacing approach. Molecular Genetics & Genomic Medicine, 2015, 3, 203-214.	1.2	21
29	Systemic infusion of IGF-I or LR(3)IGF-I stimulates visceral organ growth and proliferation of gut tissues in suckling rats. American Journal of Physiology - Renal Physiology, 1997, 272, G522-G533.	3.4	20
30	Reduced polyalanine-expanded Arx mutant protein in developing mouse subpallium alters Lmo1 transcriptional regulation. Human Molecular Genetics, 2014, 23, 1084-1094.	2.9	20
31	ARX homeodomain mutations abolish DNA binding and lead to a loss of transcriptional repression. Human Molecular Genetics, 2012, 21, 1639-1647.	2.9	19
32	Refined mapping of X-linked reticulate pigmentary disorder and sequencing of candidate genes. Human Genetics, 2008, 123, 469-476.	3.8	18
33	Histone demethylase KDM5C is a SAHA-sensitive central hub at the crossroads of transcriptional axes involved in multiple neurodevelopmental disorders. Human Molecular Genetics, 2019, 28, 4089-4102.	2.9	18
34	Polyalanine Tract Disorders and Neurocognitive Phenotypes. Advances in Experimental Medicine and Biology, 2012, 769, 185-203.	1.6	18
35	Heterozygous loss of function of IQSEC2/Iqsec2 leads to increased activated Arf6 and severe neurocognitive seizure phenotype in females. Life Science Alliance, 2019, 2, e201900386.	2.8	18
36	ARX polyalanine expansion mutations lead to migration impediment in the rostral cortex coupled with a developmental deficit of calbindin-positive cortical GABAergic interneurons. Neuroscience, 2017, 357, 220-231.	2.3	16

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37	IGFBP mRNA expression in small intestine of rat during postnatal development. American Journal of Physiology - Renal Physiology, 2001, 281, G1378-G1384.	3.4	15
38	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. Human Genetics, 2015, 134, 1163-1182.	3.8	14
39	Gonadal mosaicism of a novel IQSEC2 variant causing female limited intellectual disability and epilepsy. European Journal of Human Genetics, 2017, 25, 763-767.	2.8	14
40	Systemically but Not Orogastrically Delivered Insulin-Like Growth Factor (IGF)-I and Long [Arg3]IGF-I Stimulates Intestinal Disaccharidase Activity in Two Age Groups of Suckling Rats. Pediatric Research, 1998, 44, 663-672.	2.3	13
41	Regulating transcriptional activity by phosphorylation: A new mechanism for the ARX homeodomain transcription factor. PLoS ONE, 2018, 13, e0206914.	2.5	11
42	Reinitiation of mRNA translation in a patient with X-linked infantile spasms with a protein-truncating variant in ARX. European Journal of Human Genetics, 2016, 24, 681-689.	2.8	10
43	An Emerging Female Phenotype with Lossâ€ofâ€Function Mutations in the Aristalessâ€ Related Homeodomain Transcription Factor ARX. Human Mutation, 2017, 38, 548-555.	2.5	10
44	Is there a Mendelian transmission ratio distortion of the c.429_452dup(24bp) polyalanine tract ARX mutation?. European Journal of Human Genetics, 2012, 20, 1311-1314.	2.8	9
45	Embryonic forebrain transcriptome of mice with polyalanine expansion mutations in theARXhomeobox gene. Human Molecular Genetics, 2016, 25, ddw360.	2.9	9
46	Splice variant in ARX leading to loss of Câ€terminal region in a boy with intellectual disability and infantile onset developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1483-1490.	1.2	9
47	Extensive phenotyping of two ARX polyalanine expansion mutation mouse models that span clinical spectrum of intellectual disability and epilepsy. Neurobiology of Disease, 2017, 105, 245-256.	4.4	8
48	Early 17Î²-estradiol treatment reduces seizures but not abnormal behaviour in mice with expanded polyalanine tracts in the Aristaless related homeobox gene (ARX). Neurobiology of Disease, 2021, 153, 105329.	4.4	6
49	X-Linked Lissencephaly With Absent Corpus Callosum and Abnormal Genitalia. Child Neurology Open, 2017, 4, 2329048X1773862.	1.1	6
50	PCR Amplification and Sequence Analysis of GC-Rich Sequences: Aristaless-Related Homeobox Example. Methods in Molecular Biology, 2013, 1017, 105-120.	0.9	5
51	<scp>IQSEC2</scp>â€related encephalopathy in males due to missense variants in the pleckstrin homology domain. Clinical Genetics, 2022, 102, 72-77.	2.0	4
52	A novel ARX loss of function variant in female monozygotic twins is associated with chorea. European Journal of Medical Genetics, 2021, 64, 104315.	1.3	3
53	Production of a human epidermal growth factor fusion protein and its degradation in rat gastrointestinal flushings. Journal of Molecular Endocrinology, 1996, 16, 89-97.	2.5	2
54	Constraint and conservation of pairedâ€type homeodomains predicts the clinical outcome of missense variants of uncertain significance. Human Mutation, 2020, 41, 1407-1424.	2.5	2

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55	Challenges of â€œStickyâ€•Co-immunoprecipitation: Polyalanine Tract Proteinâ€“Protein Interactions. Methods in Molecular Biology, 2013, 1017, 121-133.	0.9	1
56	Preferential Intestinal Delivery of Long[Arg3] Insulin-Like Growth Factor (LR3IGF-I) over IGF-I in Preweaning and Adult Rats. Endocrinology, 2003, 144, 1887-1893.	2.8	0
57	Different types of diseaseâ€causing noncoding variants revealed by genomic and gene expression analyses in families with Xâ€linked intellectual disability. Human Mutation, 2021, 42, 835-847.	2.5	0