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	<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
2	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
3	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
4	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
5	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
6	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
7	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
8	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
9	IQSEC2mutation update and review of the female-specific phenotype spectrum including intellectual disability and epilepsy. Human Mutation, 2019, 40, 5-24.	2.5	36
10	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
11	Regulating transcriptional activity by phosphorylation: A new mechanism for the ARX homeodomain transcription factor. PLoS ONE, 2018, 13, e0206914.	2.5	11
12	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
13	Placental transcriptome co-expression analysis reveals conserved regulatory programs across gestation. BMC Genomics, 2017, 18, 10.	2.8	26
14	Incorrect dosage of IQSEC2, a known intellectual disability and epilepsy gene, disrupts dendritic spine morphogenesis. Translational Psychiatry, 2017, 7, e1110-e1110.	4.8	27
15	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
16	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
17	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
18	X-Linked Lissencephaly With Absent Corpus Callosum and Abnormal Genitalia. Child Neurology Open, 2017, 4, 2329048X1773862.	1.1	6

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19	Large Scale Gene Expression Meta-Analysis Reveals Tissue-Specific, Sex-Biased Gene Expression in Humans. Frontiers in Genetics, 2016, 7, 183.	2.3	91
20	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
21	Embryonic forebrain transcriptome of mice with polyalanine expansion mutations in theARXhomeobox gene. Human Molecular Genetics, 2016, 25, ddw360.	2.9	9
22	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
23	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
24	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
25	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
26	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
27	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
28	Reduced polyalanine-expanded Arx mutant protein in developing mouse subpallium alters Lmo1 transcriptional regulation. Human Molecular Genetics, 2014, 23, 1084-1094.	2.9	20
29	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
30	PCR Amplification and Sequence Analysis of GC-Rich Sequences: Aristaless-Related Homeobox Example. Methods in Molecular Biology, 2013, 1017, 105-120.	0.9	5
31	Challenges of â€œStickyâ€•Co-immunoprecipitation: Polyalanine Tract Proteinâ€“Protein Interactions. Methods in Molecular Biology, 2013, 1017, 121-133.	0.9	1
32	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
33	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
34	ARX homeodomain mutations abolish DNA binding and lead to a loss of transcriptional repression. Human Molecular Genetics, 2012, 21, 1639-1647.	2.9	19
35	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
36	Polyalanine Tract Disorders and Neurocognitive Phenotypes. Advances in Experimental Medicine and Biology, 2012, 769, 185-203.	1.6	18

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37	Screening and cell-based assessment of mutations in the Aristaless-related homeobox (ARX) gene. Clinical Genetics, 2011, 80, 510-522.	2.0	23
38	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
39	ARX spectrum disorders: making inroads into the molecular pathology. Human Mutation, 2010, 31, 889-900.	2.5	151
40	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
41	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. Nature Genetics, 2010, 42, 486-488.	21.4	134
42	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
43	The genetic landscape of intellectual disability arising from chromosome X. Trends in Genetics, 2009, 25, 308-316.	6.7	190
44	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	21.4	528
45	Refined mapping of X-linked reticulate pigmentary disorder and sequencing of candidate genes. Human Genetics, 2008, 123, 469-476.	3.8	18
46	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	21.4	397
47	Molecular pathology of expanded polyalanine tract mutations in the Aristaless-related homeobox gene. Genomics, 2007, 90, 59-71.	2.9	42
48	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
49	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
50	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
51	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
52	Enhancement of intestinal growth and repair by growth factors. Current Opinion in Pharmacology, 2001, 1, 568-574.	3.5	32
53	IGFBP mRNA expression in small intestine of rat during postnatal development. American Journal of Physiology - Renal Physiology, 2001, 281, G1378-G1384.	3.4	15
54	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

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55	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
56	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
57	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