Richard J Gibbons

List of Publications by Year
in descending order

108 papers	10,283 citations	43973 48 h-index	35952 97 g-index
113 all docs	113 docs citations	113 times ranked	11337 citing authors

#	Article	IF	Citations
1	Distinct Factors Control Histone Variant H3.3 Localization at Specific Genomic Regions. Cell, 2010, 140, 678-691.	13.5	1,069
2	Mutations in a putative global transcriptional regulator cause X-linked mental retardation with Î±-thalassemia (ATR-X syndrome). Cell, 1995, 80, 837-845.	13.5	583
3	Mutations in ATRX, encoding a SWI/SNF-like protein, cause diverse changes in the pattern of DNA methylation. Nature Genetics, 2000, 24, 368-371.	9.4	476
4	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. Nature Genetics, 2014, 46, 205-212.	9.4	417
5	ATR-X Syndrome Protein Targets Tandem Repeats and Influences Allele-Specific Expression in a Size-Dependent Manner. Cell, 2010, 143, 367-378.	13.5	365
6	The ATRX syndrome protein forms a chromatin-remodeling complex with Daxx and localizes in promyelocytic leukemia nuclear bodies. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 10635-10640.	3.3	322
7	Genetic dissection of the Î±-globin super-enhancer in vivo. Nature Genetics, 2016, 48, 895-903.	9.4	308
8	The Fanconi Anemia Pathway Maintains Genome Stability by Coordinating Replication and Transcription. Molecular Cell, 2015, 60, 351-361.	4.5	283
9	A Regulatory SNP Causes a Human Genetic Disease by Creating a New Transcriptional Promoter. Science, 2006, 312, 1215-1217.	6.0	254
10	Intragenic Enhancers Act as Alternative Promoters. Molecular Cell, 2012, 45, 447-458.	4.5	237
11	Localization of a putative transcriptional regulator (ATRX) at pericentromeric heterochromatin and the short arms of acrocentric chromosomes. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 13983-13988.	3.3	233
12	Suppression of the alternative lengthening of telomere pathway by the chromatin remodelling factor ATRX. Nature Communications, 2015, 6, 7538.	5.8	219
13	ATRX encodes a novel member of the SNF2 family of proteins: mutations point to a common mechanism underlying the ATR-X syndrome. Human Molecular Genetics, 1996, 5, 1899-1907.	1.4	217
14	Molecular-clinical spectrum of the ATR-X syndrome. American Journal of Medical Genetics Part A, 2000, 97, 204-212.	2.4	208
15	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
16	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	9.4	196
17	Combinatorial readout of histone H3 modifications specifies localization of ATRX to heterochromatin. Nature Structural and Molecular Biology, 2011, 18, 777-782.	3.6	187
18	An interspecies analysis reveals a key role for unmethylated CpG dinucleotides in vertebrate Polycomb complex recruitment. EMBO Journal, 2012, 31, 317-329.	3.5	173

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19	The chromatin-remodeling protein ATRX is critical for neuronal survival during corticogenesis. Journal of Clinical Investigation, 2005, 115, 258-267.	3.9	169
20	Mutations in the chromatin-associated protein ATRX. Human Mutation, 2008, 29, 796-802.	1.1	155
21	ATRX Plays a Key Role in Maintaining Silencing at Interstitial Heterochromatic Loci and Imprinted Genes. Cell Reports, 2015, 11, 405-418.	2.9	152
22	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	9.4	142
23	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	1.4	142
24	Histone modifying and chromatin remodelling enzymes in cancer and dysplastic syndromes. Human Molecular Genetics, 2005, 14, R85-R92.	1.4	141
25	Loss of Atrx Affects Trophoblast Development and the Pattern of X-Inactivation in Extraembryonic Tissues. PLoS Genetics, 2006, 2, e58.	1.5	140
26	Structural consequences of disease-causing mutations in the ATRX-DNMT3-DNMT3L (ADD) domain of the chromatin-associated protein ATRX. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 11939-11944.	3.3	138
27	Identification of acquired somatic mutations in the gene encoding chromatin-remodeling factor ATRX in the Î±-thalassemia myelodysplasia syndrome (ATMDS). Nature Genetics, 2003, 34, 446-449.	9.4	132
28	Alpha thalassaemia-mental retardation, X linked. Orphanet Journal of Rare Diseases, 2006, 1, 15.	1.2	130
29	The chromatin-remodeling protein ATRX is critical for neuronal survival during corticogenesis. Journal of Clinical Investigation, 2005, 115, 258-267.	3.9	119
30	Mutations in the general transcription factor TFIIH result in beta-thalassaemia in individuals with trichothiodystrophy. Human Molecular Genetics, 2001, 10, 2797-2802.	1.4	115
31	The chromatin remodeller ATRX: a repeat offender in human disease. Trends in Biochemical Sciences, 2013, 38, 461-466.	3.7	103
32	Î±-Globin as a molecular target in the treatment of Î²-thalassemia. Blood, 2015, 125, 3694-3701.	0.6	102
33	The chromatin remodelling factor <scp>ATRX</scp> suppresses Râ€loops in transcribed telomeric repeats. EMBO Reports, 2017, 18, 914-928.	2.0	99
34	Acquired Î±-thalassemia in association with myelodysplastic syndrome and other hematologic malignancies. Blood, 2005, 105, 443-452.	0.6	95
35	Mutant IDH1 Promotes Glioma Formation InÂVivo. Cell Reports, 2018, 23, 1553-1564.	2.9	91
36	Editing an Î±-globin enhancer in primary human hematopoietic stem cells as a treatment for Î²-thalassemia. Nature Communications, 2017, 8, 424.	5.8	85

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37	Acquired somatic ATRX mutations in myelodysplastic syndrome associated with Î± thalassemia (ATMDS) convey a more severe hematologic phenotype than germline ATRX mutations. Blood, 2004, 103, 2019-2026.	0.6	84
38	ATRX Dysfunction Induces Replication Defects in Primary Mouse Cells. PLoS ONE, 2014, 9, e92915.	1.1	84
39	Adventitious changes in long-range gene expression caused by polymorphic structural variation and promoter competition. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21771-21776.	3.3	77
40	Mutations in KrÃ¼ppel-like factor 1 cause transfusion-dependent hemolytic anemia and persistence of embryonic globin gene expression. Blood, 2014, 123, 1586-1595.	0.6	76
41	Absence of the XIST gene from late-replicating isodicentric X chromosomes in leukaemia. Human Molecular Genetics, 1994, 3, 1053-1059.	1.4	72
42	A nonsense mutation of theATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	2.8	72
43	Tissue-specific histone modification and transcription factor binding in Î± globin gene expression. Blood, 2007, 110, 4503-4510.	0.6	69
44	Comparison of the human and murine ATRX gene identifies highly conserved, functionally important domains. Mammalian Genome, 1998, 9, 400-403.	1.0	64
45	A newly defined X linked mental retardation syndrome associated with alpha thalassaemia.. Journal of Medical Genetics, 1991, 28, 729-733.	1.5	62
46	Somatic point mutations in RUNX1/CBFA2/AML1 are common in high-risk myelodysplastic syndrome, but not in myelofibrosis with myeloid metaplasia. European Journal of Haematology, 2005, 74, 47-53.	1.1	62
47	Neuronal Death Resulting from Targeted Disruption of the Snf2 Protein ATRX Is Mediated by p53. Journal of Neuroscience, 2008, 28, 12570-12580.	1.7	61
48	Generation of bivalent chromatin domains during cell fate decisions. Epigenetics and Chromatin, 2011, 4, 9.	1.8	54
49	A conserved truncated isoform of the ATR-X syndrome protein lacking the SWI/SNF-homology domain. Gene, 2004, 326, 23-34.	1.0	53
50	X-linked Î±-thalassemia/mental retardation (ATR-X) syndrome: A new kindred with severe genital anomalies and mild hematologic expression. American Journal of Medical Genetics Part A, 1995, 55, 302-306.	2.4	50
51	Monosomy for the most telomeric, gene-rich region of the short arm of human chromosome 16 causes minimal phenotypic effects. European Journal of Human Genetics, 2001, 9, 217-225.	1.4	47
52	Understanding Î±-Globin Gene Regulation: Aiming to Improve the Management of Thalassemia. Annals of the New York Academy of Sciences, 2005, 1054, 92-102.	1.8	47
53	Functional significance of mutations in the Snf2 domain of ATRX. Human Molecular Genetics, 2011, 20, 2603-2610.	1.4	46
54	Understanding Î±â€globin gene regulation and implications for the treatment of Î²â€thalassemia. Annals of the New York Academy of Sciences, 2016, 1368, 16-24.	1.8	44

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55	ATRX and the replication of structured DNA. Current Opinion in Genetics and Development, 2013, 23, 289-294.	1.5	40
56	Attenuation of an amino-terminal premature stop codon mutation in the ATRX gene by an alternative mode of translational initiation. Journal of Medical Genetics, 2004, 41, 951-956.	1.5	39
57	Nprl3 is required for normal development of the cardiovascular system. Mammalian Genome, 2012, 23, 404-415.	1.0	38
58	The genetic basis for mental retardation. QJM - Monthly Journal of the Association of Physicians, 1996, 89, 169-176.	0.2	36
59	The Molecular Basis of Î±-Thalassemia: A Model for Understanding Human Molecular Genetics. Hematology/Oncology Clinics of North America, 2010, 24, 1033-1054.	0.9	36
60	The macroH2A1.2 histone variant links ATRX loss to alternative telomere lengthening. Nature Structural and Molecular Biology, 2019, 26, 213-219.	3.6	36
61	JAK2V617F promotes replication fork stalling with disease-restricted impairment of the intra-S checkpoint response. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15190-15195.	3.3	35
62	Deletion of the Î±-globin gene cluster as a cause of acquired Î±-thalassemia in myelodysplastic syndrome. Blood, 2004, 103, 1518-1520.	0.6	34
63	Altered visual function and interneuron survival in Atrx knockout mice: inference for the human syndrome. Human Molecular Genetics, 2009, 18, 966-977.	1.4	34
64	Using Genomics to Study How Chromatin Influences Gene Expression. Annual Review of Genomics and Human Genetics, 2007, 8, 299-325.	2.5	33
65	Selective silencing of Î±-globin by the histone demethylase inhibitor IOX1: a potentially new pathway for treatment of Î²-thalassemia. Haematologica, 2017, 102, e80-e84.	1.7	33
66	Defining the Cause of Skewed X-Chromosome Inactivation in X-Linked Mental Retardation by Use of a Mouse Model. American Journal of Human Genetics, 2007, 80, 1138-1149.	2.6	32
67	X linked alpha thalassaemia/mental retardation: spectrum of clinical features in three related males.. Journal of Medical Genetics, 1991, 28, 738-741.	1.5	31
68	The non-deletion alpha thalassaemia/mental retardation syndrome: further support for X linkage.. Journal of Medical Genetics, 1991, 28, 742-745.	1.5	31
69	Â-Thalassemia, Mental Retardation, and Myelodysplastic Syndrome. Cold Spring Harbor Perspectives in Medicine, 2012, 2, a011759-a011759.	2.9	29
70	Germline and gonosomal mosaicism in the ATR-X syndrome. European Journal of Human Genetics, 1999, 7, 933-936.	1.4	26
71	The Î±-Thalassemia/Mental Retardation Syndromes. Medicine (United States), 1996, 75, 45-52.	0.4	24
72	Brachydactyly Type B: Linkage to Chromosome 9q22 and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 1999, 64, 578-585.	2.6	24

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73	Gastrointestinal phenotype of ATR-X syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1172-1176.	0.7	24
74	Synergistic silencing of Î±-globin and induction of Î³-globin by histone deacetylase inhibitor, vorinostat as a potential therapy for Î²-thalassaemia. Scientific Reports, 2019, 9, 11649.	1.6	21
75	The chromatin remodeller ATRX facilitates diverse nuclear processes, in a stochastic manner, in both heterochromatin and euchromatin. Nature Communications, 2022, 13, .	5.8	20
76	The non-deletion type of alpha thalassaemia/mental retardation: a recognisable dysmorphic syndrome with X linked inheritance.. Journal of Medical Genetics, 1991, 28, 724-724.	1.5	19
77	Lumping Juberg-Marsidi syndrome and X-linked Î±-thalassemia/mental retardation syndrome?. American Journal of Medical Genetics Part A, 1995, 55, 300-301.	2.4	19
78	ATRX Contributes to MeCP2-Mediated Pericentric Heterochromatin Organization during Neural Differentiation. International Journal of Molecular Sciences, 2019, 20, 5371.	1.8	19
79	Maintaining memory of silencing at imprinted differentially methylated regions. Cellular and Molecular Life Sciences, 2016, 73, 1871-1879.	2.4	18
80	A gain-of-function single nucleotide variant creates a new promoter which acts as an orientation-dependent enhancer-blocker. Nature Communications, 2021, 12, 3806.	5.8	18
81	Anaemia among females in child-bearing age: Relative contributions, effects and interactions of Î±- and Î²-thalassaemia. PLoS ONE, 2018, 13, e0206928.	1.1	17
82	Robust CRISPR/Cas9 Genome Editing of the HUDEP-2 Erythroid Precursor Line Using Plasmids and Single-Stranded Oligonucleotide Donors. Methods and Protocols, 2018, 1, 28.	0.9	17
83	An evolutionarily ancient mechanism for regulation of hemoglobin expression in vertebrate red cells. Blood, 2020, 136, 269-278.	0.6	16
84	Asplenia in ATR-X syndrome: A second report. American Journal of Medical Genetics, Part A, 2005, 139A, 37-39.	0.7	15
85	2 new cases of pontocerebellar hypoplasia type 10 identified by whole exome sequencing in a Turkish family. European Journal of Medical Genetics, 2018, 61, 273-279.	0.7	14
86	The Loss of ATRX Increases Susceptibility to Pancreatic Injury and Oncogenic KRAS in Female But Not Male Mice. Cellular and Molecular Gastroenterology and Hepatology, 2019, 7, 93-113.	2.3	14
87	Alpha thalassaemia mental retardation (ATR-X): an atypical family.. Archives of Disease in Childhood, 1994, 70, 439-440.	1.0	13
88	High-resolution analysis of cis -acting regulatory networks at the Î±-globin locus. Philosophical Transactions of the Royal Society B: Biological Sciences, 2013, 368, 20120361.	1.8	12
89	A novel mutation in the last exon of ATRX in a patient with alpha-thalassemia myelodysplastic syndrome. European Journal of Haematology, 2006, 76, 432-435.	1.1	10
90	X-linked Î± thalassaemia/mental retardation syndrome: a case with gonadal dysgenesis, caused by a novel mutation in ATRX gene. Clinical Dysmorphology, 2009, 18, 168-171.	0.1	10

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91	Analysis of Sequence Variation Underlying Tissue-specific Transcription Factor Binding and Gene Expression. Human Mutation, 2013, 34, 1140-1148.	1.1	10
92	The role of Xâ€inactivation in the gender bias of patients with acquired Î±â€thalassaemia and myelodysplastic syndrome (ATMDS). British Journal of Haematology, 2009, 144, 538-545.	1.2	9
93	Genetic and functional insights into CDA-I prevalence and pathogenesis. Journal of Medical Genetics, 2021, 58, 185-195.	1.5	9
94	Variant ATRX Syndrome with Dysfunction ofATRXandMAGT1Genes. Human Mutation, 2014, 35, 58-62.	1.1	7
95	How to Tackle Challenging ChIP-Seq, with Long-Range Cross-Linking, Using ATRX as an Example. Methods in Molecular Biology, 2018, 1832, 105-130.	0.4	7
96	Phenotypic and molecular characterization of a serum-free miniature erythroid differentiation system suitable for high-throughput screening and single-cell assays. Experimental Hematology, 2018, 60, 10-20.	0.2	6
97	ATR-X Syndrome. , 0, , 309-334.		6
98	ATRX: Taming tandem repeats. Cell Cycle, 2010, 9, 4605-4606.	1.3	4
99	A Novel Splicing Mutation in the Gene Encoding the Chromatin-Associated Factor ATRX Associated with Acquired Hemoglobin H Disease in Myelodysplastic Syndrome (ATMDS).. Blood, 2004, 104, 3606-3606.	0.6	3
100	Unusual Types of Î± Thalassemia. , 0, , 296-320.		2
101	Persistence of skewed X-chromosome inactivation in pre-B acute lymphoblastic leukemia of a female ATRX mutation carrier. Blood Advances, 2019, 3, 2627-2631.	2.5	2
102	Two Novel Somatic Mutations of the ATRX Gene in Female Patients with Acquired Alpha Thalassemia of Myelodysplastic Syndrome (ATMDS).. Blood, 2006, 108, 1765-1765.	0.6	2
103	5-Azacytidine treatment of the patient with ATMDS. European Journal of Haematology, 2006, 76, 453-453.	1.1	0
104	Research Highlights. Epigenomics, 2009, 1, 231-234.	1.0	0
105	102. Cloning and characterisation of tammar ATRX and ATRY. Reproduction, Fertility and Development, 2003, 15, 102.	0.1	0
106	Epigenetics and its Genetic Syndromes. , 2008, , 155-174.		0
107	Analysis of DNA Methylation at the Human Alpha Globin Cluster during Hematopoiesis.. Blood, 2008, 112, 1861-1861.	0.6	0
108	GENE SILENCING. , 2013, , 143-184.		0