Richard J Gibbons

List of Publications by Year
in descending order

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109 papers	10,283 citations	44069 48 h-index	36028 97 g-index
113 all docs	113 docs citations	113 times ranked	11337 citing authors

#	Article	IF	Citations
1	The chromatin remodeller ATRX facilitates diverse nuclear processes, in a stochastic manner, in both heterochromatin and euchromatin. Nature Communications, 2022, 13, .	12.8	20
2	Genetic and functional insights into CDA-I prevalence and pathogenesis. Journal of Medical Genetics, 2021, 58, 185-195.	3.2	9
3	A gain-of-function single nucleotide variant creates a new promoter which acts as an orientation-dependent enhancer-blocker. Nature Communications, 2021, 12, 3806.	12.8	18
4	An evolutionarily ancient mechanism for regulation of hemoglobin expression in vertebrate red cells. Blood, 2020, 136, 269-278.	1.4	16
5	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.	3.3	21
6	ATRX Contributes to MeCP2-Mediated Pericentric Heterochromatin Organization during Neural Differentiation. International Journal of Molecular Sciences, 2019, 20, 5371.	4.1	19
7	The macroH2A1.2 histone variant links ATRX loss to alternative telomere lengthening. Nature Structural and Molecular Biology, 2019, 26, 213-219.	8.2	36
8	Persistence of skewed X-chromosome inactivation in pre-B acute lymphoblastic leukemia of a female ATRX mutation carrier. Blood Advances, 2019, 3, 2627-2631.	5.2	2
9	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.	4.5	14
10	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
11	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.	0.4	6
12	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.	1.3	14
13	Mutant IDH1 Promotes Glioma Formation InÂVivo. Cell Reports, 2018, 23, 1553-1564.	6.4	91
14	Anaemia among females in child-bearing age: Relative contributions, effects and interactions of Î±- and Î²-thalassaemia. PLoS ONE, 2018, 13, e0206928.	2.5	17
15	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.	2.0	17
16	How to Tackle Challenging ChIP-Seq, with Long-Range Cross-Linking, Using ATRX as an Example. Methods in Molecular Biology, 2018, 1832, 105-130.	0.9	7
17	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.	3.5	33
18	The chromatin remodelling factor <scp>ATRX</scp> suppresses Râ€loops in transcribed telomeric repeats. EMBO Reports, 2017, 18, 914-928.	4.5	99

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19	Editing an Î±-globin enhancer in primary human hematopoietic stem cells as a treatment for Î²-thalassemia. Nature Communications, 2017, 8, 424.	12.8	85
20	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.	3.8	44
21	Genetic dissection of the Î±-globin super-enhancer in vivo. Nature Genetics, 2016, 48, 895-903.	21.4	308
22	Maintaining memory of silencing at imprinted differentially methylated regions. Cellular and Molecular Life Sciences, 2016, 73, 1871-1879.	5.4	18
23	Suppression of the alternative lengthening of telomere pathway by the chromatin remodelling factor ATRX. Nature Communications, 2015, 6, 7538.	12.8	219
24	ATRX Plays a Key Role in Maintaining Silencing at Interstitial Heterochromatic Loci and Imprinted Genes. Cell Reports, 2015, 11, 405-418.	6.4	152
25	Î±-Globin as a molecular target in the treatment of Î²-thalassemia. Blood, 2015, 125, 3694-3701.	1.4	102
26	The Fanconi Anemia Pathway Maintains Genome Stability by Coordinating Replication and Transcription. Molecular Cell, 2015, 60, 351-361.	9.7	283
27	ATRX Dysfunction Induces Replication Defects in Primary Mouse Cells. PLoS ONE, 2014, 9, e92915.	2.5	84
28	Mutations in KrÃ¼ppel-like factor 1 cause transfusion-dependent hemolytic anemia and persistence of embryonic globin gene expression. Blood, 2014, 123, 1586-1595.	1.4	76
29	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. Nature Genetics, 2014, 46, 205-212.	21.4	417
30	Variant ATRX Syndrome with Dysfunction ofATRXandMAGT1Genes. Human Mutation, 2014, 35, 58-62.	2.5	7
31	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.	7.1	35
32	The chromatin remodeller ATRX: a repeat offender in human disease. Trends in Biochemical Sciences, 2013, 38, 461-466.	7.5	103
33	ATRX and the replication of structured DNA. Current Opinion in Genetics and Development, 2013, 23, 289-294.	3.3	40
34	Analysis of Sequence Variation Underlying Tissue-specific Transcription Factor Binding and Gene Expression. Human Mutation, 2013, 34, 1140-1148.	2.5	10
35	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.	4.0	12
36	GENE SILENCING. , 2013, , 143-184.		0

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37	An interspecies analysis reveals a key role for unmethylated CpG dinucleotides in vertebrate Polycomb complex recruitment. EMBO Journal, 2012, 31, 317-329.	7.8	173
38	Â-Thalassemia, Mental Retardation, and Myelodysplastic Syndrome. Cold Spring Harbor Perspectives in Medicine, 2012, 2, a011759-a011759.	6.2	29
39	Intragenic Enhancers Act as Alternative Promoters. Molecular Cell, 2012, 45, 447-458.	9.7	237
40	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.	2.8	142
41	Nprl3 is required for normal development of the cardiovascular system. Mammalian Genome, 2012, 23, 404-415.	2.2	38
42	Combinatorial readout of histone H3 modifications specifies localization of ATRX to heterochromatin. Nature Structural and Molecular Biology, 2011, 18, 777-782.	8.2	187
43	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	21.4	142
44	Generation of bivalent chromatin domains during cell fate decisions. Epigenetics and Chromatin, 2011, 4, 9.	3.9	54
45	Functional significance of mutations in the Snf2 domain of ATRX. Human Molecular Genetics, 2011, 20, 2603-2610.	2.9	46
46	ATRX: Taming tandem repeats. Cell Cycle, 2010, 9, 4605-4606.	2.6	4
47	Distinct Factors Control Histone Variant H3.3 Localization at Specific Genomic Regions. Cell, 2010, 140, 678-691.	28.9	1,069
48	ATR-X Syndrome Protein Targets Tandem Repeats and Influences Allele-Specific Expression in a Size-Dependent Manner. Cell, 2010, 143, 367-378.	28.9	365
49	The Molecular Basis of Î±-Thalassemia: A Model for Understanding Human Molecular Genetics. Hematology/Oncology Clinics of North America, 2010, 24, 1033-1054.	2.2	36
50	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.	7.1	77
51	Research Highlights. Epigenomics, 2009, 1, 231-234.	2.1	0
52	Altered visual function and interneuron survival in Atrx knockout mice: inference for the human syndrome. Human Molecular Genetics, 2009, 18, 966-977.	2.9	34
53	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.	2.5	9
54	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.	0.3	10

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55	Mutations in the chromatin-associated protein ATRX. Human Mutation, 2008, 29, 796-802.	2.5	155
56	Neuronal Death Resulting from Targeted Disruption of the Snf2 Protein ATRX Is Mediated by p53. Journal of Neuroscience, 2008, 28, 12570-12580.	3.6	61
57	Epigenetics and its Genetic Syndromes. , 2008, , 155-174.		0
58	Analysis of DNA Methylation at the Human Alpha Globin Cluster during Hematopoiesis.. Blood, 2008, 112, 1861-1861.	1.4	0
59	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.	7.1	138
60	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.	6.2	32
61	Tissue-specific histone modification and transcription factor binding in Î± globin gene expression. Blood, 2007, 110, 4503-4510.	1.4	69
62	Using Genomics to Study How Chromatin Influences Gene Expression. Annual Review of Genomics and Human Genetics, 2007, 8, 299-325.	6.2	33
63	A Regulatory SNP Causes a Human Genetic Disease by Creating a New Transcriptional Promoter. Science, 2006, 312, 1215-1217.	12.6	254
64	Alpha thalassaemia-mental retardation, X linked. Orphanet Journal of Rare Diseases, 2006, 1, 15.	2.7	130
65	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.	2.2	10
66	5-Azacytidine treatment of the patient with ATMDS. European Journal of Haematology, 2006, 76, 453-453.	2.2	0
67	Gastrointestinal phenotype of ATR-X syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1172-1176.	1.2	24
68	Loss of Atrx Affects Trophoblast Development and the Pattern of X-Inactivation in Extraembryonic Tissues. PLoS Genetics, 2006, 2, e58.	3.5	140
69	Two Novel Somatic Mutations of the ATRX Gene in Female Patients with Acquired Alpha Thalassemia of Myelodysplastic Syndrome (ATMDS).. Blood, 2006, 108, 1765-1765.	1.4	2
70	Acquired Î±-thalassemia in association with myelodysplastic syndrome and other hematologic malignancies. Blood, 2005, 105, 443-452.	1.4	95
71	Understanding Î±-Globin Gene Regulation: Aiming to Improve the Management of Thalassemia. Annals of the New York Academy of Sciences, 2005, 1054, 92-102.	3.8	47
72	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.	2.2	62

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73	Asplenia in ATR-X syndrome: A second report. American Journal of Medical Genetics, Part A, 2005, 139A, 37-39.	1.2	15
74	Histone modifying and chromatin remodelling enzymes in cancer and dysplastic syndromes. Human Molecular Genetics, 2005, 14, R85-R92.	2.9	141
75	The chromatin-remodeling protein ATRX is critical for neuronal survival during corticogenesis. Journal of Clinical Investigation, 2005, 115, 258-267.	8.2	169
76	The chromatin-remodeling protein ATRX is critical for neuronal survival during corticogenesis. Journal of Clinical Investigation, 2005, 115, 258-267.	8.2	119
77	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.	3.2	39
78	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.	1.4	84
79	A conserved truncated isoform of the ATR-X syndrome protein lacking the SWI/SNF-homology domain. Gene, 2004, 326, 23-34.	2.2	53
80	Deletion of the Î±-globin gene cluster as a cause of acquired Î±-thalassemia in myelodysplastic syndrome. Blood, 2004, 103, 1518-1520.	1.4	34
81	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.	1.4	3
82	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.	21.4	132
83	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.	7.1	322
84	102. Cloning and characterisation of tammar ATRX and ATRY. Reproduction, Fertility and Development, 2003, 15, 102.	0.4	0
85	Mutations in the general transcription factor TFIIH result in beta-thalassaemia in individuals with trichothiodystrophy. Human Molecular Genetics, 2001, 10, 2797-2802.	2.9	115
86	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.	2.8	47
87	Molecular-clinical spectrum of the ATR-X syndrome. American Journal of Medical Genetics Part A, 2000, 97, 204-212.	2.4	208
88	A nonsense mutation of theATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	5.3	72
89	Mutations in ATRX, encoding a SWI/SNF-like protein, cause diverse changes in the pattern of DNA methylation. Nature Genetics, 2000, 24, 368-371.	21.4	476
90	A nonsense mutation of the ATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	5.3	0

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91	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.	7.1	233
92	Germline and gonosomal mosaicism in the ATR-X syndrome. European Journal of Human Genetics, 1999, 7, 933-936.	2.8	26
93	Brachydactyly Type B: Linkage to Chromosome 9q22 and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 1999, 64, 578-585.	6.2	24
94	Comparison of the human and murine ATRX gene identifies highly conserved, functionally important domains. Mammalian Genome, 1998, 9, 400-403.	2.2	64
95	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
96	The genetic basis for mental retardation. QJM - Monthly Journal of the Association of Physicians, 1996, 89, 169-176.	0.5	36
97	The Î±-Thalassemia/Mental Retardation Syndromes. Medicine (United States), 1996, 75, 45-52.	1.0	24
98	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.	2.9	217
99	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
100	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
101	Mutations in a putative global transcriptional regulator cause X-linked mental retardation with Î±-thalassemia (ATR-X syndrome). Cell, 1995, 80, 837-845.	28.9	583
102	Alpha thalassaemia mental retardation (ATR-X): an atypical family.. Archives of Disease in Childhood, 1994, 70, 439-440.	1.9	13
103	Absence of the XIST gene from late-replicating isodicentric X chromosomes in leukaemia. Human Molecular Genetics, 1994, 3, 1053-1059.	2.9	72
104	A newly defined X linked mental retardation syndrome associated with alpha thalassaemia.. Journal of Medical Genetics, 1991, 28, 729-733.	3.2	62
105	X linked alpha thalassaemia/mental retardation: spectrum of clinical features in three related males.. Journal of Medical Genetics, 1991, 28, 738-741.	3.2	31
106	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.	3.2	19
107	The non-deletion alpha thalassaemia/mental retardation syndrome: further support for X linkage.. Journal of Medical Genetics, 1991, 28, 742-745.	3.2	31
108	Unusual Types of Î± Thalassemia. , 0, , 296-320.		2

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109	ATR-X Syndrome. , 0, , 309-334.		6