Andrea Riccio

List of Publications by Year
in descending order

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111 papers	7,187 citations	66250 44 h-index	66518 82 g-index
112 all docs	112 docs citations	112 times ranked	7472 citing authors

#	Article	IF	Citations
1	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	1.0	8
2	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	1.8	14
3	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwithâ€“Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clinical Epigenetics, 2022, 14, .	1.8	7
4	Biallelic variant in cyclin B3 is associated with failure of maternal meiosis II and recurrent digynic triploidy. Journal of Medical Genetics, 2021, 58, 783-788.	1.5	9
5	ZBTB2 protein is a new partner of the Nucleosome Remodeling and Deacetylase (NuRD) complex. International Journal of Biological Macromolecules, 2021, 168, 67-76.	3.6	2
6	A paternally inherited 1.4â€‰kb deletion of the 11p15.5 imprinting center 2 is associated with a mild familial Silverâ€“Russell syndrome phenotype. European Journal of Human Genetics, 2021, 29, 447-454.	1.4	4
7	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. Genes, 2021, 12, 581.	1.0	5
8	The number of the CTCF binding sites of the H19/IGF2:IG-DMR correlates with DNA methylation and expression imprinting in a humanized mouse model. Human Molecular Genetics, 2021, 30, 1509-1520.	1.4	10
9	Variable Expressivity of the Beckwith-Wiedemann Syndrome in Four Pedigrees Segregating Loss-of-Function Variants of CDKN1C. Genes, 2021, 12, 706.	1.0	2
10	Zfp57 inactivation illustrates the role of ICR methylation in imprinted gene expression during neural differentiation of mouse ESCs. Scientific Reports, 2021, 11, 13802.	1.6	7
11	Both Epimutations and Chromosome Aberrations Affect Multiple Imprinted Loci in Aggressive Wilms Tumors. Cancers, 2020, 12, 3411.	1.7	6
12	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. Clinical Epigenetics, 2020, 12, 139.	1.8	40
13	Silver-Russell syndrome. Clinical and etiopathological aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2020, 118, e258-e264.	0.3	1
14	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	1.0	28
15	Wnt/Î²-catenin signaling pathway safeguards epigenetic stability and homeostasis of mouse embryonic stem cells. Scientific Reports, 2019, 9, 948.	1.6	31
16	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	1.1	11
17	A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. Genome Medicine, 2019, 11, 84.	3.6	45
18	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. Clinical Epigenetics, 2019, 11, 190.	1.8	22

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19	Transcription alterations of KCNQ1 associated with imprinted methylation defects in the Beckwithâ€“Wiedemann locus. Genetics in Medicine, 2019, 21, 1808-1820.	1.1	38
20	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. Nature Reviews Genetics, 2019, 20, 235-248.	7.7	291
21	Meg3 Non-coding RNA Expression Controls Imprinting by Preventing Transcriptional Upregulation in cis. Cell Reports, 2018, 23, 337-348.	2.9	54
22	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
23	Beckwith-Wiedemann syndrome. Clinical and etiopathogenic aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2018, 116, 368-373.	0.3	6
24	Is ZFP57 binding to H19/IGF2:IG-DMR affected in Silver-Russell syndrome?. Clinical Epigenetics, 2018, 10, 23.	1.8	25
25	Tissue-specific and mosaic imprinting defects underlie opposite congenital growth disorders in mice. PLoS Genetics, 2018, 14, e1007243.	1.5	13
26	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. Pediatrics, 2017, 140, .	1.0	87
27	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	1.0	101
28	Identification of Trombospondin-1 as a Novel Amelogenin Interactor by Functional Proteomics. Frontiers in Chemistry, 2017, 5, 74.	1.8	2
29	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome: a paradigm for genomic medicine. Clinical Genetics, 2016, 89, 403-415.	1.0	57
30	Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review and Meta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol. Journal of Pediatrics, 2016, 176, 142-149.e1.	0.9	119
31	Fetal growth patterns in Beckwithâ€“Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	1.0	34
32	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. Clinical Epigenetics, 2016, 8, 69.	1.8	9
33	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	2.9	81
34	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	1.4	68
35	Humanized H19/Igf2 locus reveals diverged imprinting mechanism between mouse and human and reflects Silverâ€“Russell syndrome phenotypes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 10938-10943.	3.3	28
36	ZFP57 maintains the parent-of-origin-specific expression of the imprinted genes and differentially affects non-imprinted targets in mouse embryonic stem cells. Nucleic Acids Research, 2016, 44, 8165-8178.	6.5	73

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37	Recommendations of the Scientific Committee of the Italian Beckwithâ€“Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome. European Journal of Medical Genetics, 2016, 59, 52-64.	0.7	76
38	ZFP57 recognizes multiple and closely spaced sequence motif variants to maintain repressive epigenetic marks in mouse embryonic stem cells. Nucleic Acids Research, 2016, 44, 1118-1132.	6.5	50
39	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	1.4	113
40	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	1.8	174
41	Insulin Like Growth Factor 2 Expression in the Rat Brain Both in Basal Condition and following Learning Predominantly Derives from the Maternal Allele. PLoS ONE, 2015, 10, e0141078.	1.1	28
42	A splicing mutation of the HMGA2 gene is associated with Silverâ€“Russell syndrome phenotype. Journal of Human Genetics, 2015, 60, 287-293.	1.1	33
43	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	1.8	23
44	A new case of de novo 6q24.2-q25.2 deletion on paternal chromosome 6 with growth hormone deficiency: a twelve-year follow-up and literature review. BMC Medical Genetics, 2015, 16, 69.	2.1	4
45	A novel large deletion of the ICR1 region including H19 and putative enhancer elements. BMC Medical Genetics, 2015, 16, 30.	2.1	9
46	Looking for CDKN1C enhancers. European Journal of Human Genetics, 2014, 22, 442-443.	1.4	19
47	Giant breast tumors in a patient with Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 182-185.	0.7	6
48	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	1.4	50
49	The PEG13-DMR and brain-specific enhancers dictate imprinted expression within the 8q24 intellectual disability risk locus. Epigenetics and Chromatin, 2014, 7, 5.	1.8	46
50	Silverâ€“Russell syndrome due to paternal H19 / IGF2 hypomethylation in a twin girl born after in vitro fertilization. American Journal of Medical Genetics, Part A, 2013, 161, 2652-2655.	0.7	22
51	Prevalence of beckwithâ€“wiedemann syndrome in North West of Italy. American Journal of Medical Genetics, Part A, 2013, 161, 2481-2486.	0.7	93
52	Genetic and epigenetic mutations affect the DNA binding capability of human ZFP57 in transient neonatal diabetes type 1. FEBS Letters, 2013, 587, 1474-1481.	1.3	25
53	The Control Region of Mitochondrial DNA Shows an Unusual CpG and Non-CpG Methylation Pattern. DNA Research, 2013, 20, 537-547.	1.5	221
54	The molecular function and clinical phenotype of partial deletions of the IGF2/H19 imprinting control region depends on the spatial arrangement of the remaining CTCF-binding sites. Human Molecular Genetics, 2013, 22, 544-557.	1.4	78

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55	Imprinting at the PLAGL1 domain is contained within a 70-kb CTCF/cohesin-mediated non-allelic chromatin loop. Nucleic Acids Research, 2013, 41, 2171-2179.	6.5	25
56	Paternal deletion of the 11p15.5 centromeric-imprinting control region is associated with alteration of imprinted gene expression and recurrent severe intrauterine growth restriction. Journal of Medical Genetics, 2013, 50, 99-103.	1.5	29
57	Gain of function in CDKN1C. Nature Genetics, 2012, 44, 737-738.	9.4	12
58	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwithâ€“Wiedemann syndrome and Silverâ€“Russell syndrome cases. Human Molecular Genetics, 2012, 21, 10-25.	1.4	135
59	Nephrological findings and genotypeâ€“phenotype correlation in Beckwithâ€“Wiedemann syndrome. Pediatric Nephrology, 2012, 27, 397-406.	0.9	55
60	A novel microdeletion in the IGF2/H19 imprinting centre region defines a recurrent mutation mechanism in familial Beckwithâ€“Wiedemann syndrome. European Journal of Medical Genetics, 2011, 54, e451-e454.	0.7	30
61	In Embryonic Stem Cells, ZFP57/KAP1 Recognize a Methylated Hexanucleotide to Affect Chromatin and DNA Methylation of Imprinting Control Regions. Molecular Cell, 2011, 44, 361-372.	4.5	503
62	22q11.2 Distal Deletion Syndrome: Description of a New Case with Truncus Arteriosus Type 2 and Review. Molecular Syndromology, 2011, 2, 35-44.	0.3	26
63	Disruption of genomic neighbourhood at the imprinted IGF2-H19 locus in Beckwithâ€“Wiedemann syndrome and Silverâ€“Russell syndrome. Human Molecular Genetics, 2011, 20, 1363-1374.	1.4	80
64	A case of Beckwith-Wiedemann syndrome caused by a cryptic 11p15 deletion encompassing the centromeric imprinted domain of the BWS locus. Journal of Medical Genetics, 2010, 47, 429-432.	1.5	30
65	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome Phenotypes Associated with 11p Duplication in a Single Family. Pediatric and Developmental Pathology, 2010, 13, 326-330.	0.5	31
66	Inherited and Sporadic Epimutations at the IGF2-H19 Locus in Beckwith-Wiedemann Syndrome and Wilms’ Tumor. Endocrine Development, 2009, 14, 1-9.	1.3	48
67	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619.	1.4	194
68	Wilms tumor and constitutional epigenetic defects. Nature Genetics, 2008, 40, 1272-1273.	9.4	2
69	MS-MLPA is a specific and sensitive technique for detecting all chromosome 11p15.5 imprinting defects of BWS and SRS in a single-tube experiment. European Journal of Human Genetics, 2008, 16, 565-571.	1.4	73
70	High frequency of loss of heterozygosity at 11p15 and IGF2 overexpression are not related to clinical outcome in childhood adrenocortical tumors positive for the R337H TP53 mutation. Cancer Genetics and Cytogenetics, 2008, 186, 19-24.	1.0	27
71	The H19 locus acts in vivo as a tumor suppressor. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 12417-12422.	3.3	300
72	Different mechanisms cause imprinting defects at the IGF2/H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumour. Human Molecular Genetics, 2008, 17, 1427-1435.	1.4	76

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73	Silver-Russell Syndrome following in Vitro Fertilization. Pediatric and Developmental Pathology, 2008, 11, 329-331.	0.5	19
74	Distinct Methylation Changes at the IGF2-H19 Locus in Congenital Growth Disorders and Cancer. PLoS ONE, 2008, 3, e1849.	1.1	93
75	Mechanisms causing imprinting defects in familial Beckwithâ€“Wiedemann syndrome with Wilms' tumour. Human Molecular Genetics, 2007, 16, 254-264.	1.4	100
76	Familial posterior helical ear pits. American Journal of Medical Genetics, Part A, 2007, 143A, 2832-2834.	0.7	2
77	Microdeletion and IGF2 loss of imprinting in a cascade causing Beckwith-Wiedemann syndrome with Wilms' tumor. Nature Genetics, 2005, 37, 785-786.	9.4	40
78	Reply to "Microdeletion and IGF2 loss of imprinting in a cascade causing Beckwith-Wiedemann syndrome with Wilms' tumor". Nature Genetics, 2005, 37, 786-787.	9.4	18
79	Molecular subtypes and phenotypic expression of Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2005, 13, 1025-1032.	1.4	284
80	Perlman syndrome: Clinical report and nine-year follow-up. American Journal of Medical Genetics, Part A, 2005, 139A, 131-135.	0.7	17
81	The two-domain hypothesis in Beckwithâ€“Wiedemann syndrome: autonomous imprinting of the telomeric domain of the distal chromosome 7 cluster. Human Molecular Genetics, 2005, 14, 503-511.	1.4	63
82	Microdeletions in the human H19 DMR result in loss of IGF2 imprinting and Beckwith-Wiedemann syndrome. Nature Genetics, 2004, 36, 958-960.	9.4	261
83	Paternal imprints can be established on the maternal Igf2-H19 locus without altering replication timing of DNA. Human Molecular Genetics, 2003, 12, 3123-3132.	1.4	19
84	Developmentally regulated functions of the H19 differentially methylated domain. Human Molecular Genetics, 2003, 13, 353-361.	1.4	11
85	The Arabidopsis SUPERMAN protein is able to specifically bind DNA through its single Cys2-His2 zinc finger motif. Nucleic Acids Research, 2002, 30, 4945-4951.	6.5	63
86	The 5â€² end of the KCNQ1OT1 gene is hypomethylated in the Beckwith-Wiedemann syndrome. Human Genetics, 2002, 111, 105-107.	1.8	19
87	The H19 endodermal enhancer is required for Igf2 activation and tumor formation in experimental liver carcinogenesis. Oncogene, 2000, 19, 6376-6385.	2.6	28
88	Relaxation of Insulin-like Growth Factor 2 Imprinting and Discordant Methylation at KvDMR1 in Two First Cousins Affected by Beckwith-Wiedemann and Klippel-Trenaunay-Weber Syndromes. American Journal of Human Genetics, 2000, 66, 841-847.	2.6	31
89	Role of histone acetylation and DNA methylation in the maintenance of the imprinted expression of theH19andIgf2genes. FEBS Letters, 1999, 458, 45-50.	1.3	73
90	Relaxation of insulin-like growth factor-2 imprinting in rat cultured cells1This paper is dedicated to the memory of Professor Gaetano Salvatore.1. Molecular and Cellular Endocrinology, 1997, 135, 153-163.	1.6	14

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91	Expression and parental imprinting of the H19 gene in human rhabdomyosarcoma. Oncogene, 1997, 14, 1503-1510.	2.6	56
92	Activation of fetal promoters of insulinlike growth factors II gene in hepatitis C virus-related chronic hepatitis, cirrhosis, and hepatocellular carcinoma. Hepatology, 1996, 23, 1304-1312.	3.6	52
93	Preferential Loss of Heterozygosity of Chromosome 7 Loci in Simian Virus 40 t/T Antigen-Induced Mouse Hepatocellular Carcinomas Does Not Involve H-ras Muatations. Acta Geneticae Medicae Et Gemellologiae, 1996, 45, 221-225.	0.2	0
94	Activation of fetal promoters of insulinlike growth factors II gene in hepatitis C virus-related chronic hepatitis, cirrhosis, and hepatocellular carcinoma. Hepatology, 1996, 23, 1304-1312.	3.6	5
95	Mono- and bi-allelic expression of insulin-like growth factor II gene in human muscle tumors. Human Molecular Genetics, 1994, 3, 1117-1121.	1.4	59
96	A common response element mediates differential effects of phorbol esters and forskolin on type-1 plasminogen activator inhibitor gene expression in human breast carcinoma cells. FEBS Journal, 1994, 220, 63-74.	0.2	40
97	Multiple levels of control of insulin-like growth factor gene expression. Molecular and Cellular Endocrinology, 1994, 101, R1-R14.	1.6	34
98	Forskolin down-regulates type-1 plasminogen activator inhibitor and tissue-type plasminogen activator and their mRNAs in human fibrosarcoma cells. Molecular and Cellular Endocrinology, 1990, 72, 103-110.	1.6	10
99	Plasminogen activator inhibitors: hormonally regulated serpins. Molecular and Cellular Endocrinology, 1990, 68, 1-19.	1.6	406
100	Tumor necrosis factor-Î± regulates mRNA for urokinase-type plasminogen activator and type-1 plasminogen activator inhibitor in human neoplastic cell lines. Molecular and Cellular Endocrinology, 1989, 61, 87-96.	1.6	22
101	The regulatory region of the human plasminogen activator inhibitor type-1 (PAI-1) gene. Nucleic Acids Research, 1988, 16, 2805-2824.	6.5	82
102	Plasminogen activator inhibitor type 1 gene is located at region q21.3-q22 of chromosome 7 and genetically linked with cystic fibrosis.. Proceedings of the National Academy of Sciences of the United States of America, 1987, 84, 8548-8552.	3.3	127
103	Plasminogen activator inhibitor type 1 biosynthesis and mRNA level are increased by dexamethasone in human fibrosarcoma cells.. Molecular and Cellular Biology, 1987, 7, 3021-3025.	1.1	56
104	Plasminogen activator inhibitor type-1 : reactive center and amino-terminal heterogeneity determined by protein and cDNA sequencing. FEBS Letters, 1986, 209, 213-218.	1.3	195
105	The Structure of the Human Plasminogen Activator (uPA) Gene. Protides of the Biological Fluids; Proceedings of the Colloquium, 1985, , 67-70.	0.1	1
106	BamHI RFLP linked to the human urokinase gene. Nucleic Acids Research, 1985, 13, 5404-5404.	6.5	10
107	Structure of the galactokinase gene ofEscherichia coli, the last (?) gene of thegaloperon. Nucleic Acids Research, 1985, 13, 1841-1853.	6.5	79
108	The human urokinase-plasminogen activator gene and its promoter. Nucleic Acids Research, 1985, 13, 2759-2771.	6.5	219

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109	Convergently functional, Rho-independent terminator in Salmonella typhimurium. Journal of Bacteriology, 1985, 163, 362-368.	1.0	47
110	Regulation of single and multicopy his operons in Escherichia coli. Journal of Bacteriology, 1985, 163, 1172-1179.	1.0	10
111	Structure and function of the internal promoter (hisBp) of the Escherichia coli K-12 histidine operon. Journal of Bacteriology, 1983, 155, 1288-1296.	1.0	46