List of Publications by Year in descending order

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		57758	58581
111	7,187	44	82
papers	citations	h-index	g-index
112	112	112	6846
all docs	docs citations	times ranked	citing authors

#	Article	IF	CITATIONS
1	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.	9.7	503
2	Plasminogen activator inhibitors: hormonally regulated serpins. Molecular and Cellular Endocrinology, 1990, 68, 1-19.	3.2	406
3	Clinical and molecular diagnosis, screening and management of Beckwith–Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	9.6	388
4	The <i>H19</i> locus acts <i>in vivo</i> as a tumor suppressor. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 12417-12422.	7.1	300
5	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. Nature Reviews Genetics, 2019, 20, 235-248.	16.3	291
6	Molecular subtypes and phenotypic expression of Beckwith–Wiedemann syndrome. European Journal of Human Genetics, 2005, 13, 1025-1032.	2.8	284
7	Microdeletions in the human H19 DMR result in loss of IGF2 imprinting and Beckwith-Wiedemann syndrome. Nature Genetics, 2004, 36, 958-960.	21.4	261
8	The Control Region of Mitochondrial DNA Shows an Unusual CpG and Non-CpG Methylation Pattern. DNA Research, 2013, 20, 537-547.	3.4	221
9	The human urokinase-plasminogen activator gene and its promoter. Nucleic Acids Research, 1985, 13, 2759-2771.	14.5	219
10	Plasminogen activator inhibitor type-1 : reactive center and amino-terminal heterogeneity determined by protein and cDNA sequencing. FEBS Letters, 1986, 209, 213-218.	2.8	195
11	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith–Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619.	2.8	194
12	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	4.1	174
13	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith–Wiedemann syndrome and Silver–Russell syndrome cases. Human Molecular Genetics, 2012, 21, 10-25.	2.9	135
14	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.	7.1	127
15	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.	1.8	119
16	(Epi)genotype–phenotype correlations in Beckwith–Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	2.8	113
17	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	2.0	101
18	Mechanisms causing imprinting defects in familial Beckwith–Wiedemann syndrome with Wilms' tumour. Human Molecular Genetics. 2007. 16. 254-264.	2.9	100

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19	Distinct Methylation Changes at the IGF2-H19 Locus in Congenital Growth Disorders and Cancer. PLoS ONE, 2008, 3, e1849.	2.5	93
20	Prevalence of beckwith–wiedemann syndrome in North West of Italy. American Journal of Medical Genetics, Part A, 2013, 161, 2481-2486.	1.2	93
21	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. Pediatrics, 2017, 140, .	2.1	87
22	The regulatory region of the human plasminogen activator inhibitor type-1 (PAI-1) gene. Nucleic Acids Research, 1988, 16, 2805-2824.	14.5	82
23	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	6.7	81
24	Disruption of genomic neighbourhood at the imprinted IGF2-H19 locus in Beckwith–Wiedemann syndrome and Silver–Russell syndrome. Human Molecular Genetics, 2011, 20, 1363-1374.	2.9	80
25	Structure of the galactokinase gene ofEscherichia coli, the last (?) gene of thegaloperon. Nucleic Acids Research, 1985, 13, 1841-1853.	14.5	79
26	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.	2.9	78
27	Different mechanisms cause imprinting defects at the IGF2/H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumour. Human Molecular Genetics, 2008, 17, 1427-1435.	2.9	76
28	Recommendations of the Scientific Committee of the Italian Beckwith–Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome. European Journal of Medical Genetics, 2016, 59, 52-64.	1.3	76
29	Role of histone acetylation and DNA methylation in the maintenance of the imprinted expression of the <i>H19</i> and <i>lgf2</i> genes. FEBS Letters, 1999, 458, 45-50.	2.8	73
30	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.	2.8	73
31	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.	14.5	73
32	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver–Russell and Beckwith–Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	2.8	68
33	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.	14.5	63
34	The two-domain hypothesis in Beckwith–Wiedemann syndrome: autonomous imprinting of the telomeric domain of the distal chromosome 7 cluster. Human Molecular Genetics, 2005, 14, 503-511.	2.9	63
35	Mono- and bi-allelic expression of insulin-like growth factor II gene in human muscle tumors. Human Molecular Genetics, 1994, 3, 1117-1121.	2.9	59
36	(Epi)genotype–phenotype correlations in Beckwith–Wiedemann syndrome: a paradigm for genomic medicine. Clinical Genetics, 2016, 89, 403-415.	2.0	57

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37	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.	2.3	56
38	Expression and parental imprinting of the H19 gene in human rhabdomyosarcoma. Oncogene, 1997, 14, 1503-1510.	5.9	56
39	Nephrological findings and genotype–phenotype correlation in Beckwith–Wiedemann syndrome. Pediatric Nephrology, 2012, 27, 397-406.	1.7	55
40	Meg3 Non-coding RNA Expression Controls Imprinting by Preventing Transcriptional Upregulation in cis. Cell Reports, 2018, 23, 337-348.	6.4	54
41	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.	7.3	52
42	Clinical utility gene card for: Beckwith–Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	50
43	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.	14.5	50
44	Inherited and Sporadic Epimutations at the <i>IGF2-H19</i> Locus in Beckwith-Wiedemann Syndrome and Wilms’ Tumor. Endocrine Development, 2009, 14, 1-9.	1.3	48
45	Convergently functional, Rho-independent terminator in Salmonella typhimurium. Journal of Bacteriology, 1985, 163, 362-368.	2.2	47
46	The PEG13-DMR and brain-specific enhancers dictate imprinted expression within the 8q24 intellectual disability risk locus. Epigenetics and Chromatin, 2014, 7, 5.	3.9	46
47	Structure and function of the internal promoter (hisBp) of the Escherichia coli K-12 histidine operon. Journal of Bacteriology, 1983, 155, 1288-1296.	2.2	46
48	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.	8.2	45
49	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
50	Microdeletion and IGF2 loss of imprinting in a cascade causing Beckwith-Wiedemann syndrome with Wilms' tumor. Nature Genetics, 2005, 37, 785-786.	21.4	40
51	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.	4.1	40
52	Transcription alterations of KCNQ1 associated with imprinted methylation defects in the Beckwith–Wiedemann locus. Genetics in Medicine, 2019, 21, 1808-1820.	2.4	38
53	Multiple levels of control of insulin-like growth factor gene expression. Molecular and Cellular Endocrinology, 1994, 101, R1-R14.	3.2	34
54	Fetal growth patterns in Beckwith–Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	2.0	34

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55	A splicing mutation of the HMGA2 gene is associated with Silver–Russell syndrome phenotype. Journal of Human Genetics, 2015, 60, 287-293.	2.3	33
56	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.	6.2	31
57	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome Phenotypes Associated with 11p Duplication in a Single Family. Pediatric and Developmental Pathology, 2010, 13, 326-330.	1.0	31
58	Wnt∫î²-catenin signaling pathway safeguards epigenetic stability and homeostasis of mouse embryonic stem cells. Scientific Reports, 2019, 9, 948.	3.3	31
59	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.	3.2	30
60	A novel microdeletion in the IGF2/H19 imprinting centre region defines a recurrent mutation mechanism in familial Beckwith–Wiedemann syndrome. European Journal of Medical Genetics, 2011, 54, e451-e454.	1.3	30
61	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.	3.2	29
62	The H19 endodermal enhancer is required for Igf2 activation and tumor formation in experimental liver carcinogenesis. Oncogene, 2000, 19, 6376-6385.	5.9	28
63	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.	2.5	28
64	Humanized <i>H19/lgf2</i> locus reveals diverged imprinting mechanism between mouse and human and reflects Silver–Russell syndrome phenotypes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 10938-10943.	7.1	28
65	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
66	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
67	22q11.2 Distal Deletion Syndrome: Description of a New Case with Truncus Arteriosus Type 2 and Review. Molecular Syndromology, 2011, 2, 35-44.	0.8	26
68	Genetic and epigenetic mutations affect the DNA binding capability of human ZFP57 in transient neonatal diabetes type 1. FEBS Letters, 2013, 587, 1474-1481.	2.8	25
69	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.	14.5	25
70	Is ZFP57 binding to H19/IGF2:IG-DMR affected in Silver-Russell syndrome?. Clinical Epigenetics, 2018, 10, 23.	4.1	25
71	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	4.1	23
72	Tumor necrosis factor-α 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.	3.2	22

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73	Silver–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.	1.2	22
74	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.	4.1	22
75	The 5′ end of the KCNQ1OT1 gene is hypomethylated in the Beckwith-Wiedemann syndrome. Human Genetics, 2002, 111, 105-107.	3.8	19
76	Paternal imprints can be established on the maternal Igf2-H19 locus without altering replication timing of DNA. Human Molecular Genetics, 2003, 12, 3123-3132.	2.9	19
77	Silver-Russell Syndrome following in Vitro Fertilization. Pediatric and Developmental Pathology, 2008, 11, 329-331.	1.0	19
78	Looking for CDKN1C enhancers. European Journal of Human Genetics, 2014, 22, 442-443.	2.8	19
79	Reply to "Microdeletion and IGF2 loss of imprinting in a cascade causing Beckwith-Wiedemann syndrome with Wilms' tumor". Nature Genetics, 2005, 37, 786-787.	21.4	18
80	Perlman syndrome: Clinical report and nine-year follow-up. American Journal of Medical Genetics, Part A, 2005, 139A, 131-135.	1.2	17
81	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.	3.2	14
82	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
83	Tissue-specific and mosaic imprinting defects underlie opposite congenital growth disorders in mice. PLoS Genetics, 2018, 14, e1007243.	3.5	13
84	Gain of function in CDKN1C. Nature Genetics, 2012, 44, 737-738.	21.4	12
85	Developmentally regulated functions of the H19 differentially methylated domain. Human Molecular Genetics, 2003, 13, 353-361.	2.9	11
86	Molecular Etiology Disclosed by Array CGH in Patients With Silver–Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	2.3	11
87	BamHI RFLP linked to the human urokinase gene. Nucleic Acids Research, 1985, 13, 5404-5404.	14.5	10
88	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.	3.2	10
89	The number of the CTCF binding sites of the <i>H19/IGF2</i> :IG-DMR correlates with DNA methylation and expression imprinting in a humanized mouse model. Human Molecular Genetics, 2021, 30, 1509-1520.	2.9	10
90	Regulation of single and multicopy his operons in Escherichia coli. Journal of Bacteriology, 1985, 163, 1172-1179.	2.2	10

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91	A novel large deletion of the ICR1 region including H19 and putative enhancer elements. BMC Medical Genetics, 2015, 16, 30.	2.1	9
92	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. Clinical Epigenetics, 2016, 8, 69.	4.1	9
93	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.	3.2	9
94	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	2.4	8
95	Zfp57 inactivation illustrates the role of ICR methylation in imprinted gene expression during neural differentiation of mouse ESCs. Scientific Reports, 2021, 11, 13802.	3.3	7
96	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwith–Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clinical Epigenetics, 2022, 14, .	4.1	7
97	Giant breast tumors in a patient with Beckwith–Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 182-185.	1.2	6
98	Beckwith-Wiedemann syndrome. Clinical and etiopathogenic aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2018, 116, 368-373.	0.2	6
99	Both Epimutations and Chromosome Aberrations Affect Multiple Imprinted Loci in Aggressive Wilms Tumors. Cancers, 2020, 12, 3411.	3.7	6
100	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. Genes, 2021, 12, 581.	2.4	5
101	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.	7.3	5
102	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
103	A paternally inherited 1.4 kb deletion of the 11p15.5 imprinting center 2 is associated with a mild familial Silver–Russell syndrome phenotype. European Journal of Human Genetics, 2021, 29, 447-454.	2.8	4
104	Familial posterior helical ear pits. American Journal of Medical Genetics, Part A, 2007, 143A, 2832-2834.	1.2	2
105	Wilms tumor and constitutional epigenetic defects. Nature Genetics, 2008, 40, 1272-1273.	21.4	2
106	Identification of Trombospondin-1 as a Novel Amelogenin Interactor by Functional Proteomics. Frontiers in Chemistry, 2017, 5, 74.	3.6	2
107	ZBTB2 protein is a new partner of the Nucleosome Remodeling and Deacetylase (NuRD) complex. International Journal of Biological Macromolecules, 2021, 168, 67-76.	7.5	2
108	Variable Expressivity of the Beckwith-Wiedemann Syndrome in Four Pedigrees Segregating Loss-of-Function Variants of CDKN1C. Genes, 2021, 12, 706.	2.4	2

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109	The Structure of the Human Plasminogen Activator (uPA) Gene. Protides of the Biological Fluids; Proceedings of the Colloquium, 1985, , 67-70.	0.1	1
110	Silver-Russell syndrome. Clinical and etiopathological aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2020, 118, e258-e264.	0.2	1
111	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