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List of Publications by Year
in descending order

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38 papers	1,851 citations	279798 23 h-index	330143 37 g-index
38 all docs	38 docs citations	38 times ranked	2358 citing authors

#	Article	IF	Citations
1	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, .	4.1	7
2	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
3	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. Genes, 2021, 12, 581.	2.4	5
4	Clinical and molecular characterization of patients with Beckwith-Wiedemann spectrum conceived through assisted reproductive technology. Molecular Genetics and Metabolism, 2021, 132, S66.	1.1	0
5	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.	2.9	10
6	Variable Expressivity of the Beckwith-Wiedemann Syndrome in Four Pedigrees Segregating Loss-of-Function Variants of CDKN1C. Genes, 2021, 12, 706.	2.4	2
7	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
8	Silver-Russell syndrome. Clinical and etiopathological aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2020, 118, e258-e264.	0.2	1
9	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
10	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
11	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
12	Transcription alterations of KCNQ1 associated with imprinted methylation defects in the Beckwithâ€“Wiedemann locus. Genetics in Medicine, 2019, 21, 1808-1820.	2.4	38
13	Beckwith-Wiedemann syndrome. Clinical and etiopathogenic aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatria, 2018, 116, 368-373.	0.2	6
14	Is ZFP57 binding to H19/IGF2:IG-DMR affected in Silver-Russell syndrome?. Clinical Epigenetics, 2018, 10, 23.	4.1	25
15	Tissue-specific and mosaic imprinting defects underlie opposite congenital growth disorders in mice. PLoS Genetics, 2018, 14, e1007243.	3.5	13
16	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. Clinical Epigenetics, 2016, 8, 69.	4.1	9
17	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
18	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

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19	A splicing mutation of the HMGA2 gene is associated with Silverâ€“Russell syndrome phenotype. Journal of Human Genetics, 2015, 60, 287-293.	2.3	33
20	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
21	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
22	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
23	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
24	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
25	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.	2.9	135
26	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.	2.9	80
27	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
28	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
29	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.	2.8	194
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	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
32	Distinct Methylation Changes at the IGF2-H19 Locus in Congenital Growth Disorders and Cancer. PLoS ONE, 2008, 3, e1849.	2.5	93
33	Mechanisms causing imprinting defects in familial Beckwithâ€“Wiedemann syndrome with Wilms' tumour. Human Molecular Genetics, 2007, 16, 254-264.	2.9	100
34	Familial posterior helical ear pits. American Journal of Medical Genetics, Part A, 2007, 143A, 2832-2834.	1.2	2
35	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
36	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.	2.9	63

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37	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
38	Two cases of misinterpretation of molecular results in incontinentia pigmenti, and a PCR-based method to discriminate NEMO/IKK? dene deletion. Human Mutation, 2003, 21, 8-11.	2.5	58