

# Angela Sparago

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Microdeletions in the human H19 DMR result in loss of IGF2 imprinting and Beckwith-Wiedemann syndrome. <i>Nature Genetics</i> , 2004, 36, 958-960.	21.4	261
2	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 611-619.	2.8	194
3	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases. <i>Human Molecular Genetics</i> , 2012, 21, 10-25.	2.9	135
4	Mechanisms causing imprinting defects in familial Beckwith-Wiedemann syndrome with Wilms' tumour. <i>Human Molecular Genetics</i> , 2007, 16, 254-264.	2.9	100
5	Distinct Methylation Changes at the IGF2-H19 Locus in Congenital Growth Disorders and Cancer. <i>PLoS ONE</i> , 2008, 3, e1849.	2.5	93
6	Disruption of genomic neighbourhood at the imprinted IGF2-H19 locus in Beckwith-Wiedemann syndrome and Silver-Russell syndrome. <i>Human Molecular Genetics</i> , 2011, 20, 1363-1374.	2.9	80
7	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. <i>Human Molecular Genetics</i> , 2013, 22, 544-557.	2.9	78
8	Different mechanisms cause imprinting defects at the IGF2/H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumour. <i>Human Molecular Genetics</i> , 2008, 17, 1427-1435.	2.9	76
9	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. <i>European Journal of Human Genetics</i> , 2008, 16, 565-571.	2.8	73
10	ZFP57 maintains the parent-of-origin-specific expression of the imprinted genes and differentially affects non-imprinted targets in mouse embryonic stem cells. <i>Nucleic Acids Research</i> , 2016, 44, 8165-8178.	14.5	73
11	The two-domain hypothesis in Beckwith-Wiedemann syndrome: autonomous imprinting of the telomeric domain of the distal chromosome 7 cluster. <i>Human Molecular Genetics</i> , 2005, 14, 503-511.	2.9	63
12	Two cases of misinterpretation of molecular results in incontinentia pigmenti, and a PCR-based method to discriminate NEMO/IKK $\gamma$ gene deletion. <i>Human Mutation</i> , 2003, 21, 8-11.	2.5	58
13	ZFP57 recognizes multiple and closely spaced sequence motif variants to maintain repressive epigenetic marks in mouse embryonic stem cells. <i>Nucleic Acids Research</i> , 2016, 44, 1118-1132.	14.5	50
14	Inherited and Sporadic Epimutations at the IGF2-H19 Locus in Beckwith-Wiedemann Syndrome and Wilms' Tumor. <i>Endocrine Development</i> , 2009, 14, 1-9.	1.3	48
15	The PEG13-DMR and brain-specific enhancers dictate imprinted expression within the 8q24 intellectual disability risk locus. <i>Epigenetics and Chromatin</i> , 2014, 7, 5.	3.9	46
16	A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. <i>Genome Medicine</i> , 2019, 11, 84.	8.2	45
17	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. <i>Clinical Epigenetics</i> , 2020, 12, 139.	4.1	40
18	Transcription alterations of KCNQ1 associated with imprinted methylation defects in the Beckwith-Wiedemann locus. <i>Genetics in Medicine</i> , 2019, 21, 1808-1820.	2.4	38

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19	A splicing mutation of the HMGA2 gene is associated with Silver-Russell syndrome phenotype. <i>Journal of Human Genetics</i> , 2015, 60, 287-293.	2.3	33
20	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome Phenotypes Associated with 11p Duplication in a Single Family. <i>Pediatric and Developmental Pathology</i> , 2010, 13, 326-330.	1.0	31
21	Paternal deletion of the 11p15.5 centromeric-imprinting control region is associated with alteration of imprinted gene expression and recurrent severe intrauterine growth restriction. <i>Journal of Medical Genetics</i> , 2013, 50, 99-103.	3.2	29
22	DNA Methylation in the Diagnosis of Monogenic Diseases. <i>Genes</i> , 2020, 11, 355.	2.4	28
23	Genetic and epigenetic mutations affect the DNA binding capability of human ZFP57 in transient neonatal diabetes type 1. <i>FEBS Letters</i> , 2013, 587, 1474-1481.	2.8	25
24	Imprinting at the PLAGL1 domain is contained within a 70-kb CTCF/cohesin-mediated non-allelic chromatin loop. <i>Nucleic Acids Research</i> , 2013, 41, 2171-2179.	14.5	25
25	Is ZFP57 binding to H19/IGF2:IG-DMR affected in Silver-Russell syndrome?. <i>Clinical Epigenetics</i> , 2018, 10, 23.	4.1	25
26	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. <i>Clinical Epigenetics</i> , 2019, 11, 190.	4.1	22
27	Reply to "Microdeletion and IGF2 loss of imprinting in a cascade causing Beckwith-Wiedemann syndrome with Wilms' tumor". <i>Nature Genetics</i> , 2005, 37, 786-787.	21.4	18
28	Tissue-specific and mosaic imprinting defects underlie opposite congenital growth disorders in mice. <i>PLoS Genetics</i> , 2018, 14, e1007243.	3.5	13
29	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. <i>Human Molecular Genetics</i> , 2021, 30, 1509-1520.	2.9	10
30	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. <i>Clinical Epigenetics</i> , 2016, 8, 69.	4.1	9
31	Biallelic variant in cyclin B3 is associated with failure of maternal meiosis II and recurrent digynic triploidy. <i>Journal of Medical Genetics</i> , 2021, 58, 783-788.	3.2	9
32	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. <i>Clinical Epigenetics</i> , 2022, 14, .	4.1	7
33	Beckwith-Wiedemann syndrome. Clinical and etiopathogenic aspects of a model genomic imprinting entity. <i>Archivos Argentinos De Pediatria</i> , 2018, 116, 368-373.	0.2	6
34	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. <i>Genes</i> , 2021, 12, 581.	2.4	5
35	Familial posterior helical ear pits. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2832-2834.	1.2	2
36	Variable Expressivity of the Beckwith-Wiedemann Syndrome in Four Pedigrees Segregating Loss-of-Function Variants of CDKN1C. <i>Genes</i> , 2021, 12, 706.	2.4	2

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37	Silver-Russell syndrome. Clinical and etiopathological aspects of a model genomic imprinting entity. Archivos Argentinos De Pediatría, 2020, 118, e258-e264.	0.2	1
38	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