Bernhard Horsthemke

List of Publications by Year
in descending order

74	5,840	117625	82547
papers	citations	34 h-index	72 g-index

83	83	83	5288
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Inherited microdeletions in the Angelman and Praderâ€“Willi syndromes define an imprinting centre on human chromosome 15. Nature Genetics, 1995, 9, 395-400.	21.4	589
2	Epigenetic changes may contribute to the formation and spontaneous regression of retinoblastoma. Human Genetics, 1989, 83, 155-158.	3.8	488
3	Cloning defined regions of the human genome by microdissection of banded chromosomes and enzymatic amplification. Nature, 1989, 338, 348-350.	27.8	351
4	Epimutations in Prader-Willi and Angelman Syndromes: A Molecular Study of 136 Patients with an Imprinting Defect. American Journal of Human Genetics, 2003, 72, 571-577.	6.2	280
5	Angelman syndrome â€” insights into a rare neurogenetic disorder. Nature Reviews Neurology, 2016, 12, 584-593.	10.1	256
6	Imprint switching on human chromosome 15 may involve alternative transcripts of the SNRPN gene. Nature Genetics, 1996, 14, 163-170.	21.4	250
7	Mechanisms of imprinting of the Praderâ€“Willi/Angelman region. American Journal of Medical Genetics, Part A, 2008, 146A, 2041-2052.	1.2	246
8	Assisted reproduction: the epigenetic perspective. Human Reproduction Update, 2005, 11, 473-482.	10.8	207
9	A critical view on transgenerational epigenetic inheritance in humans. Nature Communications, 2018, 9, 2973.	12.8	203
10	Molecular diagnosis of the Prader-Willi and Angelman syndromes by detection of parent-of-origin specific DNA methylation in 15q11-13. Human Genetics, 1992, 90, 313-5.	3.8	195
11	Maternal methylation imprints on human chromosome 15 are established during or after fertilization. Nature Genetics, 2001, 27, 341-344.	21.4	193
12	Modification of 15q11 â€” q13 DNA methylation imprints in unique Angelman and Prader â€— Willi patients. Human Molecular Genetics, 1993, 2, 1377-1382.	2.9	144
13	Sporadic Imprinting Defects in Prader-Willi Syndrome and Angelman Syndrome: Implications for Imprint-Switch Models, Genetic Counseling, and Prenatal Diagnosis. American Journal of Human Genetics, 1998, 63, 170-180.	6.2	142
14	De novo deletions of SNRPN exon 1 in early human and mouse embryos result in a paternal to maternal imprint switch. Nature Genetics, 2000, 25, 74-78.	21.4	142
15	A Single-Tube PCR Test for the Diagnosis of Angelman and Prader-Willi Syndrome Based on Allelic Methylation Differences at the SNRPN Locus. European Journal of Human Genetics, 1997, 5, 94-98.	2.8	139
16	N6-Adenosine Methylation in MiRNAs. PLoS ONE, 2015, 10, e0118438.	2.5	115
17	A paternal deletion of MKRN3, MAGEL2 and NDN does not result in Praderâ€“Willi syndrome. European Journal of Human Genetics, 2009, 17, 582-590.	2.8	112
18	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	12.8	111

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19	The Human Retinoblastoma Gene Is Imprinted. PLoS Genetics, 2009, 5, e1000790.	3.5	110
20	A previously unrecognised phenotype characterised by obesity, muscular hypotonia, and ability to speak in patients with Angelman syndrome caused by an imprinting defect. European Journal of Human Genetics, 1999, 7, 638-644.	2.8	88
21	Mechanisms of imprint dysregulation. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 321-328.	1.6	82
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	Somatic mosaicism in patients with Angelman syndrome and an imprinting defect. Human Molecular Genetics, 2004, 13, 2547-2555.	2.9	74
24	Epigenetic dynamics of monocyte-to-macrophage differentiation. Epigenetics and Chromatin, 2016, 9, 33.	3.9	73
25	Microdissection of the Prader-Willi syndrome chromosome region and identification of potential gene sequences. Genomics, 1990, 6, 521-527.	2.9	72
26	Clinical features of maternal uniparental disomy 14 in patients with an epimutation and a deletion of the imprintedDLK1/GTL2gene cluster. Human Mutation, 2008, 29, 1141-1146.	2.5	68
27	Disruption of the Bipartite Imprinting Center in a Family with Angelman Syndrome. American Journal of Human Genetics, 2001, 68, 1290-1294.	6.2	62
28	Epigenetic germline mosaicism in infertile men. Human Molecular Genetics, 2015, 24, 1295-1304.	2.9	58
29	C15orf2 and a novel noncoding transcript from the Praderâ€“Willi/Angelman syndrome region show monoallelic expression in fetal brain. Genomics, 2007, 89, 588-595.	2.9	52
30	FTO levels affect RNA modification and the transcriptome. European Journal of Human Genetics, 2013, 21, 317-323.	2.8	52
31	In Brief: Genomic imprinting and imprinting diseases. Journal of Pathology, 2014, 232, 485-487.	4.5	44
32	Identification of a Testis-Specific Gene (C15orf2) in the Praderâ€“Willi Syndrome Region on Chromosome 15. Genomics, 2000, 65, 174-183.	2.9	42
33	Heritable germline epimutations in humans. Nature Genetics, 2007, 39, 573-574.	21.4	41
34	Identification of cis- and trans-acting factors possibly modifying the risk of epimutations on chromosome 15. European Journal of Human Genetics, 2006, 14, 752-758.	2.8	37
35	New insights into the imprinted MEG8-DMR in 14q32 and clinical and molecular description of novel patients with Temple syndrome. European Journal of Human Genetics, 2017, 25, 935-945.	2.8	35
36	Expression of SNURFâ€“SNRPN upstream transcripts and epigenetic regulatory genes during human spermatogenesis. European Journal of Human Genetics, 2009, 17, 1463-1470.	2.8	33

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37	Clinical utility gene card for: Angelman Syndrome. European Journal of Human Genetics, 2015, 23, 3-3.	2.8	32
38	Pre- and Postovulatory Aging of Murine Oocytes Affect the Transcript Level and Poly(A) Tail Length of Maternal Effect Genes. PLoS ONE, 2014, 9, e108907.	2.5	30
39	Clinical phenotypes of MAGEL2 mutations and deletions. Orphanet Journal of Rare Diseases, 2014, 9, 40.	2.7	28
40	Locus-Specific DNA Methylation Analysis by Targeted Deep Bisulfite Sequencing. Methods in Molecular Biology, 2018, 1767, 351-366.	0.9	28
41	Novel microdeletions on chromosome 14q32.2 suggest a potential role for non-coding RNAs in Kagami-Ogata syndrome. European Journal of Human Genetics, 2016, 24, 1724-1729.	2.8	27
42	A germ cellâ€specific ageing pattern in otherwise healthy men. Aging Cell, 2020, 19, e13242.	6.7	27
43	Deep Bisulfite Sequencing of Aberrantly Methylated Loci in a Patient with Multiple Methylation Defects. PLoS ONE, 2013, 8, e76953.	2.5	26
44	wg-blimp: an end-to-end analysis pipeline for whole genome bisulfite sequencing data. BMC Bioinformatics, 2020, 21, 169.	2.6	26
45	The imprinted NPAP1/C15orf2 gene in the Praderâ€“Willi syndrome region encodes a nuclear pore complex associated protein. Human Molecular Genetics, 2012, 21, 4038-4048.	2.9	25
46	Preovulatory Aging In Vivo and In Vitro Affects Maturation Rates, Abundance of Selected Proteins, Histone Methylation Pattern and Spindle Integrity in Murine Oocytes. PLoS ONE, 2016, 11, e0162722.	2.5	23
47	The sperm epigenome does not display recurrent epimutations in patients with severely impaired spermatogenesis. Clinical Epigenetics, 2020, 12, 61.	4.1	23
48	The C15orf2 gene in the Praderâ€“Willi syndrome region is subject to genomic imprinting and positive selection. Neurogenetics, 2010, 11, 153-161.	1.4	22
49	The Origin of the RB1 Imprint. PLoS ONE, 2013, 8, e81502.	2.5	21
50	A maternal deletion upstream of the imprint control region 2 in 11p15 causes loss of methylation and familial Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1280-1286.	2.8	20
51	Regions of common inter-individual DNA methylation differences in human monocytes: genetic basis and potential function. Epigenetics and Chromatin, 2017, 10, 37.	3.9	20
52	Clinical utility gene card for: Prader-Willi Syndrome. European Journal of Human Genetics, 2014, 22, 1153-1153.	2.8	18
53	Altering TET dioxygenase levels within physiological range affects DNA methylation dynamics of HEK293 cells. Epigenetics, 2015, 10, 819-833.	2.7	18
54	Hormone-induced delayed ovulation affects early embryonic development. Fertility and Sterility, 2011, 95, 2390-2394.	1.0	17

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55	Evolutionary Origin and Methylation Status of Human Intronic CpG Islands that Are Not Present in Mouse. Genome Biology and Evolution, 2014, 6, 1579-1588.	2.5	16
56	Lasp1 regulates adherens junction dynamics and fibroblast transformation in destructive arthritis. Nature Communications, 2021, 12, 3624.	12.8	16
57	The adult phenotype of Schaaf-Yang syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 294.	2.7	14
58	Whole-genome methylation analysis of testicular germ cells from cryptozoospermic men points to recurrent and functionally relevant DNA methylation changes. Clinical Epigenetics, 2021, 13, 160.	4.1	12
59	The Imprinted NPAP1 Gene in the Praderâ€“Willi Syndrome Region Belongs to a POM121-Related Family of Retrogenes. Genome Biology and Evolution, 2014, 6, 344-351.	2.5	11
60	Common genetic variation in the Angelman syndrome imprinting centre affects the imprinting of chromosome 15. European Journal of Human Genetics, 2020, 28, 835-839.	2.8	10
61	Methylation analysis of SST and SSTR4 promoters in the neocortex of Alzheimer's disease patients. Neuroscience Letters, 2014, 566, 241-246.	2.1	9
62	Parental origin and functional relevance of a de novo UBE3A variant. European Journal of Medical Genetics, 2011, 54, 19-24.	1.3	8
63	Waddington's epigenetic landscape and post-Darwinian biology. BioEssays, 2012, 34, 711-712.	2.5	7
64	Genome-wide methylation analysis of retrocopy-associated CpG islands and their genomic environment. Epigenetics, 2016, 11, 216-226.	2.7	7
65	In vitro postovulatory oocyte aging affects H3K9 trimethylation in two-cell embryos after IVF. Annals of Anatomy, 2020, 227, 151424.	1.9	7
66	Genome-Wide Analysis of the Nucleosome Landscape in Individuals with Coffin-Siris Syndrome. Cytogenetic and Genome Research, 2019, 159, 1-11.	1.1	5
67	The Diagnostic Journey of a Patient with Praderâ€“Willi-Like Syndrome and a Unique Homozygous SNURF-SNRPN Variant; Bio-Molecular Analysis and Review of the Literature. Genes, 2021, 12, 875.	2.4	4
68	Human PPP1R26P1 Functions as cis-Repressive Element in Mouse Rb1. PLoS ONE, 2013, 8, e74159.	2.5	4
69	Rhythm is not enough. Nature Genetics, 2007, 39, 1190-1191.	21.4	3
70	GC-rich repeat expansions: associated disorders and mechanisms. Medizinische Genetik, 2022, 33, 325-335.	0.2	2
71	Next-Generation-Sequencing in der Epigenetik. Medizinische Genetik, 2019, 31, 205-211.	0.2	1
72	A human somatic cell culture system for modelling gene silencing by transcriptional interference. Heliyon, 2020, 6, e03261.	3.2	1

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73	Novel strategies to cure imprinting disorders. Medizinische Genetik, 2020, 32, 335-340.	0.2	1
74	Of wolves and men: the role of paternal child care in the evolution of genomic imprinting. European Journal of Human Genetics, 2009, 17, 273-274.	2.8	0