# Bernhard Horsthemke

List of Publications by Year
in descending order



| 74 | 5,840 | 117619 34 | 82542 72 |
|---|---|---|---|
| papers | citations | h-index | 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 |