Matthias Begemann

List of Publications by Year
in descending order

74	2,431	218677	233421
papers	citations	26 h-index	45 g-index

81	81	81	3413
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of transcription factor binding sites using ATAC-seq. Genome Biology, 2019, 20, 45.	8.8	346
2	Paternally InheritedIGF2Mutation and Growth Restriction. New England Journal of Medicine, 2015, 373, 349-356.	27.0	165
3	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. Nature Communications, 2015, 6, 8086.	12.8	134
4	Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Journal of Medical Genetics, 2018, 55, 497-504.	3.2	126
5	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	2.0	101
6	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	6.7	89
7	Silver-Russell syndrome: genetic basis and molecular genetic testing. Orphanet Journal of Rare Diseases, 2010, 5, 19.	2.7	74
8	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	2.8	68
9	Clinical significance of copy number variations in the 11p15.5 imprinting control regions: new cases and review of the literature. Journal of Medical Genetics, 2012, 49, 547-553.	3.2	67
10	DEGS1-associated aberrant sphingolipid metabolism impairs nervous system function in humans. Journal of Clinical Investigation, 2019, 129, 1229-1239.	8.2	65
11	Novel deletions affecting the MEG3-DMR provide further evidence for a hierarchical regulation of imprinting in 14q32. European Journal of Human Genetics, 2015, 23, 180-188.	2.8	60
12	Silver-Russell patients showing a broad range of ICR1 and ICR2 hypomethylation in different tissues. Clinical Genetics, 2011, 80, 83-88.	2.0	54
13	Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. Human Reproduction Update, 2020, 26, 197-213.	10.8	51
14	Germline GPR161 Mutations Predispose to Pediatric Medulloblastoma. Journal of Clinical Oncology, 2020, 38, 43-50.	1.6	50
15	Additional molecular findings in 11p15-associated imprinting disorders: an urgent need for multi-locus testing. Journal of Molecular Medicine, 2014, 92, 769-777.	3.9	44
16	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	2.8	44
17	Molecular Karyotyping as a Relevant Diagnostic Tool in Children with Growth Retardation with Silver-Russell Features. Journal of Pediatrics, 2012, 161, 933-942.e1.	1.8	39
18	Maternal heterozygous NLRP7 variant results in recurrent reproductive failure and imprinting disturbances in the offspring. European Journal of Human Genetics, 2017, 25, 924-929.	2.8	39

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19	Genome-wide paternal uniparental disomy mosaicism in a woman with Beckwithâ€“Wiedemann syndrome and ovarian steroid cell tumour. European Journal of Human Genetics, 2013, 21, 788-791.	2.8	37
20	Comparison of flowâ€FISH and MMâ€“qPCR telomere length assessment techniques for the screening of telomeropathies. Annals of the New York Academy of Sciences, 2020, 1466, 93-103.	3.8	35
21	Silverâ€“Russell syndrome. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 153-160.	4.7	31
22	Use of multilocus methylation-specific single nucleotide primer extension (MS-SNuPE) technology in diagnostic testing for human imprinted loci. Epigenetics, 2012, 7, 473-481.	2.7	29
23	Maternal uniparental disomy of chromosome 16 [upd(16)mat]: clinical features are rather caused by (hidden) trisomy 16 mosaicism than by upd(16)mat itself. Clinical Genetics, 2017, 92, 45-51.	2.0	29
24	Disturbed methylation at multiple imprinted loci: an increasing observation in imprinting disorders. Epigenomics, 2011, 3, 625-637.	2.1	28
25	Epigenetic and genetic diagnosis of Silverâ€“Russell syndrome. Expert Review of Molecular Diagnostics, 2012, 12, 459-471.	3.1	28
26	Deletion of the paternal allele of the imprinted MEST/PEG1 region in a patient with Silverâ€“Russell syndrome features. Clinical Genetics, 2012, 81, 298-300.	2.0	27
27	Syndromic ciliopathies: From single gene to multi gene analysis by SNP arrays and next generation sequencing. Molecular and Cellular Probes, 2015, 29, 299-307.	2.1	27
28	Recurrent somatic mutations are rare in patients with cryptic dyskeratosis congenita. Leukemia, 2018, 32, 1762-1767.	7.2	27
29	DNA methylation changes during long-term in vitro cell culture are caused by epigenetic drift. Communications Biology, 2021, 4, 598.	4.4	27
30	Phenotypic spectrum and extent of DNA methylation defects associated with multilocus imprinting disturbances. Epigenomics, 2016, 8, 801-816.	2.1	26
31	Biallelic PADI6 variants cause multilocus imprinting disturbances and miscarriages in the same family. European Journal of Human Genetics, 2021, 29, 575-580.	2.8	24
32	Targeted Next Generation Sequencing Approach in Patients Referred for Silver-Russell Syndrome Testing Increases the Mutation Detection Rate and Provides Decisive Information for Clinical Management. Journal of Pediatrics, 2017, 187, 206-212.e1.	1.8	22
33	Genetic and epigenetic findings in Silver-Russell syndrome. Pediatric Endocrinology Reviews, 2010, 8, 86-93.	1.2	22
34	Segmental maternal uniparental disomy 7q associated with DLK1/GTL2 (14q32) hypomethylation. American Journal of Medical Genetics, Part A, 2012, 158A, 423-428.	1.2	21
35	The maternal uniparental disomy of chromosome 6Â(upd(6)mat) â€œphenotypeâ€• result of placental trisomyÂ6Âmosaicism?. Molecular Genetics & Genomic Medicine, 2017, 5, 668-677.	1.2	21
36	Examinations of maternal uniparental disomy and epimutations for chromosomes 6, 14, 16 and 20 in Silver-Russell syndrome-like phenotypes. BMC Medical Genetics, 2016, 17, 20.	2.1	19

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37	Haploinsufficiency of <i>ANKRD11</i> (16q24.3) Is Not Obligatorily Associated with Cognitive Impairment but Shows a Clinical Overlap with Silver-Russell Syndrome. Molecular Syndromology, 2013, 4, 246-249.	0.8	17
38	Microdeletions of the 7q32.2 imprinted region are associated with Silverâ€“Russell syndrome features. American Journal of Medical Genetics, Part A, 2016, 170, 743-749.	1.2	17
39	Kaiso mediates human ICR1 methylation maintenance and H19 transcriptional fine regulation. Clinical Epigenetics, 2016, 8, 47.	4.1	15
40	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. Brain, 2020, 143, 2406-2420.	7.6	15
41	Need for a precise molecular diagnosis in Beckwith-Wiedemann and Silver-Russell syndrome: what has to be considered and why it is important. Journal of Molecular Medicine, 2020, 98, 1447-1455.	3.9	15
42	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
43	Novel familial distal imprinting centre 1 (11p15.5) deletion provides further insights in imprinting regulation. Clinical Epigenetics, 2019, 11, 30.	4.1	13
44	Search for cis-acting factors and maternal effect variants in Silver-Russell patients with ICR1 hypomethylation and their mothers. European Journal of Human Genetics, 2019, 27, 42-48.	2.8	13
45	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	8.2	13
46	Congenital imprinting disorders: Application of multilocus and high throughput methods to decipher new pathomechanisms and improve their management. Molecular and Cellular Probes, 2015, 29, 282-290.	2.1	12
47	First report on concordant monozygotic twins with Silverâ€“Russell syndrome and ICR1 hypomethylation. European Journal of Medical Genetics, 2016, 59, 1-4.	1.3	12
48	Familial NEDD4L variant in periventricular nodular heterotopia and in a fetus with hypokinesia and flexion contractures. Molecular Genetics & Genomic Medicine, 2018, 6, 1255-1260.	1.2	12
49	One test for all: whole exome sequencing significantly improves the diagnostic yield in growth retarded patients referred for molecular testing for Silverâ€“Russell syndrome. Orphanet Journal of Rare Diseases, 2021, 16, 42.	2.7	12
50	NLRP genes and their role in preeclampsia and multi-locus imprinting disorders. Journal of Perinatal Medicine, 2018, 46, 169-173.	1.4	11
51	Next generation sequencing and imprinting disorders: Current applications and future perspectives: Lessons from Silver-Russell syndrome. Molecular and Cellular Probes, 2019, 44, 1-7.	2.1	11
52	HMGA2 Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2401-2407.	3.6	11
53	Frequency of KCNQ1 variants causing loss of methylation of Imprinting Centre 2 in Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2020, 12, 63.	4.1	11
54	Formation of upd(7)mat by trisomic rescue: SNP array typing provides new insights in chromosomal nondisjunction. Molecular Cytogenetics, 2017, 10, 28.	0.9	10

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55	A novel homozygous splice-site mutation in the SPTBN4 gene causes axonal neuropathy without intellectual disability. European Journal of Medical Genetics, 2020, 63, 103826.	1.3	9
56	Inherited cases of <scp>CNOT3</scp>â€associated intellectual developmental disorder with speech delay, autism, and dysmorphic facies. Clinical Genetics, 2020, 98, 408-412.	2.0	9
57	Heterogeneous growth patterns in carriers of chromosome 7p12.2 imbalances affecting GRB10. American Journal of Medical Genetics, Part A, 2012, 158A, 2815-2819.	1.2	7
58	Male infant with paternal uniparental diploidy mosaicism and a 46,XX/46,XY karyotype. American Journal of Medical Genetics, Part A, 2019, 179, 2252-2256.	1.2	7
59	Biallelic loss-of-function variants in WDR11 are associated with microcephaly and intellectual disability. European Journal of Human Genetics, 2021, 29, 1663-1668.	2.8	7
60	Congenital Imprinting Disorders: A Novel Mechanism Linking Seemingly Unrelated Disorders. Journal of Pediatrics, 2013, 163, 1202-1207.	1.8	6
61	Search for altered imprinting marks in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser patients. Molecular Genetics & Genomic Medicine, 2018, 6, 1225-1228.	1.2	5
62	Contribution of GRB10 to the prenatal phenotype in Silver-Russell syndrome? Lessons from 7p12 copy number variations. European Journal of Medical Genetics, 2019, 62, 103671.	1.3	5
63	Unusual deletion of the maternal 11p15 allele in Beckwithâ€“Wiedemann syndrome with an impact on both imprinting domains. Clinical Epigenetics, 2021, 13, 30.	4.1	5
64	IGF1R mutation analysis in short children with Silver-Russell syndrome features. Journal of Pediatric Genetics, 2013, 2, 113-7.	0.7	5
65	No major contribution of IGF2 variants to the etiology of sporadic 11p15â€associated imprinting disorders. American Journal of Medical Genetics, Part A, 2016, 170, 283-284.	1.2	4
66	Biallelic CSGALNACT1-mutations cause a mild skeletal dysplasia. Bone, 2019, 127, 446-451.	2.9	4
67	Testing of Buccal Swab DNA Does Not Increase the Detection Rate for Imprinting Control Region 1 Hypomethylation in Silver-Russell Syndrome. Genetic Testing and Molecular Biomarkers, 2011, 15, 725-726.	0.7	2
68	Molecular characterization of temple syndrome families with 14q32 epimutations. European Journal of Medical Genetics, 2020, 63, 104077.	1.3	2
69	Molecular testing for imprinting disorders. Medizinische Genetik, 2020, 32, 305-319.	0.2	2
70	Genetik und Epigenetik des Silver-Russell-Syndroms. Medizinische Genetik, 2010, 22, 405-410.	0.2	1
71	Successful allogeneic stem cell transplantation of a patient with Werner syndrome and acute myeloid leukemia. Leukemia Research, 2021, 108, 106609.	0.8	1
72	Uncovering common pathogenic transcriptional dysregulations in Silver-Russell syndrome. Molecular and Cellular Pediatrics, 2014, 1, A13.	1.8	0

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73	Elucidation of Additional Mutations By Next-Generation Sequencing Is of Clinical Significance in Patients with Rare MPNs and MDS/MPN Overlap Syndromes. Blood, 2016, 128, 4260-4260.	1.4	0
74	Prevalence of Inherited Predisposition Syndromes in Young Patients with Acute Myeloid Leukemia and Aberrant Karyotype. Blood, 2020, 136, 41-42.	1.4	0