Thomas Eggermann

List of Publications by Year
in descending order

229 papers	6,925 citations	61945 43 h-index	82499 72 g-index
250 all docs	250 docs citations	250 times ranked	5635 citing authors

#	Article	IF	Citations
1	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
2	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	4.3	336
3	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. Nature Reviews Genetics, 2019, 20, 235-248.	7.7	291
4	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	4.3	224
5	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	1.8	174
6	Human GRB10 is imprinted and expressed from the paternal and maternal allele in a highly tissue- and isoform-specific fashion. Human Molecular Genetics, 2000, 9, 1587-1595.	1.4	168
7	Paternally InheritedIGF2Mutation and Growth Restriction. New England Journal of Medicine, 2015, 373, 349-356.	13.9	165
8	Molecular studies in 37 Silver-Russell syndrome patients: frequency and etiology of uniparental disomy. Human Genetics, 1997, 100, 415-419.	1.8	156
9	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. Nature Communications, 2015, 6, 8086.	5.8	134
10	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.	1.5	126
11	Determination of SMN1 and SMN2 copy number using TaqManâ„¢ technology. Human Mutation, 2003, 22, 74-78.	1.1	111
12	The centromeric 11p15 imprinting centre is also involved in Silver-Russell syndrome. Journal of Medical Genetics, 2006, 44, 59-63.	1.5	108
13	Growth retardation versus overgrowth: Silver-Russell syndrome is genetically opposite to Beckwith-Wiedemann syndrome. Trends in Genetics, 2008, 24, 195-204.	2.9	107
14	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	1.0	101
15	Russellâ€“Silver syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 355-364.	0.7	97
16	Mosaicism and uniparental disomy in prenatal diagnosis. Trends in Molecular Medicine, 2015, 21, 77-87.	3.5	95
17	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	3.5	89
18	Is maternal duplication of 11p15 associated with Silver-Russell syndrome?. Journal of Medical Genetics, 2005, 42, e26-e26.	1.5	87

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19	Cystinuria: an inborn cause of urolithiasis. Orphanet Journal of Rare Diseases, 2012, 7, 19.	1.2	84
20	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	2.9	81
21	The Endocrine Phenotype in Silver-Russell Syndrome Is Defined by the Underlying Epigenetic Alteration. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1402-1407.	1.8	79
22	Epigenetic mutations in 11p15 in Silver-Russell syndrome are restricted to the telomeric imprinting domain. Journal of Medical Genetics, 2005, 43, 615-616.	1.5	76
23	Identification of interstitial maternal uniparental disomy (UPD) (14) and complete maternal UPD(20) in a cohort of growth retarded patients. Journal of Medical Genetics, 2001, 38, 86-89.	1.5	74
24	Silver-Russell syndrome: genetic basis and molecular genetic testing. Orphanet Journal of Rare Diseases, 2010, 5, 19.	1.2	74
25	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. Epigenetics, 2018, 13, 117-121.	1.3	70
26	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	1.1	69
27	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.	1.4	68
28	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.	1.5	67
29	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	2.6	66
30	Broad Clinical Spectrum in Silver-Russell Syndrome and Consequences for Genetic Testing in Growth Retardation. Pediatrics, 2009, 123, e929-e931.	1.0	61
31	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.	1.4	60
32	Cystinuria in children: Distribution and frequencies of mutations in the SLC3A1 and SLC7A9 genes. Kidney International, 2002, 62, 1136-1142.	2.6	59
33	Silver-Russell patients showing a broad range of ICR1 and ICR2 hypomethylation in different tissues. Clinical Genetics, 2011, 80, 83-88.	1.0	54
34	A rapid microarray based whole genome analysis for detection of uniparental disomy. Human Mutation, 2005, 26, 153-159.	1.1	53
35	Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. Human Reproduction Update, 2020, 26, 197-213.	5.2	51
36	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	1.4	50

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37	Supernumerary marker chromosomes derived from chromosome 15: analysis of 32 new cases. Clinical Genetics, 2002, 62, 89-93.	1.0	49
38	Silver-Russell and Beckwith-Wiedemann Syndromes: Opposite (Epi)Mutations in 11p15 Result in Opposite Clinical Pictures. Hormone Research in Paediatrics, 2009, 71, 30-35.	0.8	48
39	MAOA-VNTR polymorphism modulates context-dependent dopamine release and aggressive behavior in males. NeuroImage, 2016, 125, 378-385.	2.1	48
40	IGF-II Serum Levels Are Normal in Children with Silver-Russell Syndrome Who Frequently Carry Epimutations at theIGF2Locus. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 4709-4712.	1.8	47
41	(Epi)mutations inÂ11p15 significantly contribute toÂSilverâ€“Russell syndrome: butÂare they generally involved inÂgrowth retardation?. European Journal of Medical Genetics, 2006, 49, 414-418.	0.7	46
42	Report of two new cases of Pallister-Killian syndrome confirmed by FISH: Tissue-specific mosaicism and loss of i(12p) by in vitro selection. , 1997, 72, 106-110.		44
43	Use of multiplex ligationâ€dependent probe amplification increases the detection rate for 11p15 epigenetic alterations in Silverâ€“Russell syndrome. Clinical Genetics, 2008, 73, 79-84.	1.0	44
44	Submicroscopic chromosomal imbalances in idiopathic Silver-Russell syndrome (SRS): the SRS phenotype overlaps with the 12q14 microdeletion syndrome. Journal of Medical Genetics, 2010, 47, 356-360.	1.5	44
45	Additional molecular findings in 11p15-associated imprinting disorders: an urgent need for multi-locus testing. Journal of Molecular Medicine, 2014, 92, 769-777.	1.7	44
46	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.	1.4	44
47	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. Hormone Research in Paediatrics, 2020, 93, 182-196.	0.8	42
48	IGF2/H19 hypomethylation in Silverâ€“Russell syndrome and isolated hemihypoplasia. European Journal of Human Genetics, 2008, 16, 328-334.	1.4	41
49	Molecular Karyotyping as a Relevant Diagnostic Tool in Children with Growth Retardation with Silver-Russell Features. Journal of Pediatrics, 2012, 161, 933-942.e1.	0.9	39
50	Maternal heterozygous NLRP7 variant results in recurrent reproductive failure and imprinting disturbances in the offspring. European Journal of Human Genetics, 2017, 25, 924-929.	1.4	39
51	Conflicting Reports of Imprinting Status of Human GRB10 in Developing Brain: How Reliable Are Somatic Cell Hybrids for Predicting Allelic Origin of Expression?. American Journal of Human Genetics, 2001, 68, 543-544.	2.6	38
52	Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome.. Journal of Medical Genetics, 1998, 35, 784-786.	1.5	37
53	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.	1.4	37
54	Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. Journal of Clinical Investigation, 2019, 129, 5123-5136.	3.9	36

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55	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.	1.4	35
56	Multilocus methylation defects in imprinting disorders. Biomolecular Concepts, 2015, 6, 47-57.	1.0	34
57	Formation of uniparental disomy 7 delineated from new cases and a UPD7 case after trisomy 7 rescue. Presentation of own results and review of the literature. Annales De GÃ©nÃ©tique, 2000, 43, 15-21.	0.4	33
58	Significant contribution of genomic rearrangements in SLC3A1 and SLC7A9 to the etiology of cystinuria. Kidney International, 2003, 64, 1564-1572.	2.6	33
59	Silverâ€“Russell syndrome. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 153-160.	2.2	31
60	Adult Height and Epigenotype in Children with Silver-Russell Syndrome Treated with GH. Hormone Research in Paediatrics, 2013, 80, 193-200.	0.8	31
61	The population-specific distribution and frequencies of genomic variants in the SLC3A1 and SLC7A9 genes and their application in molecular genetic testing of cystinuria. Urological Research, 2004, 32, 75-78.	1.5	30
62	Segmental maternal UPD(7q) in Silverâ€“Russell syndrome. Clinical Genetics, 2008, 74, 486-489.	1.0	30
63	Cancer incidence and spectrum among children with genetically confirmed Beckwith-Wiedemann spectrum in Germany: a retrospective cohort study. British Journal of Cancer, 2020, 123, 619-623.	2.9	30
64	Use of multilocus methylation-specific single nucleotide primer extension (MS-SNuPE) technology in diagnostic testing for human imprinted loci. Epigenetics, 2012, 7, 473-481.	1.3	29
65	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.	1.0	29
66	Uniparental disomy: clinical indications for testing in growth retardation. European Journal of Pediatrics, 2002, 161, 305-312.	1.3	28
67	Disturbed methylation at multiple imprinted loci: an increasing observation in imprinting disorders. Epigenomics, 2011, 3, 625-637.	1.0	28
68	Epigenetic and genetic diagnosis of Silverâ€“Russell syndrome. Expert Review of Molecular Diagnostics, 2012, 12, 459-471.	1.5	28
69	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.	1.0	27
70	Syndromic ciliopathies: From single gene to multi gene analysis by SNP arrays and next generation sequencing. Molecular and Cellular Probes, 2015, 29, 299-307.	0.9	27
71	Screening for genomic variants in ZFP57 in Silver-Russell syndrome patients with 11p15 epimutations. European Journal of Medical Genetics, 2009, 52, 415-416.	0.7	26
72	UPDtool: a tool for detection of iso- and heterodisomy in parentâ€“child trios using SNP microarrays. Bioinformatics, 2013, 29, 1562-1564.	1.8	26

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73	Phenotypic spectrum and extent of DNA methylation defects associated with multilocus imprinting disturbances. Epigenomics, 2016, 8, 801-816.	1.0	26
74	Analysis of the genes SLC7A9 and SLC3A1 in unclassified cystinurics: mutation detection rates and association between variants in SLC7A9 and the disease. Clinical Nephrology, 2002, 57, 342-348.	0.4	26
75	Screening for Mutations in the Promoter and the Coding Region of the IGFBP1 and IGFBP3 Genes in Silver-Russell Syndrome Patients. Human Heredity, 1999, 49, 123-128.	0.4	24
76	Searching for genomic variants in IGF2 and CDKN1C in Silverâ€“Russell syndrome patients. Molecular Genetics and Metabolism, 2004, 82, 246-250.	0.5	24
77	Chromosome 11p15 duplication in Silverâ€Russell syndrome due to a maternally inherited translocation t(11;15). American Journal of Medical Genetics, Part A, 2010, 152A, 1484-1487.	0.7	24
78	<scp>NSD1</scp> duplication in Silverâ€“Russell syndrome (<scp>SRS</scp>): molecular karyotyping in patients with <scp>SRS</scp> features. Clinical Genetics, 2017, 91, 73-78.	1.0	24
79	Biallelic PADI6 variants cause multilocus imprinting disturbances and miscarriages in the same family. European Journal of Human Genetics, 2021, 29, 575-580.	1.4	24
80	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	1.8	23
81	Structural and sequence variants in patients with Silver-Russell syndrome or similar features-Curation of a disease database. Human Mutation, 2018, 39, 345-364.	1.1	23
82	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.	0.9	22
83	Neural networks underlying trait aggression depend on MAOA gene alleles. Brain Structure and Function, 2018, 223, 873-881.	1.2	22
84	Growth Restriction and Genomic Imprinting-Overlapping Phenotypes Support the Concept of an Imprinting Network. Genes, 2021, 12, 585.	1.0	22
85	Maternal uniparental disomy 16 and genetic counseling: new case and survey of published cases. Genetic Counseling, 2004, 15, 183-90.	0.1	22
86	Genetic and epigenetic findings in Silver-Russell syndrome. Pediatric Endocrinology Reviews, 2010, 8, 86-93.	1.2	22
87	A new splice site mutation in the SMN1 gene causes discrepant results in SMN1 deletion screening approaches. Neuromuscular Disorders, 2008, 18, 146-149.	0.3	21
88	Segmental maternal uniparental disomy 7q associated with DLK1/GTL2 (14q32) hypomethylation. American Journal of Medical Genetics, Part A, 2012, 158A, 423-428.	0.7	21
89	The maternal uniparental disomy of chromosome 6Â(upd(6)mat) â€œphenotypeâ€• result of placental trisomyÂ6Âmosaicism?. Molecular Genetics & Genomic Medicine, 2017, 5, 668-677.	0.6	21
90	Autosomal dominant spinal muscular atrophy with lower extremity predominance: A recognizable phenotype of BICD2 mutations. Muscle and Nerve, 2016, 54, 496-500.	1.0	20

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91	Corticoâ€limbic connectivity in MAOAâ€L carriers is vulnerable to acute tryptophan depletion. Human Brain Mapping, 2017, 38, 1622-1635.	1.9	20
92	Serotonergic Contributions to Human Brain Aggression Networks. Frontiers in Neuroscience, 2019, 13, 42.	1.4	20
93	Mutations in the SLC3A1 Gene in Cystinuric Patients: Frequencies and Identification of a Novel Mutation. Genetic Testing and Molecular Biomarkers, 1999, 3, 227-231.	1.7	19
94	2p21 Deletions in hypotoniaâ€“cystinuria syndrome. European Journal of Medical Genetics, 2012, 55, 561-563.	0.7	19
95	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
96	Prenatal Detection of Uniparental Disomies (UPD): Intended and Incidental Finding in the Era of Next Generation Genomics. Genes, 2020, 11, 1454.	1.0	19
97	Adult phenotype of Russellâ€Silver syndrome: A molecular support for Barkerâ€Brenner's theory. Congenital Anomalies (discontinued), 2015, 55, 167-169.	0.3	18
98	Identification of a 21q22 duplication in a Silverâ€“Russell syndrome patient further narrows down the Down syndrome critical region. American Journal of Medical Genetics, Part A, 2010, 152A, 356-359.	0.7	17
99	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.	0.7	17
100	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3.	0.3	17
101	Characterization of Genomic Variants in CSH1 and GH2, Two Candidate Genes for Silver-Russell Syndrome in 17q24-q25. Genetic Testing and Molecular Biomarkers, 2003, 7, 259-263.	1.7	16
102	Isolated cystinuria (OMIM 238200) is not a separate entity but is caused by a mutation in the cystinuria gene SLC7A9. Clinical Genetics, 2007, 71, 597-598.	1.0	16
103	Frequency and characterization of DNA methylation defects in children born SGA. European Journal of Human Genetics, 2013, 21, 838-843.	1.4	16
104	Molecular and clinical studies in 8 patients with Temple syndrome. Clinical Genetics, 2018, 93, 1179-1188.	1.0	16
105	Mosaic rearrangement of chromosome 18: Characterization by FISH mapping and DNA studies shows trisomy 18p and monosomy 18p both of paternal origin. American Journal of Medical Genetics Part A, 2000, 92, 101-106.	2.4	15
106	Screening for Insulin-like Growth Factor-I Receptor Mutations in Patients with Silver-Russell Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2002, 15, 1167-71.	0.4	15
107	No evidence for isolated imprinting mutations in the PEG1/MEST locus in Silverâ€“Russell patients. European Journal of Medical Genetics, 2008, 51, 322-324.	0.7	15
108	A familial GLI2 deletion (2q14.2) not associated with the holoprosencephaly syndrome phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 1121-1124.	0.7	15

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109	Kaiso mediates human ICR1 methylation maintenance and H19 transcriptional fine regulation. Clinical Epigenetics, 2016, 8, 47.	1.8	15
110	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.	1.7	15
111	Genetic testing in inherited endocrine disorders: joint position paper of the European reference network on rare endocrine conditions (Endo-ERN). Orphanet Journal of Rare Diseases, 2020, 15, 144.	1.2	15
112	Molecular pathophysiology of human MICU1 deficiency. Neuropathology and Applied Neurobiology, 2021, 47, 840-855.	1.8	15
113	Somatic mosaicism for a heterozygous deletion of the survival motor neuron (SMN1) gene. European Journal of Human Genetics, 2005, 13, 309-313.	1.4	14
114	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	1.8	14
115	Clinical utility gene card for: Silverâ€“Russell syndrome. European Journal of Human Genetics, 2011, 19, 3-3.	1.4	13
116	Revisiting Wilms tumour surveillance in Beckwithâ€“Wiedemann syndrome with IC2 methylation loss, reply. European Journal of Human Genetics, 2018, 26, 471-472.	1.4	13
117	Novel familial distal imprinting centre 1 (11p15.5) deletion provides further insights in imprinting regulation. Clinical Epigenetics, 2019, 11, 30.	1.8	13
118	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.	1.4	13
119	Genetic barcoding reveals clonal dominance in iPSC-derived mesenchymal stromal cells. Stem Cell Research and Therapy, 2020, 11, 105.	2.4	13
120	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	3.9	13
121	Molecular Basis of Beckwithâ€“Wiedemann Syndrome Spectrum with Associated Tumors and Consequences for Clinical Practice. Cancers, 2022, 14, 3083.	1.7	13
122	Gene Dosage Analysis in Silver-Russell Syndrome: Use of Quantitative Competitive PCR and Dual-Color FISH to Estimate the Frequency of Duplications in 7p11.2â€“p13. Genetic Testing and Molecular Biomarkers, 2001, 5, 261-266.	1.7	12
123	Diagnostic Proceeding in Silver-Russell Syndrome. Molecular Diagnosis and Therapy, 2005, 9, 205-209.	1.2	12
124	Clinical utility gene card for: Cystinuria. European Journal of Human Genetics, 2012, 20, 3-3.	1.4	12
125	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.	0.9	12
126	First report on concordant monozygotic twins with Silverâ€“Russell syndrome and ICR1 hypomethylation. European Journal of Medical Genetics, 2016, 59, 1-4.	0.7	12

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127	The origin of imprinting defects in Temple syndrome and comparison with other imprinting disorders. Epigenetics, 2018, 13, 822-828.	1.3	12
128	Novel mutation points to a hot spot in CDKN1C causing Silverâ€“Russell syndrome. Clinical Epigenetics, 2020, 12, 152.	1.8	12
129	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype. PLoS ONE, 2020, 15, e0230003.	1.1	12
130	Balance between macrophage migration inhibitory factor and sCD74 predicts outcome in patients with acute decompensation of cirrhosis. JHEP Reports, 2021, 3, 100221.	2.6	12
131	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.	1.2	12
132	NLRP genes and their role in preeclampsia and multi-locus imprinting disorders. Journal of Perinatal Medicine, 2018, 46, 169-173.	0.6	11
133	Next generation sequencing and imprinting disorders: Current applications and future perspectives: Lessons from Silver-Russell syndrome. Molecular and Cellular Probes, 2019, 44, 1-7.	0.9	11
134	HMGA2 Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2401-2407.	1.8	11
135	Frequency of KCNQ1 variants causing loss of methylation of Imprinting Centre 2 in Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2020, 12, 63.	1.8	11
136	Formation of upd(7)mat by trisomic rescue: SNP array typing provides new insights in chromosomal nondisjunction. Molecular Cytogenetics, 2017, 10, 28.	0.4	10
137	No evidence for point mutations in the novel renal cystine transporter AGT1/SLC7A13 contributing to the etiology of cystinuria. BMC Nephrology, 2018, 19, 278.	0.8	10
138	Patient with an autosomalâ€recessive <scp>MBTPS1</scp>â€linked phenotype and clinical features of <scp>Silverâ€“Russell</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2727-2730.	0.7	10
139	Search for uniparental disomy 14 in balanced Robertsonian translocation carriers. Clinical Genetics, 1999, 56, 464-466.	1.0	9
140	Searching for genomic variants in the MESTIT1 transcript in Silver-Russell syndrome patients. Journal of Medical Genetics, 2003, 40, 65e-65.	1.5	9
141	Mosaic tetrasomy 14pter-q13 due to a supernumerary isodicentric derivate of proximal chromosome 14q. , 2005, 134A, 305-308.		9
142	Identification of novel cystinuria mutations in pediatric patients. Journal of Pediatric Urology, 2006, 2, 575-578.	0.6	9
143	Epigenetic Regulation of Growth: Lessons from Silver-Russell Syndrome. Endocrine Development, 2009, 14, 10-19.	1.3	9
144	Inherited cases of <scp>CNOT3</scp>â€associated intellectual developmental disorder with speech delay, autism, and dysmorphic facies. Clinical Genetics, 2020, 98, 408-412.	1.0	9

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145	Paternal 132 bp deletion affecting KCNQ1OT1 in 11p15.5 is associated with growth retardation but does not affect imprinting. Journal of Medical Genetics, 2021, 58, 173-176.	1.5	9
146	A case of de novo translocation 16;21: trisomy 16q phenotype and origin of the aberration. Annales De GÃ©nÃ©tique, 1998, 41, 205-8.	0.4	9
147	IRS1 and GRB2 as members of the IGF signal transduction pathway are not associated with intrauterine growth retardation and Silver-Russell syndrome. Clinical Genetics, 2001, 59, 371-373.	1.0	8
148	Evidence from skewed X inactivation for trisomy mosaicism in Silver-Russell syndrome. European Journal of Human Genetics, 2001, 9, 887-891.	1.4	8
149	Supernumerary marker chromosome 7 and maternal uniparental disomy 7 in a boy with growth retardation and triangular face. Clinical Dysmorphology, 2006, 15, 9-12.	0.1	8
150	No Evidence for Additional Imprinting Defects in Silver-Russell Syndrome Patients with Maternal Uniparental Disomy 7 or 11p15 Epimutation. Journal of Pediatric Endocrinology and Metabolism, 2007, 20, 1329-31.	0.4	8
151	ICR1 Epimutations in 11p15 are Restricted to Patients with Silver-Russell Syndrome Features. Journal of Pediatric Endocrinology and Metabolism, 2008, 21, 59-62.	0.4	8
152	Isolated hypermethylation of GRB10 (7p12.2) in a Silverâ€“Russell syndrome patient carrying a 20p13 microdeletion. Clinical Genetics, 2014, 85, 399-400.	1.0	8
153	The Frequency of Methylation Abnormalities Among Estonian Patients Selected by Clinical Diagnostic Scoring Systems for Silverâ€“Russell Syndrome and Beckwithâ€“Wiedemann Syndrome. Genetic Testing and Molecular Biomarkers, 2015, 19, 684-691.	0.3	8
154	Uniparental disomy as an unexpected cause of Meckelâ€“Gruber syndrome: report of a case. Pediatric Nephrology, 2017, 32, 1989-1992.	0.9	8
155	Molecular and Clinical Opposite Findings in 11p15.5 Associated Imprinting Disorders: Characterization of Basic Mechanisms to Improve Clinical Management. International Journal of Molecular Sciences, 2019, 20, 4219.	1.8	8
156	upd(20)mat is a rare cause of the Silverâ€Russellâ€syndromeâ€like phenotype: Two unrelated cases and screening of large cohorts. Clinical Genetics, 2020, 97, 902-907.	1.0	8
157	Search for mutations in SLC1A5 (19q13) in cystinuria patients. Journal of Inherited Metabolic Disease, 2005, 28, 1169-1171.	1.7	7
158	Heterogeneous growth patterns in carriers of chromosome 7p12.2 imbalances affecting GRB10. American Journal of Medical Genetics, Part A, 2012, 158A, 2815-2819.	0.7	7
159	In vivo Investigations of the Effect of Short- and Long-Term Recombinant Growth Hormone Treatment on DNA-Methylation in Humans. PLoS ONE, 2015, 10, e0120463.	1.1	7
160	12q14 microdeletion syndrome: A family with short stature and Silver-Russell syndrome (SRS)-like phenotype and review of the literature. European Journal of Medical Genetics, 2018, 61, 421-427.	0.7	7
161	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.	0.7	7
162	Germline variants in DNA repair genes, including BRCA1/2, may cause familial myeloproliferative neoplasms. Blood Advances, 2021, 5, 3373-3376.	2.5	7

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164	Hypomethylation in the 11p15 telomeric imprinting domain in a patient with Silver-Russell syndrome with a CSH1 deletion (17q24) renders a functional role of this alteration unlikely. Journal of Medical Genetics, 2007, 44, e77-e77.	1.5	6
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