Thomas Eggermann

List of Publications by Year in Descending Order

239 papers	5,007 citations	39 h-index	61 g-index
250 ext. papers	6,047 ext. citations	4.6 avg, IF	5.55 L-index

#	Paper	IF	Citations
239	Novel homozygous nonsense mutation in the P5PN-1 coding gene as an alternative cause for hereditary anemia with basophilic stippling.. *Clinical Case Reports (discontinued)*, **2022**, 10, e05501	0.7	
238	Childhood adversity and approach/avoidance-related behaviour in boys.. *Journal of Neural Transmission*, **2022**, 129, 421	4.3	
237	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences.. *Clinical Epigenetics*, **2022**, 14, 41	7.7	0
236	Microdeletions in 1q21 and 8q12.1 depict two additional molecular subgroups of Silver-Russell syndrome like phenotypes.. *Molecular Cytogenetics*, **2022**, 15, 19	2	1
235	Corrigendum to: Clinical spectrum and management of imprinting disorders. *Medizinische Genetik*, **2021**, 33, 61-63	0.5	
234	Paternal 132 bp deletion affecting in 11p15.5 is associated with growth retardation but does not affect imprinting. *Journal of Medical Genetics*, **2021**, 58, 173-176	5.8	2
233	Balance between macrophage migration inhibitory factor and sCD74 predicts outcome in patients with acute decompensation of cirrhosis. *JHEP Reports*, **2021**, 3, 100221	10.3	2
232	Growth Restriction and Genomic Imprinting-Overlapping Phenotypes Support the Concept of an Imprinting Network. *Genes*, **2021**, 12,	4.2	10
231	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. *Journal of Clinical Investigation*, **2021**, 131,	15.9	2
230	Overgrowth-associated partial trisomy 15q24.3-qter and mosaic 11p15.5 duplication involving Silver-Russell region in a patient with lateralized asymmetry and developmental delay. *Clinical Dysmorphology*, **2021**, 30, 189-193	0.9	0
229	Biallelic PADI6 variants cause multilocus imprinting disturbances and miscarriages in the same family. *European Journal of Human Genetics*, **2021**, 29, 575-580	5.3	12
228	Deletion of 16q22.2q23.3 in a Boy with a Phenotype Reminiscent of Silver-Russell Syndrome. *Molecular Syndromology*, **2021**, 12, 300-304	1.5	0
227	Unusual deletion of the maternal 11p15 allele in Beckwith-Wiedemann syndrome with an impact on both imprinting domains. *Clinical Epigenetics*, **2021**, 13, 30	7.7	2
226	Molecular pathophysiology of human MICU1 deficiency. *Neuropathology and Applied Neurobiology*, **2021**, 47, 840-855	5.2	2
225	Maternal Effect Mutations: A Novel Cause for Human Reproductive Failure. *Geburtshilfe Und Frauenheilkunde*, **2021**, 81, 780-788	2	2
224	Germline variants in DNA repair genes, including BRCA1/2, may cause familial myeloproliferative neoplasms. *Blood Advances*, **2021**, 5, 3373-3376	7.8	2
223	One test for all: whole exome sequencing significantly improves the diagnostic yield in growth retarded patients referred for molecular testing for Silver-Russell syndrome. *Orphanet Journal of Rare Diseases*, **2021**, 16, 42	4.2	5

222	Prenatal Detection of Uniparental Disomies (UPD): Intended and Incidental Finding in the Era of Next Generation Genomics. *Genes*, **2020**, 11,	4.2	6
221	HMGA2 Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	8
220	Frequency of KCNQ1 variants causing loss of methylation of Imprinting Centre 2 in Beckwith-Wiedemann syndrome. *Clinical Epigenetics*, **2020**, 12, 63	7.7	5
219	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	8.7	14
218	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	4.2	7
217	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype. *PLoS ONE*, **2020**, 15, e0230003	3.7	6
216	Genetic barcoding reveals clonal dominance in iPSC-derived mesenchymal stromal cells. *Stem Cell Research and Therapy*, **2020**, 11, 105	8.3	4
215	Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. *Human Reproduction Update*, **2020**, 26, 197-213	15.8	30
214	upd(20)mat is a rare cause of the Silver-Russell-syndrome-like phenotype: Two unrelated cases and screening of large cohorts. *Clinical Genetics*, **2020**, 97, 902-907	4	6
213	Imprinting disorders: novel findings and translation into diagnostics and management. *Medizinische Genetik*, **2020**, 32, 295-296	0.5	
212	Clinical spectrum and management of imprinting disorders. *Medizinische Genetik*, **2020**, 32, 321-334	0.5	1
211	Molecular testing for imprinting disorders. *Medizinische Genetik*, **2020**, 32, 305-319	0.5	1
210	Molecular characterization of temple syndrome families with 14q32 epimutations. *European Journal of Medical Genetics*, **2020**, 63, 104077	2.6	1
209	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	3.3	16
208	Novel mutation points to a hot spot in CDKN1C causing Silver-Russell syndrome. *Clinical Epigenetics* , **2020**, 12, 152	7.7	5
207	Inherited cases of CNOT3-associated intellectual developmental disorder with speech delay, autism, and dysmorphic facies. *Clinical Genetics*, **2020**, 98, 408-412	4	1
206	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	5.5	8
205	Patient with an autosomal-recessive MBTPS1-linked phenotype and clinical features of Silver-Russell syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2727-2730	2.5	4

204	Heterogeneous phenotypes in families with duplications of the paternal allele within the imprinting center 1 (H19/IGF2:TSS-DMR) in 11p15.5. *Clinical Genetics*, **2020**, 98, 418-419	4	0
203	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype **2020**, 15, e0230003		
202	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype **2020**, 15, e0230003		
201	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype **2020**, 15, e0230003		
200	Mutation analysis of multiple pilomatricomas in a patient with myotonic dystrophy type 1 suggests a DM1-associated hypermutation phenotype **2020**, 15, e0230003		
199	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,	6.3	3
198	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	2.6	4
197	Serotonergic Contributions to Human Brain Aggression Networks. *Frontiers in Neuroscience*, **2019**, 13, 42	5.1	10
196	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. *Genetical Research*, **2019**, 101, e3	1.1	9
195	Novel familial distal imprinting centre 1 (11p15.5) deletion provides further insights in imprinting regulation. *Clinical Epigenetics*, **2019**, 11, 30	7.7	10
194	Multilocus Methylation Assays in Epigenetics **2019**, 2181-2202		
193	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	2.5	3
192	Genetic Variants in the Promoter Region of the Macrophage Migration Inhibitory Factor are Associated with the Severity of Hepatitis C Virus-Induced Liver Fibrosis. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	2
191	Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. *Journal of Clinical Investigation*, **2019**, 129, 5123-5136	15.9	16
190	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	5.3	11
189	Next generation sequencing and imprinting disorders: Current applications and future perspectives: Lessons from Silver-Russell syndrome. *Molecular and Cellular Probes*, **2019**, 44, 1-7	3.3	9
188	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. *Nature Reviews Genetics*, **2019**, 20, 235-248	30.1	151
187	Molecular and clinical studies in 8 patients with Temple syndrome. *Clinical Genetics*, **2018**, 93, 1179-1188	4	14

186	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	2.6	6
185	Revisiting Wilms tumour surveillance in Beckwith-Wiedemann syndrome with IC2 methylation loss, reply. *European Journal of Human Genetics*, **2018**, 26, 471-472	5.3	7
184	Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. *Nature Reviews Endocrinology*, **2018**, 14, 229-249	15.2	234
183	Structural and sequence variants in patients with Silver-Russell syndrome or similar features-Curation of a disease database. *Human Mutation*, **2018**, 39, 345-364	4.7	17
182	Maternal variants in and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. *Journal of Medical Genetics*, **2018**, 55, 497-504	5.8	66
181	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. *Epigenetics*, **2018**, 13, 117-121	5.7	50
180	Neural networks underlying trait aggression depend on MAOA gene alleles. *Brain Structure and Function*, **2018**, 223, 873-881	4	17
179	Duplication of 11p15 Associated With Congenital Diaphragmatic Hernia. *Frontiers in Pediatrics*, **2018**, 6, 116	3.4	1
178	Uniparental Disomy and Imprinting Disorders. *OBM Genetics*, **2018**, 2, 1-1	1.7	3
177	NLRP genes and their role in preeclampsia and multi-locus imprinting disorders. *Journal of Perinatal Medicine*, **2018**, 46, 169-173	2.7	6
176	No evidence for point mutations in the novel renal cystine transporter AGT1/SLC7A13 contributing to the etiology of cystinuria. *BMC Nephrology*, **2018**, 19, 278	2.7	5
175	The origin of imprinting defects in Temple syndrome and comparison with other imprinting disorders. *Epigenetics*, **2018**, 13, 822-828	5.7	7
174	Search for altered imprinting marks in Mayer-Rokitansky-K□ter-Hauser patients. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 1225-1228	2.3	2
173	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. *Nature Reviews Endocrinology*, **2018**, 14, 476-500	15.2	132
172	NSD1 duplication in Silver-Russell syndrome (SRS): molecular karyotyping in patients with SRS features. *Clinical Genetics*, **2017**, 91, 73-78	4	14
171	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	5.3	21
170	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	3.6	13
169	Maternal heterozygous NLRP7 variant results in recurrent reproductive failure and imprinting disturbances in the offspring. *European Journal of Human Genetics*, **2017**, 25, 924-929	5.3	34

168	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	4	20
167	Cortico-limbic connectivity in MAOA-L carriers is vulnerable to acute tryptophan depletion. *Human Brain Mapping*, **2017**, 38, 1622-1635	5.9	13
166	The Diagnostic Value of IGF-2 and the IGF/IGFBP-3 System in Silver-Russell Syndrome. *Hormone Research in Paediatrics*, **2017**, 88, 201-207	3.3	5
165	Formation of upd(7)mat by trisomic rescue: SNP array typing provides new insights in chromosomal nondisjunction. *Molecular Cytogenetics*, **2017**, 10, 28	2	8
164	Diagnostik und Therapie des Silver-Russell-Syndroms. *Monatsschrift Fur Kinderheilkunde*, **2017**, 165, 895-904		
163	The maternal uniparental disomy of chromosome 6 (upd(6)mat) "phenotype": result of placental trisomy 6 mosaicism?. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 668-677	2.3	13
162	Uniparental disomy as an unexpected cause of Meckel-Gruber syndrome: report of a case. *Pediatric Nephrology*, **2017**, 32, 1989-1992	3.2	6
161	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. *Nature Reviews Endocrinology*, **2017**, 13, 105-124	15.2	224
160	Recent Advances in Imprinting Disorders. *Clinical Genetics*, **2017**, 91, 3-13	4	62
159	Imprinting Disorders in Humans **2017**, 581-592		
158	Multilocus Methylation Assays in Epigenetics **2017**, 1-22		
157	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. *Genetics in Medicine*, **2016**, 18, 309-15	8.1	52
156	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	11	41
155	Microdeletions of the 7q32.2 imprinted region are associated with Silver-Russell syndrome features. *American Journal of Medical Genetics, Part A*, **2016**, 170, 743-9	2.5	14
154	Kaiso mediates human ICR1 methylation maintenance and H19 transcriptional fine regulation. *Clinical Epigenetics*, **2016**, 8, 47	7.7	10
153	Phenotypic spectrum and extent of DNA methylation defects associated with multilocus imprinting disturbances. *Epigenomics*, **2016**, 8, 801-16	4.4	16
152	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	18
151	First report on concordant monozygotic twins with Silver-Russell syndrome and ICR1 hypomethylation. *European Journal of Medical Genetics*, **2016**, 59, 1-4	2.6	9

150	MAOA-VNTR polymorphism modulates context-dependent dopamine release and aggressive behavior in males. *NeuroImage*, **2016**, 125, 378-385	7.9	36
149	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. *European Journal of Human Genetics*, **2016**, 24, 784-93	5.3	34
148	Autosomal dominant spinal muscular atrophy with lower extremity predominance: A recognizable phenotype of BICD2 mutations. *Muscle and Nerve*, **2016**, 54, 496-500	3.4	18
147	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. *Trends in Genetics*, **2016**, 32, 444-455	8.5	51
146	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2016**, 24, 1377-87	5.3	54
145	No major contribution of IGF2 variants to the etiology of sporadic 11p15-associated imprinting disorders. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 283-4	2.5	3
144	Clinical utility gene card for: Proximal spinal muscular atrophy (SMA) - update 2015. *European Journal of Human Genetics*, **2015**, 23,	5.3	4
143	Paternally Inherited IGF2 Mutation and Growth Restriction. *New England Journal of Medicine*, **2015**, 373, 349-56	59.2	139
142	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. *Clinical Epigenetics*, **2015**, 7, 23	7.7	19
141	Chromosom 14-assoziierte Imprintingsyndrome Temple- und Kagami-Ogata-Syndrome. *Medizinische Genetik*, **2015**, 27, 247-253	0.5	2
140	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. *Nature Communications*, **2015**, 6, 8086	17.4	102
139	Congenital imprinting disorders: Application of multilocus and high throughput methods to decipher new pathomechanisms and improve their management. *Molecular and Cellular Probes*, **2015**, 29, 282-90	3.3	10
138	Multilocus methylation defects in imprinting disorders. *Biomolecular Concepts*, **2015**, 6, 47-57	3.7	31
137	Mosaicism and uniparental disomy in prenatal diagnosis. *Trends in Molecular Medicine*, **2015**, 21, 77-87	11.5	69
136	Novel deletions affecting the MEG3-DMR provide further evidence for a hierarchical regulation of imprinting in 14q32. *European Journal of Human Genetics*, **2015**, 23, 180-8	5.3	37
135	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. *Clinical Epigenetics*, **2015**, 7, 123	7.7	115
134	A familial GLI2 deletion (2q14.2) not associated with the holoprosencephaly syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1121-4	2.5	10
133	Adult phenotype of Russell-Silver syndrome: A molecular support for Barker-Brenner theory. *Congenital Anomalies (discontinued)*, **2015**, 55, 167-9	1.1	14

132	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	3.7	7
131	Syndromic ciliopathies: From single gene to multi gene analysis by SNP arrays and next generation sequencing. *Molecular and Cellular Probes*, **2015**, 29, 299-307	3.3	19
130	The Frequency of Methylation Abnormalities Among Estonian Patients Selected by Clinical Diagnostic Scoring Systems for Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome. *Genetic Testing and Molecular Biomarkers*, **2015**, 19, 684-91	1.6	8
129	Fundamental Aspects of Epigenetic in Cancer **2015**, 1-33		
128	Epigenetik. *Gynakologische Endokrinologie*, **2014**, 12, 74-78	0.1	0
127	Isolated hypermethylation of GRB10 (7p12.2) in a Silver-Russell syndrome patient carrying a 20p13 microdeletion. *Clinical Genetics*, **2014**, 85, 399-400	4	7
126	Additional molecular findings in 11p15-associated imprinting disorders: an urgent need for multi-locus testing. *Journal of Molecular Medicine*, **2014**, 92, 769-77	5.5	42
125	Uniparentale Disomien und Mosaike. *Medizinische Genetik*, **2014**, 26, 315-323	0.5	
124	CDKN1C mutations: two sides of the same coin. *Trends in Molecular Medicine*, **2014**, 20, 614-22	11.5	68
123	Patient with three euchromatic supernumerary marker chromosomes derived from chromosomes 1, 12, and 18: characterization and evaluation of the aberrations. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 736-40	2.5	2
122	Clinical utility gene card for: Beckwith-Wiedemann Syndrome. *European Journal of Human Genetics*, **2014**, 22,	5.3	43
121	Einsatz der molekularen Karyotypisierung in der Pdiatrie. *Monatsschrift Fur Kinderheilkunde*, **2013**, 161, 633-643	0.2	1
120	Molekulargenetische Diagnostik von Imprintingerkrankungen. *Medizinische Genetik*, **2013**, 25, 5-14	0.5	2
119	Frequency and characterization of DNA methylation defects in children born SGA. *European Journal of Human Genetics*, **2013**, 21, 838-43	5.3	12
118	Congenital imprinting disorders: a novel mechanism linking seemingly unrelated disorders. *Journal of Pediatrics*, **2013**, 163, 1202-7	3.6	5
117	Molekulargenetische Diagnostik von Imprinting-Erkrankungen. *BioSpektrum*, **2013**, 19, 753-758	0.1	
116	UPDtool: a tool for detection of iso- and heterodisomy in parent-child trios using SNP microarrays. *Bioinformatics*, **2013**, 29, 1562-4	7.2	21
115	Genome-wide paternal uniparental disomy mosaicism in a woman with Beckwith-Wiedemann syndrome and ovarian steroid cell tumour. *European Journal of Human Genetics*, **2013**, 21, 788-91	5.3	32

114	Adult height and epigenotype in children with Silver-Russell syndrome treated with GH. *Hormone Research in Paediatrics*, **2013**, 80, 193-200	3.3	25
113	IGF1R mutation analysis in short children with Silver-Russell syndrome features. *Journal of Pediatric Genetics*, **2013**, 2, 113-7	0.7	5
112	Multi-exon deletion in the XDH gene as a cause of classical xanthinuria. *Clinical Nephrology*, **2013**, 79, 78-80	2.1	3
111	Segmental maternal uniparental disomy 7q associated with DLK1/GTL2 (14q32) hypomethylation. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 423-8	2.5	20
110	Molecular karyotyping as a relevant diagnostic tool in children with growth retardation with Silver-Russell features. *Journal of Pediatrics*, **2012**, 161, 933-42	3.6	32
109	2p21 Deletions in hypotonia-cystinuria syndrome. *European Journal of Medical Genetics*, **2012**, 55, 561-3	2.6	13
108	Imprinting Disorders **2012**, 379-395		2
107	Epigenetic and genetic diagnosis of Silver-Russell syndrome. *Expert Review of Molecular Diagnostics* , **2012**, 12, 459-71	3.8	21
106	Heterogeneous growth patterns in carriers of chromosome 7p12.2 imbalances affecting GRB10. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2815-9	2.5	6
105	Cystinuria: an inborn cause of urolithiasis. *Orphanet Journal of Rare Diseases*, **2012**, 7, 19	4.2	63
104	Deletion of the paternal allele of the imprinted MEST/PEG1 region in a patient with Silver-Russell syndrome features. *Clinical Genetics*, **2012**, 81, 298-300	4	23
103	Clinical utility gene card for: Cystinuria. *European Journal of Human Genetics*, **2012**, 20,	5.3	10
102	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, 547-53	5.8	58
101	Use of multilocus methylation-specific single nucleotide primer extension (MS-SNuPE) technology in diagnostic testing for human imprinted loci. *Epigenetics*, **2012**, 7, 473-81	5.7	28
100	Duplication 3q13.11q23: Longitudinal study in a patient over a period of more than 7 years and refinements of the breakpoints. *Journal of Pediatric Genetics*, **2012**, 1, 143-7	0.7	
99	Mosaic tetrasomy 14pterq13.1: longitudinal study. *European Journal of Medical Genetics*, **2011**, 54, e465-7	2.6	2
98	Silver-Russell syndrome. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2011**, 25, 153-60	6.5	26
97	Disturbed methylation at multiple imprinted loci: an increasing observation in imprinting disorders. *Epigenomics*, **2011**, 3, 625-37	4.4	25

96	Molecular Genetic Testing in Cystinuria. *International Journal of Human Genetics*, **2011**, 11, 41-44	1	3
95	Imprinting Disorders in Humans **2011**, 581-593		
94	Supernumerary Asymmetric Dic(15;15) With Secondary Mosaic Formation in One of Two Developmentally Retarded Twins. *International Journal of Human Genetics*, **2011**, 11, 75-82	1	
93	Silver-Russell patients showing a broad range of ICR1 and ICR2 hypomethylation in different tissues. *Clinical Genetics*, **2011**, 80, 83-8	4	51
92	Clinical utility gene card for: Silver-Russell syndrome. *European Journal of Human Genetics*, **2011**, 19,	5.3	11
91	Silver-Russell-Kleinwuchs. *Monatsschrift Fur Kinderheilkunde*, **2011**, 159, 576-582	0.2	2
90	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, 725-6	1.6	2
89	Submicroscopic chromosomal imbalances in idiopathic Silver-Russell syndrome (SRS): the SRS phenotype overlaps with the 12q14 microdeletion syndrome. *Journal of Medical Genetics*, **2010**, 47, 356-60	5.8	38
88	Silver-Russell syndrome: genetic basis and molecular genetic testing. *Orphanet Journal of Rare Diseases*, **2010**, 5, 19	4.2	62
87	MBD3 mutations are not responsible for ICR1 hypomethylation in Silver-Russell syndrome. *European Journal of Medical Genetics*, **2010**, 53, 23-4	2.6	3
86	Uniparentale Disomien. *Medizinische Genetik*, **2010**, 22, 439-451	0.5	3
85	Genetik und Epigenetik des Silver-Russell-Syndroms. *Medizinische Genetik*, **2010**, 22, 405-410	0.5	1
84	Identification of a 21q22 duplication in a Silver-Russell syndrome patient further narrows down the Down syndrome critical region. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 356-9	2.5	12
83	Chromosome 11p15 duplication in Silver-Russell syndrome due to a maternally inherited translocation t(11;15). *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1484-7	2.5	11
82	Russell-Silver syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 355-64	3.1	77
81	Genetic and epigenetic findings in Silver-Russell syndrome. *Pediatric Endocrinology Reviews*, **2010**, 8, 86-93	1.1	20
80	Molecular Analyses of the BORIS Gene in Children with Silver-Russell Syndrome. *International Journal of Human Genetics*, **2009**, 9, 269-272	1	
79	LOT1 (ZAC1/PLAGL1) as member of an imprinted gene network does not harbor Silver-Russell specific variants. *Journal of Pediatric Endocrinology and Metabolism*, **2009**, 22, 555-9	1.6	4

78	Silver-Russell and Beckwith-Wiedemann syndromes: opposite (epi)mutations in 11p15 result in opposite clinical pictures. *Hormone Research in Paediatrics*, **2009**, 71 Suppl 2, 30-5	3.3	40
77	Epigenetic regulation of growth: lessons from Silver-Russell syndrome. *Endocrine Development*, **2009**, 14, 10-9		9
76	Broad clinical spectrum in Silver-Russell syndrome and consequences for genetic testing in growth retardation. *Pediatrics*, **2009**, 123, e929-31	7.4	51
75	Novel human pathological mutations. Gene symbol: SLC3A1. Disease: Cystinuria. *Human Genetics*, **2009**, 126, 330	6.3	
74	Screening for genomic variants in ZFP57 in Silver-Russell syndrome patients with 11p15 epimutations. *European Journal of Medical Genetics*, **2009**, 52, 415-6	2.6	21
73	Non-Mosaic Trisomy 7 in Chorionic Villi and Trisomy 18 in the Fetus: An Extreme form of Mosaic Variegated Aneuploidy?. *International Journal of Human Genetics*, **2009**, 9, 1-4	1	
72	Novel human pathological mutations. Gene symbol: SLC3A1. Disease: Cystinuria. *Human Genetics*, **2009**, 126, 329	6.3	1
71	Novel human pathological mutations. Gene symbol: SLC7A9. Disease: Cystinuria. *Human Genetics*, **2009**, 126, 330	6.3	1
70	IGF2/H19 hypomethylation in Silver-Russell syndrome and isolated hemihypoplasia. *European Journal of Human Genetics*, **2008**, 16, 328-34	5.3	33
69	Segmental maternal UPD(7q) in Silver-Russell syndrome. *Clinical Genetics*, **2008**, 74, 486-9	4	27
68	Growth retardation versus overgrowth: Silver-Russell syndrome is genetically opposite to Beckwith-Wiedemann syndrome. *Trends in Genetics*, **2008**, 24, 195-204	8.5	92
67	A new splice site mutation in the SMN1 gene causes discrepant results in SMN1 deletion screening approaches. *Neuromuscular Disorders*, **2008**, 18, 146-9	2.9	18
66	No evidence for isolated imprinting mutations in the PEG1/MEST locus in Silver-Russell patients. *European Journal of Medical Genetics*, **2008**, 51, 322-4	2.6	13
65	ICR1 epimutations in llp15 are restricted to patients with Silver-Russell syndrome features. *Journal of Pediatric Endocrinology and Metabolism*, **2008**, 21, 59-62	1.6	4
64	Are H19 variants associated with Silver-Russell syndrome?. *Journal of Pediatric Endocrinology and Metabolism*, **2008**, 21, 985-93	1.6	3
63	The endocrine phenotype in silver-russell syndrome is defined by the underlying epigenetic alteration. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 1402-7	5.6	72
62	Mosaic Trisomy 1q Due to a de novo Translocation in a Foetus with Early Developmental Abnormalities (Karyotype 46,XY, der(14),t(1;14)(p11;p11.2)/46,XY) Delineation of Parent and Cell Stage of Origin. *International Journal of Human Genetics*, **2008**, 8, 317-323	1	1
61	Use of multiplex ligation-dependent probe amplification increases the detection rate for 11p15 epigenetic alterations in Silver-Russell syndrome. *Clinical Genetics*, **2008**, 73, 79-84	4	37

60	Isolated cystinuria (OMIM 238200) is not a separate entity but is caused by a mutation in the cystinuria gene SLC7A9. *Clinical Genetics*, **2007**, 71, 597-8	4	13
59	The centromeric 11p15 imprinting centre is also involved in Silver-Russell syndrome. *Journal of Medical Genetics*, **2007**, 44, 59-63	5.8	88
58	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	1.6	6
57	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	5.8	4
56	Mutation analysis of GNAS1 and overlapping transcripts in Silver-Russell syndrome patients. *Molecular Genetics and Metabolism*, **2007**, 90, 224-6	3.7	2
55	IGF-II serum levels are normal in children with Silver-Russell syndrome who frequently carry epimutations at the IGF2 locus. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 4709-12	5.6	44
54	Identification of novel cystinuria mutations in pediatric patients. *Journal of Pediatric Urology*, **2006**, 2, 575-8	1.5	9
53	(Epi)mutations in 11p15 significantly contribute to Silver-Russell syndrome: but are they generally involved in growth retardation?. *European Journal of Medical Genetics*, **2006**, 49, 414-8	2.6	41
52	Supernumerary marker chromosome 7 and maternal uniparental disomy 7 in a boy with growth retardation and triangular face. *Clinical Dysmorphology*, **2006**, 15, 9-12	0.9	7
51	Epigenetic mutations in 11p15 in Silver-Russell syndrome are restricted to the telomeric imprinting domain. *Journal of Medical Genetics*, **2006**, 43, 615-6	5.8	63
50	Gene symbol: SLC3A1. Disease: Cystinuria. Accession #Hm0543. *Human Genetics*, **2006**, 118, 779	6.3	
49	Functional analysis of a new splice site mutation, c.605-3C>A, in the cystinuria gene SLC7A9 leading to exon skipping. *Molecular Genetics and Metabolism*, **2005**, 84, 172-5	3.7	1
48	Analysis of genomic variants in the KCNQ1OT1 transcript in Silver-Russell syndrome patients. *Molecular Genetics and Metabolism*, **2005**, 84, 376-7	3.7	4
47	Diagnostic proceeding in Silver-Russell syndrome. *Molecular Diagnosis and Therapy*, **2005**, 9, 205-9		12
46	Somatic mosaicism for a heterozygous deletion of the survival motor neuron (SMN1) gene. *European Journal of Human Genetics*, **2005**, 13, 309-13	5.3	11
45	A rapid microarray based whole genome analysis for detection of uniparental disomy. *Human Mutation*, **2005**, 26, 153-9	4.7	50
44	Mosaic tetrasomy 14pter-q13 due to a supernumerary isodicentric derivate of proximal chromosome 14q **2005**, 134, 305-8		9
43	Genetik des Silver-Russell-Syndroms. *Monatsschrift Fur Kinderheilkunde*, **2005**, 153, 264-272	0.2	2

42	Search for mutations in SLC1A5 (19q13) in cystinuria patients. *Journal of Inherited Metabolic Disease* , **2005**, 28, 1169-71	5.4	6
41	Is maternal duplication of 11p15 associated with Silver-Russell syndrome?. *Journal of Medical Genetics*, **2005**, 42, e26	5.8	68
40	Silver-Russell syndrome-like features in a patient carrying a novel NF1 mutation. *Pediatric Research*, **2005**, 58, 1265-8	3.2	1
39	Diagnostic Proceeding in Silver-Russell Syndrome **2005**, 9, 205		2
38	Ring Chromosome 18: Clinical, Cytogenetic and Molecular Genetic Studies on Four Patients. *International Journal of Human Genetics*, **2004**, 4, 197-200	1	1
37	Quantification of GRB10 in 7p12-p14 by fluorogenic 5P nuclease chemistry and application for genetic diagnosis in Silver-Russell syndrome. *Annales De G□□tique*, **2004**, 47, 99-102		4
36	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, 75-8		25
35	Searching for genomic variants in IGF2 and CDKN1C in Silver-Russell syndrome patients. *Molecular Genetics and Metabolism*, **2004**, 82, 246-50	3.7	22
34	No evidence for a role of SLC7A10 in 19q13 in the etiology of cystinuria. *Clinical Nephrology*, **2004**, 62, 71-3	2.1	5
33	Maternal uniparental disomy 16 and genetic counseling: new case and survey of published cases. *Genetic Counseling*, **2004**, 15, 183-90		19
32	Robertson-Translokationen und uniparentale Disomien: Indikationen zur pr□natalen Diagnostik?. *Reproduktionsmedizin*, **2003**, 19, 93-97		
31	Determination of SMN1 and SMN2 copy number using TaqMan technology. *Human Mutation*, **2003**, 22, 74-8	4.7	92
30	Significant contribution of genomic rearrangements in SLC3A1 and SLC7A9 to the etiology of cystinuria. *Kidney International*, **2003**, 64, 1564-72	9.9	28
29	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, 259-63		13
28	Uniparental disomy and Robertsonian translocations: risk estimation and prenatal testing. *Molecular Diagnosis and Therapy*, **2003**, 7, 113-7		1
27	Searching for genomic variants in the MESTIT1 transcript in Silver-Russell syndrome patients. *Journal of Medical Genetics*, **2003**, 40, e65	5.8	7
26	Genomic characterisation of C7orf10 in Silver-Russell syndrome patients. *Journal of Medical Genetics*, **2003**, 40, e44	5.8	4
25	Screening for mutations in the gene for phosphorylation kinase gamma1 in Silver-Russell syndrome patients. *Annales De G□□tique*, **2002**, 45, 219-21		

24	Maternale uniparentale Disomie 14 Ein weiteres Imprintingsyndrom. *Monatsschrift Fur Kinderheilkunde*, **2002**, 150, 856-865	0.2	2
23	Uniparental disomy: clinical indications for testing in growth retardation. *European Journal of Pediatrics*, **2002**, 161, 305-12	4.1	25
22	Supernumerary marker chromosomes derived from chromosome 15: analysis of 32 new cases. *Clinical Genetics*, **2002**, 62, 89-93	4	42
21	Cystinuria in children: distribution and frequencies of mutations in the SLC3A1 and SLC7A9 genes. *Kidney International*, **2002**, 62, 1136-42	9.9	50
20	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	1.6	12
19	Longitudinal Study in a Patient with Trisomy 8 Mosaicism: Cytogenetic and Molecular-Genetic Investigations over a Period of Eleven Years. *International Journal of Human Genetics*, **2002**, 2, 101-106	1	
18	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, 342-8	2.1	21
17	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, 371-3	4	6
16	Evidence from skewed X inactivation for trisomy mosaicism in Silver-Russell syndrome. *European Journal of Human Genetics*, **2001**, 9, 887-91	5.3	7
15	Gene dosage analysis in Silver-Russell syndrome: use of quantitative competitive PCR and dual-color FISH to estimate the frequency of duplications in 7p11.2-p13. *Genetic Testing and Molecular Biomarkers*, **2001**, 5, 261-6		11
14	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, 543-5	11	26
13	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, 86-9	5.8	60
12	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		13
11	Novel silent variant (c1722G>A) in the coding region of the insulin receptor substrate 1 (IRS1) gene. *Human Mutation*, **2000**, 16, 533	4.7	1
10	Familial robertsonian translocation 15;21 and rare paracentric inv(21): unexpected re-inversion in a child with translocation trisomy 21. *European Journal of Human Genetics*, **2000**, 8, 815-9	5.3	2
9	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 G□□tique*, **2000**, 43, 15-21		28
8	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, 1587-95	5.6	117
7	Search for uniparental disomy 14 in balanced Robertsonian translocation carriers. *Clinical Genetics*, **1999**, 56, 464-6	4	8

6	Mutations in the SLC3A1 gene in cystinuric patients: frequencies and identification of a novel mutation. *Genetic Testing and Molecular Biomarkers*, **1999**, 3, 227-31		18
5	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, 123-8	1.1	21
4	Paternally inherited deletion of CSH1 in a patient with Silver-Russell syndrome. *Journal of Medical Genetics*, **1998**, 35, 784-6	5.8	25
3	A case of de novo translocation 16;21: trisomy 16q phenotype and origin of the aberration. *Annales De Génétique*, **1998**, 41, 205-8		8
2	Molecular studies in 37 Silver-Russell syndrome patients: frequency and etiology of uniparental disomy. *Human Genetics*, **1997**, 100, 415-9	6.3	135
1	Report of two new cases of Pallister-Killian syndrome confirmed by FISH: tissue-specific mosaicism and loss of i(12p) by in vitro selection. *American Journal of Medical Genetics Part A*, **1997**, 72, 106-10		39