Miriam Elbracht

List of Publications by Year
in descending order

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Version: 2024-02-01

46 papers	1,204 citations	623734 14 h-index	395702 33 g-index
49 all docs	49 docs citations	49 times ranked	1751 citing authors

#	Article	IF	Citations
1	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
2	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
3	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
4	DEGS1-associated aberrant sphingolipid metabolism impairs nervous system function in humans. Journal of Clinical Investigation, 2019, 129, 1229-1239.	8.2	65
5	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
6	High mutation detection rates in cerebral cavernous malformation upon stringent inclusion criteria: oneâ€third of probands are minors. Molecular Genetics & Genomic Medicine, 2014, 2, 176-185.	1.2	53
7	Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. Human Reproduction Update, 2020, 26, 197-213.	10.8	51
8	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
9	Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. Neuropediatrics, 2017, 48, 111-114.	0.6	25
10	<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.	2.0	24
11	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
12	Structural and sequence variants in patients with Silver-Russell syndrome or similar features-Curation of a disease database. Human Mutation, 2018, 39, 345-364.	2.5	23
13	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
14	Molecular and clinical studies in 8 patients with Temple syndrome. Clinical Genetics, 2018, 93, 1179-1188.	2.0	16
15	Kaiso mediates human ICR1 methylation maintenance and H19 transcriptional fine regulation. Clinical Epigenetics, 2016, 8, 47.	4.1	15
16	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. Brain, 2020, 143, 2406-2420.	7.6	15
17	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
18	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.	2.7	15

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19	Novel familial distal imprinting centre 1 (11p15.5) deletion provides further insights in imprinting regulation. Clinical Epigenetics, 2019, 11, 30.	4.1	13
20	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
21	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	8.2	13
22	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
23	Paroxysmal tonic upgaze: A heterogeneous clinical condition responsive to carbonic anhydrase inhibition. European Journal of Paediatric Neurology, 2020, 25, 181-186.	1.6	12
24	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
25	Pure distal trisomy 2q: A rare chromosomal abnormality with recognizable phenotype. American Journal of Medical Genetics, Part A, 2009, 149A, 2547-2550.	1.2	11
26	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
27	HMGA2 Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2401-2407.	3.6	11
28	Pregnancy outcome in Charcotâ€“Marieâ€“Tooth disease: results of the CMTâ€NET cohort study in Germany. European Journal of Neurology, 2020, 27, 1390-1396.	3.3	11
29	Novel Pathogenic Variants in a Cassette Exon of CCM2 in Patients With Cerebral Cavernous Malformations. Frontiers in Neurology, 2019, 10, 1219.	2.4	10
30	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.	1.2	10
31	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
32	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
33	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
34	Germline variants in DNA repair genes, including BRCA1/2, may cause familial myeloproliferative neoplasms. Blood Advances, 2021, 5, 3373-3376.	5.2	7
35	Search for altered imprinting marks in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser patients. Molecular Genetics & Genomic Medicine, 2018, 6, 1225-1228.	1.2	5
36	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

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37	PDE10A mutation in two sisters with a hyperkinetic movement disorder - Response to levodopa. Parkinsonism and Related Disorders, 2019, 63, 240-242.	2.2	5
38	Unusual phenotypes in patients with a pathogenic germline variant in DICER1. Familial Cancer, 2021, , 1.	1.9	5
39	Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2020, 32, 321-334.	0.2	5
40	Biallelic CSGALNACT1-mutations cause a mild skeletal dysplasia. Bone, 2019, 127, 446-451.	2.9	4
41	Molecular characterization of temple syndrome families with 14q32 epimutations. European Journal of Medical Genetics, 2020, 63, 104077.	1.3	2
42	Aggressive infantile myofibromatosis with intestinal involvement. Molecular and Cellular Pediatrics, 2021, 8, 7.	1.8	2
43	Alveolar capillary dysplasia without misalignment of pulmonary veins, hyperinflammation, megalocornea and overgrowth â€“ Association with a homozygous 2bp-insertion in LTBP2?. European Journal of Medical Genetics, 2021, 64, 104209.	1.3	1
44	Successful allogeneic stem cell transplantation of a patient with Werner syndrome and acute myeloid leukemia. Leukemia Research, 2021, 108, 106609.	0.8	1
45	Charcot-Marie-Tooth neuropathy and pregnancy: general and specific issues. Medizinische Genetik, 2020, 32, 221-225.	0.2	0
46	Corrigendum to: Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2021, 33, 61-63.	0.2	0