Miriam Elbracht

List of Publications by Year
in descending order

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	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
2	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
3	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
4	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	8.2	13
5	Aggressive infantile myofibromatosis with intestinal involvement. Molecular and Cellular Pediatrics, 2021, 8, 7.	1.8	2
6	Unusual phenotypes in patients with a pathogenic germline variant in DICER1. Familial Cancer, 2021, , 1.	1.9	5
7	Successful allogeneic stem cell transplantation of a patient with Werner syndrome and acute myeloid leukemia. Leukemia Research, 2021, 108, 106609.	0.8	1
8	Germline variants in DNA repair genes, including BRCA1/2, may cause familial myeloproliferative neoplasms. Blood Advances, 2021, 5, 3373-3376.	5.2	7
9	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
10	Corrigendum to: Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2021, 33, 61-63.	0.2	0
11	Paroxysmal tonic upgaze: A heterogeneous clinical condition responsive to carbonic anhydrase inhibition. European Journal of Paediatric Neurology, 2020, 25, 181-186.	1.6	12
12	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
13	Molecular characterization of temple syndrome families with 14q32 epimutations. European Journal of Medical Genetics, 2020, 63, 104077.	1.3	2
14	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. Brain, 2020, 143, 2406-2420.	7.6	15
15	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
16	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
17	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
18	HMGA2 Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2401-2407.	3.6	11

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19	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
20	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
21	Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. Human Reproduction Update, 2020, 26, 197-213.	10.8	51
22	Charcot-Marie-Tooth neuropathy and pregnancy: general and specific issues. Medizinische Genetik, 2020, 32, 221-225.	0.2	0
23	Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2020, 32, 321-334.	0.2	5
24	Biallelic CSGALNACT1-mutations cause a mild skeletal dysplasia. Bone, 2019, 127, 446-451.	2.9	4
25	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
26	Novel familial distal imprinting centre 1 (11p15.5) deletion provides further insights in imprinting regulation. Clinical Epigenetics, 2019, 11, 30.	4.1	13
27	PDE10A mutation in two sisters with a hyperkinetic movement disorder - Response to levodopa. Parkinsonism and Related Disorders, 2019, 63, 240-242.	2.2	5
28	Novel Pathogenic Variants in a Cassette Exon of CCM2 in Patients With Cerebral Cavernous Malformations. Frontiers in Neurology, 2019, 10, 1219.	2.4	10
29	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
30	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
31	DEGS1-associated aberrant sphingolipid metabolism impairs nervous system function in humans. Journal of Clinical Investigation, 2019, 129, 1229-1239.	8.2	65
32	Molecular and clinical studies in 8 patients with Temple syndrome. Clinical Genetics, 2018, 93, 1179-1188.	2.0	16
33	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
34	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
35	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
36	Search for altered imprinting marks in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser patients. Molecular Genetics & Genomic Medicine, 2018, 6, 1225-1228.	1.2	5

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37	<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
38	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
39	Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. Neuropediatrics, 2017, 48, 111-114.	0.6	25
40	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
41	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
42	Kaiso mediates human ICR1 methylation maintenance and H19 transcriptional fine regulation. Clinical Epigenetics, 2016, 8, 47.	4.1	15
43	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
44	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
45	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
46	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