

Britt-Marie Anderlid

List of Publications by Year in descending order

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Version: 2024-02-01

48
papers

2,272
citations

279798

23
h-index

243625

44
g-index

52
all docs

52
docs citations

52
times ranked

4749
citing authors

#	ARTICLE	IF	CITATIONS
1	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , 2017, 49, 515-526.	21.4	443
2	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051.	14.8	152
3	Subtelomeric rearrangements detected in patients with idiopathic mental retardation. <i>American Journal of Medical Genetics Part A</i> , 2002, 107, 275-284.	2.4	125
4	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
5	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	12.8	105
6	Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome. <i>Molecular Autism</i> , 2018, 9, 1.	4.9	96
7	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. <i>Genome Medicine</i> , 2019, 11, 68.	8.2	88
8	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	6.2	87
9	FISH-mapping of a 100-kb terminal 22q13 deletion. <i>Human Genetics</i> , 2002, 110, 439-443.	3.8	78
10	Detailed characterization of 12 supernumerary ring chromosomes using micro-FISH and search for uniparental disomy. <i>American Journal of Medical Genetics Part A</i> , 2001, 99, 223-233.	2.4	68
11	The Roots of Autism and ADHD Twin Study in Sweden (RATSS). <i>Twin Research and Human Genetics</i> , 2014, 17, 164-176.	0.6	62
12	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. <i>Human Genetics</i> , 2001, 109, 551-558.	3.8	60
13	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. <i>Human Mutation</i> , 2017, 38, 180-192.	2.5	58
14	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	2.8	56
15	Identification of Critical Regions and Candidate Genes for Cardiovascular Malformations and Cardiomyopathy Associated with Deletions of Chromosome 1p36. <i>PLoS ONE</i> , 2014, 9, e85600.	2.5	51
16	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismature of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	6.2	48
17	Cryptic subtelomeric 6p deletion in a girl with congenital malformations and severe language impairment. <i>European Journal of Human Genetics</i> , 2003, 11, 89-92.	2.8	44
18	Single cell analysis of autism patient with bi-allelic NRXN1-alpha deletion reveals skewed fate choice in neural progenitors and impaired neuronal functionality. <i>Experimental Cell Research</i> , 2019, 383, 111469.	2.6	39

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19	<i>CTNND2</i> a candidate gene for reading problems and mild intellectual disability. <i>Journal of Medical Genetics</i> , 2015, 52, 111-122.	3.2	35
20	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. <i>European Journal of Human Genetics</i> , 2017, 25, 946-951.	2.8	33
21	Genomic screening in rare disorders: New mutations and phenotypes, highlighting <i>ALG14</i> as a novel cause of severe intellectual disability. <i>Clinical Genetics</i> , 2018, 94, 528-537.	2.0	29
22	Further evidence for specific <i>IFIH1</i> mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1396-1399.	1.2	28
23	Presynaptic dysfunction in CASK-related neurodevelopmental disorders. <i>Translational Psychiatry</i> , 2020, 10, 312.	4.8	28
24	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. <i>Twin Research and Human Genetics</i> , 2018, 21, 1-11.	0.6	27
25	Small mosaic deletion encompassing the snoRNAs and <i>SNURF</i> results in an atypical Prader-Willi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 425-431.	1.2	24
26	Minor physical anomalies in neurodevelopmental disorders: a twin study. <i>Child and Adolescent Psychiatry and Mental Health</i> , 2017, 11, 57.	2.5	24
27	Epilepsy syndromes, etiologies, and the use of next-generation sequencing in epilepsy presenting in the first 2 years of life: A population-based study. <i>Epilepsia</i> , 2020, 61, 2486-2499.	5.1	24
28	Maternal isodisomy of chromosome 9 with no impact on the phenotype in a woman with two isochromosomes: i(9p) and i(9q). <i>American Journal of Medical Genetics Part A</i> , 1999, 87, 49-52.	2.4	23
29	Toward clinical and molecular understanding of pathogenic variants in the <i>ZBTB18</i> gene. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 393-400.	1.2	22
30	Lissencephaly in an epilepsy cohort: Molecular, radiological and clinical aspects. <i>European Journal of Paediatric Neurology</i> , 2021, 30, 71-81.	1.6	22
31	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	2.3	19
32	Rare copy number variants are common in young children with autism spectrum disorder. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2015, 104, 610-618.	1.5	17
33	Benign paroxysmal torticollis of infancy does not lead to neurological sequelae. <i>Developmental Medicine and Child Neurology</i> , 2018, 60, 1251-1255.	2.1	17
34	HLA Polymorphism in Regressive and Non-Regressive Autism: A Preliminary Study. <i>Autism Research</i> , 2020, 13, 182-186.	3.8	17
35	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	2.4	13
36	Copy number variations in children with brain malformations and refractory epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 512-523.	1.2	12

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37	X-linked Malformation Deafness: Neurodevelopmental Symptoms Are Common in Children With IP3 Malformation and Mutation in POU3F4. <i>Ear and Hearing</i> , 2022, 43, 53-69.	2.1	12
38	NRXN1 Deletion and Exposure to Methylmercury Increase Astrocyte Differentiation by Different Notch-Dependent Transcriptional Mechanisms. <i>Frontiers in Genetics</i> , 2019, 10, 593.	2.3	11
39	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. <i>European Journal of Human Genetics</i> , 2016, 24, 51-58.	2.8	10
40	Clinical versus automated assessments of morphological variants in twins with and without neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1177-1189.	1.2	8
41	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. <i>Frontiers in Genetics</i> , 2019, 10, 896.	2.3	7
42	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	3.8	7
43	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 408-411.e8.	2.9	6
44	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. <i>PLoS ONE</i> , 2020, 15, e0228622.	2.5	6
45	Mowat-Wilson syndrome: Generation of two human iPS cell lines (UUIGPi004A and UUIGPi005A) from siblings with a truncating ZEB2 gene variant. <i>Stem Cell Research</i> , 2019, 39, 101518.	0.7	5
46	Variable neurodevelopmental and morphological phenotypes of carriers with 12q12 duplications. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1013.	1.2	3
47	Nasal speech in patients with 12q15 microdeletions. <i>European Journal of Human Genetics</i> , 2012, 20, 367-367.	2.8	0
48	Letter to the Editor regarding the manuscript "Lissencephaly: Update on diagnostics and clinical management" by Koenig et al. <i>Eur J Paediatr Neurol</i> . 2021; 35; 147-152. <i>European Journal of Paediatric Neurology</i> , 2022, 37, 165.	1.6	0