Britt-Marie Anderlid

List of Publications by Year
in descending order

48 papers	2,272 citations	279487 23 h-index	243296 44 g-index
52 all docs	52 docs citations	52 times ranked	4749 citing authors

#	Article	IF	Citations
1	X-linked Malformation Deafness: Neurodevelopmental Symptoms Are Common in Children With IP3 Malformation and Mutation in POU3F4. Ear and Hearing, 2022, 43, 53-69.	1.0	12
2	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	1.7	7
3	Letter to the Editor regarding the manuscript â€œLissencephaly: Update on diagnostics and clinical managementâ€•by Koenig etÂal. Eur J Paediatr Neurol. 2021; 35; 147-152. European Journal of Paediatric Neurology, 2022, 37, 165.	0.7	0
4	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
5	Lissencephaly in an epilepsy cohort: Molecular, radiological and clinical aspects. European Journal of Paediatric Neurology, 2021, 30, 71-81.	0.7	22
6	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
7	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	3.6	116
8	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	1.1	19
9	HLA Polymorphism in Regressive and Nonâ€Regressive Autism: A Preliminary Study. Autism Research, 2020, 13, 182-186.	2.1	17
10	Variable neurodevelopmental and morphological phenotypes of carriers with 12q12 duplications. Molecular Genetics & Genomic Medicine, 2020, 8, e1013.	0.6	3
11	Epilepsy syndromes, etiologies, and the use of nextâ€generation sequencing in epilepsy presenting in the first 2 years of life: A populationâ€based study. Epilepsia, 2020, 61, 2486-2499.	2.6	24
12	Presynaptic dysfunction in CASK-related neurodevelopmental disorders. Translational Psychiatry, 2020, 10, 312.	2.4	28
13	Clinical versus automated assessments of morphological variants in twins with and without neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2020, 182, 1177-1189.	0.7	8
14	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622.	1.1	6
15	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
16	Mowat-Wilson syndrome: Generation of two human iPS cell lines (UUIGPi004A and UUIGPi005A) from siblings with a truncating ZEB2 gene variant. Stem Cell Research, 2019, 39, 101518.	0.3	5
17	Single cell analysis of autism patient with bi-allelic NRXN1-alpha deletion reveals skewed fate choice in neural progenitors and impaired neuronal functionality. Experimental Cell Research, 2019, 383, 111469.	1.2	39
18	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	1.1	7

#	Article	IF	Citations
19	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. Genome Medicine, 2019, 11, 68.	3.6	88
20	NRXN1 Deletion and Exposure to Methylmercury Increase Astrocyte Differentiation by Different Notch-Dependent Transcriptional Mechanisms. Frontiers in Genetics, 2019, 10, 593.	1.1	11
21	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. Twin Research and Human Genetics, 2018, 21, 1-11.	0.3	27
22	Toward clinical and molecular understanding of pathogenic variants in the ZBTB18 gene. Molecular Genetics & Genomic Medicine, 2018, 6, 393-400.	0.6	22
23	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. Journal of Allergy and Clinical Immunology, 2018, 141, 408-411.e8.	1.5	6
24	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
25	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. Clinical Genetics, 2018, 94, 528-537.	1.0	29
26	Benign paroxysmal torticollis of infancy does not lead to neurological sequelae. Developmental Medicine and Child Neurology, 2018, 60, 1251-1255.	1.1	17
27	Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome. Molecular Autism, 2018, 9, 1.	2.6	96
28	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
29	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. European Journal of Human Genetics, 2017, 25, 946-951.	1.4	33
30	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
31	Further evidence for specific IFIH1 mutation as a cause of Singletonâ€“Merten syndrome with phenotypic heterogeneity. American Journal of Medical Genetics, Part A, 2017, 173, 1396-1399.	0.7	28
32	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	1.1	58
33	Minor physical anomalies in neurodevelopmental disorders: a twin study. Child and Adolescent Psychiatry and Mental Health, 2017, 11, 57.	1.2	24
34	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. European Journal of Human Genetics, 2016, 24, 51-58.	1.4	10
35	Rare copy number variants are common in young children with autism spectrum disorder. Acta Paediatrica, International Journal of Paediatrics, 2015, 104, 610-618.	0.7	17
36	Copy number variations in children with brain malformations and refractory epilepsy. American Journal of Medical Genetics, Part A, 2015, 167, 512-523.	0.7	12

#	Article	IF	Citations
37	CTNND2â€"a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	1.5	35
38	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	1.4	56
39	Identification of Critical Regions and Candidate Genes for Cardiovascular Malformations and Cardiomyopathy Associated with Deletions of Chromosome 1p36. PLoS ONE, 2014, 9, e85600.	1.1	51
40	The Roots of Autism and ADHD Twin Study in Sweden (RATSS). Twin Research and Human Genetics, 2014, 17, 164-176.	0.3	62
41	Small mosaic deletion encompassing the snoRNAs and SNURFâ€SNRPN results in an atypical Praderâ€"Willi syndrome phenotype. American Journal of Medical Genetics, Part A, 2014, 164, 425-431.	0.7	24
42	Nasal speech in patients with 12q15 microdeletions. European Journal of Human Genetics, 2012, 20, 367-367.	1.4	0
43	Cryptic subtelomeric 6p deletion in a girl with congenital malformations and severe language impairment. European Journal of Human Genetics, 2003, 11, 89-92.	1.4	44
44	Subtelomeric rearrangements detected in patients with idiopathic mental retardation. American Journal of Medical Genetics Part A, 2002, 107, 275-284.	2.4	125
45	FISH-mapping of a 100-kb terminal 22q13 deletion. Human Genetics, 2002, 110, 439-443.	1.8	78
46	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. Human Genetics, 2001, 109, 551-558.	1.8	60
47	Detailed characterization of 12 supernumerary ring chromosomes using microâ€FISH and search for uniparental disomy. American Journal of Medical Genetics Part A, 2001, 99, 223-233.	2.4	68
48	Maternal isodisomy of chromosome 9 with no impact on the phenotype in a woman with two isochromosomes: i(9p) and i(9q). , 1999, 87, 49-52.		23