Geert Vandeweyer

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5392990/publications.pdf

Version: 2024-02-01

45 papers	2,818 citations	279798 23 h-index	214800 47 g-index
50 all docs	50 docs citations	50 times ranked	5710 citing authors

#	Article	IF	Citations
1	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
2	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
3	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100.	1.3	157
4	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
5	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. Journal of Medical Genetics, 2010, 47, 299-311.	3.2	137
6	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	2.9	137
7	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
8	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
9	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	2.9	103
10	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	6.2	103
11	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	2.5	97
12	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. Genetics in Medicine, 2017, 19, 386-395.	2.4	94
13	Candidate Gene Resequencing in a Large Bicuspid Aortic Valve-Associated Thoracic Aortic Aneurysm Cohort: SMAD6 as an Important Contributor. Frontiers in Physiology, 2017, 8, 400.	2.8	85
14	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. Human Mutation, 2016, 37, 812-819.	2.5	76
15	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 315-326.	1.6	68
16	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	8.2	60
17	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
18	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. BMC Bioinformatics, 2011, 12, 4.	2.6	54

#	Article	IF	Citations
19	The Contribution of CLIP2 Haploinsufficiency to the Clinical Manifestations of the Williams-Beuren Syndrome. American Journal of Human Genetics, 2012, 90, 1071-1078.	6.2	41
20	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	3.5	41
21	The Compassionate Side of Neuroscience: Tony Sermoneâ€™s Undiagnosed Genetic Journeyâ€”ADNP Mutation. Journal of Molecular Neuroscience, 2015, 56, 751-757.	2.3	37
22	Osmotic stress inhibits leaf growth of Arabidopsis thaliana by enhancing ARFâ€mediated auxin responses. New Phytologist, 2020, 226, 1766-1780.	7.3	31
23	Balanced translocations in mental retardation. Human Genetics, 2009, 126, 133-147.	3.8	27
24	Novel microdeletions on chromosome 14q32.2 suggest a potential role for non-coding RNAs in Kagami-Ogata syndrome. European Journal of Human Genetics, 2016, 24, 1724-1729.	2.8	27
25	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	2.2	26
26	Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease. Scientific Reports, 2021, 11, 2515.	3.3	25
27	Copy number variation analysis in bicuspid aortic valve-related aortopathy identifies TBX20 as a contributing gene. European Journal of Human Genetics, 2019, 27, 1033-1043.	2.8	24
28	A boy with mental retardation, obesity and hypertrichosis caused by a microdeletion of 19p13.12. European Journal of Medical Genetics, 2010, 53, 291-293.	1.3	23
29	Identification of rare copy number variants in high burden schizophrenia families. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 273-282.	1.7	23
30	Mutations in ADNP affect expression and subcellular localization of the protein. Cell Cycle, 2018, 17, 1068-1075.	2.6	21
31	Haploinsufficiency of <scp>CMIP</scp> in a Girl With Autism Spectrum Disorder and Developmental Delay due to a De Novo Deletion on Chromosome 16q23.2. Autism Research, 2012, 5, 277-281.	3.8	19
32	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. Genetics in Medicine, 2019, 21, 1199-1208.	2.4	17
33	Detection and interpretation of genomic structural variation in health and disease. Expert Review of Molecular Diagnostics, 2013, 13, 61-82.	3.1	13
34	A Robust Protocol to Increase NimbleGen SeqCap EZ Multiplexing Capacity to 96 Samples. PLoS ONE, 2015, 10, e0123872.	2.5	13
35	Arrayâ€based MLPA to detect recurrent copy number variations in patients with idiopathic mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 343-348.	1.2	12
36	Insufficient evidence for a role of SERPINF1 in otosclerosis. Molecular Genetics and Genomics, 2019, 294, 1001-1006.	2.1	11

#	Article	IF	Citations
37	A de novo balanced t(2;6)(p15;p22.3) in a patient with West Syndrome disrupts a lnc-RNA. Epilepsy Research, 2012, 99, 346-349.	1.6	9
38	Whole genome sequencing of a dizygotic twin suggests a role for the serotonin receptor HTR7 in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1049-1056.	1.7	8
39	pyAmpli: an amplicon-based variant filter pipeline for targeted resequencing data. BMC Bioinformatics, 2017, 18, 554.	2.6	7
40	Multiplexed High Resolution Melting Assay for Versatile Sample Tracking in a Diagnostic and Research Setting. Journal of Molecular Diagnostics, 2016, 18, 32-38.	2.8	4
41	Overrepresentation of genetic variation in the AnkyrinG interactome is related to a range of neurodevelopmental disorders. European Journal of Human Genetics, 2020, 28, 1726-1733.	2.8	4
42	On the spot: very local chromosomal rearrangements. F1000 Biology Reports, 2012, 4, 22.	4.0	2
43	Protein interaction network analysis reveals genetic enrichment of immune system genes in frontotemporal dementia. Neurobiology of Aging, 2022, 116, 67-79.	3.1	2
44	A contemporary view on the molecular basis of neurodevelopmental disorders. , 2020, , 57-78.		0
45	The roles of patient groups in fostering cancer research. Nature Reviews Clinical Oncology, 2020, 17, 65-66.	27.6	0