Geert Vandeweyer

List of Publications by Year
in descending order

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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	Protein interaction network analysis reveals genetic enrichment of immune system genes in frontotemporal dementia. Neurobiology of Aging, 2022, 116, 67-79.	3.1	2
2	Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease. Scientific Reports, 2021, 11, 2515.	3.3	25
3	A contemporary view on the molecular basis of neurodevelopmental disorders. , 2020, , 57-78.		0
4	The roles of patient groups in fostering cancer research. Nature Reviews Clinical Oncology, 2020, 17, 65-66.	27.6	0
5	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
6	Osmotic stress inhibits leaf growth of Arabidopsis thaliana by enhancing ARFâ€mediated auxin responses. New Phytologist, 2020, 226, 1766-1780.	7.3	31
7	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
8	Insufficient evidence for a role of SERPINF1 in otosclerosis. Molecular Genetics and Genomics, 2019, 294, 1001-1006.	2.1	11
9	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
10	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
11	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
12	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
13	Mutations in ADNP affect expression and subcellular localization of the protein. Cell Cycle, 2018, 17, 1068-1075.	2.6	21
14	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
15	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
16	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	2.2	26
17	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
18	pyAmpli: an amplicon-based variant filter pipeline for targeted resequencing data. BMC Bioinformatics, 2017, 18, 554.	2.6	7

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19	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
20	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
21	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
22	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
23	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
24	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	2.5	97
25	A Robust Protocol to Increase NimbleGen SeqCap EZ Multiplexing Capacity to 96 Samples. PLoS ONE, 2015, 10, e0123872.	2.5	13
26	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
27	The Compassionate Side of Neuroscience: Tony Sermoneâ€™s Undiagnosed Genetic Journeyâ€”ADNP Mutation. Journal of Molecular Neuroscience, 2015, 56, 751-757.	2.3	37
28	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	8.2	60
29	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	3.5	41
30	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
31	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
32	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	2.9	137
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	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
35	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	2.9	103
36	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

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37	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
38	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
39	On the spot: very local chromosomal rearrangements. F1000 Biology Reports, 2012, 4, 22.	4.0	2
40	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. BMC Bioinformatics, 2011, 12, 4.	2.6	54
41	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
42	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
43	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
44	Balanced translocations in mental retardation. Human Genetics, 2009, 126, 133-147.	3.8	27
45	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