

Marjolein H Willemsen

List of Publications by Year in descending order

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Version: 2024-02-01

45
papers

5,077
citations

236925

25
h-index

243625

44
g-index

47
all docs

47
docs citations

47
times ranked

10291
citing authors

#	ARTICLE	IF	CITATIONS
1	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 880-893.	2.4	14
2	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	2.4	9
3	A Patient with Moderate Intellectual Disability and 49, XXXYY Karyotype. <i>International Journal of General Medicine</i> , 2022, Volume 15, 2799-2806.	1.8	3
4	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 1774-1780.	2.4	16
5	Human <i>KCNQ5</i> de novo mutations underlie epilepsy and intellectual disability. <i>Journal of Neurophysiology</i> , 2022, 128, 40-61.	1.8	8
6	Characterization of SETD1A haploinsufficiency in humans and <i>Drosophila</i> defines a novel neurodevelopmental syndrome. <i>Molecular Psychiatry</i> , 2021, 26, 2013-2024.	7.9	43
7	Further evidence for <i>de novo</i> variants in <i>SYNCRIP</i> as the cause of a neurodevelopmental disorder. <i>Human Mutation</i> , 2021, 42, 1094-1100.	2.5	9
8	Expanding the phenotype of <i>ASXL3</i> -related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <i>ASXL3</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3446-3458.	1.2	12
9	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. <i>Genetics in Medicine</i> , 2021, 23, 2122-2137.	2.4	16
10	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	12.8	43
11	A de novo <i>CTNNB1</i> Novel Splice Variant in an Adult Female with Severe Intellectual Disability. <i>International Medical Case Reports Journal</i> , 2020, Volume 13, 487-492.	0.8	8
12	Damaging de novo missense variants in <i>EEF1A2</i> lead to a developmental and degenerative epileptic dyskinetic encephalopathy. <i>Human Mutation</i> , 2020, 41, 1263-1279.	2.5	24
13	Epilepsy phenotype in individuals with chromosomal duplication encompassing <i>FGF12</i> . <i>Epilepsia Open</i> , 2020, 5, 301-306.	2.4	7
14	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	12.8	43
15	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. <i>Epilepsia</i> , 2019, 60, 155-164.	5.1	65
16	De novo BK channel variant causes epilepsy by affecting voltage gating but not Ca ²⁺ sensitivity. <i>European Journal of Human Genetics</i> , 2018, 26, 220-229.	2.8	47
17	<i>STAG1</i> mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 479-488.	3.2	35
18	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1821-1830.	1.2	31

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19	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	6.2	56
20	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. <i>PLoS Genetics</i> , 2017, 13, e1006864.	3.5	116
21	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies. <i>Genome Medicine</i> , 2017, 9, 118.	8.2	13
22	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	21.4	67
23	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. <i>Nature Neuroscience</i> , 2016, 19, 1194-1196.	14.8	407
24	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552.	6.2	132
25	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	2.8	108
26	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. <i>Genetical Research</i> , 2015, 97, e19.	0.9	0
27	De novo gain-of-function and loss-of-function mutations of <i>SCN8A</i> in patients with intellectual disabilities and epilepsy. <i>Journal of Medical Genetics</i> , 2015, 52, 330-337.	3.2	124
28	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2015, 96, 386-396.	6.2	27
29	Further delineation of the KBC syndrome phenotype caused by ANKRD11 aberrations. <i>European Journal of Human Genetics</i> , 2015, 23, 1176-1185.	2.8	67
30	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. <i>Genetics in Medicine</i> , 2015, 17, 460-466.	2.4	45
31	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. <i>Human Genetics</i> , 2015, 134, 97-109.	3.8	93
32	Dominant β -catenin mutations cause intellectual disability with recognizable syndromic features. <i>Journal of Clinical Investigation</i> , 2014, 124, 1468-1482.	8.2	110
33	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. <i>Nature Genetics</i> , 2014, 46, 380-384.	21.4	293
34	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. <i>Gene</i> , 2014, 538, 30-35.	2.2	11
35	Involvement of the kinesin family members <i>KIF4A</i> and <i>KIF5C</i> in intellectual disability and synaptic function. <i>Journal of Medical Genetics</i> , 2014, 51, 487-494.	3.2	90
36	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	27.8	996

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37	<i>GATAD2B</i> loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in <i>Drosophila</i> . <i>Journal of Medical Genetics</i> , 2013, 50, 507-514.	3.2	63
38	Mutations in <i>DYNC1H1</i> cause severe intellectual disability with neuronal migration defects. <i>Journal of Medical Genetics</i> , 2012, 49, 179-183.	3.2	151
39	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. <i>New England Journal of Medicine</i> , 2012, 367, 1921-1929.	27.0	1,367
40	Interpretation of clinical relevance of X-chromosome copy number variations identified in a large cohort of individuals with cognitive disorders and/or congenital anomalies. <i>European Journal of Medical Genetics</i> , 2012, 55, 586-598.	1.3	16
41	Further molecular and clinical delineation of the Wisconsin syndrome phenotype associated with interstitial 3q24q25 deletions. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 106-112.	1.2	6
42	Chromosome 1p21.3 microdeletions comprising <i>DPYD</i> and <i>MIR137</i> are associated with intellectual disability. <i>Journal of Medical Genetics</i> , 2011, 48, 810-818.	3.2	146
43	Identification of <i>ANKRD11</i> and <i>ZNF778</i> as candidate genes for autism and variable cognitive impairment in the novel 16q24.3 microdeletion syndrome. <i>European Journal of Human Genetics</i> , 2010, 18, 429-435.	2.8	99
44	Clinical and molecular characterization of two patients with a 6.75Mb overlapping deletion in 8p12p21 with two candidate loci for congenital heart defects. <i>European Journal of Medical Genetics</i> , 2009, 52, 134-139.	1.3	11
45	Females with <i>PDHA1</i> gene mutations: A diagnostic challenge. <i>Mitochondrion</i> , 2006, 6, 155-159.	3.4	27