Marjolein H Willemsen

List of Publications by Year
in descending order

45	5,077	236925	243625
papers	citations	25 h-index	44 g-index

47	47	47	10291
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	27.0	1,367
2	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
3	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
4	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
5	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	3.2	151
6	Chromosome 1p21.3 microdeletions comprising DPYD and MIR137 are associated with intellectual disability. Journal of Medical Genetics, 2011, 48, 810-818.	3.2	146
7	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
8	De novo gain-of-function and loss-of-function mutations ofSCN8Ain patients with intellectual disabilities and epilepsy. Journal of Medical Genetics, 2015, 52, 330-337.	3.2	124
9	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. PLoS Genetics, 2017, 13, e1006864.	3.5	116
10	Dominant Î²-catenin mutations cause intellectual disability with recognizable syndromic features. Journal of Clinical Investigation, 2014, 124, 1468-1482.	8.2	110
11	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
12	Identification of ANKRD11 and ZNF778 as candidate genes for autism and variable cognitive impairment in the novel 16q24.3 microdeletion syndrome. European Journal of Human Genetics, 2010, 18, 429-435.	2.8	99
13	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. Human Genetics, 2015, 134, 97-109.	3.8	93
14	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	3.2	90
15	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. European Journal of Human Genetics, 2015, 23, 1176-1185.	2.8	67
16	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	21.4	67
17	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. Epilepsia, 2019, 60, 155-164.	5.1	65
18	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	3.2	63

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19	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56
20	De novo BK channel variant causes epilepsy by affecting voltage gating but not Ca2+ sensitivity. European Journal of Human Genetics, 2018, 26, 220-229.	2.8	47
21	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
22	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
23	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
24	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
25	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	3.2	35
26	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. American Journal of Medical Genetics, Part A, 2017, 173, 1821-1830.	1.2	31
27	Females with PDHA1 gene mutations: A diagnostic challenge. Mitochondrion, 2006, 6, 155-159.	3.4	27
28	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	6.2	27
29	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
30	Interpretation of clinical relevance of X-chromosome copy number variations identified in a large cohort of individuals with cognitive disorders and/or congenital anomalies. European Journal of Medical Genetics, 2012, 55, 586-598.	1.3	16
31	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
32	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
33	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	2.4	14
34	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118.	8.2	13
35	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
36	Clinical and molecular characterization of two patients with a 6.75Mb overlapping deletion in 8p12p21 with two candidate loci for congenital heart defects. European Journal of Medical Genetics, 2009, 52, 134-139.	1.3	11

#	Article	IF	Citations
37	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. Gene, 2014, 538, 30-35.	2.2	11
38	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	2.5	9
39	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
40	A de novo CTNNB1 Novel Splice Variant in an Adult Female with Severe Intellectual Disability . International Medical Case Reports Journal, 2020, Volume 13, 487-492.	0.8	8
41	Human KCNQ5 de novo mutations underlie epilepsy and intellectual disability. Journal of Neurophysiology, 2022, 128, 40-61.	1.8	8
42	Epilepsy phenotype in individuals with chromosomal duplication encompassing FGF12. Epilepsia Open, 2020, 5, 301-306.	2.4	7
43	Further molecular and clinical delineation of the Wisconsin syndrome phenotype associated with interstitial 3q24q25 deletions. American Journal of Medical Genetics, Part A, 2011, 155, 106-112.	1.2	6
44	A Patient with Moderate Intellectual Disability and 49, XXXYY Karyotype. International Journal of General Medicine, 2022, Volume 15, 2799-2806.	1.8	3
45	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. Genetical Research, 2015, 97, e19.	0.9	0