Rob Willemsen

List of Publications by Year
in descending order

51 papers	3,017 citations	257450 24 h-index	189892 50 g-index
54 all docs	54 docs citations	54 times ranked	4121 citing authors

#	Article	IF	Citations
1	Sam68 sequestration and partial loss of function are associated with splicing alterations in FXTAS patients. EMBO Journal, 2010, 29, 1248-1261.	7.8	326
2	The FMR1 CGG repeat mouse displays ubiquitin-positive intranuclear neuronal inclusions; implications for the cerebellar tremor/ataxia syndrome. Human Molecular Genetics, 2003, 12, 949-959.	2.9	253
3	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
4	Sequestration of DROSHA and DGCR8 by Expanded CGG RNA Repeats Alters MicroRNA Processing in Fragile X-Associated Tremor/Ataxia Syndrome. Cell Reports, 2013, 3, 869-880.	6.4	216
5	Translation of Expanded CGG Repeats into FMRpolyG Is Pathogenic and May Contribute to Fragile X Tremor Ataxia Syndrome. Neuron, 2017, 93, 331-347.	8.1	194
6	Proteomic Profiling of Exosomes Leads to the Identification of Novel Biomarkers for Prostate Cancer. PLoS ONE, 2013, 8, e82589.	2.5	179
7	Clinical features of boys with fragile X premutations and intermediate alleles. American Journal of Medical Genetics Part A, 2003, 121B, 119-127.	2.4	143
8	New insights into the genetics of primary open-angle glaucoma based on meta-analyses of intraocular pressure and optic disc characteristics.. Human Molecular Genetics, 2017, 26, ddw399.	2.9	120
9	Cognitive decline, neuromotor and behavioural disturbances in a mouse model for fragile-X-associated tremor/ataxia syndrome (FXTAS). Behavioural Brain Research, 2005, 162, 233-239.	2.2	117
10	FMR1: A gene with three faces. Biochimica Et Biophysica Acta - General Subjects, 2009, 1790, 467-477.	2.4	113
11	CGGâ€repeat length and neuropathological and molecular correlates in a mouse model for fragile Xâ€associated tremor/ataxia syndrome. Journal of Neurochemistry, 2008, 107, 1671-1682.	3.9	100
12	The fragile X syndrome: From molecular genetics to neurobiology. Mental Retardation and Developmental Disabilities Research Reviews, 2004, 10, 60-67.	3.6	90
13	The GABA_A receptor is an FMRP target with therapeutic potential in fragile X syndrome. Cell Cycle, 2015, 14, 2985-2995.	2.6	87
14	DAZL Limits Pluripotency, Differentiation, and Apoptosis in Developing Primordial Germ Cells. Stem Cell Reports, 2014, 3, 892-904.	4.8	83
15	Impaired GABAergic inhibition in the hippocampus of Fmr1 knockout mice. Neuropharmacology, 2017, 116, 71-81.	4.1	58
16	Mouse models of the fragile X premutation and fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 25.	3.1	57
17	Use of model systems to understand the etiology of fragile X-associated primary ovarian insufficiency (FXPOI). Journal of Neurodevelopmental Disorders, 2014, 6, 26.	3.1	55
18	Epigenetic Characterization of the FMR1 Promoter in Induced Pluripotent Stem Cells from Human Fibroblasts Carrying an Unmethylated Full Mutation. Stem Cell Reports, 2014, 3, 548-555.	4.8	54

#	Article	IF	Citations
19	Comparing Approaches to Normalize, Quantify, and Characterize Urinary Extracellular Vesicles. Journal of the American Society of Nephrology: JASN, 2021, 32, 1210-1226.	6.1	53
20	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. Human Molecular Genetics, 2018, 27, 2039-2051.	2.9	51
21	A novel fragile X syndrome mutation reveals a conserved role for the carboxyâ€terminus in <scp>FMRP</scp> localization and function. EMBO Molecular Medicine, 2015, 7, 423-437.	6.9	41
22	Binding of NUFIP2 to Roquin promotes recognition and regulation of ICOS mRNA. Nature Communications, 2018, 9, 299.	12.8	27
23	FXTAS: A progressive neurologic syndrome associated with fragile X premutation. Current Neurology and Neuroscience Reports, 2005, 5, 405-410.	4.2	26
24	In vivo synaptic transmission and morphology in mouse models of Tuberous sclerosis, Fragile X syndrome, Neurofibromatosis type 1, and Costello syndrome. Frontiers in Cellular Neuroscience, 2015, 9, 234.	3.7	24
25	Potential pathogenic mechanisms underlying Fragile X Tremor Ataxia Syndrome: RAN translation and/or RNA gain-of-function?. European Journal of Medical Genetics, 2018, 61, 674-679.	1.3	24
26	Blood expression profiles of fragile X premutation carriers identify candidate genes involved in neurodegenerative and infertility phenotypes. Neurobiology of Disease, 2014, 65, 43-54.	4.4	23
27	Synaptic vesicle dynamic changes in a model of fragile X. Molecular Autism, 2016, 7, 17.	4.9	21
28	Heritabilities, proportions of heritabilities explained by GWAS findings, and implications of cross-phenotype effects on PR interval. Human Genetics, 2015, 134, 1211-1219.	3.8	20
29	Loss of Gap Junction Delta-2 (GJD2) gene orthologs leads to refractive error in zebrafish. Communications Biology, 2021, 4, 676.	4.4	19
30	Identification of antiparkinsonian drugs in the 6-hydroxydopamine zebrafish model. Pharmacology Biochemistry and Behavior, 2020, 189, 172828.	2.9	16
31	Selective rescue of heightened anxiety but not gait ataxia in a premutation 90CGG mouse model of Fragile X-associated tremor/ataxia syndrome. Human Molecular Genetics, 2017, 26, 2133-2145.	2.9	15
32	Combination Therapy in Fragile X Syndrome; Possibilities and Pitfalls Illustrated by Targeting the mGluR5 and GABA Pathway Simultaneously. Frontiers in Molecular Neuroscience, 2017, 10, 368.	2.9	15
33	Translational endpoints in fragile X syndrome. Neuroscience and Biobehavioral Reviews, 2014, 46, 256-269.	6.1	14
34	Astroglial-targeted expression of the fragile X CGG repeat premutation in mice yields RAN translation, motor deficits and possible evidence for cell-to-cell propagation of FXTAS pathology. Acta Neuropathologica Communications, 2019, 7, 27.	5.2	14
35	The quest for targeted therapy in fragile X syndrome. Expert Opinion on Therapeutic Targets, 2015, 19, 1277-1281.	3.4	12
36	CXorf56, a dendritic neuronal protein, identified as a new candidate gene for X-linked intellectual disability. European Journal of Human Genetics, 2018, 26, 552-560.	2.8	12

#	Article	IF	Citations
37	Paradoxical effect of baclofen on social behavior in the fragile X syndrome mouse model. Brain and Behavior, 2018, 8, e00991.	2.2	11
38	Reduction of Fmr1 mRNA Levels Rescues Pathological Features in Cortical Neurons in a Model of FXTAS. Molecular Therapy - Nucleic Acids, 2019, 18, 546-553.	5.1	11
39	Lack of a Clear Behavioral Phenotype in an Inducible FXTAS Mouse Model Despite the Presence of Neuronal FMRpolyG-Positive Aggregates. Frontiers in Molecular Biosciences, 2020, 7, 599101.	3.5	10
40	Small molecule 1a reduces FMRpolyG-mediated toxicity in in vitro and in vivo models for FMR1 premutation. Human Molecular Genetics, 2021, 30, 1632-1648.	2.9	9
41	Reduced activity-dependent protein levels in a mouse model of the fragile X premutation. Neurobiology of Learning and Memory, 2014, 109, 160-168.	1.9	7
42	Neuropathology of FMR1-premutation carriers presenting with dementia and neuropsychiatric symptoms. Brain Communications, 2021, 3, fcab007.	3.3	7
43	A missense variant in the nuclear export signal of the FMR1 gene causes intellectual disability. Gene, 2021, 768, 145298.	2.2	6
44	Zebrafish: An In Vivo Screening Model to Study Ocular Phenotypes. Translational Vision Science and Technology, 2022, 11, 17.	2.2	6
45	A combined linkage, microarray and exome analysis suggests MAP3K11 as a candidate gene for left ventricular hypertrophy. BMC Medical Genomics, 2018, 11, 22.	1.5	4
46	IDH1-mutated transgenic zebrafish lines: An in-vivo model for drug screening and functional analysis. PLoS ONE, 2018, 13, e0199737.	2.5	4
47	BC RNA Mislocalization in the Fragile X Premutation. ENeuro, 2018, 5, ENEURO.0091-18.2018.	1.9	4
48	Allopregnanolone Improves Locomotor Activity and Arousal in the Aged CGG Knock-in Mouse Model of Fragile X-Associated Tremor/Ataxia Syndrome. Frontiers in Neuroscience, 2021, 15, 752973.	2.8	1
49	Reply: PRKAR1B mutations are a rare cause of FUS negative neuronal intermediate filament inclusion disease. Brain, 2015, 138, e358-e358.	7.6	0
50	Neuropathology of FMR1â€premutation carriers presenting with dementia and neuropsychiatric symptoms. Alzheimer's and Dementia, 2020, 16, e044916.	0.8	0
51	Editorial: Proceedings of the â€œFourth International Conference of the FMR1 Premutation: Basic Mechanisms, Clinical Involvement and Therapyâ€. Frontiers in Molecular Biosciences, 2021, 8, 671875.	3.5	0