Marije E C Meuwissen

List of Publications by Year
in descending order

20	1,020	687363 13	794594 19
papers	citations	h-index	g-index

20	20	20	2442
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
2	The expanding phenotype of COL4A1 and COL4A2 mutations: clinical data on 13 newly identified families and a review of the literature. Genetics in Medicine, 2015, 17, 843-853.	2.4	204
3	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
4	Neurological findings in incontinentia pigmenti; a review. European Journal of Medical Genetics, 2012, 55, 323-331.	1.3	92
5	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
6	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
7	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	6.2	48
8	Genetic variants in the KDM6B gene are associated with neurodevelopmental delays and dysmorphic features. American Journal of Medical Genetics, Part A, 2019, 179, 1276-1286.	1.2	38
9	ACTA2 mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. American Journal of Medical Genetics, Part A, 2013, 161, 1376-1380.	1.2	36
10	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
11	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
12	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128.	2.3	24
13	Loss of USP18 in microglia induces white matter pathology. Acta Neuropathologica Communications, 2019, 7, 106.	5.2	15
14	Novel BRPF1 mutation in a boy with intellectual disability, coloboma, facial nerve palsy and hypoplasia of the corpus callosum. European Journal of Medical Genetics, 2019, 62, 103691.	1.3	15
15	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
16	Mutations in RNU7-1 Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-GoutiÃ¨res Syndrome with Severe End-Organ Involvement. Journal of Clinical Immunology, 2022, 42, 962-974.	3.8	8
17	Sleepâ€disordered breathing and nocturnal hypoventilation in children with the MECP2 duplication syndrome: A case series and review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 2437-2441.	1.2	6
18	Intracerebral hemorrhage in a neonate with an intragenic COL4A2 duplication. American Journal of Medical Genetics, Part A, 2021, 185, 571-574.	1.2	4

#	Article	IF	Citations
19	PUM1 haploinsufficiency is associated with syndromic neurodevelopmental delay and epilepsy. American Journal of Medical Genetics, Part A, 2020, 182, 591-594.	1.2	1
20	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1