Paulien A Terhal

List of Publications by Year
in descending order

22	1,056	567281 15	677142 22
papers	citations	h-index	g-index

22	22	22	2818
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
2	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
3	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81
4	A study of the clinical and radiological features in a cohort of 93 patients with a COL2A1 mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 461-475.	1.2	73
5	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
6	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	1.2	52
7	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	5.0	47
8	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	2.8	45
9	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
10	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	6.2	39
11	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
12	Non-invasive sources of cells with primary cilia from pediatric and adult patients. Cilia, 2015, 4, 8.	1.8	34
13	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
14	Mutationâ€based growth charts for SEDC and other COL2A1 related dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 205-216.	1.6	26
15	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
16	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	6.2	23
17	Biallelic variants in POLR3GL cause endosteal hyperostosis and oligodontia. European Journal of Human Genetics, 2020, 28, 31-39.	2.8	21
18	Two novel cases expanding the phenotype of SETD2â€related overgrowth syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1212-1215.	1.2	20

#	Article	IF	Citations
19	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
20	Variants in CAPZA2, a member of an F-actin capping complex, cause intellectual disability and developmental delay. Human Molecular Genetics, 2020, 29, 1537-1546.	2.9	15
21	Compound heterozygous NEK1 variants in two siblings with oral-facial-digital syndrome type II (Mohr) Tj ETQq1 1 0.784314 rgBT /Overlo	2.8	13
22	Further delineation of the GDF6 related multiple synostoses syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 225-229.	1.2	9