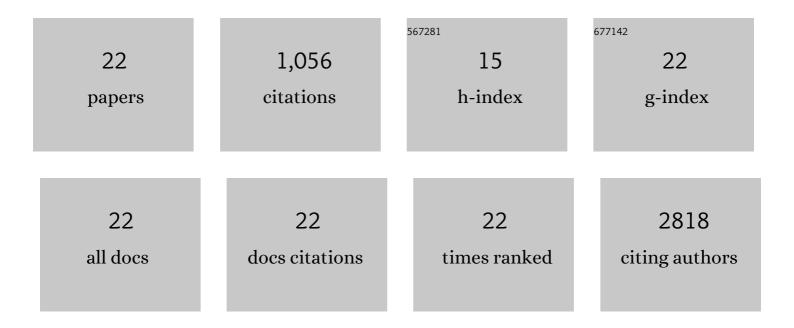
Paulien A Terhal

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
2	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
3	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
4	Biallelic variants in POLR3GL cause endosteal hyperostosis and oligodontia. European Journal of Human Genetics, 2020, 28, 31-39.	2.8	21
5	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
6	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
7	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	2.8	45
8	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
9	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
10	De Novo Mutations Affecting the Catalytic Cα 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	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	5.0	47
12	Further delineation of an entity caused by <i>CREBBP</i> and <i>EP300</i> mutations but not resembling Rubinstein–Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	1.2	52
13	Two novel cases expanding the phenotype of <i>SETD2</i> â€related overgrowth syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1212-1215.	1.2	20
14	Further delineation of the GDF6 related multiple synostoses syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 225-229.	1.2	9
15	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81
16	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
17	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

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Compound heterozygous NEK1 variants in two siblings with oral-facial-digital syndrome type II (Mohr) Tj ETQq0 0 0.28 T /Overlock 10 Tf

PAULIEN A TERHAL

#	Article	IF	CITATIONS
19	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
20	Non-invasive sources of cells with primary cilia from pediatric and adult patients. Cilia, 2015, 4, 8.	1.8	34
21	A study of the clinical and radiological features in a cohort of 93 patients with a <i>COL2A1</i> mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 461-475.	1.2	73
22	Mutationâ€based growth charts for SEDC and other <i>COL2A1</i> related dysplasias. American Journal of Medical Genetics, 2012, 160C, 205-216.	1.6	26