

# Perrine Pennamen

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2568880/publications.pdf>

Version: 2024-02-01

9  
papers

529  
citations

1478505

6  
h-index

1474206

9  
g-index

9  
all docs

9  
docs citations

9  
times ranked

790  
citing authors

#	ARTICLE	IF	CITATIONS
1	CRISPR-Cas9 genome editing induces megabase-scale chromosomal truncations. <i>Nature Communications</i> , 2019, 10, 1136.	12.8	292
2	Molecular characterization of a series of 990 index patients with albinism. <i>Pigment Cell and Melanoma Research</i> , 2018, 31, 466-474.	3.3	92
3	BLOC1S5 pathogenic variants cause a new type of Hermanskyâ€Pudlak syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1613-1622.	2.4	44
4	CRISPR-Cas9 globin editing can induce megabase-scale copy-neutral losses of heterozygosity in hematopoietic cells. <i>Nature Communications</i> , 2021, 12, 4922.	12.8	44
5	Dopachrome tautomerase variants in patients with oculocutaneous albinism. <i>Genetics in Medicine</i> , 2021, 23, 479-487.	2.4	33
6	Balanced complex chromosome rearrangement in male infertility: case report and literature review. <i>Andrologia</i> , 2015, 47, 178-185.	2.1	14
7	Novel variants in the <i>BLOC1S3</i> gene in patients presenting a mild form of Hermanskyâ€Pudlak syndrome. <i>Pigment Cell and Melanoma Research</i> , 2021, 34, 132-135.	3.3	5
8	Evidence of mosaicism in SPAST variant carriers in four French families. <i>European Journal of Human Genetics</i> , 2021, 29, 1158-1163.	2.8	3
9	Clinical and molecular findings of FRMD7 related congenital nystagmus as a differential diagnosis of ocular albinism. <i>Ophthalmic Genetics</i> , 2019, 40, 161-164.	1.2	2