Perrine Pennamen

List of Publications by Year
in descending order

9	529	1478505 6	1474206 9
papers	citations	h-index	g-index

9	9	9	790
all docs	docs citations	times ranked	citing authors

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