Mattias Van Heetvelde

List of Publications by Year
in descending order

13	895	1040056 9	1125743 13
papers	citations	h-index	g-index

16	16	16	2989
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Long-Read Sequencing to Unravel Complex Structural Variants of CEP78 Leading to Cone-Rod Dystrophy and Hearing Loss. Frontiers in Cell and Developmental Biology, 2021, 9, 664317.	3.7	11
2	Mild Leber hereditary optic neuropathy (LHON) in a Western European family due to the rare Asian m.14502T>C variant in the MT-ND6 gene. Ophthalmic Genetics, 2021, 42, 440-445.	1.2	2
3	Expanding the clinical spectrum and management of Traboulsi syndrome: report on two siblings homozygous for a novel pathogenic variant in ASPH. Ophthalmic Genetics, 2021, 42, 493-499.	1.2	5
4	Prevalence of Germline Pathogenic Variants in Cancer Predisposing Genes in Czech and Belgian Pancreatic Cancer Patients. Cancers, 2021, 13, 4430.	3.7	8
5	ISOLATED MACULOPATHY AND MODERATE RODâ€“CONE DYSTROPHY REPRESENT THE MILDER END OF THE RDH12-RELATED RETINAL DYSTROPHY SPECTRUM. Retina, 2021, 41, 1346-1355.	1.7	4
6	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
7	Accurate detection and quantification of epigenetic and genetic second hits in BRCA1 and BRCA2 -associated hereditary breast and ovarian cancer reveals multiple co-acting second hits. Cancer Letters, 2018, 425, 125-133.	7.2	12
8	Mapping the genomic landscape of inherited retinal disease genes prioritizes genes prone to coding and noncoding copy-number variations. Genetics in Medicine, 2018, 20, 202-213.	2.4	47
9	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
10	Evaluation of relative quantification of alternatively spliced transcripts using droplet digital PCR. Biomolecular Detection and Quantification, 2017, 13, 40-48.	7.0	12
11	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	1.6	152
12	Flexible, Scalable, and Efficient Targeted Resequencing on a Benchtop Sequencer for Variant Detection in Clinical Practice. Human Mutation, 2015, 36, 379-387.	2.5	43
13	Decreased DNA doubleâ€strand break repair and enhanced chromosomal radiosensitivity in irradiated nonâ€tumorigenic human breast epithelial cells with a partial BRCA1 or BRCA2 knockdown. World Academy of Sciences Journal, 0, , .	0.6	0