Richard H Van Jaarsveld

List of Publications by Year
in descending order

20 papers	1,580 citations	759233 12 h-index	713466 21 g-index
24 all docs	24 docs citations	24 times ranked	3078 citing authors

#	Article	IF	Citations
1	Sequential cancer mutations in cultured human intestinal stem cells. Nature, 2015, 521, 43-47.	27.8	853
2	Oral Mucosal Organoids as a Potential Platform for Personalized Cancer Therapy. Cancer Discovery, 2019, 9, 852-871.	9.4	222
3	Ongoing chromosomal instability and karyotype evolution in human colorectal cancer organoids. Nature Genetics, 2019, 51, 824-834.	21.4	162
4	Difference Makers: Chromosomal Instability versus Aneuploidy in Cancer. Trends in Cancer, 2016, 2, 561-571.	7.4	60
5	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
6	Nuclear chromosome locations dictate segregation error frequencies. Nature, 2022, 607, 604-609.	27.8	39
7	Genetics-first approach improves diagnostics of ESKD patients <50 years old. Nephrology Dialysis Transplantation, 2022, 37, 349-357.	0.7	27
8	<scp>HNRNPH1</scp>â€related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clinical Genetics, 2020, 98, 91-98.	2.0	25
9	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
10	Live imaging of cell division in 3D stem-cell organoid cultures. Methods in Cell Biology, 2018, 145, 91-106.	1.1	17
11	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
12	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
13	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
14	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	2.4	14
15	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. Genetics in Medicine, 2021, 23, 881-887.	2.4	13
16	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	3.8	11
17	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
18	Expanding the molecular spectrum and the neurological phenotype related to <scp>CAMTA1</scp> variants. Clinical Genetics, 2021, 99, 259-268.	2.0	6

#	Article	IF	Citations
19	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
20	Deep intronic TIMMDC1 variant delays diagnosis of rapidly progressive complex I deficiency. European Journal of Medical Genetics, 2021, 64, 104120.	1.3	2