

Richard H Van Jaarsveld

List of Publications by Year in descending order

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Version: 2024-02-01

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	Genetics-first approach improves diagnostics of ESKD patients <50 years old. Nephrology Dialysis Transplantation, 2022, 37, 349-357.	0.7	27
2	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
3	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
4	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
5	Nuclear chromosome locations dictate segregation error frequencies. Nature, 2022, 607, 604-609.	27.8	39
6	Deep intronic TIMMDC1 variant delays diagnosis of rapidly progressive complex I deficiency. European Journal of Medical Genetics, 2021, 64, 104120.	1.3	2
7	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
8	Expanding the molecular spectrum and the neurological phenotype related to <sc><i>CAMTA1</i></sc> variants. Clinical Genetics, 2021, 99, 259-268.	2.0	6
9	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
10	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
11	Pathogenic <sc><i>MAST3</i></sc> Variants in the <sc>STK</sc> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
12	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
13	Deficiency of TET3 leads to a genome-wide DNA hypermethylation epesignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	3.8	11
14	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
15	<sc><i>HNRNPH1</i></sc>-related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clinical Genetics, 2020, 98, 91-98.	2.0	25
16	Oral Mucosal Organoids as a Potential Platform for Personalized Cancer Therapy. Cancer Discovery, 2019, 9, 852-871.	9.4	222
17	Ongoing chromosomal instability and karyotype evolution in human colorectal cancer organoids. Nature Genetics, 2019, 51, 824-834.	21.4	162
18	Live imaging of cell division in 3D stem-cell organoid cultures. Methods in Cell Biology, 2018, 145, 91-106.	1.1	17

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19	Difference Makers: Chromosomal Instability versus Aneuploidy in Cancer. Trends in Cancer, 2016, 2, 561-571.	7.4	60
20	Sequential cancer mutations in cultured human intestinal stem cells. Nature, 2015, 521, 43-47.	27.8	853