Paul C Marcogliese

List of Publications by Year in descending order

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471061 580395 1,051 25 17 25 citations h-index g-index papers 31 31 31 2046 docs citations citing authors all docs times ranked

#	Article	IF	Citations
1	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. Science Advances, 2022, 8, eabl5613.	4.7	12
2	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	1.4	5
3	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
4	De novo variants in <i>EMC1</i> lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in <i>Drosophila</i> Human Molecular Genetics, 2022, 31, 3231-3244.	1.4	5
5	Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. American Journal of Human Genetics, 2022, 109, 571-586.	2.6	19
6	Missense variants in <i>CTNNB1</i> can be associated with vitreoretinopathyâ€"Seven new cases of <i>CTNNB1</i> â€associated neurodevelopmental disorder including a previously unreported retinal phenotype. Molecular Genetics & Denomic Medicine, 2021, 9, e1542.	0.6	15
7	TM2D genes regulate Notch signaling and neuronal function in Drosophila. PLoS Genetics, 2021, 17, e1009962.	1.5	5
8	Neuronal ROS-induced glial lipid droplet formation is altered by loss of Alzheimer's disease–associated genes. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	59
9	<i>Drosophila</i> Voltage-Gated Sodium Channels Are Only Expressed in Active Neurons and Are Localized to Distal Axonal Initial Segment-like Domains. Journal of Neuroscience, 2020, 40, 7999-8024.	1.7	50
10	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. Human Molecular Genetics, 2020, 29, 1568-1579.	1.4	29
11	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	2.6	23
12	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	3.8	71
13	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
14	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American Journal of Human Genetics, 2019, 105, 907-920.	2.6	22
15	<i>Lrrk2</i> alleles modulate inflammation during microbial infection of mice in a sex-dependent manner. Science Translational Medicine, 2019, 11, .	5.8	67
16	DJ-1 modulates the unfolded protein response and cell death via upregulation of ATF4 following ER stress. Cell Death and Disease, 2019, 10, 135.	2.7	29
17	Sphingolipids in the Pathogenesis of Parkinson's Disease and Parkinsonism. Trends in Endocrinology and Metabolism, 2019, 30, 106-117.	3.1	82
18	Regulation of myeloid cell phagocytosis by LRRK2 via WAVE2 complex stabilization is altered in Parkinson's disease. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E5164-E5173.	3.3	83

#	Article	IF	CITATION
19	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
20	LRRK2(I2020T) functional genetic interactors that modify eye degeneration and dopaminergic cell loss in Drosophila. Human Molecular Genetics, 2017, 26, 1247-1257.	1.4	17
21	BAG2 Gene-mediated Regulation of PINK1 Protein Is Critical for Mitochondrial Translocation of PARKIN and Neuronal Survival. Journal of Biological Chemistry, 2015, 290, 30441-30452.	1.6	52
22	Unaltered Striatal Dopamine Release Levels in Young Parkin Knockout, Pink1 Knockout, DJ-1 Knockout and LRRK2 R1441G Transgenic Mice. PLoS ONE, 2014, 9, e94826.	1.1	26
23	Progressive dopaminergic cell loss with unilateral-to-bilateral progression in a genetic model of Parkinson disease. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 15918-15923.	3.3	72
24	DJ-1 protects the nigrostriatal axis from the neurotoxin MPTP by modulation of the AKT pathway. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 3186-3191.	3.3	145
25	Loss or Gain of Function Mutations in <i>ACOX1</i> Cause Axonal Loss Via Different Mechanisms. SSRN Electronic Journal, 0, , .	0.4	0