Camilla Maffezzini

List of Publications by Year
in descending order

12 papers	492 citations	933447 10 h-index	1199594 12 g-index
12 all docs	12 docs citations	12 times ranked	1140 citing authors

#	Article	IF	Citations
1	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in *NDUFB7*. Human Mutation, 2021, 42, 378-384.	2.5	8
2	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation inÂvitro. STAR Protocols, 2021, 2, 100528.	1.2	11
3	Metabolic regulation of neurodifferentiation in the adult brain. Cellular and Molecular Life Sciences, 2020, 77, 2483-2496.	5.4	46
4	C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis. Nucleic Acids Research, 2019, 47, 9386-9399.	14.5	26
5	Defects of mitochondrial RNA turnover lead to the accumulation of double-stranded RNA in vivo. PLoS Genetics, 2019, 15, e1008240.	3.5	40
6	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711.	4.8	32
7	Mutations in the mitochondrial tryptophanylâ€tRNA synthetase cause growth retardation and progressive leukoencephalopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e654.	1.2	13
8	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. Diabetes, 2019, 68, 709-723.	0.6	22
9	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
10	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. PLoS Genetics, 2016, 12, e1006028.	3.5	43
11	Rescue of primary ubiquinone deficiency due to a novel *COQ7* defect using 2,4â€"dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	3.2	94
12	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	58