Camilla Maffezzini

List of Publications by Year
in descending order

12	492	933447 10	1199594 12
papers	citations	h-index	g-index

12	12	12	1140
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	3.2	94
3	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	58
4	Metabolic regulation of neurodifferentiation in the adult brain. Cellular and Molecular Life Sciences, 2020, 77, 2483-2496.	5.4	46
5	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. PLoS Genetics, 2016, 12, e1006028.	3.5	43
6	Defects of mitochondrial RNA turnover lead to the accumulation of double-stranded RNA in vivo. PLoS Genetics, 2019, 15, e1008240.	3.5	40
7	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711.	4.8	32
8	C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis. Nucleic Acids Research, 2019, 47, 9386-9399.	14.5	26
9	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
10	Mutations in the mitochondrial tryptophanylâ€tRNA synthetase cause growth retardation and progressive leukoencephalopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e654.	1.2	13
11	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
12	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. Human Mutation, 2021, 42, 378-384.	2.5	8