Benjamin J O'callaghan

List of Publications by Year
in descending order

6 papers	159 citations	1937685 4 h-index	1872680 6 g-index
7 all docs	7 docs citations	7 times ranked	286 citing authors

#	Article	IF	Citations
1	An update on the genetics, clinical presentation, and pathomechanisms of human riboflavin transporter deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 598-607.	3.6	65
2	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
3	A novelATP1A2mutation in a patient with hypokalaemic periodic paralysis and CNS symptoms. Brain, 2018, 141, 3308-3318.	7.6	26
4	ANGPTL6 Genetic Variants Are an Underlying Cause of Familial Intracranial Aneurysms. Neurology, 2021, 96, e947-e955.	1.1	6
5	Brownâ^'Vialettoâ^'Van Laere and Fazioâ^'Londe syndromes: SLC52A3 mutations with puzzling phenotypes and inheritance. European Journal of Neurology, 2021, 28, 945-954.	3.3	5
6	The startle disease mutation Î±1S270T predicts shortening of glycinergic synaptic currents. Journal of Physiology, 2020, 598, 3417-3438.	2.9	2