

Nicholas Jc Smith

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8334668/publications.pdf>

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9
papers

167
citations

1684188
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h-index

1474206
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g-index

9
all docs

9
docs citations

9
times ranked

465
citing authors

#	ARTICLE	IF	CITATIONS
1	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	6.2	87
2	Neuronal ceroid lipofuscinosis type 2: an Australian case series. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1210-1218.	0.8	19
3	Reduced cerebral vascularization in experimental neuronopathic Gaucher disease. <i>Journal of Pathology</i> , 2018, 244, 120-128.	4.5	17
4	Glycosphingolipid analysis in a naturally occurring ovine model of acute neuronopathic Gaucher disease. <i>Neurobiology of Disease</i> , 2016, 91, 143-154.	4.4	16
5	Quantification of plasma sulfatides by mass spectrometry: Utility for metachromatic leukodystrophy. <i>Analytica Chimica Acta</i> , 2017, 955, 79-85.	5.4	15
6	Severely impaired CTL killing is a feature of the neurological disorder Niemann-Pick disease type C1. <i>Blood</i> , 2022, 139, 1833-1849.	1.4	5
7	Nonfunctioning pituitary macroadenoma in a child with neurofibromatosis type 1. <i>ANZ Journal of Surgery</i> , 2017, 87, E220-E221.	0.7	3
8	Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations. <i>Internal Medicine Journal</i> , 2022, 52, 110-120.	0.8	3
9	Using provider-parent strategies to improve influenza vaccination in children and adolescents with special risk medical conditions: a randomised controlled trial protocol. <i>BMJ Open</i> , 2022, 12, e053838.	1.9	2