

Tommy Ståldberg

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7775735/publications.pdf>

Version: 2024-02-01

7
papers

411
citations

1478505

6
h-index

1720034

7
g-index

7
all docs

7
docs citations

7
times ranked

975
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038.	12.8	160
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
3	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
4	An LCâ€‘MS/MS-Based Method for the Quantification of Pyridox(am)ine 5â€‘Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. Analytical Chemistry, 2017, 89, 8892-8900.	6.5	24
5	Epilepsy syndromes, etiologies, and the use of nextâ€‘generation sequencing in epilepsy presenting in the first 2 years of life: A populationâ€‘based study. Epilepsia, 2020, 61, 2486-2499.	5.1	24
6	<i>SLC12A2</i> mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	1.9	20
7	Outcome at age 7 of epilepsy presenting in the first 2â€‘%years of life. A populationâ€‘based study. Epilepsia, 2022, 63, 2096-2107.	5.1	5