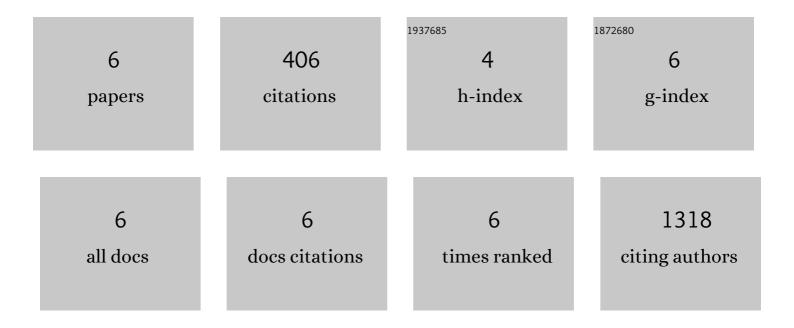
## Rory Olson

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/9887587/publications.pdf Version: 2024-02-01



ROPY OLSON

#	Article	IF	CITATIONS
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
3	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	4.1	22
4	<i>De novo DHDDS</i> variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
5	De novo <scp><i>PBX1</i></scp> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <scp>CAKUTHED</scp> phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 919-925.	1.2	6
6	Recurrent ganglioneuroma in <scp><i>PTPN11</i></scp> â€associated Noonan syndrome: A case report and literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1883-1887.	1.2	2