

Rory Olson

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9887587/publications.pdf>

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6
papers

406
citations

1937685

4
h-index

1872680

6
g-index

6
all docs

6
docs citations

6
times ranked

1318
citing authors

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