

Tomi L Toler

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3179549/publications.pdf>

Version: 2024-02-01

12
papers

417
citations

949033

11
h-index

1255698

13
g-index

14
all docs

14
docs citations

14
times ranked

1059
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	1.1	20
2	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. <i>European Journal of Human Genetics</i> , 2021, 29, 1719-1724.	1.4	2
3	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. <i>Genetics in Medicine</i> , 2020, 22, 547-556.	1.1	63
4	Phenotypic expansion of <i>KMT2D</i> -related disorder: Beyond Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1053-1065.	0.7	23
5	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	2.6	23
6	Variable cardiovascular phenotypes associated with <i>SMAD2</i> pathogenic variants. <i>Human Mutation</i> , 2018, 39, 1875-1884.	1.1	23
7	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. <i>Genetics in Medicine</i> , 2017, 19, 1040-1048.	1.1	85
8	Thyroid dysfunction in patients with Down syndrome: Results from a multi-institutional registry study. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1539-1545.	0.7	34
9	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. <i>American Journal of Human Genetics</i> , 2016, 99, 728-734.	2.6	75
10	Detecting celiac disease in patients with Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3098-3105.	0.7	14
11	National down syndrome patient database: Insights from the development of a multi-center registry study. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2520-2526.	0.7	19
12	Jaffe's Campanacci syndrome, revisited: detailed clinical and molecular analyses determine whether patients have neurofibromatosis type 1, coincidental manifestations, or a distinct disorder. <i>Genetics in Medicine</i> , 2014, 16, 448-459.	1.1	33