

Tomi L Toler

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

417
citations

840776
11
h-index

1125743
13
g-index

14
all docs

14
docs citations

14
times ranked

988
citing authors

#	ARTICLE	IF	CITATIONS
1	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.	2.4	85
2	Variants in HNRNP2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. <i>American Journal of Human Genetics</i> , 2016, 99, 728-734.	6.2	75
3	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. <i>Genetics in Medicine</i> , 2020, 22, 547-556.	2.4	63
4	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.	1.2	34
5	Jaffeâ€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.	2.4	33
6	Variable cardiovascular phenotypes associated with <i>SMAD2</i> pathogenic variants. <i>Human Mutation</i> , 2018, 39, 1875-1884.	2.5	23
7	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	6.2	23
8	Phenotypic expansion of <i>KMT2D</i> -related disorder: Beyond Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1053-1065.	1.2	23
9	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	2.4	20
10	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.	1.2	19
11	Detecting celiac disease in patients with Down syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3098-3105.	1.2	14
12	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. <i>European Journal of Human Genetics</i> , 2021, 29, 1719-1724.	2.8	2