Tomi L Toler

List of Publications by Year
in descending order

12	417	840776 11	1125743 13
papers	citations	h-index	g-index

14	14	14	988
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
2	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. European Journal of Human Genetics, 2021, 29, 1719-1724.	2.8	2
3	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	2.4	63
4	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	1.2	23
5	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
6	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	2.5	23
7	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
8	Thyroid dysfunction in patients with Down syndrome: Results from a multiâ€institutional registry study. American Journal of Medical Genetics, Part A, 2017, 173, 1539-1545.	1.2	34
9	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	6.2	75
10	Detecting celiac disease in patients with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3098-3105.	1.2	14
11	National down syndrome patient database: Insights from the development of a multiâ€center registry study. American Journal of Medical Genetics, Part A, 2015, 167, 2520-2526.	1.2	19
12	Jaffeâ€“Campanacci syndrome, revisited: detailed clinical and molecular analyses determine whether patients have neurofibromatosis type 1, coincidental manifestations, or a distinct disorder. Genetics in Medicine, 2014, 16, 448-459.	2.4	33