Tomi L Toler

List of Publications by Year
in descending order

12	417	949033 11	1255698 13
papers	citations	h-index	g-index

14	14	14	1059
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	1.1	85
2	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	2.6	75
3	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	1.1	63
4	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.	0.7	34
5	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.	1.1	33
6	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
7	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
8	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	0.7	23
9	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
10	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.	0.7	19
11	Detecting celiac disease in patients with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3098-3105.	0.7	14
12	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. European Journal of Human Genetics, 2021, 29, 1719-1724.	1.4	2