

Marina T Distefano

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6328334/publications.pdf>

Version: 2024-02-01

10
papers

1,516
citations

1040056

9
h-index

1372567

10
g-index

13
all docs

13
docs citations

13
times ranked

4065
citing authors

#	ARTICLE	IF	CITATIONS
1	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. <i>Genome Medicine</i> , 2022, 14, 6.	8.2	34
2	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. <i>Cell Genomics</i> , 2022, 2, 100131.	6.5	11
3	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2022, 24, 1392-1406.	2.4	18
4	The Gene Curation Coalition: A global effort to harmonize geneâ€‘disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	2.4	56
5	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. <i>Genetics in Medicine</i> , 2021, 23, 2208-2212.	2.4	18
6	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. <i>Genetics in Medicine</i> , 2019, 21, 2442-2452.	2.4	56
7	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. <i>Cell</i> , 2019, 177, 587-596.e9.	28.9	516
8	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. <i>Human Mutation</i> , 2018, 39, 1593-1613.	2.5	312
9	ClinVar database of global familial hypercholesterolemiaâ€‘associated DNA variants. <i>Human Mutation</i> , 2018, 39, 1631-1640.	2.5	84
10	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. <i>American Journal of Human Genetics</i> , 2017, 100, 895-906.	6.2	403