Marina T Distefano

List of Publications by Year
in descending order

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