Christina A Austin-Tse

List of Publications by Year
in descending order

19	1,320	840776 11	794594 19
papers	citations	h-index	g-index

25	25	25	3453
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	JAK inhibition in a patient with a STAT1 gain-of-function variant reveals STAT1 dysregulation as a common feature of aplastic anemia. Med, 2022, 3, 42-57.e5.	4.4	11
2	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
3	Monogenic and Polygenic Contributions to QTc Prolongation in the Population. Circulation, 2022, 145, 1524-1533.	1.6	14
4	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	3.8	48
5	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	2.5	7
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	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. Genetics in Medicine, 2021, 23, 1689-1696.	2.4	17
8	Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM). Journal of Molecular Diagnostics, 2021, 23, 589-598.	2.8	5
9	Rare Coding Variants Associated With Electrocardiographic Intervals Identify Monogenic Arrhythmia Susceptibility Genes: A Multi-Ancestry Analysis. Circulation Genomic and Precision Medicine, 2021, 14, e003300.	3.6	7
10	Genetic compensation for cilia defects in cep290 mutants by upregulation of cilia-associated small GTPases. Journal of Cell Science, 2021, 134, .	2.0	14
11	Case 6-2020: A 34-Year-Old Woman with Hyperglycemia. New England Journal of Medicine, 2020, 382, 745-753.	27.0	12
12	Assaying sensory ciliopathies using calcium biosensor expression in zebrafish ciliated olfactory neurons. Cilia, 2018, 7, 2.	1.8	17
13	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	2.4	43
14	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science, 2016, 354, .	12.6	464
15	Cytoplasmic carboxypeptidase 5 regulates tubulin glutamylation and zebrafish cilia formation and function. Molecular Biology of the Cell, 2014, 25, 1836-1844.	2.1	37
16	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184
17	Zebrafish Cilia. Methods in Enzymology, 2013, 525, 219-244.	1.0	3
18	CCDC103 mutations cause primary ciliary dyskinesia by disrupting assembly of ciliary dynein arms. Nature Genetics, 2012, 44, 714-719.	21.4	228

#	Article	IF	Citations
19	Mutation mapping and identification by whole-genome sequencing. Genome Research, 2012, 22, 1541-1548.	5.5	126