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	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science, 2016, 354, .	12.6	464
2	CCDC103 mutations cause primary ciliary dyskinesia by disrupting assembly of ciliary dynein arms. Nature Genetics, 2012, 44, 714-719.	21.4	228
3	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
4	Mutation mapping and identification by whole-genome sequencing. Genome Research, 2012, 22, 1541-1548.	5.5	126
5	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
6	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	3.8	48
7	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
8	Cytoplasmic carboxypeptidase 5 regulates tubulin glutamylation and zebrafish cilia formation and function. Molecular Biology of the Cell, 2014, 25, 1836-1844.	2.1	37
9	Assaying sensory ciliopathies using calcium biosensor expression in zebrafish ciliated olfactory neurons. Cilia, 2018, 7, 2.	1.8	17
10	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
11	Genetic compensation for cilia defects in cep290 mutants by upregulation of cilia-associated small GTPases. Journal of Cell Science, 2021, 134, .	2.0	14
12	Monogenic and Polygenic Contributions to QTc Prolongation in the Population. Circulation, 2022, 145, 1524-1533.	1.6	14
13	Case 6-2020: A 34-Year-Old Woman with Hyperglycemia. New England Journal of Medicine, 2020, 382, 745-753.	27.0	12
14	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
15	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
16	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
17	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	2.5	7
18	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

#	Article	IF	Citations
19	Zebrafish Cilia. Methods in Enzymology, 2013, 525, 219-244.	1.0	3