Ben Weisburd

List of Publications by Year
in descending order

17	18,063	623734	839539
papers	citations	14 h-index	18 g-index

21	21	21	40060
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	2.5	31
2	Questioning the Association of the STMN2 Dinucleotide Repeat With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2022, 8, e678.	1.9	1
3	WGS and RNA Studies Diagnose Noncoding DMD Variants in Males With High Creatine Kinase. Neurology: Genetics, 2021, 7, e554.	1.9	21
4	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
5	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. Nature Communications, 2021, 12, 3505.	12.8	49
6	Recurrent TTN metatranscriptâ€only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	2.5	28
7	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. Brain, 2020, 143, 2904-2910.	7.6	53
8	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
9	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
10	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	3.5	66
11	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
12	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. Human Mutation, 2017, 38, 517-523.	2.5	49
13	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
14	ClinVar data parsing. Wellcome Open Research, 2017, 2, 33.	1.8	19
15	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
16	KSHV 2.0: A Comprehensive Annotation of the Kaposi's Sarcoma-Associated Herpesvirus Genome Using Next-Generation Sequencing Reveals Novel Genomic and Functional Features. PLoS Pathogens, 2014, 10, e1003847.	4.7	264
17	Decoding Human Cytomegalovirus. Science, 2012, 338, 1088-1093.	12.6	546