

# Ben Weisburd

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2346092/publications.pdf>

Version: 2024-02-01

17  
papers

18,063  
citations

706676

14  
h-index

939365

18  
g-index

21  
all docs

21  
docs citations

21  
times ranked

43268  
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>seqr</i> : A web-based analysis and collaboration tool for rare disease genomics. <i>Human Mutation</i> , 2022, , .	1.1	31
2	Questioning the Association of the <i>STMN2</i> Dinucleotide Repeat With Amyotrophic Lateral Sclerosis. <i>Neurology: Genetics</i> , 2022, 8, e678.	0.9	1
3	WGS and RNA Studies Diagnose Noncoding <i>DMD</i> Variants in Males With High Creatine Kinase. <i>Neurology: Genetics</i> , 2021, 7, e554.	0.9	21
4	A form of muscular dystrophy associated with pathogenic variants in <i>JAG2</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	15
5	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , 2021, 12, 3505.	5.8	49
6	Recurrent <i>TTN</i> metatranscript-only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogyposis multiplex congenita and myopathy. <i>Human Mutation</i> , 2020, 41, 403-411.	1.1	28
7	A novel <i>RFC1</i> repeat motif (ACAGG) in two Asia-Pacific <i>CANVAS</i> families. <i>Brain</i> , 2020, 143, 2904-2910.	3.7	53
8	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	13.7	6,140
9	A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.	13.7	614
10	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. <i>PLoS Genetics</i> , 2018, 14, e1007329.	1.5	66
11	The ExAC browser: displaying reference data information from over 60 000 exomes. <i>Nucleic Acids Research</i> , 2017, 45, D840-D845.	6.5	587
12	Pathogenic <i>ASXL1</i> somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. <i>Human Mutation</i> , 2017, 38, 517-523.	1.1	49
13	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	516
14	ClinVar data parsing. <i>Wellcome Open Research</i> , 2017, 2, 33.	0.9	19
15	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , 2016, 536, 285-291.	13.7	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. <i>PLoS Pathogens</i> , 2014, 10, e1003847.	2.1	264
17	Decoding Human Cytomegalovirus. <i>Science</i> , 2012, 338, 1088-1093.	6.0	546