David H Ledbetter

List of Publications by Year
in descending order

58	13,655	76294	133188
papers	citations	40 h-index	59 g-index

61	61	61	20847
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	2.6	2,325
2	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
3	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
4	ClinGen â€" The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	13.9	1,016
5	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	6.5	698
6	Genetic and Pharmacologic Inactivation of ANGPTL3 and Cardiovascular Disease. New England Journal of Medicine, 2017, 377, 211-221.	13.9	633
7	Implementing genomic medicine in the clinic: the future is here. Genetics in Medicine, 2013, 15, 258-267.	1.1	472
8	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science, 2016, 354, .	6.0	464
9	Inactivating Variants in ANGPTL4 and Risk of Coronary Artery Disease. New England Journal of Medicine, 2016, 374, 1123-1133.	13.9	411
10	Clinical Genetic Testing for FamilialÂHypercholesterolemia. Journal of the American College of Cardiology, 2018, 72, 662-680.	1.2	387
11	Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 2413-2421.	1.1	378
12	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	13.7	369
13	Feasibility of blood testing combined with PET-CT to screen for cancer and guide intervention. Science, 2020, 369, .	6.0	351
14	Genetic identification of familial hypercholesterolemia within a single U.S. health care system. Science, 2016, 354, .	6.0	349
15	The Geisinger MyCode community health initiative: an electronic health recordâ€"linked biobank for precision medicine research. Genetics in Medicine, 2016, 18, 906-913.	1.1	340
16	Developmental brain dysfunction: revival and expansion of old concepts based on new genetic evidence. Lancet Neurology, The, 2013, 12, 406-414.	4.9	268
17	The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population. Biological Psychiatry, 2015, 77, 785-793.	0.7	198
18	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	6.0	195

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19	Exome Sequencingâ€“Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. JAMA Network Open, 2018, 1, e182140.	2.8	163
20	Enhanced detection of clinically relevant genomic imbalances using a targeted plus whole genome oligonucleotide microarray. Genetics in Medicine, 2008, 10, 415-429.	1.1	145
21	Using large clinical data sets to infer pathogenicity for rare copy number variants in autism cohorts. Molecular Psychiatry, 2013, 18, 1090-1095.	4.1	140
22	Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349.	1.1	134
23	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	0.7	133
24	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	1.1	125
25	The Role of Parental Cognitive, Behavioral, and Motor Profiles in Clinical Variability in Individuals With Chromosome 16p11.2 Deletions. JAMA Psychiatry, 2015, 72, 119.	6.0	112
26	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	2.6	110
27	A Cross-Disorder Method to Identify Novel Candidate Genes for Developmental Brain Disorders. JAMA Psychiatry, 2016, 73, 275.	6.0	107
28	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. Nature Communications, 2018, 9, 2252.	5.8	99
29	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	15.2	90
30	Quantifying the polygenic contribution to variable expressivity in eleven rare genetic disorders. Nature Communications, 2019, 10, 4897.	5.8	89
31	Clinical outcomes of a genomic screening program for actionable genetic conditions. Genetics in Medicine, 2020, 22, 1874-1882.	1.1	84
32	Patient-Centered Precision Health In A Learning Health Care System: Geisingerâ€™s Genomic Medicine Experience. Health Affairs, 2018, 37, 757-764.	2.5	81
33	Phenome-wide association studies demonstrating pleiotropy of genetic variants within FTO with and without adjustment for body mass index. Frontiers in Genetics, 2014, 5, 250.	1.1	66
34	Molecular Diagnostic Yield of Exome Sequencing in Patients With Cerebral Palsy. JAMA - Journal of the American Medical Association, 2021, 325, 467.	3.8	64
35	Profiling and Leveraging Relatedness in a Precision Medicine Cohort of 92,455 Exomes. American Journal of Human Genetics, 2018, 102, 874-889.	2.6	58
36	GenomeConnect: Matchmaking Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge. Human Mutation, 2015, 36, 974-978.	1.1	56

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37	The struggle to find reliable results in exome sequencing data: filtering out Mendelian errors. Frontiers in Genetics, 2014, 5, 16.	1.1	51
38	Copy Number Variants, Aneuploidies, and Human Disease. Clinics in Perinatology, 2015, 42, 227-242.	0.8	51
39	Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. Journal of Autism and Developmental Disorders, 2016, 46, 2734-2748.	1.7	47
40	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. Genetics in Medicine, 2018, 20, 554-558.	1.1	46
41	Identification of Neuropsychiatric Copy Number Variants in a Health Care System Population. JAMA Psychiatry, 2020, 77, 1276.	6.0	46
42	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.	1.1	43
43	Shift happens: family background influences clinical variability in genetic neurodevelopmental disorders. Genetics in Medicine, 2016, 18, 302-304.	1.1	39
44	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology. American Journal of Psychiatry, 2022, 179, 189-203.	4.0	29
45	Diagnostic genetic testing for neurodevelopmental psychiatric disorders: closing the gap between recommendation and clinical implementation. Current Opinion in Genetics and Development, 2021, 68, 1-8.	1.5	28
46	GWAS of serum ALT and AST reveals an association of SLC30A10 Thr95Ile with hypermanganesemia symptoms. Nature Communications, 2021, 12, 4571.	5.8	26
47	ClinGen's GenomeConnect registry enables patientâ€centered data sharing. Human Mutation, 2018, 39, 1668-1676.	1.1	25
48	Gene-level analysis of rare variants in 379,066 whole exome sequences identifies an association of GIGYF1 loss of function with type 2 diabetes. Scientific Reports, 2021, 11, 21565.	1.6	25
49	Leveraging populationâ€based exome screening to impact clinical care: The evolution of variant assessment in the <scp>Geisinger MyCode</scp> research project. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 83-94.	0.7	21
50	Genomic Variation: Lessons Learned from Whole-Genome CNV Analysis. Current Genetic Medicine Reports, 2014, 2, 146-150.	1.9	16
51	Population-scale analysis of common and rare genetic variation associated with hearing loss in adults. Communications Biology, 2022, 5, .	2.0	12
52	Long overdue: including adults with brain disorders in precision health initiatives. Current Opinion in Genetics and Development, 2020, 65, 47-52.	1.5	11
53	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. Genetics in Medicine, 2021, 23, 1738-1745.	1.1	7
54	Population Genomic Screening for Genetic Etiologies of Neurodevelopmental/Psychiatric Disorders Demonstrates Personal Utility and Positive Participant Responses. Journal of Personalized Medicine, 2021, 11, 365.	1.1	6

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55	Molecular Cytogenetic Analysis of Telomere Rearrangements. Current Protocols in Human Genetics, 2015, 84, 8.11.1-8.11.15.	3.5	3
56	Response to Buxbaum etÂal.. American Journal of Human Genetics, 2020, 107, 1004.	2.6	3
57	Medical manifestations and healthcare utilization among adult MyCode participants with neurodevelopmental psychiatric copy number variants. Genetics in Medicine, 2021, , .	1.1	2
58	Abstract 15754: The Prevalence of Electronic Health Record-Based Clinical Phenotypes in Patients With Pathogenetic Variants Associated With Arrhythmogenic Right Ventricular Cardiomyopathy. Circulation, 2015, 132, .	1.6	0