David B Goldstein

List of Publications by Year
in descending order

132 papers	11,509 citations	53660 45 h-index	34900 98 g-index
136 all docs	136 docs citations	136 times ranked	20359 citing authors

#	Article	IF	Citations
1	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	13.7	1,351
2	The â€œAll of Usâ€•Research Program. New England Journal of Medicine, 2019, 381, 668-676.	13.9	955
3	Genic Intolerance to Functional Variation and the Interpretation of Personal Genomes. PLoS Genetics, 2013, 9, e1003709.	1.5	844
4	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
6	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. Lancet, The, 2019, 393, 758-767.	6.3	368
7	Drug development in the era of precision medicine. Nature Reviews Drug Discovery, 2018, 17, 183-196.	21.5	294
8	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	1.1	284
9	Sequencing studies in human genetics: design and interpretation. Nature Reviews Genetics, 2013, 14, 460-470.	7.7	236
10	Distinct neurological disorders with ATP1A3 mutations. Lancet Neurology, The, 2014, 13, 503-514.	4.9	206
11	MicroRNAs in epilepsy: pathophysiology and clinical utility. Lancet Neurology, The, 2016, 15, 1368-1376.	4.9	200
12	Unequal representation of genetic variation across ancestry groups creates healthcare inequalityÂin the application of precision medicine. Genome Biology, 2016, 17, 157.	3.8	198
13	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	1.1	191
14	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	4.9	190
15	An Exome Sequencing Study to Assess the Role of Rare Genetic Variation in Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2017, 196, 82-93.	2.5	185
16	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	2.8	184
17	Polymorphisms of large effect explain the majority of the host genetic contribution to variation of HIV-1 virus load. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 14658-14663.	3.3	154
18	Rare-variant collapsing analyses for complex traits: guidelines and applications. Nature Reviews Genetics, 2019, 20, 747-759.	7.7	147

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19	Will tomorrow's medicines work for everyone?. Nature Genetics, 2004, 36, S34-S42.	9.4	137
20	One gene, many neuropsychiatric disorders: lessons from Mendelian diseases. Nature Neuroscience, 2014, 17, 773-781.	7.1	128
21	The Intolerance of Regulatory Sequence to Genetic Variation Predicts Gene Dosage Sensitivity. PLoS Genetics, 2015, 11, e1005492.	1.5	123
22	Annotating pathogenic non-coding variants in genic regions. Nature Communications, 2017, 8, 236.	5.8	122
23	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122
24	The intolerance to functional genetic variation of protein domains predicts the localization of pathogenic mutations within genes. Genome Biology, 2016, 17, 9.	3.8	118
25	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhoodâ€"a study of 155 patients. Orphanet Journal of Rare Diseases, 2015, 10, 123.	1.2	117
26	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102
27	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	7.1	101
28	Not the End of the Odyssey: Parental Perceptions of Whole Exome Sequencing (WES) in Pediatric Undiagnosed Disorders. Journal of Genetic Counseling, 2016, 25, 1019-1031.	0.9	91
29	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	15.2	90
30	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
31	Gaps and opportunities in refractory status epilepticus research in children: A multi-center approach by the Pediatric Status Epilepticus Research Group (pSERG). Seizure: the Journal of the British Epilepsy Association, 2014, 23, 87-97.	0.9	84
32	Disease Heritability Inferred from Familial Relationships Reported in Medical Records. Cell, 2018, 173, 1692-1704.e11.	13.5	79
33	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	2.8	73
34	Enhancer Domains Predict Gene Pathogenicity and Inform Gene Discovery in Complex Disease. American Journal of Human Genetics, 2020, 106, 215-233.	2.6	72
35	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. American Journal of Human Genetics, 2021, 108, 1350-1355.	2.6	72
36	Mosaic mutations in early-onset genetic diseases. Genetics in Medicine, 2016, 18, 746-749.	1.1	70

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37	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
38	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68
39	Causal Genetic Variants in Stillbirth. New England Journal of Medicine, 2020, 383, 1107-1116.	13.9	67
40	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	2.6	63
41	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
42	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	2.0	60
43	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	2.6	59
44	Rare loss-of-function variants in type I IFN immunity genes are not associated with severe COVID-19. Journal of Clinical Investigation, 2021, 131, .	3.9	56
45	Dominant Splice Site Mutations in PIK3R1 Cause Hyper IgM Syndrome, Lymphadenopathy and Short Stature. Journal of Clinical Immunology, 2016, 36, 462-471.	2.0	55
46	Modelling and treating GRIN2A developmental and epileptic encephalopathy in mice. Brain, 2020, 143, 2039-2057.	3.7	51
47	IFNL3 mRNA structure is remodeled by a functional non-coding polymorphism associated with hepatitis C virus clearance. Scientific Reports, 2015, 5, 16037.	1.6	49
48	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	0.7	47
49	The Importance of Synthetic Associations Will Only Be Resolved Empirically. PLoS Biology, 2011, 9, e1001008.	2.6	46
50	Epileptic encephalopathy-causing mutations in DNM1 impair synaptic vesicle endocytosis. Neurology: Genetics, 2015, 1, e4.	0.9	46
51	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	0.7	45
52	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	2.8	44
53	Natural Selection Shapes Codon Usage in the Human Genome. American Journal of Human Genetics, 2020, 107, 83-95.	2.6	44
54	A functional correlate of severity in alternating hemiplegia of childhood. Neurobiology of Disease, 2015, 77, 88-93.	2.1	43

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55	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
56	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	1.1	42
57	Reduced GABAergic Neuron Excitability, Altered Synaptic Connectivity, and Seizures in a KCNT1 Gain-of-Function Mouse Model of Childhood Epilepsy. Cell Reports, 2020, 33, 108303.	2.9	41
58	Association Between Common Variants in RBFOX1, an RNA-Binding Protein, and Brain Amyloidosis in Early and Preclinical Alzheimer Disease. JAMA Neurology, 2020, 77, 1288.	4.5	41
59	Exome-Based Rare-Variant Analyses in CKD. Journal of the American Society of Nephrology: JASN, 2019, 30, 1109-1122.	3.0	40
60	The importance of dynamic re-analysis in diagnostic whole exome sequencing. Journal of Medical Genetics, 2017, 54, 155-156.	1.5	38
61	Genetic epidemiology of motor neuron disease-associated variants in the Scottish population. Neurobiology of Aging, 2017, 51, 178.e11-178.e20.	1.5	37
62	Assessing the Role of Rare Genetic Variation in Patients With Heart Failure. JAMA Cardiology, 2021, 6, 379.	3.0	37
63	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
64	Exome sequencing in obsessiveâ€“compulsive disorder reveals a burden of rare damaging coding variants. Nature Neuroscience, 2021, 24, 1071-1076.	7.1	35
65	Inhibition of microRNA 128 promotes excitability of cultured cortical neuronal networks. Genome Research, 2016, 26, 1411-1416.	2.4	34
66	Whole-exome sequencing identifies rare variants in STAB2 associated with venous thromboembolic disease. Blood, 2020, 136, 533-541.	0.6	34
67	Clinical application of whole-genome sequencing in patients with primary immunodeficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 476-479.e6.	1.5	33
68	Serine biosynthesis defect due to haploinsufficiency of PHGDH causes retinal disease. Nature Metabolism, 2021, 3, 366-377.	5.1	32
69	Orion: Detecting regions of the human non-coding genome that are intolerant to variation using population genetics. PLoS ONE, 2017, 12, e0181604.	1.1	31
70	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
71	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genetics in Medicine, 2020, 22, 1269-1275.	1.1	30
72	Incorporating Functional Information in Tests of Excess De Novo Mutational Load. American Journal of Human Genetics, 2015, 97, 272-283.	2.6	29

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73	Improved Pathogenic Variant Localization via a Hierarchical Model of Sub-regional Intolerance. American Journal of Human Genetics, 2019, 104, 299-309.	2.6	29
74	High-impact rare genetic variants in severe schizophrenia. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	29
75	A caseâ€“control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic ABCA4 variants. Genetics in Medicine, 2019, 21, 2336-2344.	1.1	27
76	Rare genetic causes of complex kidney and urological diseases. Nature Reviews Nephrology, 2020, 16, 641-656.	4.1	27
77	Antibodies to Severe Acute Respiratory Syndrome Coronavirus 2 (SARS-CoV-2) in All of Us Research Program Participants, 2 January to 18 March 2020. Clinical Infectious Diseases, 2022, 74, 584-590.	2.9	26
78	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. PLoS Genetics, 2017, 13, e1007104.	1.5	25
79	Rare and Common Variants in KIF15 Contribute to Genetic Risk of Idiopathic Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 56-69.	2.5	25
80	Exome sequencing results in successful riboflavin treatment of a rapidly progressive neurological condition. Journal of Physical Education and Sports Management, 2015, 1, a000257.	0.5	24
81	Noncoding deletions reveal a gene that is critical for intestinal function. Nature, 2019, 571, 107-111.	13.7	24
82	meaRtools: An R package for the analysis of neuronal networks recorded on microelectrode arrays. PLoS Computational Biology, 2018, 14, e1006506.	1.5	22
83	From genetics to physiology at last. Nature, 2016, 530, 162-163.	13.7	21
84	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	2.4	21
85	De novo mutations in childhood cases of sudden unexplained death that disrupt intracellular Ca ²⁺ regulation. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	21
86	Evaluating the Impact of Functional Genetic Variation on HIV-1 Control. Journal of Infectious Diseases, 2017, 216, 1063-1069.	1.9	20
87	ATAV: a comprehensive platform for population-scale genomic analyses. BMC Bioinformatics, 2021, 22, 149.	1.2	20
88	Association of rare predicted loss-of-function variants of influenza-related type I IFN genes with critical COVID-19 pneumonia. Reply.. Journal of Clinical Investigation, 2021, 131, .	3.9	20
89	Genetic testing in individuals with cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 1448-1455.	1.1	19
90	D-DEMÃ˜, a distinct phenotype caused by ATP1A3 mutations. Neurology: Genetics, 2020, 6, e466.	0.9	18

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91	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
92	Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375.	0.5	17
93	Genome-wide association study (GWAS) of human host factors influencing viral severity of herpes simplex virus type 2 (HSV-2). Genes and Immunity, 2019, 20, 112-120.	2.2	17
94	Sudden unexpected death in asymptomatic infants due toPPA2variants. Molecular Genetics & Genomic Medicine, 2020, 8, e1008.	0.6	17
95	Lossâ€ofâ€function variants in NFIA provide further support that NFIA is a critical gene in 1p32â€p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	0.7	16
96	Utilizing Population Controls in Rare-Variant Case-Parent Association Tests. American Journal of Human Genetics, 2014, 94, 845-853.	2.6	15
97	A Transcriptomeâ€Based Drug Discovery Paradigm for Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 199-211.	2.8	14
98	Expansion of the GRIA2 phenotypic representation: a novel de novo loss of function mutation in a case with childhood onset schizophrenia. Journal of Human Genetics, 2021, 66, 339-343.	1.1	14
99	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	2.6	14
100	Phenomics and the Interpretation of Personal Genomes. Science Translational Medicine, 2014, 6, 254fs35.	5.8	13
101	Whole Exome Sequencing Reveals Severe Thrombophilia in Acute Unprovoked Idiopathic Fatal Pulmonary Embolism. EBioMedicine, 2017, 17, 95-100.	2.7	13
102	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	2.6	13
103	Growth of genome screening needs debate. Nature, 2011, 476, 27-28.	13.7	12
104	Phenotype variability in Hajdu-Cheney syndrome. European Journal of Medical Genetics, 2019, 62, 35-38.	0.7	12
105	Whole exome sequencing reveals potentially pathogenic variants in a small subset of premenopausal women with idiopathic osteoporosis. Bone, 2022, 154, 116253.	1.4	12
106	The benefit of diagnostic whole genome sequencing in schizophrenia and other psychotic disorders. Molecular Psychiatry, 2022, 27, 1435-1447.	4.1	12
107	Sustained therapeutic response to riboflavin in a child with a progressive neurological condition, diagnosed by whole-exome sequencing. Journal of Physical Education and Sports Management, 2015, 1, a000265.	0.5	11
108	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	2.6	11

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109	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	2.8	9
110	Late-onset pattern macular dystrophy mimicking ABCA4 and PRPH2 disease is caused by a homozygous frameshift mutation in ROM1. Journal of Physical Education and Sports Management, 2019, 5, a003624.	0.5	8
111	Alternating hemiplegia of childhood: evolution over time and mouse model corroboration. Brain Communications, 2021, 3, fcab128.	1.5	8
112	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	1.4	8
113	A Novel Kv7.3 Variant in the Voltage-Sensing S4 Segment in a Family With Benign Neonatal Epilepsy: Functional Characterization and in vitro Rescue by Î²-Hydroxybutyrate. Frontiers in Physiology, 2020, 11, 1040.	1.3	7
114	The phenotype of bilateral hippocampal sclerosis and its management in â€œreal lifeâ€•clinical settings. Epilepsia, 2018, 59, 1410-1420.	2.6	6
115	Neuropsychiatric genomics in precision medicine: diagnostics, gene discovery, and translation. Dialogues in Clinical Neuroscience, 2016, 18, 237-252.	1.8	6
116	Incorporating external information to improve sparse signal detection in rareâ€variant geneâ€setâ€based analyses. Genetic Epidemiology, 2020, 44, 330-338.	0.6	5
117	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	1.1	5
118	Hormonal intervention for the treatment of veterans with COVID-19 requiring hospitalization (HITCH): a multicenter, phase 2 randomized controlled trial of best supportive care vs best supportive care plus degarelix: study protocol for a randomized controlled trial. Trials, 2021, 22, 431.	0.7	5
119	Precision genetic cellular models identify therapies protective against ER stress. Cell Death and Disease, 2021, 12, 770.	2.7	5
120	Encephalopathy-causing mutations in GÎ²1 (GNB1) alter regulation of neuronal GIRK channels. IScience, 2021, 24, 103018.	1.9	4
121	Exome Sequencing in Venous Thromboembolic Disease Identifies Excess Mutation Burden in PROS1, PROC, SERPINC1 and STAB2. Blood, 2016, 128, 3794-3794.	0.6	4
122	Diagnostic sequencing to support genetically stratified medicine in a tertiary care setting. Genetics in Medicine, 2022, 24, 862-869.	1.1	4
123	Academicâ€“industrial partnerships in drug discovery in the age of genomics. Trends in Biotechnology, 2015, 33, 320-322.	4.9	3
124	Ancestry adjustment improves genome-wide estimates of regional intolerance. Genetics, 2022, , .	1.2	2
125	Translating amyotrophic lateral sclerosis genes into drug development leads. Nature Genetics, 2021, 53, 1624-1626.	9.4	2
126	A genome-wide screen for variants influencing certolizumab pegol response in a moderate to severe rheumatoid arthritis population. PLoS ONE, 2022, 17, e0261165.	1.1	2

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127	Reply. Annals of Neurology, 2017, 81, 161-162.	2.8	0
128	P1â€170: WHOLEâ€EXOME SEQUENCING IN 20,197 INDIVIDUALS IDENTIFIES ULTRAâ€RARE SORL1 LOSSâ€OFâ€FUNCTION VARIANTS IN LATEâ€ONSET ALZHEIMER'S DISEASE. Alzheimer's and Dementia, 2018, 14, P344.	0.4	0
129	Association of CYP2B6 Single-Nucleotide Polymorphisms Altering Efavirenz Metabolism With Hepatitis C Virus (HCV) Treatment Relapse Among Human Immunodeficiency Virus/HCVâ€“Coinfected African Americans Receiving Ledipasvir/Sofosbuvir in the ION-4 Trial. Clinical Infectious Diseases, 2018, 66, 1953-1956.	2.9	0
130	Genetic associations with brain amyloidosis. Alzheimer's and Dementia, 2020, 16, e042191.	0.4	0
131	Genome-Wide Association Study of the Human Genetic Factors Influencing the Risk of Adverse Events during Idelalislib Therapy in Patients with Relapsed Indolent Lymphoma. Blood, 2016, 128, 5284-5284.	0.6	0
132	Focused goodness of fit tests for gene set analyses. Briefings in Bioinformatics, 2021, , .	3.2	0