

Jonathan A Bernstein

List of Publications by Year in descending order

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Version: 2024-02-01

102
papers

8,958
citations

94269

37
h-index

48187

88
g-index

104
all docs

104
docs citations

104
times ranked

16790
citing authors

#	ARTICLE	IF	CITATIONS
1	Assembly of functionally integrated human forebrain spheroids. <i>Nature</i> , 2017, 545, 54-59.	13.7	931
2	Global analysis of mRNA decay and abundance in <i>Escherichia coli</i> at single-gene resolution using two-color fluorescent DNA microarrays. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 9697-9702.	3.3	782
3	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. <i>Nature Genetics</i> , 2016, 48, 1581-1586.	9.4	654
4	Using induced pluripotent stem cells to investigate cardiac phenotypes in Timothy syndrome. <i>Nature</i> , 2011, 471, 230-234.	13.7	624
5	Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. <i>Nature Medicine</i> , 2011, 17, 1657-1662.	15.2	521
6	Clinical Interpretation and Implications of Whole-Genome Sequencing. <i>JAMA - Journal of the American Medical Association</i> , 2014, 311, 1035.	3.8	398
7	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. <i>Genetics in Medicine</i> , 2017, 19, 209-214.	1.1	261
8	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	13.9	261
9	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
10	Association of <i>MTOR</i> Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. <i>JAMA Neurology</i> , 2016, 73, 836.	4.5	234
11	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. <i>Nature Medicine</i> , 2019, 25, 911-919.	15.2	221
12	Reanalysis of Clinical Exome Sequencing Data. <i>New England Journal of Medicine</i> , 2019, 380, 2478-2480.	13.9	205
13	Global analysis of <i>Escherichia coli</i> RNA degradosome function using DNA microarrays. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 2758-2763.	3.3	199
14	Mutations in <i>NGLY1</i> cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway. <i>Genetics in Medicine</i> , 2014, 16, 751-758.	1.1	191
15	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
16	Inhibiting Plasma Kallikrein for Hereditary Angioedema Prophylaxis. <i>New England Journal of Medicine</i> , 2017, 376, 717-728.	13.9	138
17	De Novo Mutations in Protein Kinase Genes <i>CAMK2A</i> and <i>CAMK2B</i> Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	136
18	Inappropriate p53 activation during development induces features of CHARGE syndrome. <i>Nature</i> , 2014, 514, 228-232.	13.7	117

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19	Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. <i>New England Journal of Medicine</i> , 2022, 386, 700-702.	13.9	116
20	Neuronal defects in a human cellular model of 22q11.2 deletion syndrome. <i>Nature Medicine</i> , 2020, 26, 1888-1898.	15.2	113
21	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. <i>American Journal of Human Genetics</i> , 2016, 99, 934-941.	2.6	111
22	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)â€“associated inflammatory diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1337-1345.	1.5	103
23	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. <i>Brain</i> , 2017, 140, 2610-2622.	3.7	102
24	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. <i>European Journal of Human Genetics</i> , 2015, 23, 1473-1481.	1.4	101
25	A deleterious Nav1.1 mutation selectively impairs telencephalic inhibitory neurons derived from Dravet Syndrome patients. <i>ELife</i> , 2016, 5, .	2.8	101
26	<i>RASA1</i> somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1450-1454.	0.7	85
27	Clinical Course of Six Children With GNAO1 Mutations Causing aâ€“Severe and Distinctive Movement Disorder. <i>Pediatric Neurology</i> , 2016, 59, 81-84.	1.0	78
28	Cold-aggravated pain in humans caused by a hyperactive Nav1.9 channel mutant. <i>Nature Communications</i> , 2015, 6, 10049.	5.8	71
29	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. <i>Genetics in Medicine</i> , 2019, 21, 1585-1593.	1.1	67
30	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 101, 503-515.	2.6	61
31	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
32	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. <i>Science Translational Medicine</i> , 2020, 12, .	5.8	60
33	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
34	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. <i>American Journal of Human Genetics</i> , 2018, 103, 1009-1021.	2.6	57
35	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2018, 103, 154-162.	2.6	56
36	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. <i>Nature Genetics</i> , 2019, 51, 755-763.	9.4	56

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37	Automated syndrome diagnosis by three-dimensional facial imaging. <i>Genetics in Medicine</i> , 2020, 22, 1682-1693.	1.1	47
38	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. <i>Nature Biotechnology</i> , 2022, 40, 1035-1041.	9.4	45
39	Clinical delineation of the <i>PACS1</i>-related syndrome—Report on 19 patients. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 670-675.	0.7	44
40	New insights into mitral valve dystrophy: a Filamin-A genotype—phenotype and outcome study. <i>European Heart Journal</i> , 2018, 39, 1269-1277.	1.0	44
41	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. <i>Journal of Genetic Counseling</i> , 2019, 28, 1107-1118.	0.9	42
42	Mutation update for the <i>SATB2</i> gene. <i>Human Mutation</i> , 2019, 40, 1013-1029.	1.1	38
43	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. <i>Acta Neuropathologica</i> , 2020, 139, 415-442.	3.9	38
44	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	2.6	37
45	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. <i>Neuron</i> , 2018, 100, 1354-1368.e5.	3.8	35
46	Clinical and molecular characterization of de novo loss of function variants in <i>HNRNPU</i>. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2680-2689.	0.7	34
47	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. <i>Human Mutation</i> , 2018, 39, 666-675.	1.1	34
48	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	0.7	34
49	Familial cardiac valvulopathy due to filamin A mutation. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2236-2241.	0.7	33
50	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. <i>Genetics in Medicine</i> , 2019, 21, 464-470.	1.1	33
51	Strong evidence for genotype—phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. <i>Human Molecular Genetics</i> , 2022, 31, 625-637.	1.4	32
52	Clinical, cytogenetic, and molecular outcomes in a series of 66 patients with Pierre Robin sequence and literature review: 22q11.2 deletion is less common than other chromosomal anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 870-880.	0.7	31
53	De novo and rare mutations in the HSPA1L heat shock gene associated with inflammatory bowel disease. <i>Genome Medicine</i> , 2017, 9, 8.	3.6	27
54	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. <i>Genetics in Medicine</i> , 2020, 22, 362-370.	1.1	24

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55	Predominant and novel de novo variants in 29 individuals with <i>ALG13</i> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1333-1348.	1.7	24
56	High-frequency actionable pathogenic exome variants in an average-risk cohort. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003178.	0.5	23
57	Impaired Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia. <i>Journal of Genetic Counseling</i> , 2016, 25, 936-944.	0.9	22
58	Sleep Disturbances in Individuals With Phelan-McDermid Syndrome: Correlation With Caregivers' Sleep Quality and Daytime Functioning. <i>Sleep</i> , 2017, 40, .	0.6	21
59	Exploring the Medical and Psychosocial Concerns of Adolescents and Young Adults With Craniofacial Microsomia. <i>Cleft Palate-Craniofacial Journal</i> , 2018, 55, 1430-1439.	0.5	21
60	Functional analysis of novel <i>DEAF1</i> variants identified through clinical exome sequencing expands <i>DEAF1</i> -associated neurodevelopmental disorder (DAND) phenotype. <i>Human Mutation</i> , 2017, 38, 1774-1785.	1.1	20
61	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 966-977.	0.7	20
62	Functional and structural analysis of cytokine-selective <i>IL6ST</i> defects that cause recessive hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 585-598.	1.5	20
63	Perceived utility and disutility of genomic sequencing for pediatric patients: Perspectives from parents with diverse sociodemographic characteristics. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1088-1101.	0.7	20
64	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. <i>Pediatric Neurology</i> , 2019, 90, 37-43.	1.0	19
65	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in <i>LARS1</i> . <i>Genetics in Medicine</i> , 2020, 22, 1863-1873.	1.1	19
66	It seems like COVID-19 now is the only disease present on Earth, living with a rare or undiagnosed disease during the COVID-19 pandemic. <i>Genetics in Medicine</i> , 2021, 23, 837-844.	1.1	19
67	Whole genome sequencing reveals novel <i>IGHMBP2</i> variant leading to unique cryptic splice site and Charcot-Marie-Tooth phenotype with early onset symptoms. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00676.	0.6	18
68	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	1.1	18
69	Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons. <i>Biological Psychiatry</i> , 2021, 89, 497-509.	0.7	17
70	Variable clinical severity in <i>TANGO2</i> deficiency: Case series and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 473-487.	0.7	17
71	<i>WISP3</i> mutation associated with pseudorheumatoid dysplasia. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a001990.	0.5	16
72	Allelic loss of function <i>WNT5A</i> mutations in an infant with severe and atypical manifestations of Robinow syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1030-1036.	0.7	15

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73	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. <i>Journal of Pediatrics</i> , 2018, 196, 291-297.e2.	0.9	15
74	An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease. <i>European Journal of Human Genetics</i> , 2018, 26, 1810-1818.	1.4	15
75	Association of AHSG with alopecia and mental retardation (APMR) syndrome. <i>Human Genetics</i> , 2017, 136, 287-296.	1.8	14
76	Isolated Congenital Anosmia and CNGA2 Mutation. <i>Scientific Reports</i> , 2017, 7, 2667.	1.6	14
77	Psychometric Study of the Social Responsiveness Scale in Phelan-McDermid Syndrome. <i>Autism Research</i> , 2020, 13, 1383-1396.	2.1	14
78	Factors Associated with Uptake of Genetics Services for Hypertrophic Cardiomyopathy. <i>Journal of Genetic Counseling</i> , 2015, 24, 797-809.	0.9	12
79	Prenatal treatment of ornithine transcarbamylase deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 297-300.	0.5	12
80	Respiratory system involvement in Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1849-1857.	0.7	11
81	A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. <i>Journal of Genetic Counseling</i> , 2019, 28, 213-228.	0.9	11
82	<i>NSD1</i> mutations deregulate transcription and DNA methylation of bivalent developmental genes in Sotos syndrome. <i>Human Molecular Genetics</i> , 2022, 31, 2164-2184.	1.4	11
83	"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey. <i>Journal of Genetic Counseling</i> , 2021, 30, 1707-1718.	0.9	10
84	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. <i>Pediatric Neurology</i> , 2020, 106, 24-31.	1.0	9
85	Functional and structural analyses of novel Smith-Kingsmore Syndrome-Associated MTOR variants reveal potential new mechanisms and predictors of pathogenicity. <i>PLoS Genetics</i> , 2021, 17, e1009651.	1.5	9
86	TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006198.	0.5	9
87	Combined Genome Sequencing and RNA Analysis Reveals and Characterizes a Deep Intronic Variant in IGHMBP2 in a Patient With Spinal Muscular Atrophy With Respiratory Distress Type 1. <i>Pediatric Neurology</i> , 2021, 114, 16-20.	1.0	7
88	Clinical and radiographic delineation of Bent Bone Dysplasia-FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angel-shaped Phalanges. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2652-2661.	0.7	6
89	A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis. <i>Journal of General Internal Medicine</i> , 2019, 34, 1058-1062.	1.3	6
90	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2021, 13, 53.	1.5	6

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91	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 361-372.	2.6	6
92	Change in Prevalence of Orofacial Clefts in California between 1987 and 2010. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1910-1916.	0.7	5
93	46,XY disorders of sex development and congenital diaphragmatic hernia: A case with dysmorphic facies, truncus arteriosus, bifid thymus, gut malrotation, rhizomelia, and adactyly. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1360-1364.	0.7	4
94	A recurrent fibrillin-1 mutation in severe early onset Marfan syndrome. <i>Journal of Pediatric Genetics</i> , 2015, 03, 157-162.	0.3	4
95	Teaching Biochemistry and Genetics to Students of Medicine, Pharmacy, and Dentistry. <i>Medical Science Educator</i> , 2017, 27, 855-859.	0.7	4
96	Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period. <i>Journal of Ultrasound in Medicine</i> , 2016, 35, 1353-1358.	0.8	3
97	Identification of a novel mutation in the APTX gene associated with ataxia-oculomotor apraxia. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002014.	0.5	3
98	Candidate variants in TUB are associated with familial tremor. <i>PLoS Genetics</i> , 2020, 16, e1009010.	1.5	3
99	Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, CIRCGEN121003591.	1.6	3
100	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. <i>Genetics in Medicine</i> , 2021, 23, 661-668.	1.1	2
101	RNase G complementation of rne null mutation identifies functional interrelationships with RNase E in <i>E. coli</i> . <i>Molecular Microbiology</i> , 2002, 46, 295-295.	1.2	1
102	InpherNet accelerates monogenic disease diagnosis using patients' candidate genes' neighbors. <i>Genetics in Medicine</i> , 2021, 23, 1984-1992.	1.1	1