Jonathan A Bernstein

List of Publications by Year
in descending order

102 papers	8,958 citations	94415 37 h-index	48312 88 g-index
104 all docs	104 docs citations	104 times ranked	16790 citing authors

#	Article	IF	Citations
1	Strong evidence for genotypeâ€“phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. Human Molecular Genetics, 2022, 31, 625-637.	2.9	32
2	Variable clinical severity in <scp>TANGO2</scp> deficiency: Case series and literature review. American Journal of Medical Genetics, Part A, 2022, 188, 473-487.	1.2	17
3	Perceived utility and disutility of genomic sequencing for pediatric patients: Perspectives from parents with diverse sociodemographic characteristics. American Journal of Medical Genetics, Part A, 2022, 188, 1088-1101.	1.2	20
4	NSD1 mutations deregulate transcription and DNA methylation of bivalent developmental genes in Sotos syndrome. Human Molecular Genetics, 2022, 31, 2164-2184.	2.9	11
5	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
6	Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. New England Journal of Medicine, 2022, 386, 700-702.	27.0	116
7	Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003591.	3.6	3
8	TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION. Journal of Physical Education and Sports Management, 2022, , mcs.a006198.	1.2	9
9	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. Nature Biotechnology, 2022, 40, 1035-1041.	17.5	45
10	Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons. Biological Psychiatry, 2021, 89, 497-509.	1.3	17
11	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. Pediatric Neurology, 2021, 114, 16-20.	2.1	7
12	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
13	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
14	â€œIt seems like COVID-19 now is the only disease present on Earthâ€: living with a rare or undiagnosed disease during the COVID-19 pandemic. Genetics in Medicine, 2021, 23, 837-844.	2.4	19
15	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
16	â€œDoctors can read about it, they can know about it, but they've never lived with itâ€: How parents use social media throughout the diagnostic odyssey. Journal of Genetic Counseling, 2021, 30, 1707-1718.	1.6	10
17	Functional and structural analyses of novel Smith-Kingsmore Syndrome-Associated MTOR variants reveal potential new mechanisms and predictors of pathogenicity. PLoS Genetics, 2021, 17, e1009651.	3.5	9
18	InpherNet accelerates monogenic disease diagnosis using patientsâ€™ candidate genesâ€™ neighbors. Genetics in Medicine, 2021, 23, 1984-1992.	2.4	1

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19	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2021, 148, 585-598.	2.9	20
20	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 53.	3.1	6
21	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370.	2.4	24
22	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. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
23	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	3.6	24
24	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
25	Candidate variants in TUB are associated with familial tremor. PLoS Genetics, 2020, 16, e1009010.	3.5	3
26	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60
27	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	2.4	47
28	Psychometric Study of the Social Responsiveness Scale in Phelanâ€“McDermid Syndrome. Autism Research, 2020, 13, 1383-1396.	3.8	14
29	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
30	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. Pediatric Neurology, 2020, 106, 24-31.	2.1	9
31	Neuronal defects in a human cellular model of 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1888-1898.	30.7	113
32	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
33	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. Genetics in Medicine, 2019, 21, 464-470.	2.4	33
34	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. Journal of Genetic Counseling, 2019, 28, 1107-1118.	1.6	42
35	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
36	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	30.7	221

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37	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38
38	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	1.2	18
39	A Patient with Sjogrenâ€™s Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis. Journal of General Internal Medicine, 2019, 34, 1058-1062.	2.6	6
40	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. American Journal of Medical Genetics, Part A, 2019, 179, 966-977.	1.2	20
41	A toolkit for genetics providers in followâ€up of patients with nonâ€diagnostic exome sequencing. Journal of Genetic Counseling, 2019, 28, 213-228.	1.6	11
42	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	21.4	56
43	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. Genetics in Medicine, 2019, 21, 1585-1593.	2.4	67
44	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. Pediatric Neurology, 2019, 90, 37-43.	2.1	19
45	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
46	Biallelic lossâ€ofâ€function WNT5A mutations in an infant with severe and atypical manifestations of Robinow syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1030-1036.	1.2	15
47	Exploring the Medical and Psychosocial Concerns of Adolescents and Young Adults With Craniofacial Microsomia. Cleft Palate-Craniofacial Journal, 2018, 55, 1430-1439.	0.9	21
48	Prenatal treatment of ornithine transcarbamylase deficiency. Molecular Genetics and Metabolism, 2018, 123, 297-300.	1.1	12
49	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
50	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. Journal of Pediatrics, 2018, 196, 291-297.e2.	1.8	15
51	New insights into mitral valve dystrophy: a Filamin-A genotypeâ€“phenotype and outcome study. European Heart Journal, 2018, 39, 1269-1277.	2.2	44
52	WISP3 mutation associated with pseudorheumatoid dysplasia. Journal of Physical Education and Sports Management, 2018, 4, a001990.	1.2	16
53	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	8.1	35
54	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. American Journal of Human Genetics, 2018, 103, 1009-1021.	6.2	57

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55	High-frequency actionable pathogenic exome variants in an average-risk cohort. Journal of Physical Education and Sports Management, 2018, 4, a003178.	1.2	23
56	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
57	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
58	Change in Prevalence of Orofacial Clefts in California between 1987 and 2010. American Journal of Medical Genetics, Part A, 2018, 176, 1910-1916.	1.2	5
59	An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease. European Journal of Human Genetics, 2018, 26, 1810-1818.	2.8	15
60	Association of AHSG with alopecia and mental retardation (APMR) syndrome. Human Genetics, 2017, 136, 287-296.	3.8	14
61	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
62	Inhibiting Plasma Kallikrein for Hereditary Angioedema Prophylaxis. New England Journal of Medicine, 2017, 376, 717-728.	27.0	138
63	De novo and rare mutations in the HSPA1L heat shock gene associated with inflammatory bowel disease. Genome Medicine, 2017, 9, 8.	8.2	27
64	Assembly of functionally integrated human forebrain spheroids. Nature, 2017, 545, 54-59.	27.8	931
65	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
66	Functional analysis of novelDEAF1variants identified through clinical exome sequencing expandsDEAF1-associated neurodevelopmental disorder (DAND) phenotype. Human Mutation, 2017, 38, 1774-1785.	2.5	20
67	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
68	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
69	Identification of a novel mutation in the APTX gene associated with ataxia-oculomotor apraxia. Journal of Physical Education and Sports Management, 2017, 3, a002014.	1.2	3
70	Isolated Congenital Anosmia and CNGA2 Mutation. Scientific Reports, 2017, 7, 2667.	3.3	14
71	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	1.2	34
72	Teaching Biochemistry and Genetics to Students of Medicine, Pharmacy, and Dentistry. Medical Science Educator, 2017, 27, 855-859.	1.5	4

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73	Sleep Disturbances in Individuals With Phelan-McDermid Syndrome: Correlation With Caregiversâ€™ Sleep Quality and Daytime Functioning. Sleep, 2017, 40, .	1.1	21
74	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
75	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. Genetics in Medicine, 2017, 19, 209-214.	2.4	261
76	Clinical and radiographic delineation of Bent Bone Dysplasiaâ€FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angelâ€shaped Phalanges. American Journal of Medical Genetics, Part A, 2016, 170, 2652-2661.	1.2	6
77	Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period. Journal of Ultrasound in Medicine, 2016, 35, 1353-1358.	1.7	3
78	RASA1 somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1450-1454.	1.2	85
79	Respiratory system involvement in Costello syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1849-1857.	1.2	11
80	Impaired Healthâ€Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia. Journal of Genetic Counseling, 2016, 25, 936-944.	1.6	22
81	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
82	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
83	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	21.4	654
84	Clinical Course of Six Children With GNAO1 Mutations Causing aÂSevere and Distinctive Movement Disorder. Pediatric Neurology, 2016, 59, 81-84.	2.1	78
85	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. American Journal of Medical Genetics, Part A, 2016, 170, 870-880.	1.2	31
86	Clinical delineation of the PACS1â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
87	A deleterious Nav1.1 mutation selectively impairs telencephalic inhibitory neurons derived from Dravet Syndrome patients. ELife, 2016, 5, .	6.0	101
88	46,XY disorders of sex development and congenital diaphragmatic hernia: A case with dysmorphic facies, truncus arteriosus, bifid thymus, gut malrotation, rhizomelia, and adactyly. American Journal of Medical Genetics, Part A, 2015, 167, 1360-1364.	1.2	4
89	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)â€“associated inflammatory diseases. Journal of Allergy and Clinical Immunology, 2015, 136, 1337-1345.	2.9	103
90	Cold-aggravated pain in humans caused by a hyperactive NaV1.9 channel mutant. Nature Communications, 2015, 6, 10049.	12.8	71

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91	Factors Associated with Uptake of Genetics Services for Hypertrophic Cardiomyopathy. Journal of Genetic Counseling, 2015, 24, 797-809.	1.6	12
92	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101
93	A recurrent fibrillin-1 mutation in severe early onset Marfan syndrome. Journal of Pediatric Genetics, 2015, 03, 157-162.	0.7	4
94	Clinical Interpretation and Implications of Whole-Genome Sequencing. JAMA - Journal of the American Medical Association, 2014, 311, 1035.	7.4	398
95	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
96	Inappropriate p53 activation during development induces features of CHARGE syndrome. Nature, 2014, 514, 228-232.	27.8	117
97	Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. Nature Medicine, 2011, 17, 1657-1662.	30.7	521
98	Using induced pluripotent stem cells to investigate cardiac phenotypes in Timothy syndrome. Nature, 2011, 471, 230-234.	27.8	624
99	Familial cardiac valvulopathy due to filamin A mutation. American Journal of Medical Genetics, Part A, 2011, 155, 2236-2241.	1.2	33
100	Global analysis of Escherichia coli RNA degradosome function using DNA microarrays. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 2758-2763.	7.1	199
101	Global analysis of mRNA decay and abundance in Escherichia coli at single-gene resolution using two-color fluorescent DNA microarrays. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 9697-9702.	7.1	782
102	RNase G complementation of rne null mutation identifies functional interrelationships with RNase E in E. coli. Molecular Microbiology, 2002, 46, 295-295.	2.5	1