Jonathan A Bernstein

List of Publications by Year in Descending Order

99 papers	5,939 citations	30 h-index	76 g-index
104 ext. papers	7,681 ext. citations	11.8 avg, IF	5.24 L-index

#	Paper	IF	Citations
99	NSD1 mutations deregulate transcription and DNA methylation of bivalent developmental genes in Sotos syndrome.. *Human Molecular Genetics*, **2022**,	5.6	1
98	Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.. *New England Journal of Medicine*, **2022**,	59.2	10
97	Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.. *Circulation Genomic and Precision Medicine*, **2022**, CIRCGEN121003591	5.2	1
96	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**,	2.5	2
95	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.. *Nature Biotechnology*, **2022**,	44.5	4
94	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 53	4.6	0
93	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1649-1665	2.5	0
92	"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. *Journal of Genetic Counseling*, **2021**, 30, 1707-1718	2.5	1
91	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	6	1
90	InpherNet accelerates monogenic disease diagnosis using patients candidate genes neighbors. *Genetics in Medicine*, **2021**, 23, 1984-1992	8.1	0
89	Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons. *Biological Psychiatry*, **2021**, 89, 497-509	7.9	5
88	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	2.9	2
87	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. *Genetics in Medicine*, **2021**, 23, 259-271	8.1	6
86	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. *Genetics in Medicine*, **2021**, 23, 661-668	8.1	1
85	"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. *Genetics in Medicine*, **2021**, 23, 837-844	8.1	5
84	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	11.5	5
83	Strong evidence for genotype-phenotype correlations in Phelan-McDermid syndrome: Results from the developmental Synaptopathies consortium. *Human Molecular Genetics*, **2021**,	5.6	2

82	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. *Science Translational Medicine*, **2020**, 12,	17.5	11
81	Automated syndrome diagnosis by three-dimensional facial imaging. *Genetics in Medicine*, **2020**, 22, 1682-1693	8.1	17
80	Psychometric Study of the Social Responsiveness Scale in Phelan-McDermid Syndrome. *Autism Research*, **2020**, 13, 1383-1396	5.1	4
79	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. *American Journal of Human Genetics*, **2020**, 106, 570-583	11	21
78	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. *Pediatric Neurology*, **2020**, 106, 24-31	2.9	4
77	Neuronal defects in a human cellular model of 22q11.2 deletion syndrome. *Nature Medicine*, **2020**, 26, 1888-1898	50.5	27
76	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	14.3	22
75	Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1333-1348	5.4	10
74	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. *Genetics in Medicine*, **2020**, 22, 1863-1873	8.1	9
73	Candidate variants in TUB are associated with familial tremor. *PLoS Genetics*, **2020**, 16, e1009010	6	1
72	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. *Genetics in Medicine*, **2020**, 22, 362-370	8.1	12
71	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
70	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. *Nature Medicine*, **2019**, 25, 911-919	50.5	116
69	Mutation update for the SATB2 gene. *Human Mutation*, **2019**, 40, 1013-1029	4.7	22
68	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic splice-site and Charcot-Marie-Tooth phenotype with early onset symptoms. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00676	2.3	11
67	A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis. *Journal of General Internal Medicine*, **2019**, 34, 1058-1062	4	3
66	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. *American Journal of Medical Genetics, Part A*, **2019**, 179, 966-977	2.5	10
65	A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. *Journal of Genetic Counseling*, **2019**, 28, 213-228	2.5	4

64	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. *Nature Genetics*, **2019**, 51, 755-763	36.3	25
63	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
62	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. *Genetics in Medicine*, **2019**, 21, 464-470	8.1	12
61	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. *Journal of Genetic Counseling*, **2019**, 28, 1107-1118	2.5	20
60	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. *Genetics in Medicine*, **2019**, 21, 1585-1593	8.1	32
59	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. *Pediatric Neurology*, **2019**, 90, 37-43	2.9	14
58	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. *American Journal of Human Genetics*, **2018**, 102, 494-504	11	44
57	Biallelic loss-of-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	2.5	8
56	Exploring the Medical and Psychosocial Concerns of Adolescents and Young Adults With Craniofacial Microsomia: A Qualitative Study. *Cleft Palate-Craniofacial Journal*, **2018**, 55, 1430-1439	1.9	12
55	Prenatal treatment of ornithine transcarbamylase deficiency. *Molecular Genetics and Metabolism*, **2018**, 123, 297-300	3.7	8
54	Genotype-phenotype correlations in individuals with pathogenic RERE variants. *Human Mutation*, **2018**, 39, 666-675	4.7	19
53	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. *Journal of Pediatrics*, **2018**, 196, 291-297.e2	3.6	12
52	New insights into mitral valve dystrophy: a Filamin-A genotype-phenotype and outcome study. *European Heart Journal*, **2018**, 39, 1269-1277	9.5	26
51	Change in Prevalence of Orofacial Clefts in California between 1987 and 2010. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1910-1916	2.5	2
50	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	5.3	10
49	mutation associated with pseudorheumatoid dysplasia. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	10
48	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. *Neuron*, **2018**, 100, 1354-1368.e5	13.9	20
47	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	11	30

46	High-frequency actionable pathogenic exome variants in an average-risk cohort. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	15
45	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. *New England Journal of Medicine*, **2018**, 379, 2131-2139	59.2	129
44	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. *American Journal of Human Genetics*, **2018**, 103, 154-162	11	25
43	Association of AHSG with alopecia and mental retardation (APMR) syndrome. *Human Genetics*, **2017** , 136, 287-296	6.3	8
42	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
41	Inhibiting Plasma Kallikrein for Hereditary Angioedema Prophylaxis. *New England Journal of Medicine*, **2017**, 376, 717-728	59.2	99
40	De novo and rare mutations in the HSPA1L heat shock gene associated with inflammatory bowel disease. *Genome Medicine*, **2017**, 9, 8	14.4	22
39	Assembly of functionally integrated human forebrain spheroids. *Nature*, **2017**, 545, 54-59	50.4	604
38	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
37	Functional analysis of novel DEAF1 variants identified through clinical exome sequencing expands DEAF1-associated neurodevelopmental disorder (DAND) phenotype. *Human Mutation*, **2017**, 38, 1774-1785	4.7	15
36	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	11	37
35	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. *Brain*, **2017**, 140, 2610-2622	11.2	63
34	Identification of a novel mutation in the gene associated with ataxia-oculomotor apraxia. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	1
33	Isolated Congenital Anosmia and CNGA2 Mutation. *Scientific Reports*, **2017**, 7, 2667	4.9	8
32	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2680-2689	2.5	23
31	Teaching Biochemistry and Genetics to Students of Dentistry, Medicine, and Pharmacy 6 International Conference of the Association of Biochemistry Educators (ABE) Clearwater Beach, FL, USA, May 7-11, 2017. *Medical Science Educator*, **2017**, 27, 855-859	0.7	4
30	Sleep Disturbances in Individuals With Phelan-McDermid Syndrome: Correlation With CaregiversT Sleep Quality and Daytime Functioning. *Sleep*, **2017**, 40,	1.1	9
29	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45	36.3	172

28	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. *Genetics in Medicine*, **2017**, 19, 209-214	8.1	161
27	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	11	68
26	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. *Nature Genetics*, **2016**, 48, 1581-1586	36.3	423
25	Clinical Course of Six Children With GNAO1 Mutations Causing a Severe and Distinctive Movement Disorder. *Pediatric Neurology*, **2016**, 59, 81-4	2.9	59
24	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**, 170A, 870-80	2.5	21
23	Clinical delineation of the PACS1-related syndrome--Report on 19 patients. *American Journal of Medical Genetics, Part A*, **2016**, 170, 670-5	2.5	28
22	A deleterious Nav1.1 mutation selectively impairs telencephalic inhibitory neurons derived from Dravet Syndrome patients. *ELife*, **2016**, 5,	8.9	79
21	Clinical and radiographic delineation of Bent Bone Dysplasia-FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angel-shaped Phalanges. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2652-61	2.5	6
20	Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period. *Journal of Ultrasound in Medicine*, **2016**, 35, 1353-8	2.9	3
19	RASA1 somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1450-4	2.5	68
18	Respiratory system involvement in Costello syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1849-57	2.5	6
17	Impaired Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia. *Journal of Genetic Counseling*, **2016**, 25, 936-44	2.5	12
16	Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurology*, **2016**, 73, 836-845	17.2	166
15	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. *European Journal of Human Genetics*, **2015**, 23, 1473-81	5.3	63
14	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, 1360-4	2.5	2
13	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1337-45	11.5	73
12	Cold-aggravated pain in humans caused by a hyperactive NaV1.9 channel mutant. *Nature Communications*, **2015**, 6, 10049	17.4	52
11	Factors Associated with Uptake of Genetics Services for Hypertrophic Cardiomyopathy. *Journal of Genetic Counseling*, **2015**, 24, 797-809	2.5	9

10	A recurrent fibrillin-1 mutation in severe early onset Marfan syndrome. *Journal of Pediatric Genetics* , **2014**, 3, 157-62	0.7	3
9	Clinical interpretation and implications of whole-genome sequencing. *JAMA - Journal of the American Medical Association*, **2014**, 311, 1035-45	27.4	333
8	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway. *Genetics in Medicine*, **2014**, 16, 751-8	8.1	138
7	Inappropriate p53 activation during development induces features of CHARGE syndrome. *Nature*, **2014**, 514, 228-32	50.4	101
6	Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. *Nature Medicine*, **2011**, 17, 1657-62	50.5	451
5	Using induced pluripotent stem cells to investigate cardiac phenotypes in Timothy syndrome. *Nature*, **2011**, 471, 230-4	50.4	538
4	Familial cardiac valvulopathy due to filamin A mutation. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2236-41	2.5	28
3	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, 2758-63	11.5	178
2	RNase G complementation of rne null mutation identifies functional interrelationships with RNase E in E. coli. *Molecular Microbiology*, **2002**, 46, 295-295	4.1	1
1	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, 9697-702	11.5	652