Michael J Bamshad

List of Publications by Year
in descending order

121 papers	10,394 citations	70961 41 h-index	40881 93 g-index
124 all docs	124 docs citations	124 times ranked	21356 citing authors

#	Article	IF	Citations
1	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. Journal of Genetic Counseling, 2022, 31, 326-337.	0.9	1
2	Comparing encounter-based and annualized chronic pseudomonas infection definitions in cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 40-44.	0.3	3
3	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. Journal of Cystic Fibrosis, 2022, 21, 463-470.	0.3	13
4	Leveraging TOPMed imputation server and constructing a cohort-specific imputation reference panel to enhance genotype imputation among cystic fibrosis patients. Human Genetics and Genomics Advances, 2022, 3, 100090.	1.0	6
5	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
6	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.0	5
7	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
8	Club cell secretory protein and lung function in children with cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 811-820.	0.3	8
9	Exome sequencing identifies variants in infants with sacral agenesis. Birth Defects Research, 2022, 114, 215-227.	0.8	2
10	Expanding <scp>ACTA2</scp> genotypes with corresponding phenotypes overlapping with smooth muscle dysfunction syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2389-2396.	0.7	5
11	Exome-wide rare variant analysis in familial essential tremor. Parkinsonism and Related Disorders, 2021, 82, 109-116.	1.1	11
12	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. Human Genetics and Genomics Advances, 2021, 2, 100016.	1.0	7
13	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
14	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
15	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	0.7	5
16	Germline SAMD9L truncation variants trigger global translational repression. Journal of Experimental Medicine, 2021, 218, .	4.2	20
17	Expanding the phenotype, genotype and biochemical knowledge of <scp>ALG3â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 987-1000.	1.7	4
18	Deletion of CTCF sites in the SHH locus alters enhancerâ€“promoter interactions and leads to acheiropodia. Nature Communications, 2021, 12, 2282.	5.8	37

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19	A mutation in SLC37A4 causes a dominantly inherited congenital disorder of glycosylation characterized by liver dysfunction. American Journal of Human Genetics, 2021, 108, 1040-1052.	2.6	7
20	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	2.6	105
21	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	4.5	33
22	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
23	The pleiotropy associated with de novo variants in CHD4, CNOT3, and SETD5 extends to moyamoya angiopathy. Genetics in Medicine, 2020, 22, 427-431.	1.1	34
24	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
25	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
26	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.	1.7	24
27	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0
28	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	2.6	21
29	Rare deleterious variants of NOTCH1, GATA4, SMAD6, and ROBO4 are enriched in BAV with early onset complications but not in BAV with heritable thoracic aortic disease. Molecular Genetics & Genomic Medicine, 2020, 8, e1406.	0.6	17
30	Multiplexed Functional Assessment of Genetic Variants in CARD11. American Journal of Human Genetics, 2020, 107, 1029-1043.	2.6	38
31	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
32	Mutations in GET4 disrupt the transmembrane domain recognition complex pathway. Journal of Inherited Metabolic Disease, 2020, 43, 1037-1045.	1.7	4
33	8q24 genetic variation and comprehensive haplotypes altering familial risk of prostate cancer. Nature Communications, 2020, 11, 1523.	5.8	10
34	SOS1 Gain-of-Function Variants in Dilated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e002892.	1.6	10
35	Genetic Ancestry Testing. JAMA - Journal of the American Medical Association, 2020, 323, 1089.	3.8	15
36	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	3.9	43

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37	Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. Birth Defects Research, 2019, 111, 1618-1632.	0.8	9
38	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
39	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	2.6	61
40	Mendelian Gene Discovery: Fast and Furious with No End in Sight. American Journal of Human Genetics, 2019, 105, 448-455.	2.6	166
41	Mutations in GDF11 and the extracellular antagonist, Follistatin, as a likely cause of Mendelian forms of orofacial clefting in humans. Human Mutation, 2019, 40, 1813-1825.	1.1	26
42	Mutations in the transloconâ€associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2019, 42, 993-997.	1.7	18
43	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
44	Front Cover, Volume 40, Issue 10. Human Mutation, 2019, 40, i.	1.1	0
45	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
46	Loss of function, missense, and intronic variants in NOTCH1 confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts. Genetic Epidemiology, 2019, 43, 215-226.	0.6	25
47	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	2.6	81
48	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. Genetics in Medicine, 2019, 21, 1611-1620.	1.1	88
49	A presynaptic congenital myasthenic syndrome attributed to a homozygous sequence variant in LAMA5. Annals of the New York Academy of Sciences, 2018, 1413, 119-125.	1.8	7
50	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
51	Genetic counselors on the frontline of precision health. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 5-9.	0.7	16
52	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
53	Complex signatures of natural selection at GYPA. Human Genetics, 2018, 137, 151-160.	1.8	20
54	Mutations in the fourth Î²-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. Human Mutation, 2018, 39, 811-815.	1.1	17

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55	ERCC4 variants identified in a cohort of patients with segmental progeroid syndromes. Human Mutation, 2018, 39, 255-265.	1.1	23
56	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	1.5	79
57	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.	2.6	57
58	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. American Journal of Human Genetics, 2018, 103, 968-975.	2.6	43
59	A content analysis of the views of genetics professionals on race, ancestry, and genetics. AJOB Empirical Bioethics, 2018, 9, 222-234.	0.8	22
60	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. American Journal of Human Genetics, 2018, 102, 1143-1157.	2.6	94
61	Whole exome sequencing analysis in severe chronic obstructive pulmonary disease. Human Molecular Genetics, 2018, 27, 3801-3812.	1.4	32
62	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. Mitochondrion, 2017, 34, 84-90.	1.6	21
63	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
64	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	2.6	74
65	Presynaptic congenital myasthenic syndrome with a homozygous sequence variant in LAMA5 combines myopia, facial tics, and failure of neuromuscular transmission. American Journal of Medical Genetics, Part A, 2017, 173, 2240-2245.	0.7	29
66	Sequencing of sporadic Attentionâ€Deficit Hyperactivity Disorder (ADHD) identifies novel and potentially pathogenic de novo variants and excludes overlap with genes associated with autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 381-389.	1.1	44
67	Survival beyond the perinatal period expands the phenotypes caused by mutations in GLE1. American Journal of Medical Genetics, Part A, 2017, 173, 3098-3103.	0.7	10
68	The Epithelial Sodium Channel Is a Modifier of the Long-Term Nonprogressive Phenotype Associated with F508del CFTR Mutations. American Journal of Respiratory Cell and Molecular Biology, 2017, 57, 711-720.	1.4	27
69	A Qualitative Analysis of How Anthropologists Interpret the Race Construct. American Anthropologist, 2017, 119, 422-434.	0.7	25
70	My46: a Web-based tool for self-guided management of genomic test results in research and clinical settings. Genetics in Medicine, 2017, 19, 467-475.	1.1	30
71	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. Genes, 2017, 8, 381.	1.0	19
72	Whole genome sequencing of extreme phenotypes identifies variants in CD101 and UBE2V1 associated with increased risk of sexually acquired HIV-1. PLoS Pathogens, 2017, 13, e1006703.	2.1	16

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73	A second family with CATSHL syndrome: Confirmatory report of another unique FGFR3 syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1908-1911.	0.7	15
74	Elevated plasma dihydroorotate in Miller syndrome: Biochemical, diagnostic and clinical implications, and treatment with uridine. Molecular Genetics and Metabolism, 2016, 119, 83-90.	0.5	15
75	Contractile properties of developing human fetal cardiac muscle. Journal of Physiology, 2016, 594, 437-452.	1.3	63
76	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	2.6	100
77	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	2.6	43
78	Guidelines for Large-Scale Sequence-Based Complex Trait Association Studies: Lessons Learned from the NHLBI Exome Sequencing Project. American Journal of Human Genetics, 2016, 99, 791-801.	2.6	79
79	Development of a subset of forelimb muscles and their attachment sites requires the ulnar-mammary syndrome geneTbx3. DMM Disease Models and Mechanisms, 2016, 9, 1257-1269.	1.2	38
80	An inactivating mutation in intestinal cell kinase,ICK, impairs hedgehog signalling and causes short rib-polydactyly syndrome. Human Molecular Genetics, 2016, 25, 3998-4011.	1.4	44
81	Use of metaphors about exome and whole genome sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 1127-1133.	0.7	4
82	The Simons Genome Diversity Project: 300 genomes from 142 diverse populations. Nature, 2016, 538, 201-206.	13.7	1,216
83	A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. Nature Communications, 2016, 7, 12522.	5.8	136
84	Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features. Genetics in Medicine, 2016, 18, 788-795.	1.1	88
85	Identification of Rare Variants in ATP8B4 as a Risk Factor for Systemic Sclerosis by Wholeâ€Exome Sequencing. Arthritis and Rheumatology, 2016, 68, 191-200.	2.9	32
86	Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. American Journal of Human Genetics, 2016, 98, 58-74.	2.6	248
87	Exome Sequencing Analysis in Severe, Early-Onset Chronic Obstructive Pulmonary Disease. American Journal of Respiratory and Critical Care Medicine, 2016, 193, 1353-1363.	2.5	46
88	Developments in our understanding of the genetic basis of birth defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 680-691.	1.6	30
89	Mitochondrial DNA Copy Number in Sleep Duration Discordant Monozygotic Twins. Sleep, 2015, 38, 1655-1658.	0.6	8
90	Rare Variation Facilitates Inferences of Fine-Scale Population Structure in Humans. Molecular Biology and Evolution, 2015, 32, 653-660.	3.5	38

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91	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473.	2.6	124
92	Estimates of Continental Ancestry Vary Widely among Individuals with the Same mtDNA Haplogroup. American Journal of Human Genetics, 2015, 96, 183-193.	2.6	40
93	Global diversity, population stratification, and selection of human copy-number variation. Science, 2015, 349, aab3761.	6.0	293
94	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
95	Speech and language in a genotyped cohort of individuals with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1483-1492.	0.7	33
96	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
97	The embryonic myosin R672C mutation that underlies Freeman-Sheldon syndrome impairs cross-bridge detachment and cycling in adult skeletal muscle. Human Molecular Genetics, 2015, 24, 3348-3358.	1.4	47
98	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	2.4	313
99	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	13.7	581
100	Exome Sequencing of Phenotypic Extremes Identifies CAV2 and TMC6 as Interacting Modifiers of Chronic Pseudomonas aeruginosa Infection in Cystic Fibrosis. PLoS Genetics, 2015, 11, e1005273.	1.5	39
101	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
102	Solving Glycosylation Disorders: Fundamental Approaches Reveal Complicated Pathways. American Journal of Human Genetics, 2014, 94, 161-175.	2.6	222
103	Characteristics of Neutral and Deleterious Protein-Coding Variation among Individuals and Populations. American Journal of Human Genetics, 2014, 95, 421-436.	2.6	89
104	Genotypeâ€phenotype relationships in Freemanâ€“Sheldon syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2808-2813.	0.7	45
105	Attitudes of Genetics Professionals Toward the Return of Incidental Results from Exome and Whole-Genome Sequencing. American Journal of Human Genetics, 2014, 95, 77-84.	2.6	109
106	Pathogenic Variants for Mendelian and Complex Traits in Exomes of 6,517 European and African Americans: Implications for the Return of Incidental Results. American Journal of Human Genetics, 2014, 95, 183-193.	2.6	78
107	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	2.6	171
108	A Non-Active-Site SET Domain Surface Crucial for the Interaction of MLL1 and the RbBP5/Ash2L Heterodimer within MLL Family Core Complexes. Journal of Molecular Biology, 2014, 426, 2283-2299.	2.0	46

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109	Attitudes of African Americans Toward Return of Results From Exome and Whole Genome Sequencing. , 2013, 161, n/a-n/a.		1
110	Practices and Policies of Clinical Exome Sequencing Providers: Analysis and Implications. , 2013, 161, n/a-n/a.		2
111	Kabuki syndrome missense mutations disrupt the formation and histone methyltransferase activity of the MLL2 core complex. FASEB Journal, 2013, 27, 772.1.	0.2	0
112	Unanticipated results from exome sequencing/whole genome sequencing: The sky won't fall. American Journal of Medical Genetics, Part A, 2012, 158A, 2643-2644.	0.7	2
113	The Centers for Mendelian Genomics: A new largeâ€scale initiative to identify the genes underlying rare Mendelian conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 1523-1525.	0.7	110
114	Exome sequencing as a tool for Mendelian disease gene discovery. Nature Reviews Genetics, 2011, 12, 745-755.	7.7	1,484
115	Arthrogryposis: A Review and Update. Journal of Bone and Joint Surgery - Series A, 2009, 91, 40-46.	1.4	311
116	Lost in translation: Meaningful policies for writing about genetics and race. American Journal of Medical Genetics, Part A, 2007, 143A, 971-972.	0.7	4
117	Clinical Characteristics and Natural History of Freeman-Sheldon Syndrome. Pediatrics, 2006, 117, 754-762.	1.0	104
118	Mutations in embryonic myosin heavy chain (MYH3) cause Freeman-Sheldon syndrome and Sheldon-Hall syndrome. Nature Genetics, 2006, 38, 561-565.	9.4	233
119	Clinical analysis of a variant of Freeman-Sheldon syndrome (DA2B). , 1998, 76, 93-98.		54
120	A revised and extended classification of the distal arthrogryposes. , 1996, 65, 277-281.		209
121	Distal arthrogryposis type 1: Clinical analysis of a large kindred. , 1996, 65, 282-285.		36