Michael J Bamshad

List of Publications by Year in Descending Order

116 papers	7,290 citations	36 h-index	84 g-index
124 ext. papers	9,196 ext. citations	8.7 avg, IF	5.4 L-index

#	Paper	IF	Citations
116	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	0.8	
115	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
114	Variants in cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology.. *Human Genetics and Genomics Advances*, **2022**, 3, 100102	0.8	0
113	Variant-level matching for diagnosis and discovery: challenges and opportunities.. *Human Mutation*, **2022**,	4.7	1
112	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. *Journal of Cystic Fibrosis*, **2021**,	4.1	2
111	Further delineation of van den Ende-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	2.5	0
110	Germline SAMD9L truncation variants trigger global translational repression. *Journal of Experimental Medicine*, **2021**, 218,	16.6	6
109	Expanding the phenotype, genotype and biochemical knowledge of ALG3-CDG. *Journal of Inherited Metabolic Disease*, **2021**, 44, 987-1000	5.4	1
108	Deletion of CTCF sites in the SHH locus alters enhancer-promoter interactions and leads to acheiropodia. *Nature Communications*, **2021**, 12, 2282	17.4	8
107	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	11	2
106	Exome-wide rare variant analysis in familial essential tremor. *Parkinsonism and Related Disorders*, **2021**, 82, 109-116	3.6	5
105	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. *Human Genetics and Genomics Advances*, **2021**, 2, 100016-100016	0.8	1
104	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 119-133	2.5	6
103	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
102	Targeted long-read sequencing identifies missing disease-causing variation. *American Journal of Human Genetics*, **2021**, 108, 1436-1449	11	16
101	DIAPH1 Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. *JAMA Neurology*, **2021**, 78, 993-1003	17.2	7
100	Response to Hamosh et al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	

99	Multiplexed Functional Assessment of Genetic Variants in CARD11. *American Journal of Human Genetics*, **2020**, 107, 1029-1043	11	8
98	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1338-1347	8.1	9
97	Mutations in GET4 disrupt the transmembrane domain recognition complex pathway. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1037-1045	5.4	0
96	8q24 genetic variation and comprehensive haplotypes altering familial risk of prostate cancer. *Nature Communications*, **2020**, 11, 1523	17.4	4
95	Gain-of-Function Variants in Dilated Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2020**, 13, e002892	5.2	5
94	Genetic Ancestry Testing: What Is It and Why Is It Important?. *JAMA - Journal of the American Medical Association*, **2020**, 323, 1089-1090	27.4	3
93	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4423-4439	15.9	19
92	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. *Genetics in Medicine*, **2020**, 22, 538-546	8.1	14
91	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
90	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
89	Response to Hall et al. *American Journal of Human Genetics*, **2020**, 107, 1188-1189	11	
88	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. *American Journal of Human Genetics*, **2020**, 107, 293-310	11	6
87	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	2.3	5
86	The pleiotropy associated with de novo variants in CHD4, CNOT3, and SETD5 extends to moyamoya angiopathy. *Genetics in Medicine*, **2020**, 22, 427-431	8.1	10
85	Redefining the Etiologic Landscape of Cerebellar Malformations. *American Journal of Human Genetics*, **2019**, 105, 606-615	11	40
84	Mendelian Gene Discovery: Fast and Furious with No End in Sight. *American Journal of Human Genetics*, **2019**, 105, 448-455	11	73
83	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	4.7	14
82	Mutations in the translocon-associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. *Journal of Inherited Metabolic Disease*, **2019**, 42, 993-997	5.4	10

81	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	2.9	4
80	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. *American Journal of Human Genetics*, **2019**, 105, 302-316	11	19
79	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
78	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
77	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	2.6	12
76	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	11	47
75	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. *Genetics in Medicine*, **2019**, 21, 1611-1620	8.1	52
74	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	6.5	3
73	Plain-language medical vocabulary for precision diagnosis. *Nature Genetics*, **2018**, 50, 474-476	36.3	20
72	Genetic counselors on the frontline of precision health. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 5-9	3.1	7
71	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. *American Journal of Human Genetics*, **2018**, 102, 309-320	11	85
70	Complex signatures of natural selection at GYPA. *Human Genetics*, **2018**, 137, 151-160	6.3	7
69	Mutations in the fourth β-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. *Human Mutation*, **2018**, 39, 811-815	4.7	11
68	Whole exome sequencing analysis in severe chronic obstructive pulmonary disease. *Human Molecular Genetics*, **2018**, 27, 3801-3812	5.6	19
67	ERCC4 variants identified in a cohort of patients with segmental progeroid syndromes. *Human Mutation*, **2018**, 39, 255-265	4.7	17
66	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	6	47
65	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
64	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. *American Journal of Human Genetics*, **2018**, 103, 968-975	11	28

63	A content analysis of the views of genetics professionals on race, ancestry, and genetics. *AJOB Empirical Bioethics*, **2018**, 9, 222-234	3	11
62	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	11	52
61	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. *Mitochondrion*, **2017**, 34, 84-90	4.9	16
60	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
59	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	11	52
58	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	2.5	26
57	Sequencing of sporadic Attention-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	3.5	22
56	Survival beyond the perinatal period expands the phenotypes caused by mutations in GLE1. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3098-3103	2.5	8
55	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	5.7	22
54	A Qualitative Analysis of How Anthropologists Interpret the Race Construct. *American Anthropologist*, **2017**, 119, 422-434	1.5	20
53	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	8.1	23
52	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. *Genes*, **2017**, 8,	4.2	14
51	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	7.6	9
50	The Simons Genome Diversity Project: 300 genomes from 142 diverse populations. *Nature*, **2016**, 538, 201-206	50.4	759
49	A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. *Nature Communications*, **2016**, 7, 12522	17.4	90
48	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, 788-95	8.1	67
47	Identification of Rare Variants in ATP8B4 as a Risk Factor for Systemic Sclerosis by Whole-Exome Sequencing. *Arthritis and Rheumatology*, **2016**, 68, 191-200	9.5	28
46	Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. *American Journal of Human Genetics*, **2016**, 98, 58-74	11	189

45	Exome Sequencing Analysis in Severe, Early-Onset Chronic Obstructive Pulmonary Disease. *American Journal of Respiratory and Critical Care Medicine*, **2016**, 193, 1353-63	10.2	35
44	A second family with CATSHL syndrome: Confirmatory report of another unique FGFR3 syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1908-11	2.5	12
43	Elevated plasma dihydroorotate in Miller syndrome: Biochemical, diagnostic and clinical implications, and treatment with uridine. *Molecular Genetics and Metabolism*, **2016**, 119, 83-90	3.7	12
42	Contractile properties of developing human fetal cardiac muscle. *Journal of Physiology*, **2016**, 594, 437-53	5.9	34
41	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	11	70
40	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. *American Journal of Human Genetics*, **2016**, 98, 772-81	11	29
39	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	11	67
38	Development of a subset of forelimb muscles and their attachment sites requires the ulnar-mammary syndrome gene Tbx3. *DMM Disease Models and Mechanisms*, **2016**, 9, 1257-1269	4.1	24
37	An inactivating mutation in intestinal cell kinase, ICK, impairs hedgehog signalling and causes short rib-polydactyly syndrome. *Human Molecular Genetics*, **2016**, 25, 3998-4011	5.6	31
36	Use of metaphors about exome and whole genome sequencing. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1127-33	2.5	3
35	Estimates of continental ancestry vary widely among individuals with the same mtDNA haplogroup. *American Journal of Human Genetics*, **2015**, 96, 183-93	11	19
34	Global diversity, population stratification, and selection of human copy-number variation. *Science*, **2015**, 349, aab3761	33.3	224
33	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
32	Speech and language in a genotyped cohort of individuals with Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1483-92	2.5	27
31	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. *American Journal of Human Genetics*, **2015**, 96, 841-9	11	36
30	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, 3348-58	5.6	37
29	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. *Genome Research*, **2015**, 25, 305-15	9.7	252
28	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. *Nature*, **2015**, 518, 102-6	50.4	463

27	Developments in our understanding of the genetic basis of birth defects. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2015**, 103, 680-91		22
26	Mitochondrial DNA Copy Number in Sleep Duration Discordant Monozygotic Twins. *Sleep*, **2015**, 38, 1655-8	5.1	5
25	Rare variation facilitates inferences of fine-scale population structure in humans. *Molecular Biology and Evolution*, **2015**, 32, 653-60	8.3	31
24	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, 462-73	11	91
23	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	6	29
22	Characteristics of neutral and deleterious protein-coding variation among individuals and populations. *American Journal of Human Genetics*, **2014**, 95, 421-36	11	74
21	Genotype-phenotype relationships in Freeman-Sheldon syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2808-13	2.5	36
20	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	11	98
19	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, 183-93	11	68
18	Mutations in PIEZO2 cause Gordon syndrome, Marden-Walker syndrome, and distal arthrogryposis type 5. *American Journal of Human Genetics*, **2014**, 94, 734-44	11	124
17	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, 2283-99	6.5	34
16	RNF213 rare variants in an ethnically diverse population with Moyamoya disease. *Stroke*, **2014**, 45, 3200-7	6.7	103
15	Solving glycosylation disorders: fundamental approaches reveal complicated pathways. *American Journal of Human Genetics*, **2014**, 94, 161-75	11	187
14	Kabuki syndrome missense mutations disrupt the formation and histone methyltransferase activity of the MLL2 core complex. *FASEB Journal*, **2013**, 27, 772.1	0.9	
13	Attitudes of African Americans Toward Return of Results From Exome and Whole Genome Sequencing **2013**, 161, 1064		1
12	Practices and Policies of Clinical Exome Sequencing Providers: Analysis and Implications **2013**, 161, 935		2
11	Unanticipated results from exome sequencing/whole genome sequencing: The sky won't fall **2012**, 158A, 2643-2644		2
10	The Centers for Mendelian Genomics: a new large-scale initiative to identify the genes underlying rare Mendelian conditions. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1523-5	2.5	92

9	Exome sequencing as a tool for Mendelian disease gene discovery. *Nature Reviews Genetics*, **2011**, 12, 745-55	30.1	1265
8	Arthrogryposis: a review and update. *Journal of Bone and Joint Surgery - Series A*, **2009**, 91 Suppl 4, 40-6	5.6	232
7	Lost in translation: meaningful policies for writing about genetics and race. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 971-2	2.5	3
6	Clinical characteristics and natural history of Freeman-Sheldon syndrome. *Pediatrics*, **2006**, 117, 754-62	7.4	87
5	Mutations in embryonic myosin heavy chain (MYH3) cause Freeman-Sheldon syndrome and Sheldon-Hall syndrome. *Nature Genetics*, **2006**, 38, 561-5	36.3	196
4	Clinical analysis of a variant of Freeman-Sheldon syndrome (DA2B). *American Journal of Medical Genetics Part A*, **1998**, 76, 93-8		45
3	A revised and extended classification of the distal arthrogryposes. *American Journal of Medical Genetics Part A*, **1996**, 65, 277-81		171
2	Distal arthrogryposis type 1: clinical analysis of a large kindred. *American Journal of Medical Genetics Part A*, **1996**, 65, 282-5		28
1	Mutations inMYLPFcause a novel segmental amyoplasia that manifests as distal arthrogryposis		1