Daniel G Macarthur

List of Publications by Year in Descending Order

144 papers	40,287 citations	57 h-index	159 g-index
159 ext. papers	55,306 ext. citations	17.7 avg, IF	5.97 L-index

#	Paper	IF	Citations
144	Mitochondrial DNA variation across 56,434 individuals in gnomAD.. *Genome Research*, **2022**,	9.7	3
143	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
142	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation.. *Cell Stem Cell*, **2022**,	18	1
141	seqr: a web-based analysis and collaboration tool for rare disease genomics.. *Human Mutation*, **2022** ,	4.7	1
140	Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. *European Journal of Human Genetics*, **2021**, 29, 816-826	5.3	1
139	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. *Cell*, **2021**, 184, 2633-2648.e19	56.2	20
138	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications*, **2021**, 12, 3505	17.4	5
137	Leveraging supervised learning for functionally‐informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. *Nature Communications*, **2021**, 12, 3394	17.4	9
136	Pathogenic deep intronic MTM1 variant activates a pseudo-exon encoding a nonsense codon resulting in severe X-linked myotubular myopathy. *European Journal of Human Genetics*, **2021**, 29, 61-66	5.3	4
135	WGS and RNA Studies Diagnose Noncoding Variants in Males With High Creatine Kinase. *Neurology: Genetics*, **2021**, 7, e554	3.8	4
134	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. *Genetics in Medicine*, **2021**, 23, 881-887	8.1	1
133	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3
132	From variant to function in human disease genetics. *Science*, **2021**, 373, 1464-1468	33.3	11
131	Autosomal recessive variants in alter the ε-tubulin ring complex leading to neurodevelopmental disease. *IScience*, **2021**, 24, 101948	6.1	1
130	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. *American Journal of Human Genetics*, **2020**, 107, 727-742	11	2
129	Evaluating drug targets through human loss-of-function genetic variation. *Nature*, **2020**, 581, 459-464	50.4	53
128	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278

127	Characterising the loss-of-function impact of 5Quntranslated region variants in 15,708 individuals. *Nature Communications*, **2020**, 11, 2523	17.4	35
126	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. *Nature Communications*, **2020**, 11, 2539	17.4	51
125	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
124	Transcript expression-aware annotation improves rare variant interpretation. *Nature*, **2020**, 581, 452-458	50.4	55
123	The effect of LRRK2 loss-of-function variants in humans. *Nature Medicine*, **2020**, 26, 869-877	50.5	47
122	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. *Genetics in Medicine*, **2020**, 22, 1478-1488	8.1	25
121	Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients. *Neuromuscular Disorders*, **2020**, 30, 310-314	2.9	5
120	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. *Clinical Genetics*, **2020**, 97, 764-769	4	7
119	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. *ELife*, **2020**, 9,	8.9	19
118	Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. *International Journal of Epidemiology*, **2020**, 49, 20-21i	7.8	19
117	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. *Human Mutation*, **2020**, 41, 403-411	4.7	10
116	A brief history of human disease genetics. *Nature*, **2020**, 577, 179-189	50.4	181
115	A Quantitative Proteome Map of the Human Body. *Cell*, **2020**, 183, 269-283.e19	56.2	73
114	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. *European Journal of Human Genetics*, **2020**, 28, 383-387	5.3	4
113	Identification of a Novel Deep Intronic Mutation in CAPN3 Presenting a Promising Target for Therapeutic Splice Modulation. *Journal of Neuromuscular Diseases*, **2019**, 6, 475-483	5	4
112	Recessive DES cardio/myopathy without myofibrillar aggregates: intronic splice variant silences one allele leaving only missense L190P-desmin. *European Journal of Human Genetics*, **2019**, 27, 1267-1273	5.3	6
111	Resolving the full spectrum of human genome variation using Linked-Reads. *Genome Research*, **2019**, 29, 635-645	9.7	86
110	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. *Genetics in Medicine*, **2019**, 21, 694-704	8.1	14

109	Pathogenic Abnormal Splicing Due to Intronic Deletions that Induce Biophysical Space Constraint for Spliceosome Assembly. *American Journal of Human Genetics*, **2019**, 105, 573-587	11	14
108	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. *Science*, **2019**, 366, 351-356	33.3	42
107	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. *JCI Insight*, **2019**, 4,	9.9	23
106	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. *American Journal of Human Genetics*, **2019**, 104, 187-190	11	8
105	Reply to Selective effects of heterozygous protein-truncating variants*Nature Genetics*, **2019**, 51, 3-4	36.3	1
104	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
103	Extending the clinical and mutational spectrum of -related myopathies in a non-Hutterite population. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 490-493	5.5	6
102	Quantitative analysis of population-scale family trees with millions of relatives. *Science*, **2018**, 360, 171-175	33.3	94
101	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
100	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessive TNNT3 splice variant. *Human Mutation*, **2018**, 39, 383-388	4.7	30
99	MEGF10 related myopathies: A new case with adult onset disease with prominent respiratory failure and review of reported phenotypes. *Neuromuscular Disorders*, **2018**, 28, 48-53	2.9	9
98	and Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2123-2138	12.7	21
97	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. *Skeletal Muscle*, **2018**, 8, 23	5.1	26
96	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2348-2361	12.7	75
95	STRetch: detecting and discovering pathogenic short tandem repeat expansions. *Genome Biology*, **2018**, 19, 121	18.3	66
94	Insights into the genetic epidemiology of Crohn and rare diseases in the Ashkenazi Jewish population. *PLoS Genetics*, **2018**, 14, e1007329	6	41
93	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. *Neuromuscular Disorders*, **2018**, 28, 614-618	2.9	7
92	A synthetic-diploid benchmark for accurate variant-calling evaluation. *Nature Methods*, **2018**, 15, 595-597	21.6	83

91	Limb girdle muscular dystrophy due to mutations in. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 506-512	5.5	12
90	The Genetic Landscape of Diamond-Blackfan Anemia. *American Journal of Human Genetics*, **2018**, 103, 930-947	11	108
89	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. *Nature Communications*, **2018**, 9, 1929	17.4	48
88	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. *American Journal of Human Genetics*, **2018**, 102, 1204-1211	11	59
87	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. *Genome Research*, **2018**, 28, 968-974	9.7	23
86	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. *Nature Genetics*, **2017**, 49, 238-248	36.3	88
85	The ExAC browser: displaying reference data information from over 60 000 exomes. *Nucleic Acids Research*, **2017**, 45, D840-D845	20.1	348
84	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. *Human Mutation*, **2017**, 38, 517-523	4.7	34
83	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. *Nature Genetics*, **2017**, 49, 504-510	36.3	203
82	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. *Nature*, **2017**, 544, 235-239	50.4	208
81	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. *Science Translational Medicine*, **2017**, 9,	17.5	338
80	Using high-resolution variant frequencies to empower clinical genome interpretation. *Genetics in Medicine*, **2017**, 19, 1151-1158	8.1	208
79	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. *Nature Genetics*, **2017**, 49, 806-810	36.3	84
78	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. *Cell*, **2017**, 169, 6-12	56.2	81
77	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. *European Journal of Human Genetics*, **2017**, 25, 572-581	5.3	14
76	Landscape of X chromosome inactivation across human tissues. *Nature*, **2017**, 550, 244-248	50.4	417
75	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. *Orphanet Journal of Rare Diseases*, **2017**, 12, 151	4.2	27
74	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. *Orphanet Journal of Rare Diseases*, **2017**, 12, 173	4.2	18

73	A Loss-of-Function Splice Acceptor Variant in Is Protective for Type 2 Diabetes. *Diabetes*, **2017**, 66, 2903-2914	29	32
72	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. *Nature Communications*, **2017**, 8, 382	17.4	19
71	A second truncation in TTN causes early onset recessive muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 1009-1017	2.9	9
70	Human genetic variation alters CRISPR-Cas9 on- and off-targeting specificity at therapeutically implicated loci. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E11257-E11266	11.5	66
69	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. *Nature Communications*, **2017**, 8, 16015	17.4	80
68	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. *Npj Genomic Medicine*, **2017**, 2,	6.2	48
67	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. *Genetics in Medicine*, **2017**, 19, 192-203	8.1	386
66	Estimated disease incidence of RAG1/2 mutations: A case report and querying the Exome Aggregation Consortium. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 690-692.e3	11.5	8
65	ClinVar data parsing. *Wellcome Open Research*, **2017**, 2, 33	4.8	16
64	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. *American Journal of Human Genetics*, **2016**, 99, 711-719	11	44
63	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
62	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. *Nature Genetics*, **2016**, 48, 1107-11	36.3	135
61	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. *Neuron*, **2016**, 92, 336-338	13.9	14
60	High-throughput discovery of novel developmental phenotypes. *Nature*, **2016**, 537, 508-514	50.4	608
59	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. *Nature Communications*, **2016**, 7, 13293	17.4	27
58	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. *Annals of Neurology*, **2016**, 80, 101-11	9.4	39
57	Quantifying prion disease penetrance using large population control cohorts. *Science Translational Medicine*, **2016**, 8, 322ra9	17.5	205
56	Health and population effects of rare gene knockouts in adult humans with related parents. *Science*, **2016**, 352, 474-7	33.3	185

55	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. *European Journal of Human Genetics*, **2016**, 24, 1216-9	5.3	2
54	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. *Neurology*, **2016**, 86, 391-8	6.5	83
53	Analysis of the ACTN3 heterozygous genotype suggests that α-actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. *Human Molecular Genetics*, **2016**, 25, 866-77	5.6	26
52	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. *Neuromuscular Disorders*, **2016**, 26, 500-3	2.9	25
51	Superheroes of disease resistance. *Nature Biotechnology*, **2016**, 34, 512-3	44.5	3
50	Variants in SLC18A3, vesicular acetylcholine transporter, cause congenital myasthenic syndrome. *Neurology*, **2016**, 87, 1442-1448	6.5	35
49	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. *American Journal of Human Genetics*, **2016**, 99, 1086-1105	11	29
48	RNAseq analysis for the diagnosis of muscular dystrophy. *Annals of Clinical and Translational Neurology*, **2016**, 3, 55-60	5.3	58
47	The landscape of genomic imprinting across diverse adult human tissues. *Genome Research*, **2015**, 25, 927-36	9.7	139
46	Expanding the phenotype of GMPPB mutations. *Brain*, **2015**, 138, 836-44	11.2	45
45	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans. *Science*, **2015**, 348, 648-60	33.3	3242
44	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
43	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
42	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy: Outcomes and Lessons Learned. *JAMA Neurology*, **2015**, 72, 1424-32	17.2	121
41	A respiratory chain controlled signal transduction cascade in the mitochondrial intermembrane space mediates hydrogen peroxide signaling. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E5679-88	11.5	47
40	Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. *Human Molecular Genetics*, **2015**, 24, 6146-59	5.6	56
39	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
38	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. *Genome Medicine*, **2015**, 7, 90	14.4	38

37	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
36	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcot-Marie-Tooth disease. *Neuromuscular Disorders*, **2014**, 24, 666-70	2.9	14
35	Biallelic variants in TTLL5, encoding a tubulin glutamylase, cause retinal dystrophy. *American Journal of Human Genetics*, **2014**, 94, 760-9	11	52
34	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. *Journal of Neuromuscular Diseases*, **2014**, 1, 135-149	5	20
33	Distribution and medical impact of loss-of-function variants in the Finnish founder population. *PLoS Genetics*, **2014**, 10, e1004494	6	243
32	Allelic expression of deleterious protein-coding variants across human tissues. *PLoS Genetics*, **2014**, 10, e1004304	6	43
31	Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. *JAMA - Journal of the American Medical Association*, **2014**, 311, 2305-14	27.4	164
30	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. *Journal of Neuromuscular Diseases*, **2014**, 1, 135-149	5	10
29	Integrative annotation of variants from 1092 humans: application to cancer genomics. *Science*, **2013** , 342, 1235587	33.3	281
28	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
27	The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. *Genome Research*, **2013**, 23, 749-61	9.7	150
26	The Genotype-Tissue Expression (GTEx) project. *Nature Genetics*, **2013**, 45, 580-5	36.3	4179
25	The uncertain road towards genomic medicine. *Trends in Genetics*, **2012**, 28, 303-5	8.5	8
24	Challenges in clinical genomics. *Genome Medicine*, **2012**, 4, 43	14.4	5
23	A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, **2012**, 335, 823-8	33.3	880
22	Methods: Face up to false positives. *Nature*, **2012**, 487, 427-8	50.4	42
21	Origins and functional impact of copy number variation in the human genome. *Nature*, **2010**, 464, 704-12	50.4	1467
20	Loss-of-function variants in the genomes of healthy humans. *Human Molecular Genetics*, **2010**, 19, R125-30	5.6	141

19	The ACTN3 Gene and Human Performance **2010**, 204-214		3
18	Human Y chromosome base-substitution mutation rate measured by direct sequencing in a deep-rooting pedigree. *Current Biology*, **2009**, 19, 1453-7	6.3	154
17	An Actn3 knockout mouse provides mechanistic insights into the association between alpha-actinin-3 deficiency and human athletic performance. *Human Molecular Genetics*, **2008**, 17, 1076-86	5.6	218
16	Loss of ACTN3 gene function alters mouse muscle metabolism and shows evidence of positive selection in humans. *Nature Genetics*, **2007**, 39, 1261-5	36.3	223
15	ACTN3: A genetic influence on muscle function and athletic performance. *Exercise and Sport Sciences Reviews*, **2007**, 35, 30-4	6.7	94
14	Genes and human elite athletic performance. *Human Genetics*, **2005**, 116, 331-9	6.3	142
13	A gene for speed? The evolution and function of alpha-actinin-3. *BioEssays*, **2004**, 26, 786-95	4.1	166
12	Systematic evaluation of genome sequencing as a first-tier diagnostic test for prenatal and pediatric disorders		2
11	seqr : a web-based analysis and collaboration tool for rare disease genomics		1
10	Regional missense constraint improves variant deleteriousness prediction		102
9	STRetch: detecting and discovering pathogenic short tandem repeat expansions		7
8	Scaling accurate genetic variant discovery to tens of thousands of samples		392
7	Leveraging supervised learning for functionally-informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs		2
6	Evaluating potential drug targets through human loss-of-function genetic variation		12
5	The mutational constraint spectrum quantified from variation in 141,456 humans		381
4	Transcript expression-aware annotation improves rare variant discovery and interpretation		8
3	Systematic single-variant and gene-based association testing of 3,700 phenotypes in 281,850 UK Biobank exomes		6
2	Mitochondrial DNA variation across 56,434 individuals in gnomAD		2