Daniel G Macarthur

List of Publications by Year
in descending order

133 papers	64,253 citations	17440 63 h-index	11052 137 g-index
159 all docs	159 docs citations	159 times ranked	86114 citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
3	The Genotype-Tissue Expression (GTEx) project. Nature Genetics, 2013, 45, 580-585.	21.4	6,815
4	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
5	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. Science, 2015, 348, 648-660.	12.6	4,659
6	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	27.8	1,857
7	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	27.8	1,721
8	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	12.6	1,095
9	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
10	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	27.8	1,001
11	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	21.4	943
12	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	27.8	764
13	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
14	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
15	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203.	2.4	585
16	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
17	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	27.8	441
18	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355

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19	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	3.5	351
20	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	12.6	341
21	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	21.4	298
22	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	27.8	292
23	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
24	Loss of ACTN3 gene function alters mouse muscle metabolism and shows evidence of positive selection in humans. Nature Genetics, 2007, 39, 1261-1265.	21.4	278
25	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	12.6	272
26	An Actn3 knockout mouse provides mechanistic insights into the association between Â-actinin-3 deficiency and human athletic performance. Human Molecular Genetics, 2008, 17, 1076-1086.	2.9	266
27	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
28	A Quantitative Proteome Map of the Human Body. Cell, 2020, 183, 269-283.e19.	28.9	243
29	Association of a Low-Frequency Variant inHNF1AWith Type 2 Diabetes in a Latino Population. JAMA - Journal of the American Medical Association, 2014, 311, 2305.	7.4	230
30	The landscape of genomic imprinting across diverse adult human tissues. Genome Research, 2015, 25, 927-936.	5.5	216
31	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	5.5	206
32	A gene for speed? The evolution and function of ?-actinin-3. BioEssays, 2004, 26, 786-795.	2.5	197
33	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
34	Variant interpretation using population databases: Lessons from gnomAD. Human Mutation, 2022, 43, 1012-1030.	2.5	184
35	Resolving the full spectrum of human genome variation using Linked-Reads. Genome Research, 2019, 29, 635-645.	5.5	182
36	Human Y Chromosome Base-Substitution Mutation Rate Measured by Direct Sequencing in a Deep-Rooting Pedigree. Current Biology, 2009, 19, 1453-1457.	3.9	180

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37	Loss-of-function variants in the genomes of healthy humans. Human Molecular Genetics, 2010, 19, R125-R130.	2.9	172
38	Genes and human elite athletic performance. Human Genetics, 2005, 116, 331-339.	3.8	171
39	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	21.4	167
40	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	9.0	164
41	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
42	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	21.4	157
43	Quantitative analysis of population-scale family trees with millions of relatives. Science, 2018, 360, 171-175.	12.6	157
44	A synthetic-diploid benchmark for accurate variant-calling evaluation. Nature Methods, 2018, 15, 595-597.	19.0	154
45	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015.	12.8	149
46	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.	6.1	147
47	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	27.8	142
48	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
49	ACTN3. Exercise and Sport Sciences Reviews, 2007, 35, 30-34.	3.0	118
50	STRetch: detecting and discovering pathogenic short tandem repeat expansions. Genome Biology, 2018, 19, 121.	8.8	117
51	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	27.8	115
52	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
53	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. Cell, 2017, 169, 6-12.	28.9	103
54	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	6.2	102

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55	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. Science, 2019, 366, 351-356.	12.6	99
56	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	12.8	99
57	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	12.8	98
58	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.	7.1	96
59	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	28.9	94
60	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
61	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.	6.2	81
62	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	30.7	79
63	From variant to function in human disease genetics. Science, 2021, 373, 1464-1468.	12.6	75
64	<scp>RNA</scp> seq analysis for the diagnosis of muscular dystrophy. Annals of Clinical and Translational Neurology, 2016, 3, 55-60.	3.7	73
65	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	12.8	73
66	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.	1.9	71
67	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
68	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, .	3.8	67
69	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	3.5	66
70	Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159.	2.9	64
71	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
72	Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues. PLoS Genetics, 2014, 10, e1004304.	3.5	60

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73	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	5.5	59
74	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.	7.1	58
75	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	5.3	57
76	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	7.6	54
77	A Loss-of-Function Splice Acceptor Variant in IGF2 Is Protective for Type 2 Diabetes. Diabetes, 2017, 66, 2903-2914.	0.6	52
78	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	8.2	49
79	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. Human Mutation, 2017, 38, 517-523.	2.5	49
80	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. Nature Communications, 2021, 12, 3505.	12.8	49
81	Face up to false positives. Nature, 2012, 487, 427-428.	27.8	48
82	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessiveÂTNNT3Âsplice variant. Human Mutation, 2018, 39, 383-388.	2.5	48
83	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.1	46
84	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
85	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
86	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	6.0	45
87	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.	2.7	44
88	Leveraging supervised learning for functionallyÂinformed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. Nature Communications, 2021, 12, 3394.	12.8	44
89	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
90	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	6.1	42

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91	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	5.5	41
92	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. Nature Communications, 2017, 8, 382.	12.8	40
93	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.	4.2	40
94	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, 500-503.	0.6	38
95	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	12.8	35
96	Analysis of theACTN3heterozygous genotype suggests that Î±-actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. Human Molecular Genetics, 2016, 25, 866-877.	2.9	35
97	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, .	5.0	33
98	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	2.5	31
99	Recurrent TTN metatranscriptâ€only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	2.5	28
100	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. Genetics in Medicine, 2019, 21, 694-704.	2.4	27
101	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	11.1	27
102	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	2.6	25
103	Pathogenic Abnormal Splicing Due to Intronic Deletions that Induce Biophysical Space Constraint for Spliceosome Assembly. American Journal of Human Genetics, 2019, 105, 573-587.	6.2	25
104	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
105	Human disease genomics: from variants to biology. Genome Biology, 2017, 18, 20.	8.8	23
106	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. Neuron, 2016, 92, 336-338.	8.1	21
107	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.	2.7	21
108	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	1.9	21

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109	WGS and RNA Studies Diagnose Noncoding DMD Variants in Males With High Creatine Kinase. Neurology: Genetics, 2021, 7, e554.	1.9	21
110	ClinVar data parsing. Wellcome Open Research, 2017, 2, 33.	1.8	19
111	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.	2.8	18
112	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.6	18
113	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2014, 24, 666-670.	0.6	17
114	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. Clinical Genetics, 2020, 97, 764-769.	2.0	17
115	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	6.2	15
116	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.	2.8	14
117	Estimated disease incidence of RAG1/2 mutations: AÂcase report and querying the Exome Aggregation Consortium. Journal of Allergy and Clinical Immunology, 2017, 139, 690-692.e3.	2.9	13
118	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.	0.6	13
119	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. Genetics in Medicine, 2021, 23, 881-887.	2.4	13
120	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.	2.8	13
121	Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients. Neuromuscular Disorders, 2020, 30, 310-314.	0.6	12
122	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.	0.6	11
123	Extending the clinical and mutational spectrum ofTRIM32-related myopathies in a non-Hutterite population. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 490-493.	1.9	11
124	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.	2.8	10
125	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	2.6	10
126	The uncertain road towards genomic medicine. Trends in Genetics, 2012, 28, 303-305.	6.7	8

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127	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.	2.6	6
128	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	21.4	6
129	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	2.8	6
130	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	4.1	6
131	Challenges in clinical genomics. Genome Medicine, 2012, 4, 43.	8.2	5
132	Superheroes of disease resistance. Nature Biotechnology, 2016, 34, 512-513.	17.5	4
133	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. European Journal of Human Genetics, 2016, 24, 1216-1219.	2.8	2