Daniel G Macarthur

List of Publications by Year
in descending order

133 papers	64,253 citations	20036 63 h-index	12638 137 g-index
159 all docs	159 docs citations	159 times ranked	93528 citing authors

#	Article	IF	Citations
1	Variant interpretation using population databases: Lessons from gnomAD. Human Mutation, 2022, 43, 1012-1030.	1.1	184
2	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	2.4	59
3	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
4	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	5.2	27
5	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	1.1	31
6	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.	1.4	10
7	WGS and RNA Studies Diagnose Noncoding DMD Variants in Males With High Creatine Kinase. Neurology: Genetics, 2021, 7, e554.	0.9	21
8	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. Genetics in Medicine, 2021, 23, 881-887.	1.1	13
9	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.	1.4	13
10	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. Cell, 2021, 184, 2633-2648.e19.	13.5	94
11	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. Nature Communications, 2021, 12, 3505.	5.8	49
12	Leveraging supervised learning for functionallyÂinformed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. Nature Communications, 2021, 12, 3394.	5.8	44
13	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
14	From variant to function in human disease genetics. Science, 2021, 373, 1464-1468.	6.0	75
15	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	1.9	6
16	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	1.4	6
17	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.	0.9	71
18	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.	1.1	28

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19	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
20	A Quantitative Proteome Map of the Human Body. Cell, 2020, 183, 269-283.e19.	13.5	243
21	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	2.6	25
22	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
23	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
24	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	5.8	99
25	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	5.8	98
26	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
27	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
28	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	15.2	79
29	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	1.1	62
30	Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients. Neuromuscular Disorders, 2020, 30, 310-314.	0.3	12
31	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. Clinical Genetics, 2020, 97, 764-769.	1.0	17
32	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	2.8	45
33	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. Genetics in Medicine, 2019, 21, 694-704.	1.1	27
34	Pathogenic Abnormal Splicing Due to Intronic Deletions that Induce Biophysical Space Constraint for Spliceosome Assembly. American Journal of Human Genetics, 2019, 105, 573-587.	2.6	25
35	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. Science, 2019, 366, 351-356.	6.0	99
36	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.	1.1	6

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37	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.	1.4	14
38	Resolving the full spectrum of human genome variation using Linked-Reads. Genome Research, 2019, 29, 635-645.	2.4	182
39	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	2.6	15
40	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	9.4	6
41	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
42	Extending the clinical and mutational spectrum ofTRIM32-related myopathies in a non-Hutterite population. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 490-493.	0.9	11
43	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, .	2.3	33
44	Quantitative analysis of population-scale family trees with millions of relatives. Science, 2018, 360, 171-175.	6.0	157
45	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
46	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessiveÂTNNT3Âsplice variant. Human Mutation, 2018, 39, 383-388.	1.1	48
47	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.	0.3	13
48	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	0.9	21
49	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	2.6	184
50	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. Nature Communications, 2018, 9, 1929.	5.8	73
51	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	2.6	102
52	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	2.4	41
53	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	3.0	42
54	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.	1.9	40

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55	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.	3.0	147
56	STRetch: detecting and discovering pathogenic short tandem repeat expansions. Genome Biology, 2018, 19, 121.	3.8	117
57	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
58	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.	0.3	11
59	A synthetic-diploid benchmark for accurate variant-calling evaluation. Nature Methods, 2018, 15, 595-597.	9.0	154
60	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
61	Human disease genomics: from variants to biology. Genome Biology, 2017, 18, 20.	3.8	23
62	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	6.5	587
63	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. Human Mutation, 2017, 38, 517-523.	1.1	49
64	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	9.4	298
65	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	13.7	292
66	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	5.8	516
67	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	1.1	355
68	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	9.4	157
69	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. Cell, 2017, 169, 6-12.	13.5	103
70	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.	1.4	18
71	Landscape of X chromosome inactivation across human tissues. Nature, 2017, 550, 244-248.	13.7	764
72	A Loss-of-Function Splice Acceptor Variant in IGF2 Is Protective for Type 2 Diabetes. Diabetes, 2017, 66, 2903-2914.	0.3	52

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73	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. Nature Communications, 2017, 8, 382.	5.8	40
74	A â€˜second truncationâ€™ in TTN causes early onset recessive muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1009-1017.	0.3	18
75	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.	3.3	96
76	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015.	5.8	149
77	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, .	1.7	67
78	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203.	1.1	585
79	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.	1.5	13
80	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.	1.2	44
81	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.	1.2	21
82	ClinVar data parsing. Wellcome Open Research, 2017, 2, 33.	0.9	19
83	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, 500-503.	0.3	38
84	Superheroes of disease resistance. Nature Biotechnology, 2016, 34, 512-513.	9.4	4
85	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.5	46
86	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	2.6	45
87	<scp>RNA</scp> seq analysis for the diagnosis of muscular dystrophy. Annals of Clinical and Translational Neurology, 2016, 3, 55-60.	1.7	73
88	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.	2.6	81
89	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
90	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	9.4	167

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91	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. Neuron, 2016, 92, 336-338.	3.8	21
92	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	13.7	1,001
93	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	5.8	35
94	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	2.8	57
95	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	5.8	289
96	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	6.0	272
97	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. European Journal of Human Genetics, 2016, 24, 1216-1219.	1.4	2
98	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.5	107
99	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.	1.4	35
100	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	3.6	49
101	The landscape of genomic imprinting across diverse adult human tissues. Genome Research, 2015, 25, 927-936.	2.4	216
102	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	3.7	54
103	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. Science, 2015, 348, 648-660.	6.0	4,659
104	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
105	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
106	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	4.5	164
107	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.	3.3	58
108	Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159.	1.4	64

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109	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	13.7	1,014
110	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	1.1	25
111	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	1.5	351
112	Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues. PLoS Genetics, 2014, 10, e1004304.	1.5	60
113	Association of a Low-Frequency Variant inHNF1AWith Type 2 Diabetes in a Latino Population. JAMA - Journal of the American Medical Association, 2014, 311, 2305.	3.8	230
114	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
115	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2014, 24, 666-670.	0.3	17
116	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	2.6	67
117	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	1.1	10
118	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	6.0	341
119	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
120	The origin, evolution, and functional impact of short insertionâ€“deletion variants identified in 179 human genomes. Genome Research, 2013, 23, 749-761.	2.4	206
121	The Genotype-Tissue Expression (GTEx) project. Nature Genetics, 2013, 45, 580-585.	9.4	6,815
122	Face up to false positives. Nature, 2012, 487, 427-428.	13.7	48
123	Challenges in clinical genomics. Genome Medicine, 2012, 4, 43.	3.6	5
124	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
125	The uncertain road towards genomic medicine. Trends in Genetics, 2012, 28, 303-305.	2.9	8
126	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	13.7	1,721

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127	Loss-of-function variants in the genomes of healthy humans. Human Molecular Genetics, 2010, 19, R125-R130.	1.4	172
128	Human Y Chromosome Base-Substitution Mutation Rate Measured by Direct Sequencing in a Deep-Rooting Pedigree. Current Biology, 2009, 19, 1453-1457.	1.8	180
129	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.	1.4	266
130	ACTN3. Exercise and Sport Sciences Reviews, 2007, 35, 30-34.	1.6	118
131	Loss of ACTN3 gene function alters mouse muscle metabolism and shows evidence of positive selection in humans. Nature Genetics, 2007, 39, 1261-1265.	9.4	278
132	Genes and human elite athletic performance. Human Genetics, 2005, 116, 331-339.	1.8	171
133	A gene for speed? The evolution and function of ?-actinin-3. BioEssays, 2004, 26, 786-795.	1.2	197