

# Daniel G Macarthur

## List of Publications by Citations

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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	A global reference for human genetic variation. <i>Nature</i> , <b>2015</b> , 526, 68-74	50.4	8599
143	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , <b>2016</b> , 536, 285-91	50.4	6940
142	The Genotype-Tissue Expression (GTEx) project. <i>Nature Genetics</i> , <b>2013</b> , 45, 580-5	36.3	4179
141	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans. <i>Science</i> , <b>2015</b> , 348, 648-60	33.3	3242
140	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , <b>2020</b> , 581, 434-443	50.4	2278
139	Origins and functional impact of copy number variation in the human genome. <i>Nature</i> , <b>2010</b> , 464, 704-12	50.4	1467
138	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , <b>2013</b> , 501, 506-31	31.4	1323
137	A systematic survey of loss-of-function variants in human protein-coding genes. <i>Science</i> , <b>2012</b> , 335, 823-8	33.3	880
136	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , <b>2015</b> , 526, 82-90	50.4	776
135	A framework for the interpretation of de novo mutation in human disease. <i>Nature Genetics</i> , <b>2014</b> , 46, 944-50	36.3	656
134	High-throughput discovery of novel developmental phenotypes. <i>Nature</i> , <b>2016</b> , 537, 508-514	50.4	608
133	Landscape of X chromosome inactivation across human tissues. <i>Nature</i> , <b>2017</b> , 550, 244-248	50.4	417
132	Scaling accurate genetic variant discovery to tens of thousands of samples		392
131	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 192-203	8.1	386
130	The mutational constraint spectrum quantified from variation in 141,456 humans		381
129	The ExAC browser: displaying reference data information from over 60 000 exomes. <i>Nucleic Acids Research</i> , <b>2017</b> , 45, D840-D845	20.1	348
128	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. <i>Science Translational Medicine</i> , <b>2017</b> , 9,	17.5	338

127	Integrative annotation of variants from 1092 humans: application to cancer genomics. <i>Science</i> , <b>2013</b> , 342, 1235587	33.3	281
126	Distribution and medical impact of loss-of-function variants in the Finnish founder population. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004494	6	243
125	A structural variation reference for medical and population genetics. <i>Nature</i> , <b>2020</b> , 581, 444-451	50.4	223
124	Loss of ACTN3 gene function alters mouse muscle metabolism and shows evidence of positive selection in humans. <i>Nature Genetics</i> , <b>2007</b> , 39, 1261-5	36.3	223
123	An Actn3 knockout mouse provides mechanistic insights into the association between alpha-actinin-3 deficiency and human athletic performance. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 1076-86	5.6	218
122	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. <i>Nature</i> , <b>2017</b> , 544, 235-239	50.4	208
121	Using high-resolution variant frequencies to empower clinical genome interpretation. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1151-1158	8.1	208
120	Quantifying prion disease penetrance using large population control cohorts. <i>Science Translational Medicine</i> , <b>2016</b> , 8, 322ra9	17.5	205
119	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. <i>Nature Genetics</i> , <b>2017</b> , 49, 504-510	36.3	203
118	Health and population effects of rare gene knockouts in adult humans with related parents. <i>Science</i> , <b>2016</b> , 352, 474-7	33.3	185
117	A brief history of human disease genetics. <i>Nature</i> , <b>2020</b> , 577, 179-189	50.4	181
116	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , <b>2015</b> , 348, 666-9	33.3	170
115	A gene for speed? The evolution and function of alpha-actinin-3. <i>BioEssays</i> , <b>2004</b> , 26, 786-95	4.1	166
114	Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. <i>JAMA - Journal of the American Medical Association</i> , <b>2014</b> , 311, 2305-14	27.4	164
113	Human Y chromosome base-substitution mutation rate measured by direct sequencing in a deep-rooting pedigree. <i>Current Biology</i> , <b>2009</b> , 19, 1453-7	6.3	154
112	The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. <i>Genome Research</i> , <b>2013</b> , 23, 749-61	9.7	150
111	Genes and human elite athletic performance. <i>Human Genetics</i> , <b>2005</b> , 116, 331-9	6.3	142
110	Loss-of-function variants in the genomes of healthy humans. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, R125-306	30.6	141

109	The landscape of genomic imprinting across diverse adult human tissues. <i>Genome Research</i> , <b>2015</b> , 25, 927-36	9.7	139
108	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. <i>Nature Genetics</i> , <b>2016</b> , 48, 1107-11	36.3	135
107	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy: Outcomes and Lessons Learned. <i>JAMA Neurology</i> , <b>2015</b> , 72, 1424-32	17.2	121
106	The Genetic Landscape of Diamond-Blackfan Anemia. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 930-947	11	108
105	Regional missense constraint improves variant deleteriousness prediction		102
104	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 798-812	8.1	100
103	Quantitative analysis of population-scale family trees with millions of relatives. <i>Science</i> , <b>2018</b> , 360, 171-175	37.3	94
102	ACTN3: A genetic influence on muscle function and athletic performance. <i>Exercise and Sport Sciences Reviews</i> , <b>2007</b> , 35, 30-4	6.7	94
101	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , <b>2017</b> , 49, 238-248	36.3	88
100	Resolving the full spectrum of human genome variation using Linked-Reads. <i>Genome Research</i> , <b>2019</b> , 29, 635-645	9.7	86
99	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. <i>Nature Genetics</i> , <b>2017</b> , 49, 806-810	36.3	84
98	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. <i>Neurology</i> , <b>2016</b> , 86, 391-8	6.5	83
97	A synthetic-diploid benchmark for accurate variant-calling evaluation. <i>Nature Methods</i> , <b>2018</b> , 15, 595-597	21.6	83
96	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. <i>Cell</i> , <b>2017</b> , 169, 6-12	56.2	81
95	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. <i>Nature Communications</i> , <b>2017</b> , 8, 16015	17.4	80
94	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2018</b> , 29, 2348-2361	12.7	75
93	A Quantitative Proteome Map of the Human Body. <i>Cell</i> , <b>2020</b> , 183, 269-283.e19	56.2	73
92	STRetch: detecting and discovering pathogenic short tandem repeat expansions. <i>Genome Biology</i> , <b>2018</b> , 19, 121	18.3	66

91	Human genetic variation alters CRISPR-Cas9 on- and off-targeting specificity at therapeutically implicated loci. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, E11257-E11266	11.5	66
90	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , <b>2018</b> , 83, 1105-1124	9.4	59
89	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 1204-1211	11	59
88	RNAseq analysis for the diagnosis of muscular dystrophy. <i>Annals of Clinical and Translational Neurology</i> , <b>2016</b> , 3, 55-60	5.3	58
87	Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6146-59	5.6	56
86	Transcript expression-aware annotation improves rare variant interpretation. <i>Nature</i> , <b>2020</b> , 581, 452-458	50.4	55
85	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , <b>2020</b> , 581, 459-464	50.4	53
84	Biallelic variants in TLL5, encoding a tubulin glutamylase, cause retinal dystrophy. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 760-9	11	52
83	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. <i>Nature Communications</i> , <b>2020</b> , 11, 2539	17.4	51
82	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. <i>Npj Genomic Medicine</i> , <b>2017</b> , 2,	6.2	48
81	A whole-genome sequence study identifies genetic risk factors for neuromyelitis optica. <i>Nature Communications</i> , <b>2018</b> , 9, 1929	17.4	48
80	A respiratory chain controlled signal transduction cascade in the mitochondrial intermembrane space mediates hydrogen peroxide signaling. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2015</b> , 112, E5679-88	11.5	47
79	The effect of LRRK2 loss-of-function variants in humans. <i>Nature Medicine</i> , <b>2020</b> , 26, 869-877	50.5	47
78	Expanding the phenotype of GMPPB mutations. <i>Brain</i> , <b>2015</b> , 138, 836-44	11.2	45
77	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 711-719	11	44
76	Allelic expression of deleterious protein-coding variants across human tissues. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004304	6	43
75	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. <i>Science</i> , <b>2019</b> , 366, 351-356	33.3	42
74	Methods: Face up to false positives. <i>Nature</i> , <b>2012</b> , 487, 427-8	50.4	42

73	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007329	6	41
72	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. <i>Annals of Neurology</i> , <b>2016</b> , 80, 101-111	9.4	39
71	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. <i>Genome Medicine</i> , <b>2015</b> , 7, 90	14.4	38
70	Characterising the loss-of-function impact of 5' untranslated region variants in 15,708 individuals. <i>Nature Communications</i> , <b>2020</b> , 11, 2523	17.4	35
69	Variants in SLC18A3, vesicular acetylcholine transporter, cause congenital myasthenic syndrome. <i>Neurology</i> , <b>2016</b> , 87, 1442-1448	6.5	35
68	Pathogenic ASXL1 somatic variants in reference databases complicate germline variant interpretation for Bohring-Opitz Syndrome. <i>Human Mutation</i> , <b>2017</b> , 38, 517-523	4.7	34
67	A Loss-of-Function Splice Acceptor Variant in Irf1 is Protective for Type 2 Diabetes. <i>Diabetes</i> , <b>2017</b> , 66, 2903-2914	2.9	32
66	Nemaline myopathy and distal arthrogyria associated with an autosomal recessive TNNT3 splice variant. <i>Human Mutation</i> , <b>2018</b> , 39, 383-388	4.7	30
65	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 1086-1105	11	29
64	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 151	4.2	27
63	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. <i>Nature Communications</i> , <b>2016</b> , 7, 13293	17.4	27
62	Analysis of the ACTN3 heterozygous genotype suggests that $\alpha$ -actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 866-879	5.6	26
61	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. <i>Skeletal Muscle</i> , <b>2018</b> , 8, 23	5.1	26
60	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1478-1488	8.1	25
59	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 500-3	2.9	25
58	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. <i>JCI Insight</i> , <b>2019</b> , 4,	9.9	23
57	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. <i>Genome Research</i> , <b>2018</b> , 28, 968-974	9.7	23
56	and Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2018</b> , 29, 2123-2138	12.7	21

55	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. <i>Journal of Neuromuscular Diseases</i> , <b>2014</b> , 1, 135-149	5	20
54	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. <i>Cell</i> , <b>2021</b> , 184, 2633-2648.e19	56.2	20
53	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. <i>Nature Communications</i> , <b>2017</b> , 8, 382	17.4	19
52	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. <i>ELife</i> , <b>2020</b> , 9,	8.9	19
51	Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. <i>International Journal of Epidemiology</i> , <b>2020</b> , 49, 20-21i	7.8	19
50	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 173	4.2	18
49	ClinVar data parsing. <i>Wellcome Open Research</i> , <b>2017</b> , 2, 33	4.8	16
48	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 572-581	5.3	14
47	Publicly Available Data Provide Evidence against NR1H3 R415Q Causing Multiple Sclerosis. <i>Neuron</i> , <b>2016</b> , 92, 336-338	13.9	14
46	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 694-704	8.1	14
45	Pathogenic Abnormal Splicing Due to Intronic Deletions that Induce Biophysical Space Constraint for Spliceosome Assembly. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 573-587	11	14
44	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , <b>2014</b> , 24, 666-70	2.9	14
43	Evaluating potential drug targets through human loss-of-function genetic variation		12
42	Limb girdle muscular dystrophy due to mutations in. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 506-512	5.5	12
41	From variant to function in human disease genetics. <i>Science</i> , <b>2021</b> , 373, 1464-1468	33.3	11
40	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogyrosis multiplex congenita and myopathy. <i>Human Mutation</i> , <b>2020</b> , 41, 403-411	4.7	10
39	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. <i>Journal of Neuromuscular Diseases</i> , <b>2014</b> , 1, 135-149	5	10
38	MEGF10 related myopathies: A new case with adult onset disease with prominent respiratory failure and review of reported phenotypes. <i>Neuromuscular Disorders</i> , <b>2018</b> , 28, 48-53	2.9	9

37	A second truncation in TTN causes early onset recessive muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2017</b> , 27, 1009-1017	2.9	9
36	Leveraging supervised learning for functionally informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. <i>Nature Communications</i> , <b>2021</b> , 12, 3394	17.4	9
35	The uncertain road towards genomic medicine. <i>Trends in Genetics</i> , <b>2012</b> , 28, 303-5	8.5	8
34	Estimated disease incidence of RAG1/2 mutations: A case report and querying the Exome Aggregation Consortium. <i>Journal of Allergy and Clinical Immunology</i> , <b>2017</b> , 139, 690-692.e3	11.5	8
33	Transcript expression-aware annotation improves rare variant discovery and interpretation		8
32	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 187-190	11	8
31	The genetic and clinical landscape of nanophthalmos and posterior microphthalmos in an Australian cohort. <i>Clinical Genetics</i> , <b>2020</b> , 97, 764-769	4	7
30	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. <i>Neuromuscular Disorders</i> , <b>2018</b> , 28, 614-618	2.9	7
29	STRetch: detecting and discovering pathogenic short tandem repeat expansions		7
28	Recessive DES cardio/myopathy without myofibrillar aggregates: intronic splice variant silences one allele leaving only missense L190P-desmin. <i>European Journal of Human Genetics</i> , <b>2019</b> , 27, 1267-1273	5.3	6
27	Systematic single-variant and gene-based association testing of 3,700 phenotypes in 281,850 UK Biobank exomes		6
26	Extending the clinical and mutational spectrum of -related myopathies in a non-Hutterite population. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2019</b> , 90, 490-493	5.5	6
25	Transcriptome variation in human tissues revealed by long-read sequencing		6
24	Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients. <i>Neuromuscular Disorders</i> , <b>2020</b> , 30, 310-314	2.9	5
23	Challenges in clinical genomics. <i>Genome Medicine</i> , <b>2012</b> , 4, 43	14.4	5
22	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. <i>Genetics in Medicine</i> , <b>2022</b> ,	8.1	5
21	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , <b>2021</b> , 12, 3505	17.4	5
20	Identification of a Novel Deep Intronic Mutation in CAPN3 Presenting a Promising Target for Therapeutic Splice Modulation. <i>Journal of Neuromuscular Diseases</i> , <b>2019</b> , 6, 475-483	5	4



19	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. <i>European Journal of Human Genetics</i> , <b>2020</b> , 28, 383-387	5.3	4
18	Pathogenic deep intronic MTM1 variant activates a pseudo-exon encoding a nonsense codon resulting in severe X-linked myotubular myopathy. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 61-66	5.3	4
17	WGS and RNA Studies Diagnose Noncoding Variants in Males With High Creatine Kinase. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e554	3.8	4
16	The ACTN3 Gene and Human Performance <b>2010</b> , 204-214		3
15	Mitochondrial DNA variation across 56,434 individuals in gnomAD.. <i>Genome Research</i> , <b>2022</b> ,	9.7	3
14	Superheroes of disease resistance. <i>Nature Biotechnology</i> , <b>2016</b> , 34, 512-3	44.5	3
13	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , <b>2021</b> , 597, E3-E4	50.4	3
12	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 727-742	11	2
11	Prominent scapulae mimicking an inherited myopathy expands the phenotype of CHD7-related disease. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 1216-9	5.3	2
10	Systematic evaluation of genome sequencing as a first-tier diagnostic test for prenatal and pediatric disorders		2
9	Leveraging supervised learning for functionally-informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs		2
8	Mitochondrial DNA variation across 56,434 individuals in gnomAD		2
7	seqr : a web-based analysis and collaboration tool for rare disease genomics		1
6	Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 816-826	5.3	1
5	Reply to Selective effects of heterozygous protein-truncating variants <i>Nature Genetics</i> , <b>2019</b> , 51, 3-4	36.3	1
4	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 881-887	8.1	1
3	Autosomal recessive variants in alter the Tubulin ring complex leading to neurodevelopmental disease. <i>IScience</i> , <b>2021</b> , 24, 101948	6.1	1
2	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation.. <i>Cell Stem Cell</i> , <b>2022</b> ,	18	1

1 seqr: a web-based analysis and collaboration tool for rare disease genomics.. *Human Mutation*, **2022**

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