Deborah A Nickerson

List of Publications by Citations

162 papers	33,546 citations	73 h-index	167 g-index
167 ext. papers	38,597 ext. citations	14 avg, IF	6.45 L-index

#	Paper	IF	Citations
162	Biological, clinical and population relevance of 95 loci for blood lipids. *Nature*, **2010**, 466, 707-13	50.4	2742
161	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. *Nature*, **2012**, 485, 246-50	50.4	1587
160	Exome sequencing identifies the cause of a mendelian disorder. *Nature Genetics*, **2010**, 42, 30-5	36.3	1573
159	Targeted capture and massively parallel sequencing of 12 human exomes. *Nature*, **2009**, 461, 272-6	50.4	1573
158	The contribution of de novo coding mutations to autism spectrum disorder. *Nature*, **2014**, 515, 216-21	50.4	1470
157	Selecting a maximally informative set of single-nucleotide polymorphisms for association analyses using linkage disequilibrium. *American Journal of Human Genetics*, **2004**, 74, 106-20	11	1334
156	Evolution and functional impact of rare coding variation from deep sequencing of human exomes. *Science*, **2012**, 337, 64-9	33.3	1280
155	Exome sequencing as a tool for Mendelian disease gene discovery. *Nature Reviews Genetics*, **2011**, 12, 745-55	30.1	1265
154	Effect of VKORC1 haplotypes on transcriptional regulation and warfarin dose. *New England Journal of Medicine*, **2005**, 352, 2285-93	59.2	1149
153	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. *Nature Genetics*, **2010**, 42, 790-3	36.3	1041
152	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nature Genetics*, **2011**, 43, 585-9	36.3	899
151	Multiplex targeted sequencing identifies recurrently mutated genes in autism spectrum disorders. *Science*, **2012**, 338, 1619-22	33.3	892
150	Mapping and sequencing of structural variation from eight human genomes. *Nature*, **2008**, 453, 56-64	50.4	878
149	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. *Nature*, **2013**, 493, 216-20	50.4	723
148	Loss-of-function mutations in APOC3, triglycerides, and coronary disease. *New England Journal of Medicine*, **2014**, 371, 22-31	59.2	721
147	Mapping complex disease loci in whole-genome association studies. *Nature*, **2004**, 429, 446-52	50.4	500
146	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. *Nature*, **2015**, 518, 102-6	50.4	463

145	Population analysis of large copy number variants and hotspots of human genetic disease. *American Journal of Human Genetics*, **2009**, 84, 148-61	11	454
144	DNA sequence diversity in a 9.7-kb region of the human lipoprotein lipase gene. *Nature Genetics*, **1998**, 19, 233-40	36.3	436
143	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
142	Sequence variation in the human angiotensin converting enzyme. *Nature Genetics*, **1999**, 22, 59-62	36.3	408
141	Copy number variation detection and genotyping from exome sequence data. *Genome Research*, **2012**, 22, 1525-32	9.7	406
140	Population history and natural selection shape patterns of genetic variation in 132 genes. *PLoS Biology*, **2004**, 2, e286	9.7	383
139	Concept, design and implementation of a cardiovascular gene-centric 50 k SNP array for large-scale genomic association studies. *PLoS ONE*, **2008**, 3, e3583	3.7	321
138	Inactivating mutations in NPC1L1 and protection from coronary heart disease. *New England Journal of Medicine*, **2014**, 371, 2072-82	59.2	307
137	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. *Nature*, **2021**, 590, 290-299	50.4	268
136	Genome-wide studies of copy number variation and exome sequencing identify rare variants in BAG3 as a cause of dilated cardiomyopathy. *American Journal of Human Genetics*, **2011**, 88, 273-82	11	264
135	Additional SNPs and linkage-disequilibrium analyses are necessary for whole-genome association studies in humans. *Nature Genetics*, **2003**, 33, 518-21	36.3	262
134	Polymorphisms within the C-reactive protein (CRP) promoter region are associated with plasma CRP levels. *American Journal of Human Genetics*, **2005**, 77, 64-77	11	258
133	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics*, **2012**, 44, 916-21	36.3	257
132	Definition and clinical importance of haplotypes. *Annual Review of Medicine*, **2005**, 56, 303-20	17.4	243
131	Toll-like receptor 1 polymorphisms affect innate immune responses and outcomes in sepsis. *American Journal of Respiratory and Critical Care Medicine*, **2008**, 178, 710-20	10.2	226
130	De novo rates and selection of large copy number variation. *Genome Research*, **2010**, 20, 1469-81	9.7	221
129	Exome sequencing identifies SMAD3 mutations as a cause of familial thoracic aortic aneurysm and dissection with intracranial and other arterial aneurysms. *Circulation Research*, **2011**, 109, 680-6	15.7	221
128	Exome sequencing identifies a spectrum of mutation frequencies in advanced and lethal prostate cancers. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 17087-92	11.5	211

127	Mutational and selective effects on copy-number variants in the human genome. *Nature Genetics*, **2007**, 39, S22-9	36.3	193
126	Genomic regions exhibiting positive selection identified from dense genotype data. *Genome Research*, **2005**, 15, 1553-65	9.7	191
125	Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. *American Journal of Human Genetics*, **2016**, 98, 58-74	11	189
124	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. *Nature Genetics*, **2008**, 40, 1199-203	36.3	174
123	Identification, replication, and functional fine-mapping of expression quantitative trait loci in primary human liver tissue. *PLoS Genetics*, **2011**, 7, e1002078	6	171
122	Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. *American Journal of Human Genetics*, **2014**, 94, 233-45	11	170
121	Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis. *Nature Genetics*, **2012**, 44, 886-9	36.3	170
120	Massively parallel exon capture and library-free resequencing across 16 genomes. *Nature Methods*, **2009**, 6, 315-6	21.6	164
119	A statin-dependent QTL for GATM expression is associated with statin-induced myopathy. *Nature*, **2013**, 502, 377-80	50.4	160
118	Pharmacogenetic meta-analysis of genome-wide association studies of LDL cholesterol response to statins. *Nature Communications*, **2014**, 5, 5068	17.4	160
117	Polymorphisms of the HNF1A gene encoding hepatocyte nuclear factor-1 alpha are associated with C-reactive protein. *American Journal of Human Genetics*, **2008**, 82, 1193-201	11	155
116	Completing the map of human genetic variation. *Nature*, **2007**, 447, 161-5	50.4	153
115	Massively parallel sequencing and rare disease. *Human Molecular Genetics*, **2010**, 19, R119-24	5.6	147
114	Pattern of sequence variation across 213 environmental response genes. *Genome Research*, **2004**, 14, 1821-31	9.7	147
113	Haplotype diversity across 100 candidate genes for inflammation, lipid metabolism, and blood pressure regulation in two populations. *American Journal of Human Genetics*, **2004**, 74, 610-22	11	146
112	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. *Nature Methods*, **2010**, 7, 250-1	21.6	139
111	Haploinsufficiency of SF3B4, a component of the pre-mRNA spliceosomal complex, causes Nager syndrome. *American Journal of Human Genetics*, **2012**, 90, 925-33	11	135
110	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1511-6	2.5	132

109	Genetic variation is associated with C-reactive protein levels in the Third National Health and Nutrition Examination Survey. *Circulation*, **2006**, 114, 2458-65	16.7	126
108	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
107	Comprehensive identification and characterization of diallelic insertion-deletion polymorphisms in 330 human candidate genes. *Human Molecular Genetics*, **2005**, 14, 59-69	5.6	124
106	Variation in the 3-hydroxyl-3-methylglutaryl coenzyme a reductase gene is associated with racial differences in low-density lipoprotein cholesterol response to simvastatin treatment. *Circulation*, **2008**, 117, 1537-44	16.7	122
105	A multivariate genome-wide association analysis of 10 LDL subfractions, and their response to statin treatment, in 1868 Caucasians. *PLoS ONE*, **2015**, 10, e0120758	3.7	118
104	denovo-db: a compendium of human de novo variants. *Nucleic Acids Research*, **2017**, 45, D804-D811	20.1	113
103	Exome sequencing identifies mutations in CCDC114 as a cause of primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 92, 99-106	11	111
102	Mutations in SPAG1 cause primary ciliary dyskinesia associated with defective outer and inner dynein arms. *American Journal of Human Genetics*, **2013**, 93, 711-20	11	109
101	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. *Genome Research*, **2009**, 19, 1579-85	9.7	106
100	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
99	Methods for genomic partitioning. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 263-84	9.7	98
98	The patterns of natural variation in human genes. *Annual Review of Genomics and Human Genetics*, **2005**, 6, 287-312	9.7	96
97	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
96	Altered splicing of ATP6AP2 causes X-linked parkinsonism with spasticity (XPDS). *Human Molecular Genetics*, **2013**, 22, 3259-68	5.6	89
95	Autosomal dominant familial dyskinesia and facial myokymia: single exome sequencing identifies a mutation in adenylyl cyclase 5. *Archives of Neurology*, **2012**, 69, 630-5		87
94	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. *American Journal of Human Genetics*, **2018**, 102, 309-320	11	85
93	Cladistic structure within the human Lipoprotein lipase gene and its implications for phenotypic association studies. *Genetics*, **2000**, 156, 1259-75	4	77
92	PGRNseq: a targeted capture sequencing panel for pharmacogenetic research and implementation. *Pharmacogenetics and Genomics*, **2016**, 26, 161-168	1.9	74

91	Mendelian Gene Discovery: Fast and Furious with No End in Sight. *American Journal of Human Genetics*, **2019**, 105, 448-455	11	73
90	Automating resequencing-based detection of insertion-deletion polymorphisms. *Nature Genetics*, **2006**, 38, 1457-62	36.3	69
89	Quantifying rare, deleterious variation in 12 human cytochrome P450 drug-metabolism genes in a large-scale exome dataset. *Human Molecular Genetics*, **2014**, 23, 1957-63	5.6	68
88	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
87	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program		68
86	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
85	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
84	Mutations in TBC1D24, a gene associated with epilepsy, also cause nonsyndromic deafness DFNB86. *American Journal of Human Genetics*, **2014**, 94, 144-52	11	66
83	Allele frequency matching between SNPs reveals an excess of linkage disequilibrium in genic regions of the human genome. *PLoS Genetics*, **2006**, 2, e142	6	62
82	A human homeotic transformation resulting from mutations in PLCB4 and GNAI3 causes auriculocondylar syndrome. *American Journal of Human Genetics*, **2012**, 90, 907-14	11	60
81	Common and rare von Willebrand factor (VWF) coding variants, VWF levels, and factor VIII levels in African Americans: the NHLBI Exome Sequencing Project. *Blood*, **2013**, 122, 590-7	2.2	60
80	Efficient selection of tagging single-nucleotide polymorphisms in multiple populations. *Human Genetics*, **2006**, 120, 58-68	6.3	60
79	"Mandibulofacial dysostosis with microcephaly" caused by EFTUD2 mutations: expanding the phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 108-13	2.5	56
78	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
77	Combined influence of LDLR and HMGCR sequence variation on lipid-lowering response to simvastatin. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2010**, 30, 1485-92	9.4	52
76	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. *Genetics in Medicine*, **2019**, 21, 1611-1620	8.1	52
75	Sequence variation and linkage disequilibrium in the human T-cell receptor beta (TCRB) locus. *American Journal of Human Genetics*, **2001**, 69, 381-95	11	51
74	Oligonucleotide ligation assay for detecting mutations in the human immunodeficiency virus type 1 pol gene that are associated with resistance to zidovudine, didanosine, and lamivudine. *Journal of Clinical Microbiology*, **1998**, 36, 569-72	9.7	51

73	Rare-variant extensions of the transmission disequilibrium test: application to autism exome sequence data. *American Journal of Human Genetics*, **2014**, 94, 33-46	11	48
72	Exome sequencing reveals novel rare variants in the ryanodine receptor and calcium channel genes in malignant hyperthermia families. *Anesthesiology*, **2013**, 119, 1054-65	4.3	45
71	A 32 kb critical region excluding Y402H in CFH mediates risk for age-related macular degeneration. *PLoS ONE*, **2011**, 6, e25598	3.7	41
70	The effects of scale: variation in the APOA1/C3/A4/A5 gene cluster. *Human Genetics*, **2004**, 115, 36-56	6.3	40
69	Exome sequencing identifies a recurrent de novo ZSWIM6 mutation associated with acromelic frontonasal dysostosis. *American Journal of Human Genetics*, **2014**, 95, 235-40	11	39
68	Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke: The NHLBI Exome Sequence Project. *JAMA Neurology*, **2015**, 72, 781-8	17.2	37
67	Maternal Modifiers and Parent-of-Origin Bias of the Autism-Associated 16p11.2 CNV. *American Journal of Human Genetics*, **2016**, 98, 45-57	11	36
66	Exome sequencing in suspected monogenic dyslipidemias. *Circulation: Cardiovascular Genetics*, **2015**, 8, 343-50		36
65	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. *American Journal of Human Genetics*, **2016**, 98, 331-8	11	35
64	Joint linkage and association analysis with exome sequence data implicates SLC25A40 in hypertriglyceridemia. *American Journal of Human Genetics*, **2013**, 93, 1035-45	11	35
63	Rare variation facilitates inferences of fine-scale population structure in humans. *Molecular Biology and Evolution*, **2015**, 32, 653-60	8.3	31
62	Association of exome sequences with plasma C-reactive protein levels in >9000 participants. *Human Molecular Genetics*, **2015**, 24, 559-71	5.6	31
61	Imputation of coding variants in African Americans: better performance using data from the exome sequencing project. *Bioinformatics*, **2013**, 29, 2744-9	7.2	30
60	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. *European Journal of Human Genetics*, **2015**, 23, 1207-15	5.3	29
59	Variation in LPA is associated with Lp(a) levels in three populations from the Third National Health and Nutrition Examination Survey. *PLoS ONE*, **2011**, 6, e16604	3.7	29
58	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
57	Direct detection of null alleles in SNP genotyping data. *Human Molecular Genetics*, **2006**, 15, 1931-7	5.6	28
56	Meta-analysis of genome-wide association studies of HDL cholesterol response to statins. *Journal of Medical Genetics*, **2016**, 53, 835-845	5.8	28

55	LPA and PLG sequence variation and kringle IV-2 copy number in two populations. *Human Heredity*, **2008**, 66, 199-209	1.1	26
54	Successes and challenges of using whole exome sequencing to identify novel genes underlying an inherited predisposition for thoracic aortic aneurysms and acute aortic dissections. *Trends in Cardiovascular Medicine*, **2014**, 24, 53-60	6.9	25
53	The environmental genome project: reference polymorphisms for drug metabolism genes and genome-wide association studies. *Drug Metabolism Reviews*, **2008**, 40, 241-61	7	25
52	High-throughput genotyping of intermediate-size structural variation. *Human Molecular Genetics*, **2006**, 15, 1159-67	5.6	25
51	Sequence variation in the human T-cell receptor loci. *Immunological Reviews*, **2002**, 190, 26-39	11.3	25
50	A common VLDLR polymorphism interacts with APOE genotype in the prediction of carotid artery disease risk. *Journal of Lipid Research*, **2008**, 49, 588-96	6.3	24
49	Whole Exome Sequencing in Atrial Fibrillation. *PLoS Genetics*, **2016**, 12, e1006284	6	24
48	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. *Human Genetics*, **2018**, 137, 735-752	6.3	24
47	Calling Star Alleles With Stargazer in 28 Pharmacogenes With Whole Genome Sequences. *Clinical Pharmacology and Therapeutics*, **2019**, 106, 1328-1337	6.1	23
46	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
45	Patient genotypes impact survival after surgery for isolated congenital heart disease. *Annals of Thoracic Surgery*, **2014**, 98, 104-10; discussion 110-1	2.7	22
44	D-Dimer in African Americans: Whole Genome Sequence Analysis and Relationship to Cardiovascular Disease Risk in the Jackson Heart Study. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2017**, 37, 2220-2227	9.4	22
43	Rare coding variation in paraoxonase-1 is associated with ischemic stroke in the NHLBI Exome Sequencing Project. *Journal of Lipid Research*, **2014**, 55, 1173-8	6.3	19
42	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. *Journal of Clinical Investigation*, **2020**, 130, 4423-4439	15.9	19
41	Preconception Carrier Screening by Genome Sequencing: Results from the Clinical Laboratory. *American Journal of Human Genetics*, **2018**, 102, 1078-1089	11	18
40	Genomic characterization of the RH locus detects complex and novel structural variation in multi-ethnic cohorts. *Genetics in Medicine*, **2019**, 21, 477-486	8.1	17
39	Massively parallel sequencing: the new frontier of hematologic genomics. *Blood*, **2013**, 122, 3268-75	2.2	17
38	Linkage and association of phospholipid transfer protein activity to LASS4. *Journal of Lipid Research* , **2011**, 52, 1837-46	6.3	17

37	Allelic spectrum of the natural variation in CRP. *Human Genetics*, **2006**, 119, 496-504	6.3	17
36	Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. *Mitochondrion*, **2017**, 34, 84-90	4.9	16
35	Targeted enrichment of specific regions in the human genome by array hybridization. *Current Protocols in Human Genetics*, **2010**, Chapter 18, Unit 18.3	3.2	16
34	Sequence diversity, natural selection and linkage disequilibrium in the human T cell receptor alpha/delta locus. *Human Genetics*, **2006**, 119, 255-66	6.3	16
33	Targeted long-read sequencing identifies missing disease-causing variation. *American Journal of Human Genetics*, **2021**, 108, 1436-1449	11	16
32	TagSNP evaluation for the association of 42 inflammation loci and vascular disease: evidence of IL6, FGB, ALOX5, NFKBIA, and IL4R loci effects. *Human Genetics*, **2007**, 121, 65-75	6.3	15
31	MED resulting from recessively inherited mutations in the gene encoding calcium-activated nucleotidase CANT1. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2415-2421	2.5	14
30	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
29	A variational Bayes discrete mixture test for rare variant association. *Genetic Epidemiology*, **2014**, 38, 21-30	2.6	12
28	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. *Human Genetics*, **2019**, 138, 593-600	6.3	11
27	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
26	Whole-exome imputation of sequence variants identified two novel alleles associated with adult body height in African Americans. *Human Molecular Genetics*, **2014**, 23, 6607-15	5.6	11
25	Influence of common and rare genetic variation on warfarin dose among African-Americans and European-Americans using the exome array. *Pharmacogenomics*, **2017**, 18, 1059-1073	2.6	10
24	Hypertension and single nucleotide polymorphisms. *Current Hypertension Reports*, **2000**, 2, 44-9	4.7	8
23	Identifylng DNA polymorphisms in humanTCRA/D variable genes by direct sequencing of PCR products. *Immunogenetics*, **1996**, 44, 121-127	3.2	8
22	Association Between Absolute Neutrophil Count and Variation at TCIRG1: The NHLBI Exome Sequencing Project. *Genetic Epidemiology*, **2016**, 40, 470-4	2.6	8
21	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. *Circulation: Cardiovascular Genetics*, **2016**, 9, 368-74		7
20	DIAPH1 Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. *JAMA Neurology*, **2021**, 78, 993-1003	17.2	7

19	Targeted interrogation of copy number variation using SCIMMkit. *Bioinformatics*, **2010**, 26, 120-2	7.2	6
18	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
17	Rare loss of function variants in candidate genes and risk of colorectal cancer. *Human Genetics*, **2018**, 137, 795-806	6.3	6
16	Integrating host genomics with surveillance for invasive bacterial diseases. *Emerging Infectious Diseases*, **2008**, 14, 1138-40	10.2	5
15	Common genetic variation in the prothrombin gene, hormone therapy, and incident nonfatal myocardial infarction in postmenopausal women. *American Journal of Epidemiology*, **2006**, 163, 600-7	3.8	5
14	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
13	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
12	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
11	Genetic analysis ofde novovariants reveals sex differences in complex and isolated congenital diaphragmatic hernia and indicatesMYRFas a candidate gene		3
10	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. *European Journal of Human Genetics*, **2016**, 24, 1181-7	5.3	2
9	Genotyping by ligation assays. *Current Protocols in Human Genetics*, **2001**, Chapter 2, Unit 2.6	3.2	2
8	Identifying DNA polymorphisms in human TCRA/D variable genes by direct sequencing of PCR products. *Immunogenetics*, **1996**, 44, 121-127	3.2	2
7	Genetic interactions drive heterogeneity in causal variant effect sizes for gene expression and complex traits		1
6	ARMC9 and TOGARAM1 define a Joubert syndrome-associated protein module that regulates axonemal post-translational modifications and cilium stability		1
5	Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features		1
4	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. *Human Genetics and Genomics Advances*, **2021**, 2, 100016-100016	0.8	1
3	Analysis of Sequence Variations. *Methods and Principles in Medicinal Chemistry*, **2001**, 49-68	0.4	
2	Whole-Exome Sequencing and hiPSC Cardiomyocyte Models Identify , , and of Potential Importance to Left Ventricular Hypertrophy in an African Ancestry Population. *Frontiers in Genetics*, **2021**, 12, 588452	4.5	

1	Novel biallelic variants affecting the OTU domain of the gene OTUD6B associate with severe intellectual disability syndrome and molecular dynamics simulations.. *European Journal of Medical Genetics*, **2022**, 104497	2.6