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List of Publications by Year in descending order

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81743 149479 30,262 53 39 56 citations g-index h-index papers 59 59 59 36645 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature, 2007, 447, 661-678.	13.7	8,895
2	A Common Variant in the FTO Gene Is Associated with Body Mass Index and Predisposes to Childhood and Adult Obesity. Science, 2007, 316, 889-894.	6.0	3,884
3	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	9.4	2,634
4	Replication of Genome-Wide Association Signals in UK Samples Reveals Risk Loci for Type 2 Diabetes. Science, 2007, 316, 1336-1341.	6.0	2,040
5	Hundreds of variants clustered in genomic loci and biological pathways affect human height. Nature, 2010, 467, 832-838.	13.7	1,789
6	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. Nature Genetics, 2008, 40, 638-645.	9.4	1,683
7	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. Nature Genetics, 2007, 39, 1329-1337.	9.4	1,298
8	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. Nature Genetics, 2010, 42, 949-960.	9.4	836
9	Genome-wide association analysis identifies 20 loci that influence adult height. Nature Genetics, 2008, 40, 575-583.	9.4	742
10	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	13.7	737
11	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072.	1.5	415
12	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. Nature Genetics, 2014, 46, 812-814.	9.4	411
13	A common variant of HMGA2 is associated with adult and childhood height in the general population. Nature Genetics, 2007, 39, 1245-1250.	9.4	373
14	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
15	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
16	Common variants in WFS1 confer risk of type 2 diabetes. Nature Genetics, 2007, 39, 951-953.	9.4	333
17	Assessing the Combined Impact of 18 Common Genetic Variants of Modest Effect Sizes on Type 2 Diabetes Risk. Diabetes, 2008, 57, 3129-3135.	0.3	279
18	Common Variation in the <i>FTO</i> Gene Alters Diabetes-Related Metabolic Traits to the Extent Expected Given Its Effect on BMI. Diabetes, 2008, 57, 1419-1426.	0.3	277

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19	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	9.4	255
20	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet, The, 2015, 386, 957-963.	6.3	250
21	GATA6 haploinsufficiency causes pancreatic agenesis in humans. Nature Genetics, 2012, 44, 20-22.	9.4	249
22	Improved genetic testing for monogenic diabetes using targeted next-generation sequencing. Diabetologia, 2013, 56, 1958-1963.	2.9	248
23	Loss-of-function nuclear factor \hat{I}^{g} B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. Journal of Allergy and Clinical Immunology, 2018, 142, 1285-1296.	1.5	185
24	Loss of the interleukin-6 receptor causes immunodeficiency, atopy, and abnormal inflammatory responses. Journal of Experimental Medicine, 2019, 216, 1986-1998.	4.2	153
25	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. Nature Genetics, 2013, 45, 947-950.	9.4	151
26	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	13.7	148
27	Type 2 Diabetes Risk Alleles Are Associated With Reduced Size at Birth. Diabetes, 2009, 58, 1428-1433.	0.3	135
28	Analysis of Transcription Factors Key for Mouse Pancreatic Development Establishes NKX2-2 and MNX1 Mutations as Causes of Neonatal Diabetes in Man. Cell Metabolism, 2014, 19, 146-154.	7.2	123
29	<i>GATA4</i> Mutations Are a Cause of Neonatal and Childhood-Onset Diabetes. Diabetes, 2014, 63, 2888-2894.	0.3	108
30	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. Journal of the American Society of Nephrology: JASN, 2017, 28, 2529-2539.	3.0	99
31	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. Journal of Allergy and Clinical Immunology, 2020, 146, 901-911.	1.5	78
32	A <i>CACNA1D</i> mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. Pediatric Diabetes, 2017, 18, 320-323.	1.2	67
33	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	1.1	64
34	Recessively Inherited <i>LRBA </i> Mutations Cause Autoimmunity Presenting as Neonatal Diabetes. Diabetes, 2017, 66, 2316-2322.	0.3	59
35	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	1.4	51
36	What will whole genome searches for susceptibility genes for common complex disease offer to clinical practice?. Journal of Internal Medicine, 2008, 263, 16-27.	2.7	49

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37	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	2.6	46
38	Next generation sequencing of chromosomal rearrangements in patients with split-hand/split-foot malformation provides evidence for <i>DYNC1I1</i> exonic enhancers of <i>DLX5/6</i> expression in humans. Journal of Medical Genetics, 2014, 51, 264-267.	1.5	43
39	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. American Journal of Human Genetics, 2019, 104, 985-989.	2.6	43
40	Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated with the risk of type 2 diabetes. Diabetologia, 2008, 51, 2205-2213.	2.9	41
41	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 2303-2306.	1.5	40
42	Polygenic Risk Variants for Type 2 Diabetes Susceptibility Modify Age at Diagnosis in Monogenic <i>HNF1A</i> Diabetes. Diabetes, 2010, 59, 266-271.	0.3	37
43	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
44	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. European Journal of Human Genetics, 2015, 23, 1744-1748.	1.4	34
45	A rare SNP in pre-miR-34a is associated with increased levels of miR-34a in pancreatic beta cells. Acta Diabetologica, 2014, 51, 325-329.	1.2	30
46	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	2.6	26
47	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. American Journal of Human Genetics, 2016, 98, 857-868.	2.6	21
48	Thousands of missing variants in the UK Biobank are recoverable by genome realignment. Annals of Human Genetics, 2020, 84, 214-220.	0.3	20
49	<i><scp>SOS</scp>1</i> frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. Clinical Endocrinology, 2016, 84, 715-719.	1.2	11
50	ADA2 deficiency complicated by EBV-driven lymphoproliferative disease. Clinical Immunology, 2020, 215, 108443.	1.4	9
51	Copy number variation of <i>LINGO1</i> in familial dystonic tremor. Neurology: Genetics, 2019, 5, e307.	0.9	8
52	Pathogenic NFKB2 variant in the ankyrin repeat domain (R635X) causes a variable antibody deficiency. Clinical Immunology, 2019, 203, 23-27.	1.4	5
53	Predicting the Occurrence of Variants in RAG1 and RAG2. Journal of Clinical Immunology, 2019, 39, 688-701.	2.0	3