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List of Publications by Year
in descending order

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125	12,761	50276	29157
papers	citations	46 h-index	104 g-index

129	129	129	12604
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional Genomic Screening in Human Pluripotent Stem Cells Reveals New Roadblocks in Early Pancreatic Endoderm Formation. Cells, 2022, 11, 582.	4.1	2
2	Familial autoimmunity in neurological patients with GAD65 antibodies: an interview-based study. Journal of Neurology, 2021, 268, 2515-2522.	3.6	4
3	DNAJC3 deficiency induces Î²-cell mitochondrial apoptosis and causes syndromic young-onset diabetes. European Journal of Endocrinology, 2021, 184, 455-468.	3.7	29
4	Human Pluripotent Stem Cells Go Diabetic: A Glimpse on Monogenic Variants. Frontiers in Endocrinology, 2021, 12, 648284.	3.5	2
5	Mutations and variants of ONECUT1 in diabetes. Nature Medicine, 2021, 27, 1928-1940.	30.7	24
6	Transcriptional changes and the role of ONECUT1 in hPSC pancreatic differentiation. Communications Biology, 2021, 4, 1298.	4.4	16
7	General regression model: A â€œmodelâ€freeâ€•association test for quantitative traits allowing to test for the underlying genetic model. Annals of Human Genetics, 2020, 84, 280-290.	0.8	0
8	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, 2020, 130, 6338-6353.	8.2	58
9	Molecular genetics of the transcription factor GLIS3 identifies its dual function in beta cells and neurons. Genomics, 2018, 110, 98-111.	2.9	22
10	A systematic review of nonâ€genetic predictors and genetic factors of glycated haemoglobin in type 1 diabetes one year after diagnosis. Diabetes/Metabolism Research and Reviews, 2018, 34, e3051.	4.0	11
11	dUTPase (DUT) Is Mutated in a Novel Monogenic Syndrome With Diabetes and Bone Marrow Failure. Diabetes, 2017, 66, 1086-1096.	0.6	22
12	Juvenile-Onset Diabetes and Congenital Cataract: â€œDouble-Geneâ€•Mutations Mimicking a Syndromic Diabetes Presentation. Genes, 2017, 8, 309.	2.4	8
13	A Missense Mutation in PPP1R15B Causes a Syndrome Including Diabetes, Short Stature, and Microcephaly. Diabetes, 2015, 64, 3951-3962.	0.6	71
14	Early-onset diabetes mellitus and neurodevelopmental retardation: the first Greek case of Wolcott-Rallison syndrome. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 967-970.	0.9	5
15	A novel ALMS1 splice mutation in a non-obese juvenile-onset insulin-dependent syndromic diabetic patient. European Journal of Human Genetics, 2014, 22, 140-143.	2.8	19
16	SLC29A3 mutation in a patient with syndromic diabetes with features of pigmented hypertrichotic dermatosis with insulin-dependent diabetes, H syndrome and Faisalabad histiocytosis. Diabetes and Metabolism, 2013, 39, 281-285.	2.9	24
17	GLIS3, a Susceptibility Gene for Type 1 and Type 2 Diabetes, Modulates Pancreatic Beta Cell Apoptosis via Regulation of a Splice Variant of the BH3-Only Protein Bim. PLoS Genetics, 2013, 9, e1003532.	3.5	151
18	tRNA Methyltransferase Homolog Gene TRMT10A Mutation in Young Onset Diabetes and Primary Microcephaly in Humans. PLoS Genetics, 2013, 9, e1003888.	3.5	103

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19	Evidence of Gene-Gene Interaction and Age-at-Diagnosis Effects in Type 1 Diabetes. Diabetes, 2012, 61, 3012-3017.	0.6	60
20	Confirmation of novel type 1 diabetes risk loci in families. Diabetologia, 2012, 55, 996-1000.	6.3	50
21	Tests for Genetic Interactions in Type 1 Diabetes. Diabetes, 2011, 60, 1030-1040.	0.6	43
22	Wolcott-Rallison syndrome due to the same mutation (W522X) in EIF2AK3 in two unrelated families and review of the literature*. Pediatric Diabetes, 2010, 11, 279-285.	2.9	43
23	A Novel Hypomorphic PDX1 Mutation Responsible for Permanent Neonatal Diabetes With Subclinical Exocrine Deficiency. Diabetes, 2010, 59, 733-740.	0.6	85
24	Genetics of Type 1 Diabetes: What's Next?. Diabetes, 2010, 59, 1561-1571.	0.6	256
25	Wolcott-Rallison syndrome. Orphanet Journal of Rare Diseases, 2010, 5, 29.	2.7	164
26	Genome-Wide Scan for Linkage to Type 1 Diabetes in 2,496 Multiplex Families From the Type 1 Diabetes Genetics Consortium. Diabetes, 2009, 58, 1018-1022.	0.6	87
27	The large form of human 2â€²,5â€²-Oligoadenylate Synthetase (OAS3) exerts antiviral effect against Chikungunya virus. Virology, 2009, 384, 216-222.	2.4	90
28	Current status and the future for the genetics of type I diabetes. Genes and Immunity, 2009, 10, S128-S131.	4.1	8
29	Overview of the Type I Diabetes Genetics Consortium. Genes and Immunity, 2009, 10, S1-S4.	4.1	57
30	rs2476601 T allele (R620W) defines high-risk PTPN22 type I diabetes-associated haplotypes with preliminary evidence for an additional protective haplotype. Genes and Immunity, 2009, 10, S21-S26.	4.1	27
31	Association analysis of SNPs in the IL4R locus with type I diabetes. Genes and Immunity, 2009, 10, S33-S41.	4.1	16
32	Evidence for association of the TCF7 locus with type I diabetes. Genes and Immunity, 2009, 10, S54-S59.	4.1	28
33	Evaluation of IL12B as a candidate type I diabetes susceptibility gene using data from the Type I Diabetes Genetics Consortium. Genes and Immunity, 2009, 10, S64-S68.	4.1	23
34	The Type I Diabetes Genetics Consortium â€˜Rapid Responseâ€™ family-based candidate gene study: strategy, genes selection, and main outcome. Genes and Immunity, 2009, 10, S121-S127.	4.1	15
35	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nature Genetics, 2009, 41, 703-707.	21.4	1,513
36	FCRL3 â˜’169CT functional polymorphism in type 1 diabetes and autoimmunity traits. Biomedicine and Pharmacotherapy, 2008, 62, 153-157.	5.6	16

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37	WFS1 mutations are frequent monogenic causes of juvenile-onset diabetes mellitus in Lebanon. Human Molecular Genetics, 2008, 17, 4012-4021.	2.9	48
38	A Human Type 1 Diabetes Susceptibility Locus Maps to Chromosome 21q22.3. Diabetes, 2008, 57, 2858-2861.	0.6	103
39	Genetic Determination and Linkage Mapping of Plasmodium falciparum Malaria Related Traits in Senegal. PLoS ONE, 2008, 3, e2000.	2.5	49
40	PTPN22 R620W Functional Variant in Type 1 Diabetes and Autoimmunity Related Traits. Diabetes, 2007, 56, 522-526.	0.6	57
41	Severe FOXP3+ and NaÃ¯ve T Lymphopenia in a Non-IPEX Form of Autoimmune Enteropathy Combined With an Immunodeficiency. Gastroenterology, 2007, 132, 1694-1704.	1.3	26
42	Mutations in GLIS3 are responsible for a rare syndrome with neonatal diabetes mellitus and congenital hypothyroidism. Nature Genetics, 2006, 38, 682-687.	21.4	327
43	The Type 1 Diabetes Genetics Consortium. Annals of the New York Academy of Sciences, 2006, 1079, 1-8.	3.8	116
44	Microcephaly and simplified gyral pattern of the brain associated with early onset insulin-dependent diabetes mellitus. Neurogenetics, 2006, 7, 259-263.	1.4	24
45	Genetic study of ICAM1 in clinical malaria in Senegal. Tissue Antigens, 2005, 65, 474-480.	1.0	15
46	A variant in the CD209 promoter is associated with severity of dengue disease. Nature Genetics, 2005, 37, 507-513.	21.4	267
47	Reverse cascade screening of newborns for hereditary haemochromatosis: a model for other late onset diseases?. Journal of Medical Genetics, 2005, 42, 390-395.	3.2	23
48	Recessive mutations in PTHR1 cause contrasting skeletal dysplasias in Eiken and Blomstrand syndromes. Human Molecular Genetics, 2005, 14, 1-5.	2.9	116
49	Type 1 Diabetes. Diabetes, 2005, 54, 2995-3001.	0.6	221
50	Crohnâ€™s disease associated CARD15 (NOD2) variants are not involved in the susceptibility to type 1 diabetes. Molecular Genetics and Metabolism, 2005, 86, 379-383.	1.1	12
51	Wolcott-Rallison Syndrome. Diabetes, 2004, 53, 1876-1883.	0.6	165
52	Genetic and functional evaluation of an interleukin-12 polymorphism (IDDM18) in families with type 1 diabetes. Journal of Medical Genetics, 2004, 41, e39-e39.	3.2	33
53	Lost in translation. EMBO Reports, 2004, 5, 1015-1015.	4.5	0
54	Genotype at a promoter polymorphism of the interleukin-6 gene is associated with baseline levels of plasma C-reactive protein. Cardiovascular Research, 2002, 53, 1029-1034.	3.8	227

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55	Trans-ethnic fine mapping of a quantitative trait locus for circulating angiotensin I-converting enzyme (ACE). Human Molecular Genetics, 2001, 10, 1077-1084.	2.9	89
56	EIF2AK3, encoding translation initiation factor 2-Î± kinase 3, is mutated in patients with Wolcott-Rallison syndrome. Nature Genetics, 2000, 25, 406-409.	21.4	733
57	Screening for the GRA mutation in Jamaica. Journal of Human Hypertension, 2000, 14, 157-158.	2.2	5
58	Screening for Hereditary Hemochromatosis. American Journal of Gastroenterology, 2000, 95, 1368-1369.	0.4	7
59	Linkage mapping for hypertension susceptibility genes. Current Hypertension Reports, 1999, 1, 15-24.	3.5	8
60	Measured Haplotype Analysis of the Angiotensin-I Converting Enzyme Gene. Human Molecular Genetics, 1998, 7, 1745-1751.	2.9	197
61	Genetic analysis of chromosome 2 in type 1 diabetes: analysis of putative loci IDDM7, IDDM12, and IDDM13 and candidate genes NRAMP1 and IA-2 and the interleukin-1 gene cluster. IMDIAB Group. Diabetes, 1998, 47, 1797-1799.	0.6	66
62	Genetic Susceptibility for Human Familial Essential Hypertension in a Region of Homology with Blood Pressure Linkage on Rat Chromosome 10. Human Molecular Genetics, 1997, 6, 2077-2085.	2.9	172
63	Comparative mapping of novel simple sequence repeat markers in a hypertension-related region on rat chromosome 1. Mammalian Genome, 1997, 8, 215-217.	2.2	22
64	GÃ©nÃ©tique du diabÃ¨te insulino-dÃ©pendant. Annales De L'Institut Pasteur / ActualitÃ©s, 1996, 7, 13-19.	0.1	0
65	Contribution of genes of the major histocompatibility complex to susceptibility and disease phenotype in inflammatory bowel disease. Lancet, The, 1996, 347, 1212-1217.	13.7	356
66	Genetics of insulin-dependent diabetes mellitus. Current Opinion in Genetics and Development, 1996, 6, 354-360.	3.3	7
67	Genetics of Insulin-Dependent Diabetes Mellitus.. Endocrine Journal, 1996, 43, 605-613.	1.6	65
68	Distribution of HLAâ€DQA1 and â€DQB1 alleles and DQA1â€DQB1 genotypes among Senegalese patients with insulinâ€dependent diabetes mellitus. Tissue Antigens, 1996, 47, 333-337.	1.0	10
69	A missense mutation in the glucagon receptor gene is associated with nonâ€“insulinâ€“dependent diabetes mellitus. Nature Genetics, 1995, 9, 299-304.	21.4	177
70	Regulation of insulin gene expression by the IDDM associated, insulin locus haplotype. Human Molecular Genetics, 1995, 4, 501-506.	2.9	98
71	The CEPH Consortium Linkage Map of Human Chromosome 11. Genomics, 1995, 27, 101-112.	2.9	19
72	Evaluation of the SA Locus in Human Hypertension. Hypertension, 1995, 25, 6-13.	2.7	40

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73	Genetic mapping of a susceptibility locus for insulin-dependent diabetes mellitus on chromosome llq. Nature, 1994, 371, 161-164.	27.8	412
74	Systematic study of human Î±Î² T cell receptor V segments shows allelic variations resulting in a large number of distinct T cell receptor haplotypes. European Journal of Immunology, 1993, 23, 1277-1283.	2.9	53
75	Genetic mapping through the use of synthetic tandem repeats in the mouse genome. Mammalian Genome, 1993, 4, 135-140.	2.2	4
76	Susceptibility to insulin dependent diabetes mellitus maps to a 4.1 kb segment of DNA spanning the insulin gene and associated VNTR. Nature Genetics, 1993, 4, 305-310.	21.4	253
77	A Radiation Hybrid Map of Human Chromosome 11q22-q23 Containing the Ataxia-Telangiectasia Disease Locus. Genomics, 1993, 17, 1-5.	2.9	23
78	Genetic analysis of diabetes and insulitis in an interspecific cross of the nonobese diabetic mouse with Mus spretus.. Proceedings of the National Academy of Sciences of the United States of America, 1993, 90, 1877-1881.	7.1	55
79	Dinucleotide repeat polymorphism at the PI locus. Human Molecular Genetics, 1993, 2, 2203-2203.	2.9	0
80	Precise Localization of a Gene Responsible for Ataxia-Telangiectasia on Chromosome 11q. , 1993, , 23-35.		2
81	Maternal inheritance of atopic IgE responsiveness on chromosome 11 q. Lancet, The, 1992, 340, 381-384.	13.7	357
82	Identification of a CA repeat at the TCRA locus using yeast artificial chromosomes: A general method for generating highly polymorphic markers at chosen loci. Genomics, 1992, 13, 820-825.	2.9	43
83	Ataxia-telangiectasia: Linkage analysis in highly inbred Arab and Druze families and differentiation from an ataxia-microcephaly-cataract syndrome. Human Genetics, 1992, 88, 619-626.	3.8	23
84	Linkage analysis in juvenile neuronal ceroid lipofuscinosis. American Journal of Medical Genetics Part A, 1992, 42, 542-545.	2.4	10
85	Chromosomal mapping of two genetic loci associated with blood-pressure regulation in hereditary hypertensive rats. Nature, 1991, 353, 521-529.	27.8	633
86	Insulin-IGF2 region on chromosome 11p encodes a gene implicated in HLA-DR4-dependent diabetes susceptibility. Nature, 1991, 354, 155-159.	27.8	388
87	Simultaneous localization of cosmids and chromosome R-banding by fluorescence microscopy: application to regional mapping of human chromosome 11.. Proceedings of the National Academy of Sciences of the United States of America, 1990, 87, 6639-6643.	7.1	114
88	In situ hybridization ascertains the presence of a translocation t(6;11) in an acute monocytic leukemia. Genes Chromosomes and Cancer, 1990, 2, 341-344.	2.8	16
89	Minisatellite linkage maps in the mouse by cross-hybridization with human probes containing tandem repeats.. Proceedings of the National Academy of Sciences of the United States of America, 1990, 87, 4585-4589.	7.1	73
90	Batten disease (Spielmeyer-Vogt disease, juvenile onset neuronal ceroid-lipofuscinosis) gene (CLN3) maps to human chromosome 16. Genomics, 1990, 8, 387-390.	2.9	100

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91	A detailed genetic map of the long arm of chromosome 11. Genomics, 1990, 7, 335-345.	2.9	110
92	Mapping of a gene determining tuberous sclerosis to human chromosome 11q14î—¸11q23. Genomics, 1990, 6, 105-114.	2.9	109
93	A primary map of 24 loci on human chromosome 16. Genomics, 1990, 6, 419-427.	2.9	44
94	Isolation and mapping of a polymorphic DNA sequence (CJ52.197) on chromosome 16 [D16S156]. Nucleic Acids Research, 1989, 17, 8015-8015.	14.5	0
95	Isolation and mapping of a polymorphic DNA sequence (pCJS2.94T1) on chromosome 16 [D16S159]. Nucleic Acids Research, 1989, 17, 7543-7543.	14.5	0
96	Isolation and mapping of a polymorphic DNA sequence (CJ52.199) on chromosome 16 [D16S155]. Nucleic Acids Research, 1989, 17, 8020-8020.	14.5	0
97	Isolation and mapping of a polymorphic DNA sequence (pCJ52.99M2) on chromosome 11 [D11S389]. Nucleic Acids Research, 1989, 17, 9504-9504.	14.5	1
98	Isolation and mapping of a polymorphic DNA sequence (CJ52.10) on chromosome 16 [D16S153]. Nucleic Acids Research, 1989, 17, 8012-8012.	14.5	0
99	Isolation and mapping of a polymorphic DNA sequence (CJ52.27) on chromosome 16 [D16S149]. Nucleic Acids Research, 1989, 17, 8013-8013.	14.5	0
100	Isolation and mapping of a polymorphic DNA sequence (CJ52.1) on chromosome 16 [D16S152]. Nucleic Acids Research, 1989, 17, 8014-8014.	14.5	0
101	Isolation and mapping of a polymorphic DNA sequence (pCJ52.102T1) on chromosome 11 [D11S387]. Nucleic Acids Research, 1989, 17, 9509-9509.	14.5	0
102	Isolation and mapping of a polymorphic DNA sequence (HBI18P1) on chromosome 11 [D11S147]. Nucleic Acids Research, 1989, 17, 9510-9510.	14.5	2
103	Isolation and mapping of a polymorphic DNA sequence (p1CJ52.208M2) on chromosome 11 [D11S351]. Nucleic Acids Research, 1989, 17, 9508-9508.	14.5	0
104	Isolation and mapping of a polymorphic DNA sequence (pCJ52.75M1) on chromosome 11 [D11S385]. Nucleic Acids Research, 1989, 17, 9505-9505.	14.5	1
105	Isolation and mapping of a polymorphic DNA sequence (pCJ52.209M1) on chromosome 16 [D16S151]. Nucleic Acids Research, 1989, 17, 7542-7542.	14.5	0
106	Isolation and mapping of a polymorphic DNA sequence (CJ52.5) on chromosome 11 [D11S386]. Nucleic Acids Research, 1989, 17, 9503-9503.	14.5	1
107	Isolation and mapping of a polymorphic DNA sequence (2CJ52.208M2) on chromosome 11 [D11S351]. Nucleic Acids Research, 1989, 17, 9507-9507.	14.5	0
108	Isolation and mapping of a polymorphic DNA sequence (CJ52.105) on chromosome 16 [D16S154]. Nucleic Acids Research, 1989, 17, 8016-8016.	14.5	0

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109	Isolation and mapping of a polymorphic DNA sequence (pCJ52.95M1) on chromosome 16 [D16S148]. Nucleic Acids Research, 1989, 17, 8017-8017.	14.5	0
110	Isolation and mapping of a polymorphic DNA sequence (CJ52.112) on chromosome 16 [D16S158]. Nucleic Acids Research, 1989, 17, 8019-8019.	14.5	0
111	Isolation and mapping of a polymorphic DNA sequence (pCJ52.209M2) on chromosome 16 [D16S151]. Nucleic Acids Research, 1989, 17, 8021-8021.	14.5	0
112	Isolation and mapping of a polymorphic DNA sequence (pCJ52.196M1) on chromosome 16 [D16S160]. Nucleic Acids Research, 1989, 17, 8018-8018.	14.5	0
113	Isolation and mapping of a polymorphic DNA sequence (CJ52.96) on chromosome 16 [D16S157]. Nucleic Acids Research, 1989, 17, 9506-9506.	14.5	0
114	Isolation and mapping of a polymorphic DNA sequence (CJ52.161) on chromosome 16 [D16S150]. Nucleic Acids Research, 1989, 17, 8402-8402.	14.5	0
115	Isolation and mapping of a polymorphic DNA sequence (CJ52.4) on chromosome 11 [D11S388]. Nucleic Acids Research, 1989, 17, 9502-9502.	14.5	0
116	Isolation and mapping of a polymorphic DNA sequence (CJ52.193) on chromosome 11 [D11S384]. Nucleic Acids Research, 1989, 17, 9501-9501.	14.5	3
117	Isolation and mapping of a polymorphic DNA sequence (CJ52.15) on chromosome 11 [D11S383]. Nucleic Acids Research, 1989, 17, 10149-10149.	14.5	0
118	Linkage maps of human chromosomes. Genome, 1989, 31, 1066-1072.	2.0	11
119	Linkage studies in X-linked Alport's syndrome. Human Genetics, 1988, 81, 85-87.	3.8	39
120	Characterization of a human â€˜midisatelliteâ€™ sequence. Nucleic Acids Research, 1987, 15, 2537-2547.	14.5	64
121	Human chromosome 22.. Journal of Medical Genetics, 1987, 24, 65-78.	3.2	45
122	Complete cDNA sequence of human complement C1s and close physical linkage of the homologous genes C1s and C1r. Biochemistry, 1987, 26, 8516-8524.	2.5	128
123	Construction of Human Genetic Linkage Maps: I. Progress and Perspectives. Cold Spring Harbor Symposia on Quantitative Biology, 1986, 51, 29-38.	1.1	13
124	The beta chorionic gonadotropin-beta luteinizing gene cluster maps to human chromosome 19. Human Genetics, 1984, 67, 174-177.	3.8	42
125	Strategies for multilocus linkage analysis in humans.. Proceedings of the National Academy of Sciences of the United States of America, 1984, 81, 3443-3446.	7.1	2,517