Yalda Jamshidi

List of Publications by Year
in descending order

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89 papers	6,970 citations	87888 38 h-index	66911 78 g-index
97 all docs	97 docs citations	97 times ranked	14838 citing authors

#	Article	IF	Citations
1	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
2	Meta-Analysis of Genome-Wide Association Studies in >80 000 Subjects Identifies Multiple Loci for C-Reactive Protein Levels. Circulation, 2011, 123, 731-738.	1.6	461
3	Genome Analyses of >200,000 Individuals Identify 58 Loci for Chronic Inflammation and Highlight Pathways that Link Inflammation and Complex Disorders. American Journal of Human Genetics, 2018, 103, 691-706.	6.2	326
4	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. Nature Genetics, 2010, 42, 1068-1076.	21.4	308
5	Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel. Nature Communications, 2015, 6, 8111.	12.8	300
6	Identification of heart rateâ€“associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	21.4	282
7	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
8	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	6.2	197
9	Elite swimmers and the D allele of the ACE I/D polymorphism. Human Genetics, 2001, 108, 230-232.	3.8	185
10	Variation in the PPARÎ± gene is associated with altered function in vitro and plasma lipid concentrations in Type II diabetic subjects. Diabetologia, 2000, 43, 673-680.	6.3	180
11	A Large Candidate Gene Survey Identifies the KCNE1 D85N Polymorphism as a Possible Modulator of Drug-Induced Torsades de Pointes. Circulation: Cardiovascular Genetics, 2012, 5, 91-99.	5.1	150
12	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. PLoS Medicine, 2016, 13, e1001976.	8.4	150
13	Peroxisome Proliferatorâ€“Activated Receptor Î± Gene Regulates Left Ventricular Growth in Response to Exercise and Hypertension. Circulation, 2002, 105, 950-955.	1.6	149
14	Characterization of the Human PPARÎ± Promoter: Identification of a Functional Nuclear Receptor Response Element. Molecular Endocrinology, 2002, 16, 1013-1028.	3.7	144
15	Peroxisome Proliferator-Activated Receptor Î± Gene Variants Influence Progression of Coronary Atherosclerosis and Risk of Coronary Artery Disease. Circulation, 2002, 105, 1440-1445.	1.6	136
16	Genome-wide association analysis identifies multiple loci related to resting heart rate. Human Molecular Genetics, 2010, 19, 3885-3894.	2.9	133
17	Meta-analysis of Dense Genecentric Association Studies Reveals Common and Uncommon Variants Associated with Height. American Journal of Human Genetics, 2011, 88, 6-18.	6.2	122
18	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	2.8	113

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19	Role of common and rare variants in SCN10A: results from the Brugada syndrome QRS locus gene discovery collaborative study. Cardiovascular Research, 2015, 106, 520-529.	3.8	108
20	Common Variation in the NOS1AP Gene Is Associated With Drug-Induced QT Prolongation and Ventricular Arrhythmia. Journal of the American College of Cardiology, 2012, 60, 841-850.	2.8	101
21	Characterization of the Human PPARÂ Promoter: Identification of a Functional Nuclear Receptor Response Element. Molecular Endocrinology, 2002, 16, 1013-1028.	3.7	85
22	Mining the Human Phenome Using Allelic Scores That Index Biological Intermediates. PLoS Genetics, 2013, 9, e1003919.	3.5	84
23	Insertion/Deletion Polymorphism of the Angiotensin I-Converting Enzyme Gene and Arterial Oxygen Saturation at High Altitude. American Journal of Respiratory and Critical Care Medicine, 2002, 166, 362-366.	5.6	82
24	Gene-centric meta-analyses of 108 912 individuals confirm known body mass index loci and reveal three novel signals. Human Molecular Genetics, 2013, 22, 184-201.	2.9	82
25	Whole-genome sequence-based analysis of thyroid function. Nature Communications, 2015, 6, 5681.	12.8	75
26	Endurance enhancement related to the human angiotensin I-converting enzyme I-D polymorphism is not due to differences in the cardiorespiratory response to training. European Journal of Applied Physiology, 2002, 86, 240-244.	2.5	72
27	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. Nature Communications, 2018, 9, 2904.	12.8	71
28	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	6.2	67
29	A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. Nature Communications, 2014, 5, 4871.	12.8	62
30	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	12.8	59
31	Genome Wide Analysis of Drug-Induced Torsades de Pointes: Lack of Common Variants with Large Effect Sizes. PLoS ONE, 2013, 8, e78511.	2.5	57
32	Common Genetic Variation Near the Phospholamban Gene Is Associated with Cardiac Repolarisation: Meta-Analysis of Three Genome-Wide Association Studies. PLoS ONE, 2009, 4, e6138.	2.5	53
33	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
34	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394.	3.3	50
35	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. Genome Biology, 2018, 19, 87.	8.8	47
36	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45

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37	The SH2B Gene is Associated with Serum Leptin and Body Fat in Normal Female Twins. Obesity, 2007, 15, 5-9.	3.0	44
38	Pleiotropy among Common Genetic Loci Identified for Cardiometabolic Disorders and C-Reactive Protein. PLoS ONE, 2015, 10, e0118859.	2.5	43
39	MicroRNA-153 targeting of KCNQ4 contributes to vascular dysfunction in hypertension. Cardiovascular Research, 2016, 112, 581-589.	3.8	43
40	Hypertrophic effects of urocortin homologous peptides are mediated via activation of the Akt pathway. Biochemical and Biophysical Research Communications, 2005, 328, 442-448.	2.1	39
41	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790.	12.8	39
42	The use of polygenic risk scores in pre-implantation genetic testing: an unproven, unethical practice. European Journal of Human Genetics, 2022, 30, 493-495.	2.8	38
43	Fine-mapping, novel loci identification, and SNP association transferability in a genome-wide association study of QRS duration in African Americans. Human Molecular Genetics, 2016, 25, 4350-4368.	2.9	37
44	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.1	35
45	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. Human Molecular Genetics, 2019, 28, 1919-1929.	2.9	35
46	Heritability of QT Interval: How Much Is Explained by Genes for Resting Heart Rate?. Journal of Cardiovascular Electrophysiology, 2008, 19, 386-391.	1.7	34
47	Tribal ethnicity and CYP2B6 genetics in Ugandan and Zimbabwean populations in the UK: implications for efavirenz dosing in HIV infection. Journal of Antimicrobial Chemotherapy, 2010, 65, 2614-2619.	3.0	34
48	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
49	Genome-wide association study of circulating interleukin 6 levels identifies novel loci. Human Molecular Genetics, 2021, 30, 393-409.	2.9	32
50	Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. Nature Communications, 2019, 10, 357.	12.8	30
51	Discovery of novel heart rate-associated loci using the Exome Chip. Human Molecular Genetics, 2017, 26, 2346-2363.	2.9	29
52	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. European Journal of Human Genetics, 2019, 27, 952-962.	2.8	29
53	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
54	ExomeChip-Wide Analysis of 95 626 Individuals Identifies 10 Novel Loci Associated With QT and JT Intervals. Circulation Genomic and Precision Medicine, 2018, 11, e001758.	3.6	27

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55	The age-dependency of genetic and environmental influences on serum cytokine levels: A twin study. Cytokine, 2012, 60, 108-113.	3.2	24
56	Genetic determinants of the response to bezafibrate treatment in the lower extremity arterial disease event reduction (LEADER) trial. Atherosclerosis, 2002, 163, 183-192.	0.8	23
57	Digenic inheritance of mutations in the cardiac troponin (TNNT2) and cardiac beta myosin heavy chain (MYH7) as the cause of severe dilated cardiomyopathy. European Journal of Medical Genetics, 2017, 60, 485-488.	1.3	23
58	Testicular expression of TDRD1, TDRD5, TDRD9 and TDRD12 in azoospermia. BMC Medical Genetics, 2020, 21, 33.	2.1	23
59	Common polymorphisms in SOCS3 are not associated with body weight, insulin sensitivity or lipid profile in normal female twins. Diabetologia, 2006, 49, 306-310.	6.3	22
60	Phosphatidylinositol 3-kinase p85Î± regulatory subunit gene PIK3R1 haplotype is associated with body fat and serum leptin in a female twin population. Diabetologia, 2006, 49, 2659-2667.	6.3	22
61	Multiple roles of integrin-Î±3 at the neuromuscular junction. Journal of Cell Science, 2017, 130, 1772-1784.	2.0	22
62	Analysis of enriched rare variants in JPH2-encoded junctophilin-2 among Greater Middle Eastern individuals reveals a novel homozygous variant associated with neonatal dilated cardiomyopathy. Scientific Reports, 2019, 9, 9038.	3.3	22
63	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
64	SHP-2 and PI3-kinase genes PTPN11 and PIK3R1 may influence serum apoB and LDL cholesterol levels in normal women. Atherosclerosis, 2007, 194, e26-e33.	0.8	19
65	Genetic Risk for Primary Open-Angle Glaucoma Determined byLMX1BHaplotypes. , 2009, 50, 1522.		19
66	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. Circulation Genomic and Precision Medicine, 2018, 11, e002037.	3.6	19
67	Coagulation Gene Expression Profiling in Infants With Necrotizing Enterocolitis. Journal of Pediatric Gastroenterology and Nutrition, 2016, 63, e169-e175.	1.8	18
68	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
69	Autosomal recessive cardiomyopathy and sudden cardiac death associated with variants in MYL3. Genetics in Medicine, 2021, 23, 787-792.	2.4	16
70	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. JCI Insight, 2019, 4, .	5.0	15
71	A Comparison of Heritability Estimates by Classical Twin Modeling and Based on Genome-Wide Genetic Relatedness for Cardiac Conduction Traits. Twin Research and Human Genetics, 2017, 20, 489-498.	0.6	14
72	Genetic and environmental influences on stability and change in baseline levels of C-reactive protein: A longitudinal twin study. Atherosclerosis, 2017, 265, 172-178.	0.8	13

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73	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118.	8.2	13
74	Sequencing of SCN5A Identifies Rare and Common Variants Associated With Cardiac Conduction: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium. Circulation: Cardiovascular Genetics, 2014, 7, 365-373.	5.1	12
75	Signal-transduction pathways involved in the hypertrophic effect of hsp56 in neonatal cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2004, 36, 381-392.	1.9	11
76	Novel genes for QTc interval. How much heritability is explained, and how much is left to find?. Genome Medicine, 2010, 2, 35.	8.2	11
77	Common STAT3 Variants Are Not Associated With Obesity or Insulin Resistance in Female Twins*. Obesity, 2007, 15, 1634-1639.	3.0	9
78	Whole exome sequence analysis reveals a homozygous mutation in PNPLA2 as the cause of severe dilated cardiomyopathy secondary to neutral lipid storage disease. International Journal of Cardiology, 2016, 210, 41-44.	1.7	8
79	Undernutrition in adolescence and risk of cardiovascular disease. European Heart Journal, 2012, 33, 433-435.	2.2	7
80	The genetics of proâ€arrhythmic adverse drug reactions. British Journal of Clinical Pharmacology, 2014, 77, 618-625.	2.4	7
81	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Due to MTMR2 Mutations and Implications in Membrane Trafficking. Frontiers in Neuroscience, 2019, 13, 974.	2.8	7
82	The narrow-sense and common single nucleotide polymorphism heritability of early repolarization. International Journal of Cardiology, 2019, 279, 135-140.	1.7	7
83	Causal graphs for the analysis of genetic cohort data. Physiological Genomics, 2020, 52, 369-378.	2.3	4
84	Novel mutation identification and copy number variant detection via exome sequencing in congenital muscular dystrophy. Molecular Genetics & Genomic Medicine, 2020, 8, e1387.	1.2	3
85	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
86	142â€…Update on familial thoracic aortic aneurysm disease in the 100,000 genomes project: space for discovery. , 2019, , .		1
87	Common TGFÎ²2, BMP4, and FOXC1 variants are not associated with primary open-angle glaucoma. Molecular Vision, 2012, 18, 1526-39.	1.1	1
88	Meta-analysis of Dense Genecentric Association Studies Reveals Common and Uncommon Variants Associated with Height. American Journal of Human Genetics, 2012, 90, 1116-1117.	6.2	0
89	Multiple roles of integrin-Î±3 at the neuromuscular junction. Development (Cambridge), 2017, 144, e1.1-e1.1.	2.5	0