Yalda Jamshidi

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7253323/publications.pdf

Version: 2024-02-01

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 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
2	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
3	Autosomal recessive cardiomyopathy and sudden cardiac death associated with variants in MYL3. Genetics in Medicine, 2021, 23, 787-792.	2.4	16
4	Genome-wide association study of circulating interleukin 6 levels identifies novel loci. Human Molecular Genetics, 2021, 30, 393-409.	2.9	32
5	Causal graphs for the analysis of genetic cohort data. Physiological Genomics, 2020, 52, 369-378.	2.3	4
6	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
7	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
8	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
9	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	12.8	59
10	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
11	Testicular expression of TDRD1, TDRD5, TDRD9 and TDRD12 in azoospermia. BMC Medical Genetics, 2020, 21, 33.	2.1	23
12	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
13	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790.	12.8	39
14	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
15	Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. Nature Communications, 2019, 10, 357.	12.8	30
16	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
17	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
18	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45

#	Article	IF	Citations
19	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. Human Molecular Genetics, 2019, 28, 1919-1929.	2.9	35
20	142â€...Update on familial thoracic aortic aneurysm disease in the 100,000 genomes project: space for discovery. , 2019, , .		1
21	The narrow-sense and common single nucleotide polymorphism heritability of early repolarization. International Journal of Cardiology, 2019, 279, 135-140.	1.7	7
22	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. JCI Insight, 2019, 4, .	5.0	15
23	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
24	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
25	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. Circulation Genomic and Precision Medicine, 2018, 11, e002037.	3.6	19
26	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.1	35
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	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. Genome Biology, 2018, 19, 87.	8.8	47
29	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
30	Discovery of novel heart rate-associated loci using the Exome Chip. Human Molecular Genetics, 2017, 26, 2346-2363.	2.9	29
31	Multiple roles of integrin-Î±3 at the neuromuscular junction. Journal of Cell Science, 2017, 130, 1772-1784.	2.0	22
32	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
33	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
34	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
35	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394.	3.3	50
36	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

#	Article	IF	Citations
37	Multiple roles of integrin-Î±3 at the neuromuscular junction. Development (Cambridge), 2017, 144, e1.1-e1.1.	2.5	0
38	MicroRNA-153 targeting of KCNQ4 contributes to vascular dysfunction in hypertension. Cardiovascular Research, 2016, 112, 581-589.	3.8	43
39	Coagulation Gene Expression Profiling in Infants With Necrotizing Enterocolitis. Journal of Pediatric Gastroenterology and Nutrition, 2016, 63, e169-e175.	1.8	18
40	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
41	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	2.8	113
42	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
43	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
44	Pleiotropy among Common Genetic Loci Identified for Cardiometabolic Disorders and C-Reactive Protein. PLoS ONE, 2015, 10, e0118859.	2.5	43
45	Whole-genome sequence-based analysis of thyroid function. Nature Communications, 2015, 6, 5681.	12.8	75
46	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
47	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
48	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
49	Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel. Nature Communications, 2015, 6, 8111.	12.8	300
50	The genetics of proâ€arrhythmic adverse drug reactions. British Journal of Clinical Pharmacology, 2014, 77, 618-625.	2.4	7
51	A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. Nature Communications, 2014, 5, 4871.	12.8	62
52	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
53	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
54	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

#	Article	IF	Citations
55	Identification of heart rateâ€“associated loci and their effects on cardiac conduction and rhythm disorders. Nature Genetics, 2013, 45, 621-631.	21.4	282
56	Mining the Human Phenome Using Allelic Scores That Index Biological Intermediates. PLoS Genetics, 2013, 9, e1003919.	3.5	84
57	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
58	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
59	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
60	Undernutrition in adolescence and risk of cardiovascular disease. European Heart Journal, 2012, 33, 433-435.	2.2	7
61	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
62	The age-dependency of genetic and environmental influences on serum cytokine levels: A twin study. Cytokine, 2012, 60, 108-113.	3.2	24
63	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
64	Common TGFÎ²2, BMP4, and FOXC1 variants are not associated with primary open-angle glaucoma. Molecular Vision, 2012, 18, 1526-39.	1.1	1
65	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
66	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
67	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. Nature Genetics, 2010, 42, 1068-1076.	21.4	308
68	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
69	Genome-wide association analysis identifies multiple loci related to resting heart rate. Human Molecular Genetics, 2010, 19, 3885-3894.	2.9	133
70	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
71	Genetic Risk for Primary Open-Angle Glaucoma Determined byLMX1BHaplotypes. , 2009, 50, 1522.		19
72	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

#	Article	IF	Citations
73	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
74	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
75	Common STAT3 Variants Are Not Associated With Obesity or Insulin Resistance in Female Twins*. Obesity, 2007, 15, 1634-1639.	3.0	9
76	The SH2B Gene is Associated with Serum Leptin and Body Fat in Normal Female Twins. Obesity, 2007, 15, 5-9.	3.0	44
77	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
78	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
79	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
80	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
81	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
82	Peroxisome Proliferatorâ€“Activated Receptor Î± Gene Regulates Left Ventricular Growth in Response to Exercise and Hypertension. Circulation, 2002, 105, 950-955.	1.6	149
83	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
84	Characterization of the Human PPARÎ± Promoter: Identification of a Functional Nuclear Receptor Response Element. Molecular Endocrinology, 2002, 16, 1013-1028.	3.7	144
85	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
86	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
87	Characterization of the Human PPARÂ Promoter: Identification of a Functional Nuclear Receptor Response Element. Molecular Endocrinology, 2002, 16, 1013-1028.	3.7	85
88	Elite swimmers and the D allele of the ACE I/D polymorphism. Human Genetics, 2001, 108, 230-232.	3.8	185
89	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