

# Santiago Rodriguez

## List of Publications by Year in descending order

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Version: 2024-02-01

50  
papers

2,091  
citations

430754

18  
h-index

243529

44  
g-index

52  
all docs

52  
docs citations

52  
times ranked

4680  
citing authors

#	ARTICLE	IF	CITATIONS
1	Confirmed causal effect of obesity on asthma and new insights on potential underlying shared genetic mechanisms. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 484-486.	1.5	2
2	The Y Chromosome: A Complex Locus for Genetic Analyses of Complex Human Traits. <i>Genes</i> , 2020, 11, 1273.	1.0	12
3	A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. <i>Human Genomics</i> , 2019, 13, 6.	1.4	25
4	Associations of Mitochondrial and Nuclear Mitochondrial Variants and Genes with Seven Metabolic Traits. <i>American Journal of Human Genetics</i> , 2019, 104, 112-138.	2.6	106
5	Low platelet count: Predictor of death and graft loss after liver transplantation. <i>World Journal of Hepatology</i> , 2019, 11, 99-108.	0.8	8
6	Association of copy number variation across the genome with neuropsychiatric traits in the general population. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 489-502.	1.1	26
7	Dobutamine stress echocardiography, myocardial perfusion scintigraphy, invasive coronary angiography, and post-liver transplantation events: Systematic review and meta-analysis. <i>Clinical Transplantation</i> , 2018, 32, e13222.	0.8	22
8	Cardiometabolic phenotypes and mitochondrial DNA copy number in two cohorts of UK women. <i>Mitochondrion</i> , 2018, 39, 9-19.	1.6	13
9	Unravelling the Roles of Susceptibility Loci for Autoimmune Diseases in the Post-GWAS Era. <i>Genes</i> , 2018, 9, 377.	1.0	28
10	Using Y-Chromosomal Haplogroups in Genetic Association Studies and Suggested Implications. <i>Genes</i> , 2018, 9, 45.	1.0	4
11	Associations of Y chromosomal haplogroups with cardiometabolic risk factors and subclinical vascular measures in males during childhood and adolescence. <i>Atherosclerosis</i> , 2018, 274, 94-103.	0.4	19
12	Mitochondrial DNA Haplogroups and Breast Cancer Risk Factors in the Avon Longitudinal Study of Parents and Children (ALSPAC). <i>Genes</i> , 2018, 9, 395.	1.0	9
13	Education and myopia: assessing the direction of causality by mendelian randomisation. <i>BMJ: British Medical Journal</i> , 2018, 361, k2022.	2.4	184
14	Cardiac stress testing and coronary artery disease in liver transplantation candidates: Meta-analysis. <i>World Journal of Hepatology</i> , 2018, 10, 877-886.	0.8	20
15	Y Chromosome, Mitochondrial DNA and Childhood Behavioural Traits. <i>Scientific Reports</i> , 2017, 7, 11655.	1.6	4
16	Frequency of KLK3 gene deletions in the general population. <i>Annals of Clinical Biochemistry</i> , 2017, 54, 472-480.	0.8	0
17	Early life adiposity and telomere length across the life course: a systematic review and meta-analysis. <i>Wellcome Open Research</i> , 2017, 2, 118.	0.9	3
18	Collapsed methylation quantitative trait loci analysis for low frequency and rare variants. <i>Human Molecular Genetics</i> , 2016, 25, 4339-4349.	1.4	11

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19	Cost-effectiveness of whole-body bone scans in the pre-liver transplant assessment of patients with hepatocellular carcinoma in Southern Brazil. <i>Clinical Transplantation</i> , 2016, 30, 399-406.	0.8	5
20	Importance of Genetic Studies in Consanguineous Populations for the Characterization of Novel Human Gene Functions. <i>Annals of Human Genetics</i> , 2016, 80, 187-196.	0.3	41
21	A study of common Mendelian disease carriers across ageing British cohorts: meta-analyses reveal heterozygosity for alpha 1-antitrypsin deficiency increases respiratory capacity and height. <i>Journal of Medical Genetics</i> , 2016, 53, 280-288.	1.5	9
22	Lipids, obesity and gallbladder disease in women: insights from genetic studies using the cardiovascular gene-centric 50K SNP array. <i>European Journal of Human Genetics</i> , 2016, 24, 106-112.	1.4	23
23	Proxy Molecular Diagnosis from Whole-Exome Sequencing Reveals Papillon-Lefevre Syndrome Caused by a Missense Mutation in CTSC. <i>PLoS ONE</i> , 2015, 10, e0121351.	1.1	4
24	Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. <i>BioMed Research International</i> , 2015, 2015, 1-16.	0.9	7
25	Variation in the SLC23A1 gene does not influence cardiometabolic outcomes to the extent expected given its association with f-ascorbic acid. <i>American Journal of Clinical Nutrition</i> , 2015, 101, 202-209.	2.2	13
26	Haptoglobin Duplicon, Hemoglobin, and Vitamin C: Analyses in the British Women's Heart and Health Study and Caerphilly Prospective Study. <i>Disease Markers</i> , 2014, 2014, 1-5.	0.6	5
27	Nonsense Mutation in Coiled-Coil Domain Containing 151 Gene ( <i>CCDC151</i> ) Causes Primary Ciliary Dyskinesia. <i>Human Mutation</i> , 2014, 35, 1446-1448.	1.1	33
28	Very Low PSA Concentrations and Deletions of the KLK3 Gene. <i>Clinical Chemistry</i> , 2013, 59, 234-244.	1.5	12
29	Dependence of Deodorant Usage on ABCC11 Genotype: Scope for Personalized Genetics in Personal Hygiene. <i>Journal of Investigative Dermatology</i> , 2013, 133, 1760-1767.	0.3	18
30	Molecular and Population Analysis of Natural Selection on the Human Haptoglobin Duplication. <i>Annals of Human Genetics</i> , 2012, 76, 352-362.	0.3	30
31	Complexity of a complex trait locus: HP, HPR, haemoglobin and cholesterol. <i>Gene</i> , 2012, 499, 8-13.	1.0	20
32	Combined analysis of <i>CHRNA5</i> , <i>CHRNA3</i> and <i>CYP2A6</i> in relation to adolescent smoking behaviour. <i>Journal of Psychopharmacology</i> , 2011, 25, 915-923.	2.0	16
33	Genetic Variants Associated with von Willebrand Factor Levels in Healthy Men and Women Identified Using the HumanCVD BeadChip. <i>Annals of Human Genetics</i> , 2011, 75, 456-467.	0.3	28
34	Amplification ratio control system for copy number variation genotyping. <i>Nucleic Acids Research</i> , 2011, 39, e54-e54.	6.5	10
35	Carrier Status for the Common R501X and 2282del4 Filaggrin Mutations Is Not Associated with Hearing Phenotypes in 5377 Children from the ALSPAC Cohort. <i>PLoS ONE</i> , 2009, 4, e5784.	1.1	8
36	Hardy-Weinberg Equilibrium Testing of Biological Ascertainment for Mendelian Randomization Studies. <i>American Journal of Epidemiology</i> , 2009, 169, 505-514.	1.6	886

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37	Comment on: Marchand and Polychronakos (2007) Evaluation of Polymorphic Splicing in the Mechanism of the Association of the Insulin Gene with Diabetes: Diabetes 56:709-713. Diabetes, 2007, 56, e16-e16.	0.3	4
38	Molecular genetics of human growth hormone, insulin-like growth factors and their pathways in common disease. Human Genetics, 2007, 122, 1-21.	1.8	63
39	Questioning INS VNTR role in obesity and diabetes: subclasses tag IGF2-INS-TH haplotypes; and -23HphI as a STEP (splicing and translational efficiency polymorphism). Physiological Genomics, 2006, 28, 113-113.	1.0	7
40	A study of TH01 and IGF2-INS-TH haplotypes in relation to smoking initiation in three independent surveys. Pharmacogenetics and Genomics, 2006, 16, 15-23.	0.7	11
41	Replication of IGF2-INS-TH*5 haplotype effect on obesity in older men and study of related phenotypes. European Journal of Human Genetics, 2006, 14, 109-116.	1.4	26
42	MIDAS: software for analysis and visualisation of interallelic disequilibrium between multiallelic markers. BMC Bioinformatics, 2006, 7, 227.	1.2	110
43	Variants in the Human Insulin Gene That Affect Pre-mRNA Splicing: Is -23HphI a Functional Single Nucleotide Polymorphism at IDDM2?. Diabetes, 2006, 55, 260-264.	0.3	56
44	Non-recombining chromosome Y haplogroups and centromeric HindIII RFLP in relation to blood pressure in 2,743 middle-aged Caucasian men from the UK. Human Genetics, 2005, 116, 311-318.	1.8	15
45	Late Life Metabolic Syndrome, Early Growth, and Common Polymorphism in the Growth Hormone and Placental Lactogen Gene Cluster. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 5569-5576.	1.8	29
46	Haplotypic analyses of the IGF2-INS-TH gene cluster in relation to cardiovascular risk traits. Human Molecular Genetics, 2004, 13, 715-725.	1.4	57
47	Evidence of Admixture from Haplotyping in an Epidemiological Study of UK Caucasian Males: Implications for Association Analyses. Human Heredity, 2004, 57, 142-155.	0.4	13
48	Typing dinucleotide repeat loci using microplate array diagonal gel electrophoresis: Proof of principle. Electrophoresis, 2004, 25, 975-979.	1.3	1
49	Paucimorphic Alleles versus Polymorphic Alleles and Rare Mutations in Disease Causation: Theory, Observation and Detection. Current Genomics, 2004, 5, 431-438.	0.7	7
50	Spectrum of Nonrandom Associations Between Microsatellite Loci on Human Chromosome 11p15. Genetics, 2001, 158, 1235-1251.	1.2	26