

# Augusto Rendon

## List of Publications by Year in descending order

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Version: 2024-02-01

55  
papers

8,023  
citations

109137

35  
h-index

161609

54  
g-index

57  
all docs

57  
docs citations

57  
times ranked

16920  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study. <i>Lancet Neurology</i> , The, 2022, 21, 234-245.	4.9	74
2	Newborn Screening by Genomic Sequencing: Opportunities and Challenges. <i>International Journal of Neonatal Screening</i> , 2022, 8, 40.	1.2	25
3	An ancestral 10-bp repeat expansion in <i>VWA1</i> causes recessive hereditary motor neuropathy. <i>Brain</i> , 2021, 144, 584-600.	3.7	20
4	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. <i>Genetics in Medicine</i> , 2021, 23, 2360-2368.	1.1	13
5	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. <i>American Journal of Human Genetics</i> , 2021, 108, 1551-1557.	2.6	36
6	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care – Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	13.9	352
7	GA4GH: International policies and standards for data sharing across genomic research and healthcare. <i>Cell Genomics</i> , 2021, 1, 100029.	3.0	94
8	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. <i>Blood Advances</i> , 2020, 4, 3495-3506.	2.5	31
9	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	13.7	338
10	PanelApp crowdsources expert knowledge to establish consensus diagnostic gene panels. <i>Nature Genetics</i> , 2019, 51, 1560-1565.	9.4	294
11	Germline selection shapes human mitochondrial DNA diversity. <i>Science</i> , 2019, 364, .	6.0	178
12	The 100,000 Genomes Project: bringing whole genome sequencing to the NHS. <i>BMJ: British Medical Journal</i> , 2018, 361, k1687.	2.4	312
13	Missense variants in the X-linked gene <i>PRPS1</i> cause retinal degeneration in females. <i>Human Mutation</i> , 2018, 39, 80-91.	1.1	23
14	HGVA: the Human Genome Variation Archive. <i>Nucleic Acids Research</i> , 2017, 45, W189-W194.	6.5	6
15	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 995-1005.	2.6	23
16	Platelet function is modified by common sequence variation in megakaryocyte super enhancers. <i>Nature Communications</i> , 2017, 8, 16058.	5.8	50
17	A dominant gain-of-function mutation in universal tyrosine kinase <i>SRC</i> causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. <i>Science Translational Medicine</i> , 2016, 8, 328ra30.	5.8	87
18	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2016, 127, 2791-2803.	0.6	157

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19	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. <i>Blood</i> , 2016, 127, 2903-2914.	0.6	121
20	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. <i>Cell</i> , 2016, 167, 1398-1414.e24.	13.5	573
21	Identifying High-Risk CLL to Predict Early Relapse after FCR Based Treatment Using Whole Genome Sequencing: First Results from the Genomics England CLL Pilot. <i>Blood</i> , 2016, 128, 2022-2022.	0.6	5
22	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. <i>Genome Medicine</i> , 2015, 7, 36.	3.6	119
23	± variants defined by next-generation sequencing: Predicting variants likely to cause Glanzmann thrombasthenia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E1898-907.	3.3	36
24	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. <i>Nature Communications</i> , 2014, 5, 4926.	5.8	192
25	Familial pseudohyperkalemia in blood donors: a novel mutation with implications for transfusion practice. <i>Transfusion</i> , 2014, 54, 3043-3050.	0.8	40
26	Transcriptional diversity during lineage commitment of human blood progenitors. <i>Science</i> , 2014, 345, 1251033.	6.0	253
27	Gray platelet syndrome: proinflammatory megakaryocytes and ±-granule loss cause myelofibrosis and confer metastasis resistance in mice. <i>Blood</i> , 2014, 124, 3624-3635.	0.6	79
28	± Variants Defined By Next Generation Sequencing: Implications for Predicting Variants Likely to Cause Glanzmann Thrombasthenia and Alloimmune Disorders. <i>Blood</i> , 2014, 124, 4151-4151.	0.6	1
29	Meta-analysis of Gene-Level Associations for Rare Variants Based on Single-Variant Statistics. <i>American Journal of Human Genetics</i> , 2013, 93, 236-248.	2.6	60
30	SMIM1 underlies the Vel blood group and influences red blood cell traits. <i>Nature Genetics</i> , 2013, 45, 542-545.	9.4	96
31	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. <i>Nature Genetics</i> , 2013, 45, 501-512.	9.4	578
32	Maps of open chromatin highlight cell type–restricted patterns of regulatory sequence variation at hematological trait loci. <i>Genome Research</i> , 2013, 23, 1130-1141.	2.4	34
33	Transcription factor and chromatin features predict genes associated with eQTLs. <i>Nucleic Acids Research</i> , 2013, 41, 1450-1463.	6.5	28
34	Canonical Wnt signaling in megakaryocytes regulates proplatelet formation. <i>Blood</i> , 2013, 121, 188-196.	0.6	42
35	A GWAS sequence variant for platelet volume marks an alternative DNMT3 promoter in megakaryocytes near a MEIS1 binding site. <i>Blood</i> , 2012, 120, 4859-4868.	0.6	44
36	Interleukin-6 receptor pathways in coronary heart disease: a collaborative meta-analysis of 82 studies. <i>Lancet</i> , 2012, 379, 1205-1213.	6.3	668

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37	Seventy-five genetic loci influencing the human red blood cell. <i>Nature</i> , 2012, 492, 369-375.	13.7	320
38	Transcription factor co-localization patterns affect human cell type-specific gene expression. <i>BMC Genomics</i> , 2012, 13, 263.	1.2	12
39	Comparison of Methods for Competitive Tests of Pathway Analysis. <i>PLoS ONE</i> , 2012, 7, e41018.	1.1	40
40	Monocyte Gene Expression Signature of Patients with Early Onset Coronary Artery Disease. <i>PLoS ONE</i> , 2012, 7, e32166.	1.1	34
41	Exome sequencing identifies NBEAL2 as the causative gene for gray platelet syndrome. <i>Nature Genetics</i> , 2011, 43, 735-737.	9.4	245
42	New gene functions in megakaryopoiesis and platelet formation. <i>Nature</i> , 2011, 480, 201-208.	13.7	401
43	Multiple Loci Are Associated with White Blood Cell Phenotypes. <i>PLoS Genetics</i> , 2011, 7, e1002113.	1.5	106
44	Integrating Genome-Wide Genetic Variations and Monocyte Expression Data Reveals Trans-Regulated Gene Modules in Humans. <i>PLoS Genetics</i> , 2011, 7, e1002367.	1.5	126
45	Maps of Open Chromatin Guide the Functional Follow-Up of Genome-Wide Association Signals: Application to Hematological Traits. <i>PLoS Genetics</i> , 2011, 7, e1002139.	1.5	38
46	Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. <i>Nature Genetics</i> , 2009, 41, 1191-1198.	9.4	324
47	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. <i>Nature Genetics</i> , 2009, 41, 1182-1190.	9.4	481
48	A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. <i>Blood</i> , 2009, 113, 3831-3837.	0.6	117
49	A HaemAtlas: characterizing gene expression in differentiated human blood cells. <i>Blood</i> , 2009, 113, e1-e9.	0.6	215
50	Treatment planning using tailored and standard cylindrical light diffusers for photodynamic therapy of the prostate. <i>Physics in Medicine and Biology</i> , 2008, 53, 1131-1149.	1.6	20
51	Developing Conformal Therapy Treatment Planning for Photodynamic Therapy. , 2008, , .		0
52	The HaemAtlas: Characterising Gene Expression in Differentiated Human Blood Cells. <i>Blood</i> , 2008, 112, 2453-2453.	0.6	0
53	A Common Single Nucleotide Polymorphism in the Chromosome 7q22.3 Region, Which Is Frequently Deleted in Myeloid Malignancies, Is Associated with Mean Platelet Volume and Platelet Function in Healthy Individuals. <i>Blood</i> , 2008, 112, 86-86.	0.6	1
54	Towards conformal light delivery using tailored cylindrical diffusers: attainable light dose distributions. <i>Physics in Medicine and Biology</i> , 2006, 51, 5967-5975.	1.6	16

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55	The Distribution of the Anticancer Drug Doxorubicin in Relation to Blood Vessels in Solid Tumors. Clinical Cancer Research, 2005, 11, 8782-8788.	3.2	428