Augusto Rendon

List of Publications by Year
in descending order

55 papers	8,023 citations	109321 35 h-index	161849 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. Lancet Neurology, The, 2022, 21, 234-245.	10.2	74
2	Newborn Screening by Genomic Sequencing: Opportunities and Challenges. International Journal of Neonatal Screening, 2022, 8, 40.	3.2	25
3	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
4	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. Genetics in Medicine, 2021, 23, 2360-2368.	2.4	13
5	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
6	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
7	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
8	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. Blood Advances, 2020, 4, 3495-3506.	5.2	31
9	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
10	PanelApp crowdsources expert knowledge to establish consensus diagnostic gene panels. Nature Genetics, 2019, 51, 1560-1565.	21.4	294
11	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
12	The 100â€‰000 Genomes Project: bringing whole genome sequencing to the NHS. BMJ: British Medical Journal, 2018, 361, k1687.	2.3	312
13	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
14	HGVA: the Human Genome Variation Archive. Nucleic Acids Research, 2017, 45, W189-W194.	14.5	6
15	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. American Journal of Human Genetics, 2017, 101, 995-1005.	6.2	23
16	Platelet function is modified by common sequence variation in megakaryocyte super enhancers. Nature Communications, 2017, 8, 16058.	12.8	50
17	A dominant gain-of-function mutation in universal tyrosine kinase SRC causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. Science Translational Medicine, 2016, 8, 328ra30.	12.4	87
18	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157

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19	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. Blood, 2016, 127, 2903-2914.	1.4	121
20	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. Cell, 2016, 167, 1398-1414.e24.	28.9	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. Blood, 2016, 128, 2022-2022.	1.4	5
22	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. Genome Medicine, 2015, 7, 36.	8.2	119
23	Î±IIbÎ²3 variants defined by next-generation sequencing: Predicting variants likely to cause Glanzmann thrombasthenia. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E1898-907.	7.1	36
24	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	12.8	192
25	Familial pseudohyperkalemia in blood donors: a novel mutation with implications for transfusion practice. Transfusion, 2014, 54, 3043-3050.	1.6	40
26	Transcriptional diversity during lineage commitment of human blood progenitors. Science, 2014, 345, 1251033.	12.6	253
27	Gray platelet syndrome: proinflammatory megakaryocytes and Î±-granule loss cause myelofibrosis and confer metastasis resistance in mice. Blood, 2014, 124, 3624-3635.	1.4	79
28	Î±IIbÎ²3 Variants Defined By Next Generation Sequencing: Implications for Predicting Variants Likely to Cause Glanzmann Thrombasthenia and Alloimmune Disorders. Blood, 2014, 124, 4151-4151.	1.4	1
29	Meta-analysis of Gene-Level Associations for Rare Variants Based on Single-Variant Statistics. American Journal of Human Genetics, 2013, 93, 236-248.	6.2	60
30	SMIM1 underlies the Vel blood group and influences red blood cell traits. Nature Genetics, 2013, 45, 542-545.	21.4	96
31	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. Nature Genetics, 2013, 45, 501-512.	21.4	578
32	Maps of open chromatin highlight cell typeâ€“restricted patterns of regulatory sequence variation at hematological trait loci. Genome Research, 2013, 23, 1130-1141.	5.5	34
33	Transcription factor and chromatin features predict genes associated with eQTLs. Nucleic Acids Research, 2013, 41, 1450-1463.	14.5	28
34	Canonical Wnt signaling in megakaryocytes regulates proplatelet formation. Blood, 2013, 121, 188-196.	1.4	42
35	A GWAS sequence variant for platelet volume marks an alternative DNM3 promoter in megakaryocytes near a MEIS1 binding site. Blood, 2012, 120, 4859-4868.	1.4	44
36	Interleukin-6 receptor pathways in coronary heart disease: a collaborative meta-analysis of 82 studies. Lancet, The, 2012, 379, 1205-1213.	13.7	668

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37	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
38	Transcription factor co-localization patterns affect human cell type-specific gene expression. BMC Genomics, 2012, 13, 263.	2.8	12
39	Comparison of Methods for Competitive Tests of Pathway Analysis. PLoS ONE, 2012, 7, e41018.	2.5	40
40	Monocyte Gene Expression Signature of Patients with Early Onset Coronary Artery Disease. PLoS ONE, 2012, 7, e32166.	2.5	34
41	Exome sequencing identifies NBEAL2 as the causative gene for gray platelet syndrome. Nature Genetics, 2011, 43, 735-737.	21.4	245
42	New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208.	27.8	401
43	Multiple Loci Are Associated with White Blood Cell Phenotypes. PLoS Genetics, 2011, 7, e1002113.	3.5	106
44	Integrating Genome-Wide Genetic Variations and Monocyte Expression Data Reveals Trans-Regulated Gene Modules in Humans. PLoS Genetics, 2011, 7, e1002367.	3.5	126
45	Maps of Open Chromatin Guide the Functional Follow-Up of Genome-Wide Association Signals: Application to Hematological Traits. PLoS Genetics, 2011, 7, e1002139.	3.5	38
46	Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. Nature Genetics, 2009, 41, 1191-1198.	21.4	324
47	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. Nature Genetics, 2009, 41, 1182-1190.	21.4	481
48	A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. Blood, 2009, 113, 3831-3837.	1.4	117
49	A HaemAtlas: characterizing gene expression in differentiated human blood cells. Blood, 2009, 113, e1-e9.	1.4	215
50	Treatment planning using tailored and standard cylindrical light diffusers for photodynamic therapy of the prostate. Physics in Medicine and Biology, 2008, 53, 1131-1149.	3.0	20
51	Developing Conformal Therapy Treatment Planning for Photodynamic Therapy. , 2008, , .		0
52	The HaemAtlas: Characterising Gene Expression in Differentiated Human Blood Cells. Blood, 2008, 112, 2453-2453.	1.4	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. Blood, 2008, 112, 86-86.	1.4	1
54	Towards conformal light delivery using tailored cylindrical diffusers: attainable light dose distributions. Physics in Medicine and Biology, 2006, 51, 5967-5975.	3.0	16

#	Article	IF	Citations
55	The Distribution of the Anticancer Drug Doxorubicin in Relation to Blood Vessels in Solid Tumors. Clinical Cancer Research, 2005, 11, 8782-8788.	7.0	428