Alejandro A SchÃ¤ffer

List of Publications by Year
in descending order

72 papers	7,018 citations	101543 36 h-index	91884 69 g-index
330 all docs	330 docs citations	330 times ranked	12316 citing authors

#	Article	IF	Citations
1	STAT3Mutations in the Hyper-IgE Syndrome. New England Journal of Medicine, 2007, 357, 1608-1619.	27.0	1,098
2	Autosomal dominant immune dysregulation syndrome in humans with CTLA4 mutations. Nature Medicine, 2014, 20, 1410-1416.	30.7	723
3	Deleterious Mutations in LRBA Are Associated with a Syndrome of Immune Deficiency and Autoimmunity. American Journal of Human Genetics, 2012, 90, 986-1001.	6.2	452
4	Genetic Linkage of Hyper-IgE Syndrome to Chromosome 4. American Journal of Human Genetics, 1999, 65, 735-744.	6.2	360
5	Virus Variation Resource â€“ improved response to emergent viral outbreaks. Nucleic Acids Research, 2017, 45, D482-D490.	14.5	342
6	The evolution of tumour phylogenetics: principles and practice. Nature Reviews Genetics, 2017, 18, 213-229.	16.3	240
7	Inherited CARD9 deficiency in otherwise healthy children and adults with Candida speciesâ€“induced meningoencephalitis, colitis, or both. Journal of Allergy and Clinical Immunology, 2015, 135, 1558-1568.e2.	2.9	208
8	Heritability of life span in the Old Order Amish. American Journal of Medical Genetics Part A, 2001, 102, 346-352.	2.4	175
9	The extended clinical phenotype of 64 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 402-412.	2.9	163
10	Digenic inheritance in medical genetics. Journal of Medical Genetics, 2013, 50, 641-652.	3.2	162
11	Hypomorphic homozygous mutations in phosphoglucomutase 3 (PGM3) impair immunity and increase serum IgE levels. Journal of Allergy and Clinical Immunology, 2014, 133, 1410-1419.e13.	2.9	160
12	Integrated genome-wide analysis of expression quantitative trait loci aids interpretation of genomic association studies. Genome Biology, 2017, 18, 16.	8.8	151
13	Large-Scale Study of Antibody Titer Decay following BNT162b2 mRNA Vaccine or SARS-CoV-2 Infection. Vaccines, 2022, 10, 64.	4.4	144
14	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
15	JAGN1 deficiency causes aberrant myeloid cell homeostasis and congenital neutropenia. Nature Genetics, 2014, 46, 1021-1027.	21.4	119
16	Hypomorphic caspase activation and recruitment domain 11 (CARD11) mutations associated with diverse immunologic phenotypes with or without atopic disease. Journal of Allergy and Clinical Immunology, 2019, 143, 1482-1495.	2.9	116
17	ZNF341 controls STAT3 expression and thereby immunocompetence. Science Immunology, 2018, 3, .	11.9	113
18	Elapsed time since BNT162b2 vaccine and risk of SARS-CoV-2 infection: test negative design study. BMJ, The, 2021, 375, e067873.	6.0	110

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19	Genomic imbalances in the progression of endocrine pancreatic tumors. Genes Chromosomes and Cancer, 2001, 32, 364-372.	2.8	105
20	Single-Cell Genetic Analysis of Ductal Carcinoma in Situ and Invasive Breast Cancer Reveals Enormous Tumor Heterogeneity yet Conserved Genomic Imbalances and Gain of MYC during Progression. American Journal of Pathology, 2012, 181, 1807-1822.	3.8	104
21	Distance-Based Reconstruction of Tree Models for Oncogenesis. Journal of Computational Biology, 2000, 7, 789-803.	1.6	96
22	A Hereditary Form of Small Intestinal Carcinoid Associated With a Germline Mutation in Inositol Polyphosphate Multikinase. Gastroenterology, 2015, 149, 67-78.	1.3	96
23	A homozygous single-base deletion in MLPH causes the dilute coat color phenotype in the domestic cat. Genomics, 2006, 88, 698-705.	2.9	89
24	Chromatin-remodeling factor SMARCD2 regulates transcriptional networks controlling differentiation of neutrophil granulocytes. Nature Genetics, 2017, 49, 742-752.	21.4	87
25	Inherited biallelic CSF3R mutations in severe congenital neutropenia. Blood, 2014, 123, 3811-3817.	1.4	79
26	DCLRE1C(ARTEMIS) mutations causing phenotypes ranging from atypical severe combined immunodeficiency to mere antibody deficiency. Human Molecular Genetics, 2015, 24, 7361-7372.	2.9	72
27	Endogenous Retrovirus Insertion in theKITOncogene DeterminesWhiteandWhite spottingin Domestic Cats. G3: Genes, Genomes, Genetics, 2014, 4, 1881-1891.	1.8	66
28	Plasma cell deficiency in human subjects with heterozygous mutations in Sec61 translocon alpha 1 subunit (SEC61A1). Journal of Allergy and Clinical Immunology, 2018, 141, 1427-1438.	2.9	63
29	Deconstructing common variable immunodeficiency by genetic analysis. Current Opinion in Genetics and Development, 2007, 17, 201-212.	3.3	60
30	Chromosomal Alterations and Gene Expression Changes Associated with the Progression of Leukoplakia to Advanced Gingivobuccal Cancer. Translational Oncology, 2017, 10, 396-409.	3.7	60
31	In vitro and inÂvivo identification of clinically approved drugs that modify <scp>ACE</scp> 2 expression. Molecular Systems Biology, 2020, 16, e9628.	7.2	47
32	Linkage of autosomal-dominant common variable immunodeficiency to chromosome 4q. European Journal of Human Genetics, 2006, 14, 867-875.	2.8	46
33	Algorithms to Model Single Gene, Single Chromosome, and Whole Genome Copy Number Changes Jointly in Tumor Phylogenetics. PLoS Computational Biology, 2014, 10, e1003740.	3.2	46
34	VADR: validation and annotation of virus sequence submissions to GenBank. BMC Bioinformatics, 2020, 21, 211.	2.6	46
35	Chromosome abnormalities in ovarian adenocarcinoma: III. Using breakpoint data to infer and test mathematical models for oncogenesis. Genes Chromosomes and Cancer, 2000, 28, 106-120.	2.8	45
36	Potential non-B DNA regions in the human genome are associated with higher rates of nucleotide mutation and expression variation. Nucleic Acids Research, 2014, 42, 12367-12379.	14.5	45

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37	The Chromatin-binding Protein HMGN1 Regulates the Expression of Methyl CpG-binding Protein 2 (MECP2) and Affects the Behavior of Mice. Journal of Biological Chemistry, 2011, 286, 42051-42062.	3.4	42
38	Genetic heterogeneity in severe congenital neutropenia: how many aberrant pathways can kill a neutrophil?. Current Opinion in Allergy and Clinical Immunology, 2007, 7, 481-494.	2.3	40
39	Identification of drugs associated with reduced severity of COVID-19 â€“ a case-control study in a large population. ELife, 2021, 10, .	6.0	32
40	Analysis of families with common variable immunodeficiency (CVID) and IgA deficiency suggests linkage of CVID to chromosome 16q. Human Genetics, 2006, 118, 725-729.	3.8	31
41	Multiple microRNAs within the 14q32 cluster target the mRNAs of major type 1 diabetes autoantigens IAâ€2, IAâ€2Î², and GAD65. FASEB Journal, 2015, 29, 4374-4383.	0.5	31
42	An Integrative CGH, MSI and Candidate Genes Methylation Analysis of Colorectal Tumors. PLoS ONE, 2014, 9, e82185.	2.5	29
43	Single-Cell Genetic Analysis Reveals Insights into Clonal Development of Prostate Cancers and Indicates Loss of PTEN as a Marker of Poor Prognosis. American Journal of Pathology, 2014, 184, 2671-2686.	3.8	29
44	A commonSLC26A4-linked haplotype underlying non-syndromic hearing loss with enlargement of the vestibular aqueduct. Journal of Medical Genetics, 2017, 54, 665-673.	3.2	29
45	Beyond Synthetic Lethality: Charting the Landscape of Pairwise Gene Expression States Associated with Survival in Cancer. Cell Reports, 2019, 28, 938-948.e6.	6.4	29
46	Inferring models of multiscale copy number evolution for single-tumor phylogenetics. Bioinformatics, 2015, 31, i258-i267.	4.1	28
47	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	3.2	28
48	Deconvolving Clinically Relevant Cellular Immune Cross-talk from Bulk Gene Expression Using CODEFACS and LIRICS Stratifies Patients with Melanoma to Antiâ€“PD-1 Therapy. Cancer Discovery, 2022, 12, 1088-1105.	9.4	28
49	Aneuploidy, TP53 mutation, and amplification of MYC correlate with increased intratumor heterogeneity and poor prognosis of breast cancer patients. Genes Chromosomes and Cancer, 2018, 57, 165-175.	2.8	27
50	VecScreen_plus_taxonomy: imposing a tax(onomy) increase on vector contamination screening. Bioinformatics, 2018, 34, 755-759.	4.1	23
51	Coordinated Conditional Simulation with SLINK and SUP of Many Markers Linked or Associated to a Trait in Large Pedigrees. Human Heredity, 2011, 71, 126-134.	0.8	20
52	The evolution of single cell-derived colorectal cancer cell lines is dominated by the continued selection of tumor-specific genomic imbalances, despite random chromosomal instability. Carcinogenesis, 2018, 39, 993-1005.	2.8	20
53	Quickly identifying identical and closely related subjects in large databases using genotype data. PLoS ONE, 2017, 12, e0179106.	2.5	20
54	rh_tsp_map 3.0: end-to-end radiation hybrid mapping with improved speed and quality control. Bioinformatics, 2007, 23, 1156-1158.	4.1	19

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55	Immune Determinants of the Association between Tumor Mutational Burden and Immunotherapy Response across Cancer Types. Cancer Research, 2022, 82, 2076-2083.	0.9	18
56	Targeted genomic analysis reveals widespread autoimmune disease association with regulatory variants in the TNF superfamily cytokine signalling network. Genome Medicine, 2016, 8, 76.	8.2	17
57	Phylogenetic analysis of multiple FISH markers in oral tongue squamous cell carcinoma suggests that a diverse distribution of copy number changes is associated with poor prognosis. International Journal of Cancer, 2016, 138, 98-109.	5.1	16
58	Genome-Wide Changes in Protein Translation Efficiency Are Associated with Autism. Genome Biology and Evolution, 2018, 10, 1902-1919.	2.5	15
59	FISHtrees 3.0: Tumor Phylogenetics Using a Ploidy Probe. PLoS ONE, 2016, 11, e0158569.	2.5	13
60	The landscape of receptor-mediated precision cancer combination therapy via a single-cell perspective. Nature Communications, 2022, 13, 1613.	12.8	12
61	Animal Models of Human Granulocyte Diseases. Hematology/Oncology Clinics of North America, 2013, 27, 129-148.	2.2	10
62	Recurring Amplification at 11q22.1-q22.2 Locus Plays an Important Role in Lymph Node Metastasis and Radioresistance in OSCC. Scientific Reports, 2017, 7, 16051.	3.3	10
63	High Levels of Chromosomal Copy Number Alterations and TP53 Mutations Correlate with Poor Outcome in Younger Breast Cancer Patients. American Journal of Pathology, 2020, 190, 1643-1656.	3.8	10
64	Tumor Copy Number Deconvolution Integrating Bulk and Single-Cell Sequencing Data. Journal of Computational Biology, 2020, 27, 565-598.	1.6	10
65	Mutations in COMP cause familial carpal tunnel syndrome. Nature Communications, 2020, 11, 3642.	12.8	8
66	Tumor heterogeneity assessed by sequencing and fluorescence in situ hybridization (FISH) data. Bioinformatics, 2021, 37, 4704-4711.	4.1	5
67	The GENDULF algorithm: mining transcriptomics to uncover modifier genes for monogenic diseases. Molecular Systems Biology, 2020, 16, e9701.	7.2	2
68	Deficiency Of JAGN1 Causes Severe Congenital Neutropenia Associated With Defective Secretory Pathway and Aberrant Myeloid Cell Homeostasis. Blood, 2013, 122, 439-439.	1.4	2
69	A Calculator for COVID-19 Severity Prediction Based on Patient Risk Factors and Number of Vaccines Received. Microorganisms, 2022, 10, 1238.	3.6	2
70	Tumor Copy Number Data Deconvolution Integrating Bulk and Single-cell Sequencing Data. , 2018, , .		0
71	Inherited Biallelic Loss-Of-Function Mutations In CSF3R Define a Novel Type Of Severe Congenital Neutropenia With Full Myeloid Cell Maturation and Refractoriness To RhG-CSF. Blood, 2013, 122, 1025-1025.	1.4	0
72	Clinical and Laboratory Features in the Israeli Population with COVID-19 Infection after Pfizer-BioNTech mRNA Booster Vaccination. Vaccines, 2022, 10, 636.	4.4	0