

Roger Colobran

List of Publications by Year in descending order

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Version: 2024-02-01

82
papers

6,383
citations

186209

28
h-index

79644

73
g-index

87
all docs

87
docs citations

87
times ranked

11006
citing authors

#	ARTICLE	IF	CITATIONS
1	VEXAS syndrome: relapsing polychondritis and myelodysplastic syndrome with associated immunoglobulin A vasculitis. <i>Rheumatology</i> , 2022, 61, e69-e71.	0.9	10
2	Case Report: X-Linked SASH3 Deficiency Presenting as a Common Variable Immunodeficiency. <i>Frontiers in Immunology</i> , 2022, 13, 881206.	2.2	7
3	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2200413119.	3.3	110
4	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	59
5	Epigenetic profiling linked to multisystem inflammatory syndrome in children (MIS-C): A multicenter, retrospective study. <i>EClinicalMedicine</i> , 2022, 50, 101515.	3.2	11
6	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. <i>Journal of Clinical Immunology</i> , 2021, 41, 639-657.	2.0	30
7	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. <i>EBioMedicine</i> , 2021, 65, 103246.	2.7	52
8	Case Report: Partial Uniparental Disomy Unmasks a Novel Recessive Mutation in the LYST Gene in a Patient With a Severe Phenotype of ChÅ©diak-Higashi Syndrome. <i>Frontiers in Immunology</i> , 2021, 12, 625591.	2.2	5
9	Epigenome-wide association study of COVID-19 severity with respiratory failure. <i>EBioMedicine</i> , 2021, 66, 103339.	2.7	90
10	SARS-CoV-2â€related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	100
11	Activation-induced deaminase is critical for the establishment of DNA methylation patterns prior to the germinal center reaction. <i>Nucleic Acids Research</i> , 2021, 49, 5057-5073.	6.5	5
12	Coordinated Response to Imported Vaccine-Derived Poliovirus Infection, Barcelona, Spain, 2019â€2020. <i>Emerging Infectious Diseases</i> , 2021, 27, 1513-1516.	2.0	2
13	Atypical Inflammatory Syndrome Triggered by SARS-CoV-2 in Infants with Down Syndrome. <i>Journal of Clinical Immunology</i> , 2021, 41, 1457-1462.	2.0	9
14	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-Î². <i>Journal of Clinical Immunology</i> , 2021, 41, 1425-1442.	2.0	39
15	Newborn Screening for SCID: Experience in Spain (Catalonia). <i>International Journal of Neonatal Screening</i> , 2021, 7, 46.	1.2	4
16	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	5.6	357
17	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	267
18	Case Report: Characterizing the Role of the STXBP2-R190C Monoallelic Mutation Found in a Patient With Hemophagocytic Syndrome and Langerhans Cell Histiocytosis. <i>Frontiers in Immunology</i> , 2021, 12, 723836.	2.2	4

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19	Pre-existing Autoantibodies Neutralizing High Concentrations of Type I Interferons in Almost 10% of COVID-19 Patients Admitted to Intensive Care in Barcelona. <i>Journal of Clinical Immunology</i> , 2021, 41, 1733-1744.	2.0	66
20	Early Diagnosis and Treatment of Purine Nucleoside Phosphorylase (PNP) Deficiency through TREC-Based Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2021, 7, 62.	1.2	2
21	Inborn errors of TLR3- or MDA5-dependent type I IFN immunity in children with enterovirus rhombencephalitis. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	12
22	Molecular analysis of the novel L243R mutation in STXBP2 reveals impairment of degranulation activity. <i>International Journal of Hematology</i> , 2020, 111, 440-450.	0.7	2
23	The IL-2R β R328X nonsense mutation allows partial STAT-5 phosphorylation and defines a critical region involved in the leaky-SCID phenotype. <i>Clinical and Experimental Immunology</i> , 2020, 200, 61-72.	1.1	2
24	Polyendocrine autoimmune syndromes reveal mechanisms of tolerance and autoimmunity. <i>Medicina Clínica (English Edition)</i> , 2020, 154, 444-446.	0.1	0
25	Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hyperomorphic PLAG2 Mutations. <i>Journal of Clinical Immunology</i> , 2020, 40, 987-1000.	2.0	41
26	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,749
27	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,983
28	FHLdb: A Comprehensive Database on the Molecular Basis of Familial Hemophagocytic Lymphohistiocytosis. <i>Frontiers in Immunology</i> , 2020, 11, 107.	2.2	4
29	Uncovering Low-Level Maternal Gonosomal Mosaicism in X-Linked Agammaglobulinemia: Implications for Genetic Counseling. <i>Frontiers in Immunology</i> , 2020, 11, 46.	2.2	5
30	Commentary: Bradykinin-Mediated Angioedema: An Update of the Genetic Causes and the Impact of Genomics. <i>Frontiers in Genetics</i> , 2020, 11, 304.	1.1	0
31	Síndromes poliendocrinos autoinmunes que revelan mecanismos de tolerancia y autoinmunidad. <i>Medicina Clínica</i> , 2020, 154, 444-446.	0.3	0
32	Regulation of TSHR Expression in the Thyroid and Thymus May Contribute to TSHR Tolerance Failure in Graves' Disease Patients via Two Distinct Mechanisms. <i>Frontiers in Immunology</i> , 2019, 10, 1695.	2.2	11
33	Expanding the Clinical and Genetic Spectra of Primary Immunodeficiency-Related Disorders With Clinical Exome Sequencing: Expected and Unexpected Findings. <i>Frontiers in Immunology</i> , 2019, 10, 2325.	2.2	41
34	First Universal Newborn Screening Program for Severe Combined Immunodeficiency in Europe. Two-Years' Experience in Catalonia (Spain). <i>Frontiers in Immunology</i> , 2019, 10, 2406.	2.2	45
35	Analysis of the PD-1/PD-L1 axis in human autoimmune thyroid disease: Insights into pathogenesis and clues to immunotherapy associated thyroid autoimmunity. <i>Journal of Autoimmunity</i> , 2019, 103, 102285.	3.0	62
36	eDiVA Classification and prioritization of pathogenic variants for clinical diagnostics. <i>Human Mutation</i> , 2019, 40, 865-878.	1.1	19

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37	Serum protein electrophoresis and complement deficiencies: a veteran but very versatile test in clinical laboratories. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, e179-e182.	1.4	3
38	Identification of 22q11.2 deletion syndrome via newborn screening for severe combined immunodeficiency. Two yearsâ€™ experience in Catalonia (Spain). <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e1016.	0.6	8
39	Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 359-368.	1.5	53
40	Extended immunophenotyping reference values in a healthy pediatric population. <i>Cytometry Part B - Clinical Cytometry</i> , 2019, 96, 223-233.	0.7	79
41	TNFAIP3 haploinsufficiency is the cause of autoinflammatory manifestations in a patient with a deletion of 13Mb on chromosome 6. <i>Clinical Immunology</i> , 2018, 191, 44-51.	1.4	40
42	Central Tolerance Mechanisms to TSHR in Gravesâ€™ Disease: Contributions to Understand the Genetic Association. <i>Hormone and Metabolic Research</i> , 2018, 50, 863-870.	0.7	13
43	LRBA Deficiency in a Patient With a Novel Homozygous Mutation Due to Chromosome 4 Segmental Uniparental Isodisomy. <i>Frontiers in Immunology</i> , 2018, 9, 2397.	2.2	37
44	Th1-skewed profile and excessive production of proinflammatory cytokines in a NFKB1-deficient patient with CVID and severe gastrointestinal manifestations. <i>Clinical Immunology</i> , 2018, 195, 49-58.	1.4	30
45	Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond. <i>Frontiers in Immunology</i> , 2018, 9, 636.	2.2	142
46	Lethal Influenza in Two Related Adults with Inherited GATA2 Deficiency. <i>Journal of Clinical Immunology</i> , 2018, 38, 513-526.	2.0	29
47	Complement factor 5 (C5) p.A252T mutation is prevalent in, but not restricted to, sub-Saharan Africa: implications for the susceptibility to meningococcal disease. <i>Clinical and Experimental Immunology</i> , 2017, 189, 226-231.	1.1	8
48	Early Versus Late Diagnosis of Complement Factor I Deficiency: Clinical Consequences Illustrated in Two Families with Novel Homozygous CFI Mutations. <i>Journal of Clinical Immunology</i> , 2017, 37, 781-789.	2.0	13
49	Complement factor 5 (C5) p.A252T mutation is prevalent in, but not restricted to, Sub-Saharan Africa: Implications for the susceptibility to meningococcal disease. <i>Molecular Immunology</i> , 2017, 89, 158-159.	1.0	0
50	CD26/DPPIV inhibition alters the expression of immune response-related genes in the thymi of NOD mice. <i>Molecular and Cellular Endocrinology</i> , 2016, 426, 101-112.	1.6	7
51	Novel Mutations Causing C5 Deficiency in Three North-African Families. <i>Journal of Clinical Immunology</i> , 2016, 36, 388-396.	2.0	13
52	AIRE genetic variants and predisposition to polygenic autoimmune disease: The case of Gravesâ€™ disease and a systematic literature review. <i>Human Immunology</i> , 2016, 77, 643-651.	1.2	20
53	Clinical laboratory standard capillary protein electrophoresis alerted of a low C3 state and lead to the identification of a Factor I deficiency due to a novel homozygous mutation. <i>Immunology Letters</i> , 2016, 174, 19-22.	1.1	7
54	Clinical and structural impact of mutations affecting the residue Phe367 of FOXP3 in patients with IPEX syndrome. <i>Clinical Immunology</i> , 2016, 163, 60-65.	1.4	14

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55	Type 1 Diabetes Prevention in NOD Mice by Targeting DPPiV/CD26 Is Associated with Changes in CD8+T Effector Memory Subset. PLoS ONE, 2015, 10, e0142186.	1.1	8
56	Central T cell tolerance: Identification of tissue-restricted autoantigens in the thymus HLA-DR peptidome. Journal of Autoimmunity, 2015, 60, 12-19.	3.0	27
57	Gravesâ€™ Disease TSHR-Stimulating Antibodies (TSABs) Induce the Activation of Immature Thymocytes: A Clue to the Riddle of TSABs Generation?. Journal of Immunology, 2015, 194, 4199-4206.	0.4	28
58	Genetics of Gravesâ€™ Disease: Special Focus on the Role of TSHR Gene. Hormone and Metabolic Research, 2015, 47, 753-766.	0.7	38
59	Novel and atypical splicing mutation in a compound heterozygous UNC13D defect presenting in Familial Hemophagocytic Lymphohistiocytosis triggered by EBV infection. Clinical Immunology, 2014, 153, 292-297.	1.4	6
60	Identification and characterization of a novel splice site mutation in the SERPING1 gene in a family with hereditary angioedema. Clinical Immunology, 2014, 150, 143-148.	1.4	10
61	Autoimmune Predisposition in Down Syndrome May Result from a Partial Central Tolerance Failure due to Insufficient Intrathymic Expression of <i>AIRE</i> and Peripheral Antigens. Journal of Immunology, 2014, 193, 3872-3879.	0.4	88
62	A Novel Splice Site Mutation in the SERPING1 Gene Leads to Haploinsufficiency by Complete Degradation of the Mutant Allele mRNA in a Case of Familial Hereditary Angioedema. Journal of Clinical Immunology, 2014, 34, 521-523.	2.0	11
63	Una nueva p�gina web para todos. Immunologia (Barcelona, Spain: 1987), 2013, 32, 121-122.	0.1	1
64	Bisulfite genomic sequencing to uncover variability in DNA methylation: Optimized protocol applied to human T cell differentiation genes. Immunologia (Barcelona, Spain: 1987), 2012, 31, 97-105.	0.1	1
65	Decreased AIRE and promiscuous gene expression in thymus from Down syndrome individuals may explain predisposition to autoimmunity. Journal of Translational Medicine, 2012, 10, .	1.8	0
66	Analysis of the cumulative changes in Gravesâ€™ disease thyroid glands points to IFN signature, plasmacytoid DCs and alternatively activated macrophages as chronicity determining factors. Journal of Autoimmunity, 2011, 36, 189-200.	3.0	34
67	A SNP in intron 1 of TSHR controls its thymic expression and susceptibility to Gravesâ€™ disease suggesting central tolerance failure in pathogenesis. Journal of Translational Medicine, 2011, 9, .	1.8	0
68	Association of an SNP with intrathymic transcription of TSHR and Graves' disease: a role for defective thymic tolerance. Human Molecular Genetics, 2011, 20, 3415-3423.	1.4	74
69	CCL4L Polymorphisms and CCL4/CCL4L Serum Levels Are Associated with Psoriasis Severity. Journal of Investigative Dermatology, 2011, 131, 1830-1837.	0.3	25
70	Type 1 Diabetes and Gravesâ€™ disease transcriptomic analysis show common contributing disease pathways. New Biotechnology, 2010, 27, S51.	2.4	0
71	Copy number variation in chemokine superfamily: the complex scene of <i>CCL3L</i> and <i>CCL4L</i> genes in health and disease. Clinical and Experimental Immunology, 2010, 162, 41-52.	1.1	36
72	CCL4L polymorphisms and serum levels are associated with psoriasis severity. Journal of Translational Medicine, 2010, 8, .	1.8	1

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73	S.103. Detection of Interferon Signature, Plasmacytoid Dendritic Cells (pDCs) and Alternatively Activated Macrophages (AAM) in Graves' Disease Thyroid as Chronicity Factors. <i>Clinical Immunology</i> , 2009, 131, S161.	1.4	0
74	Copy number variation in the CCL4L gene is associated with susceptibility to acute rejection in lung transplantation. <i>Genes and Immunity</i> , 2009, 10, 254-259.	2.2	24
75	Population structure in copy number variation and SNPs in the CCL4L chemokine gene. <i>Genes and Immunity</i> , 2008, 9, 279-288.	2.2	19
76	The chemokine network. I. How the genomic organization of chemokines contains clues for deciphering their functional complexity. <i>Clinical and Experimental Immunology</i> , 2007, 148, 208-217.	1.1	85
77	The chemokine network. II. On how polymorphisms and alternative splicing increase the number of molecular species and configure intricate patterns of disease susceptibility. <i>Clinical and Experimental Immunology</i> , 2007, 150, 1-12.	1.1	55
78	Multiple Products Derived from Two CCL4 Loci: High Incidence of a New Polymorphism in HIV+ Patients. <i>Journal of Immunology</i> , 2005, 174, 5655-5664.	0.4	45
79	Development of a new HLA-DRB real-time PCR typing method. <i>Human Immunology</i> , 2005, 66, 85-91.	1.2	14
80	HLA-B27 genotyping by Fluorescent Resonance Emission Transfer (FRET) probes in real-time PCR. <i>Human Immunology</i> , 2004, 65, 826-838.	1.2	22
81	Islet transplantation in seminal vesicles restores glycemia in diabetic rats: a preliminary study. <i>Transplantation Proceedings</i> , 2002, 34, 196-199.	0.3	2
82	Common Variable Immunodeficiency and Neurodevelopmental Delay Due to a 13Mb Deletion on Chromosome 4 Including the NFKB1 Gene: A Case Report. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	1