Roger Colobran

List of Publications by Year
in descending order

82 papers	6,383 citations	186265 28 h-index	79698 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. Rheumatology, 2022, 61, e69-e71.	1.9	10
2	Case Report: X-Linked SASH3 Deficiency Presenting as a Common Variable Immunodeficiency. Frontiers in Immunology, 2022, 13, 881206.	4.8	7
3	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	7.1	110
4	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	8.5	59
5	Epigenetic profiling linked to multisystem inflammatory syndrome in children (MIS-C): A multicenter, retrospective study. EClinicalMedicine, 2022, 50, 101515.	7.1	11
6	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	3.8	30
7	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
8	Case Report: Partial Uniparental Disomy Unmasks a Novel Recessive Mutation in the LYST Gene in a Patient With a Severe Phenotype of ChÃ©diak-Higashi Syndrome. Frontiers in Immunology, 2021, 12, 625591.	4.8	5
9	Epigenome-wide association study of COVID-19 severity with respiratory failure. EBioMedicine, 2021, 66, 103339.	6.1	90
10	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	8.5	100
11	Activation-induced deaminase is critical for the establishment of DNA methylation patterns prior to the germinal center reaction. Nucleic Acids Research, 2021, 49, 5057-5073.	14.5	5
12	Coordinated Response to Imported Vaccine-Derived Poliovirus Infection, Barcelona, Spain, 2019â€“2020. Emerging Infectious Diseases, 2021, 27, 1513-1516.	4.3	2
13	Atypical Inflammatory Syndrome Triggered by SARS-CoV-2 in Infants with Down Syndrome. Journal of Clinical Immunology, 2021, 41, 1457-1462.	3.8	9
14	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-Î². Journal of Clinical Immunology, 2021, 41, 1425-1442.	3.8	39
15	Newborn Screening for SCID: Experience in Spain (Catalonia). International Journal of Neonatal Screening, 2021, 7, 46.	3.2	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. Science Immunology, 2021, 6, .	11.9	357
17	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	11.9	267
18	Case Report: Characterizing the Role of the STXBP2-R190C Monoallelic Mutation Found in a Patient With Hemophagocytic Syndrome and Langerhans Cell Histiocytosis. Frontiers in Immunology, 2021, 12, 723836.	4.8	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. Journal of Clinical Immunology, 2021, 41, 1733-1744.	3.8	66
20	Early Diagnosis and Treatment of Purine Nucleoside Phosphorylase (PNP) Deficiency through TREC-Based Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 62.	3.2	2
21	Inborn errors of TLR3- or MDA5-dependent type I IFN immunity in children with enterovirus rhombencephalitis. Journal of Experimental Medicine, 2021, 218, .	8.5	12
22	Molecular analysis of the novel L243R mutation in STXBP2 reveals impairment of degranulation activity. International Journal of Hematology, 2020, 111, 440-450.	1.6	2
23	The ILâ€2RG R328X nonsense mutation allows partial STATâ€5 phosphorylation and defines a critical region involved in the leakyâ€SCID phenotype. Clinical and Experimental Immunology, 2020, 200, 61-72.	2.6	2
24	Polyendocrine autoimmune syndromes reveal mechanisms of tolerance and autoimmunity. Medicina ClÃnica (English Edition), 2020, 154, 444-446.	0.2	0
25	Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hypermorphic PLCG2 Mutations. Journal of Clinical Immunology, 2020, 40, 987-1000.	3.8	41
26	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
27	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,983
28	FHLdb: A Comprehensive Database on the Molecular Basis of Familial Hemophagocytic Lymphohistiocytosis. Frontiers in Immunology, 2020, 11, 107.	4.8	4
29	Uncovering Low-Level Maternal Gonosomal Mosaicism in X-Linked Agammaglobulinemia: Implications for Genetic Counseling. Frontiers in Immunology, 2020, 11, 46.	4.8	5
30	Commentary: Bradykinin-Mediated Angioedema: An Update of the Genetic Causes and the Impact of Genomics. Frontiers in Genetics, 2020, 11, 304.	2.3	0
31	SÃndromes poliendocrinos autoinmunes que revelan mecanismos de tolerancia y autoinmunidad. Medicina ClÃnica, 2020, 154, 444-446.	0.6	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. Frontiers in Immunology, 2019, 10, 1695.	4.8	11
33	Expanding the Clinical and Genetic Spectra of Primary Immunodeficiency-Related Disorders With Clinical Exome Sequencing: Expected and Unexpected Findings. Frontiers in Immunology, 2019, 10, 2325.	4.8	41
34	First Universal Newborn Screening Program for Severe Combined Immunodeficiency in Europe. Two-Years' Experience in Catalonia (Spain). Frontiers in Immunology, 2019, 10, 2406.	4.8	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. Journal of Autoimmunity, 2019, 103, 102285.	6.5	62
36	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	2.5	19

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37	Serum protein electrophoresis and complement deficiencies: a veteran but very versatile test in clinical laboratories. Clinical Chemistry and Laboratory Medicine, 2019, 57, e179-e182.	2.3	3
38	Identification of 22q11.2 deletion syndrome via newborn screening for severe combined immunodeficiency. Two yearsâ€™ experience in Catalonia (Spain). Molecular Genetics & Genomic Medicine, 2019, 7, e1016.	1.2	8
39	Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. Journal of Allergy and Clinical Immunology, 2019, 143, 359-368.	2.9	53
40	Extended immunophenotyping reference values in a healthy pediatric population. Cytometry Part B - Clinical Cytometry, 2019, 96, 223-233.	1.5	79
41	TNFAIP3 haploinsufficiency is the cause of autoinflammatory manifestations in a patient with a deletion of 13Mb on chromosome 6. Clinical Immunology, 2018, 191, 44-51.	3.2	40
42	Central Tolerance Mechanisms to TSHR in Gravesâ€™ Disease: Contributions to Understand the Genetic Association. Hormone and Metabolic Research, 2018, 50, 863-870.	1.5	13
43	LRBA Deficiency in a Patient With a Novel Homozygous Mutation Due to Chromosome 4 Segmental Uniparental Isodisomy. Frontiers in Immunology, 2018, 9, 2397.	4.8	37
44	Th1-skewed profile and excessive production of proinflammatory cytokines in a NFKB1-deficient patient with CVID and severe gastrointestinal manifestations. Clinical Immunology, 2018, 195, 49-58.	3.2	30
45	Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond. Frontiers in Immunology, 2018, 9, 636.	4.8	142
46	Lethal Influenza in Two Related Adults with Inherited GATA2 Deficiency. Journal of Clinical Immunology, 2018, 38, 513-526.	3.8	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. Clinical and Experimental Immunology, 2017, 189, 226-231.	2.6	8
48	Early Versus Late Diagnosis of Complement Factor I Deficiency: Clinical Consequences Illustrated in Two Families with Novel Homozygous CFI Mutations. Journal of Clinical Immunology, 2017, 37, 781-789.	3.8	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. Molecular Immunology, 2017, 89, 158-159.	2.2	0
50	CD26/DPPIV inhibition alters the expression of immune response-related genes in the thymi of NOD mice. Molecular and Cellular Endocrinology, 2016, 426, 101-112.	3.2	7
51	Novel Mutations Causing C5 Deficiency in Three North-African Families. Journal of Clinical Immunology, 2016, 36, 388-396.	3.8	13
52	AIRE genetic variants and predisposition to polygenic autoimmune disease: The case of Gravesâ€™ disease and a systematic literature review. Human Immunology, 2016, 77, 643-651.	2.4	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. Immunology Letters, 2016, 174, 19-22.	2.5	7
54	Clinical and structural impact of mutations affecting the residue Phe367 of FOXP3 in patients with IPEX syndrome. Clinical Immunology, 2016, 163, 60-65.	3.2	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.	2.5	8
56	Central T cell tolerance: Identification of tissue-restricted autoantigens in the thymus HLA-DR peptidome. Journal of Autoimmunity, 2015, 60, 12-19.	6.5	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.	0.8	28
58	Genetics of Gravesâ€™ Disease: Special Focus on the Role of TSHR Gene. Hormone and Metabolic Research, 2015, 47, 753-766.	1.5	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.	3.2	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.	3.2	10
61	Autoimmune Predisposition in Down Syndrome May Result from a Partial Central Tolerance Failure due to Insufficient Intrathymic Expression of AIRE and Peripheral Antigens. Journal of Immunology, 2014, 193, 3872-3879.	0.8	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.	3.8	11
63	Una nueva pÃ¡gina web para todos. Inmunologia (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. Inmunologia (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, .	4.4	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.	6.5	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, .	4.4	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.	2.9	74
69	CCL4L Polymorphisms and CCL4/CCL4L Serum Levels Are Associated with Psoriasis Severity. Journal of Investigative Dermatology, 2011, 131, 1830-1837.	0.7	25
70	Type 1 Diabetes and Gravesâ€™ disease transcriptomic analysis show common contributing disease pathways. New Biotechnology, 2010, 27, S51.	4.4	0
71	Copy number variation in chemokine superfamily: the complex scene ofCCL3Lâ€“CCL4Lgenes in health and disease. Clinical and Experimental Immunology, 2010, 162, 41-52.	2.6	36
72	CCL4L polymorphisms and serum levels are associated with psoriasis severity. Journal of Translational Medicine, 2010, 8, .	4.4	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. Clinical Immunology, 2009, 131, S161.	3.2	0
74	Copy number variation in the CCL4L gene is associated with susceptibility to acute rejection in lung transplantation. Genes and Immunity, 2009, 10, 254-259.	4.1	24
75	Population structure in copy number variation and SNPs in the CCL4L chemokine gene. Genes and Immunity, 2008, 9, 279-288.	4.1	19
76	The chemokine network. I. How the genomic organization of chemokines contains clues for deciphering their functional complexity. Clinical and Experimental Immunology, 2007, 148, 208-217.	2.6	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. Clinical and Experimental Immunology, 2007, 150, 1-12.	2.6	55
78	Multiple Products Derived from Two CCL4 Loci: High Incidence of a New Polymorphism in HIV+ Patients. Journal of Immunology, 2005, 174, 5655-5664.	0.8	45
79	Development of a new HLA-DRB real-time PCR typing method. Human Immunology, 2005, 66, 85-91.	2.4	14
80	HLA-B27 genotyping by Fluorescent Resonance Emission Transfer (FRET) probes in real-time PCR. Human Immunology, 2004, 65, 826-838.	2.4	22
81	Islet transplantation in seminal vesicles restores glycemia in diabetic rats: a preliminary study. Transplantation Proceedings, 2002, 34, 196-199.	0.6	2
82	Common Variable Immunodeficiency and Neurodevelopmental Delay Due to a 13Mb Deletion on Chromosome 4 Including the NFKB1 Gene: A Case Report. Frontiers in Immunology, 0, 13, .	4.8	1