Marie-Louise FrÃ©mond

List of Publications by Year
in descending order

54 papers	2,716 citations	279798 23 h-index	243625 44 g-index
58 all docs	58 docs citations	58 times ranked	4097 citing authors

#	Article	IF	Citations
1	Lung Inflammation in STING-Associated Vasculopathy with Onset in Infancy (SAVI). Cells, 2022, 11, 318.	4.1	28
2	Systemic inflammatory syndrome in children with <scp>FARSA</scp> deficiency. Clinical Genetics, 2022, 101, 552-558.	2.0	7
3	Complex Allele with Additive Gain-of-Function STING1 Variants in a Patient with Cavitating Lung Lesions and Aspergillosis. Journal of Clinical Immunology, 2022, 42, 1156-1159.	3.8	4
4	COPA syndrome, 5 years after: Where are we?. Joint Bone Spine, 2021, 88, 105070.	1.6	21
5	Rheumatoid factor positive polyarticular juvenile idiopathic arthritis associated with a novel COPA mutation. Rheumatology, 2021, 60, e171-e173.	1.9	6
6	Overview of STING-Associated Vasculopathy with Onset in Infancy (SAVI) Among 21 Patients. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 803-818.e11.	3.8	98
7	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. Journal of Clinical Immunology, 2021, 41, 603-609.	3.8	16
8	Life-Saving, Dose-Adjusted, Targeted Therapy in a Patient with a STAT3 Gain-of-Function Mutation. Journal of Clinical Immunology, 2021, 41, 807-810.	3.8	10
9	JAK inhibitors are effective in a subset of patients with juvenile dermatomyositis: a monocentric retrospective study. Rheumatology, 2021, 60, 5801-5808.	1.9	52
10	STING-Mediated Lung Inflammation and Beyond. Journal of Clinical Immunology, 2021, 41, 501-514.	3.8	48
11	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	8.5	185
12	Syndrome COPA, quoi de neuf cinq ans aprÃ¨sÂ?. Revue Du Rhumatisme (Edition Francaise), 2021, 88, 183-189.	0.0	0
13	JAK inhibition in the type I interferonopathies. Journal of Allergy and Clinical Immunology, 2021, 148, 991-993.	2.9	19
14	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
15	Emerging Place of JAK Inhibitors in the Treatment of Inborn Errors of Immunity. Frontiers in Immunology, 2021, 12, 717388.	4.8	23
16	Use of ruxolitinib in COPA syndrome manifesting as life-threatening alveolar haemorrhage. Thorax, 2020, 75, 92-95.	5.6	36
17	Bone Marrow Transplantation in Congenital Erythropoietic Porphyria: Sustained Efficacy but Unexpected Liver Dysfunction. Biology of Blood and Marrow Transplantation, 2020, 26, 704-711.	2.0	10
18	Anti-MDA5 juvenile idiopathic inflammatory myopathy: a specific subgroup defined by differentially enhanced interferon-Î± signalling. Rheumatology, 2020, 59, 1927-1937.	1.9	26

#	Article	IF	Citations
19	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	21.4	105
20	JAK Inhibition in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2020, 383, 2190-2193.	27.0	24
21	Mutations in COPA lead to abnormal trafficking of STING to the Golgi and interferon signaling. Journal of Experimental Medicine, 2020, 217, .	8.5	130
22	Type I Interferonopathies: from a Novel Concept to Targeted Therapeutics. Current Rheumatology Reports, 2020, 22, 32.	4.7	30
23	Mendelian disorders of immunity related to an upregulation of type I interferon. , 2020, , 751-772.		2
24	Comment on: â€˜Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitorsâ€™ by Giannelou et al: mutations in TRNT1 result in a constitutive activation of type I interferon signalling. Annals of the Rheumatic Diseases, 2019, 78, e86-e86.	0.9	12
25	COPA Syndrome as a Cause of Lupus Nephritis. Kidney International Reports, 2019, 4, 1187-1189.	0.8	19
26	Haploidentical Hematopoietic Stem Cell Transplantation with Post-Transplant Cyclophosphamide for Primary Immunodeficiencies and Inherited Disorders in Children. Biology of Blood and Marrow Transplantation, 2019, 25, 1363-1373.	2.0	78
27	OP0107â€…HETEROZYGOUS MUTATIONS IN COPA ARE ASSOCIATED WITH ENHANCED TYPE I INTERFERON SIGNALLING. , 2019, , .		0
28	Efficacy of JAK1/2 inhibition in the treatment of chilblain lupus due to TREX1 deficiency. Annals of the Rheumatic Diseases, 2019, 78, 431-433.	0.9	53
29	Inherited Immunodeficiency: A New Association With Early-Onset Childhood Panniculitis. Pediatrics, 2018, 141, S496-S500.	2.1	24
30	Successful haematopoietic stem cell transplantation in a case of pulmonary alveolar proteinosis due to GM-CSF receptor deficiency. Thorax, 2018, 73, 590-592.	5.6	24
31	Reverse-Transcriptase Inhibitors in the Aicardiâ€“GoutiÃ¨res Syndrome. New England Journal of Medicine, 2018, 379, 2275-2277.	27.0	106
32	A child with severe juvenile dermatomyositis treated with ruxolitinib. Brain, 2018, 141, e80-e80.	7.6	58
33	COPA syndrome restricted to life-threatening alveolar hemorrhages: clinical, pathological, molecular and biological characterization. , 2018, , .		1
34	Disease-associated mutations identify a novel region in human STING necessary for the control of type I interferon signaling. Journal of Allergy and Clinical Immunology, 2017, 140, 543-552.e5.	2.9	159
35	Brief Report: Blockade of TANKâ€Binding Kinase 1/IKKÉ› Inhibits Mutant Stimulator of Interferon Genes (STING)â€“Mediated Inflammatory Responses in Human Peripheral Blood Mononuclear Cells. Arthritis and Rheumatology, 2017, 69, 1495-1501.	5.6	22
36	Detection of interferon alpha protein reveals differential levels and cellular sources in disease. Journal of Experimental Medicine, 2017, 214, 1547-1555.	8.5	288

#	Article	IF	Citations
37	Assessment of Type I Interferon Signaling in Pediatric Inflammatory Disease. Journal of Clinical Immunology, 2017, 37, 123-132.	3.8	163
38	Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176.	12.8	164
39	JAK inhibition in STING-associated interferonopathy. Annals of the Rheumatic Diseases, 2016, 75, e75-e75.	0.9	22
40	011 Monogenic type I interferonopathies: from diagnosis to treatment. Journal of Investigative Dermatology, 2016, 136, S162.	0.7	0
41	Efficacy of the Janus kinase 1/2 inhibitor ruxolitinib in the treatment of vasculopathy associated with TMEM173 -activating mutations in 3 children. Journal of Allergy and Clinical Immunology, 2016, 138, 1752-1755.	2.9	192
42	SAT0514â€...Mosaic Tetrasomy 9P: A Mendelian Condition Associated with Pediatric-Onset Overlap Myositis. Annals of the Rheumatic Diseases, 2015, 74, 846.2-846.	0.9	0
43	Mosaic tetrasomy 9p: a mendelian interferonopathy associated with pediatric-onset overlap myositis. Pediatric Rheumatology, 2015, 13, P140.	2.1	0
44	Next-Generation Sequencing for Diagnosis and Tailored Therapy: A Case Report of Astrovirus-Associated Progressive Encephalitis. Journal of the Pediatric Infectious Diseases Society, 2015, 4, e53-e57.	1.3	116
45	Mosaic Tetrasomy 9p: A Mendelian Condition Associated With Pediatric-Onset Overlap Myositis. Pediatrics, 2015, 136, e544-e547.	2.1	10
46	Inherited CARD9 Deficiency in 2 Unrelated Patients With Invasive Exophiala Infection. Journal of Infectious Diseases, 2015, 211, 1241-1250.	4.0	141
47	Reiterated Therapeutic Drug Monitoring (TDM) Dosing to Significantly Improve the Control of Exposure to IV Busulfan in Infants and Older Children Undergoing Hematopoietic Stem-Cell Transplantation (HSCT). Blood, 2015, 126, 4326-4326.	1.4	0
48	Digestive perianastomotic ulcerations and Crohn's disease. Journal of Crohn's and Colitis, 2014, 8, 1624-1631.	1.3	17
49	Clinical spectrum and long-term follow-up of 14 cases with G6PC3 mutations from the French severe congenital neutropenia registry. Orphanet Journal of Rare Diseases, 2014, 9, 183.	2.7	48
50	Circulating endothelial cells as markers of endothelial dysfunction during hematopoietic stem cell transplantation for pediatric primary immunodeficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 1203-1206.	2.9	12
51	Antiâ€“TNF-Î± Therapy May Cause Neonatal Neutropenia. Pediatrics, 2014, 134, e1189-e1193.	2.1	54
52	Cellular and humoral immunity elicited by influenza vaccines in pediatric hematopoietic-stem cell transplantation. Human Immunology, 2012, 73, 884-890.	2.4	10
53	CD4 T-Cell Response to Pandemic H1N1 Specific Vaccine After HSCT In Pediatric Recipients. Blood, 2010, 116, 2338-2338.	1.4	0
54	A child with severe juvenile dermatomyositis treated with ruxolitinib. Journal of Financial Econometrics, 0, , .	1.5	0