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List of Publications by Year in descending order

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papers

4,532
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218677

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#	ARTICLE	IF	CITATIONS
1	Comprehensive analysis of ADA2 genetic variants and estimation of carrier frequency driven by a function-based approach. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 379-387.	2.9	27
2	TNF inhibition in vasculitis management in adenosine deaminase 2 deficiency (DADA2). <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 1812-1816.e6.	2.9	24
3	Haploinsufficiency of PSMD12 Causes Proteasome Dysfunction and Subclinical Autoinflammation. <i>Arthritis and Rheumatology</i> , 2022, 74, 1083-1090.	5.6	13
4	Mechanisms of vascular inflammation in deficiency of adenosine deaminase 2 (DADA2). <i>Seminars in Immunopathology</i> , 2022, 44, 269-280.	6.1	20
5	Low-ratio somatic NLRC4 mutation causes late-onset autoinflammatory disease. <i>Annals of the Rheumatic Diseases</i> , 2022, 81, 1173-1178.	0.9	10
6	Deficiency of Adenosine Deaminase 2 in Adults and Children: Experience From India. <i>Arthritis and Rheumatology</i> , 2021, 73, 276-285.	5.6	43
7	Novel ADA2 Compound Heterozygous Mutations Resulting in Deficiency of Adenosine Deaminase 2 in a Pair of Siblings. <i>Journal of Clinical Immunology</i> , 2021, 41, 837-842.	3.8	2
8	Complicated Diagnosis and Treatment of HA20 due to Contiguous Gene Deletions involving 6q23.3. <i>Journal of Clinical Immunology</i> , 2021, 41, 1420-1423.	3.8	6
9	RIPK1-Associated Inborn Errors of Innate Immunity. <i>Frontiers in Immunology</i> , 2021, 12, 676946.	4.8	9
10	Deubiquitination of proteasome subunits by OTULIN regulates type I IFN production. <i>Science Advances</i> , 2021, 7, eabi6794.	10.3	8
11	Haploinsufficiency of A20 (HA20): updates on the genetics, phenotype, pathogenesis and treatment. <i>World Journal of Pediatrics</i> , 2020, 16, 575-584.	1.8	49
12	A dominant autoinflammatory disease caused by non-cleavable variants of RIPK1. <i>Nature</i> , 2020, 577, 109-114.	27.8	163
13	Mutations that prevent caspase cleavage of RIPK1 cause autoinflammatory disease. <i>Nature</i> , 2020, 577, 103-108.	27.8	198
14	Interstitial Lung Disease and Psoriasis in a Child With Aicardi-Goutières Syndrome. <i>Frontiers in Immunology</i> , 2020, 11, 985.	4.8	22
15	New Horizons in the Genetic Etiology of Systemic Lupus Erythematosus and Lupus-Like Disease: Monogenic Lupus and Beyond. <i>Journal of Clinical Medicine</i> , 2020, 9, 712.	2.4	81
16	Deficiency of Adenosine Deaminase 2 (DADA2): Hidden Variants, Reduced Penetrance, and Unusual Inheritance. <i>Journal of Clinical Immunology</i> , 2020, 40, 917-926.	3.8	32
17	Genotype and functional correlates of disease phenotype in deficiency of adenosine deaminase 2 (DADA2). <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 1664-1672.e10.	2.9	95
18	Juvenile Onset Splenomegaly and Oculopathy Due to Germline Mutation in ALPK1. <i>Journal of Clinical Immunology</i> , 2020, 40, 350-358.	3.8	10

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19	Treatment Strategies for Deficiency of Adenosine Deaminase 2. <i>New England Journal of Medicine</i> , 2019, 380, 1582-1584.	27.0	138
20	Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors. <i>Annals of the Rheumatic Diseases</i> , 2018, 77, 612-619.	0.9	49
21	Disrupted N-linked glycosylation as a disease mechanism in deficiency of ADA2. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1363-1365.e8.	2.9	28
22	ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study. <i>Annals of the Rheumatic Diseases</i> , 2017, 76, 1648-1656.	0.9	199
23	Brief Report: Deficiency of Complement 1r Subcomponent in Early-Onset Systemic Lupus Erythematosus: The Role of Disease-Modifying Alleles in a Monogenic Disease. <i>Arthritis and Rheumatology</i> , 2017, 69, 1832-1839.	5.6	38
24	The monogenic autoinflammatory diseases define new pathways in human innate immunity and inflammation. <i>Nature Immunology</i> , 2017, 18, 832-842.	14.5	301
25	Human adenosine deaminases ADA1 and ADA2 bind to different subsets of immune cells. <i>Cellular and Molecular Life Sciences</i> , 2017, 74, 555-570.	5.4	87
26	NF- κ B Pathway in Autoinflammatory Diseases: Dysregulation of Protein Modifications by Ubiquitin Defines a New Category of Autoinflammatory Diseases. <i>Frontiers in Immunology</i> , 2017, 8, 399.	4.8	168
27	Biallelic hypomorphic mutations in a linear deubiquitinase define otulipenia, an early-onset autoinflammatory disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 10127-10132.	7.1	206
28	Loss-of-function mutations in TNFAIP3 leading to A20 haploinsufficiency cause an early-onset autoinflammatory disease. <i>Nature Genetics</i> , 2016, 48, 67-73.	21.4	513
29	Adenosine deaminase 2 deficiency presenting as spastic paraplegia and systemic vasculitis. <i>Journal of Neurology</i> , 2016, 263, 818-820.	3.6	21
30	Brief Report: Cryopyrin-Associated Periodic Syndrome Caused by a Myeloid-Restricted Somatic <i>NLRP3</i> Mutation. <i>Arthritis and Rheumatology</i> , 2015, 67, 2482-2486.	5.6	82
31	Additive loss-of-function proteasome subunit mutations in CANDLE/PRAAS patients promote type I IFN production. <i>Journal of Clinical Investigation</i> , 2015, 125, 4196-4211.	8.2	258
32	Mutant ADA2 in Vasculopathies. <i>New England Journal of Medicine</i> , 2014, 371, 478-481.	27.0	122
33	When less is more. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2014, 14, 491-500.	2.3	29
34	Early-Onset Stroke and Vasculopathy Associated with Mutations in ADA2. <i>New England Journal of Medicine</i> , 2014, 370, 911-920.	27.0	687
35	Targeted resequencing implicates the familial Mediterranean fever gene <i>MEFV</i> and the toll-like receptor 4 gene <i>TLR4</i> in Behçet disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 8134-8139.	7.1	140
36	Antibody deficiency associated with an inherited autosomal dominant mutation in TWEAK. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 5127-5132.	7.1	72

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37	A Hypermorphic Missense Mutation in PLCG2 , Encoding Phospholipase C ^β 2, Causes a Dominantly Inherited Autoinflammatory Disease with Immunodeficiency. American Journal of Human Genetics, 2012, 91, 713-720.	6.2	327
38	Transposable-Element Associated Small RNAs in Bombyx mori Genome. PLoS ONE, 2012, 7, e36599.	2.5	22
39	Novel microRNAs in silkworm (Bombyx mori). Functional and Integrative Genomics, 2010, 10, 405-415.	3.5	37
40	A discovery of novel microRNAs in the silkworm (Bombyx mori) genome. Genomics, 2009, 94, 438-444.	2.9	32
41	The Silkworm (Bombyx mori) microRNAs and Their Expressions in Multiple Developmental Stages. PLoS ONE, 2008, 3, e2997.	2.5	130