Janna Saarela

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2803533/publications.pdf

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43	6,150	361413	254184
papers	citations	20 h-index	43 g-index

46	46	46	12487
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Implementing a Functional Precision Medicine Tumor Board for Acute Myeloid Leukemia. Cancer Discovery, 2022, 12, 388-401.	9.4	73
2	RhoG deficiency abrogates cytotoxicity of human lymphocytes and causes hemophagocytic lymphohistiocytosis. Blood, 2021, 137, 2033-2045.	1.4	27
3	Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients. Journal of Clinical Immunology, 2021, 41, 1633-1647.	3.8	43
4	Do monogenic inborn errors of immunity cause susceptibility to severe COVID-19?. Journal of Clinical Investigation, 2021, 131, .	8.2	3
5	Loss of DIAPH1 causes SCBMS, combined immunodeficiency, and mitochondrial dysfunction. Journal of Allergy and Clinical Immunology, 2021, 148, 599-611.	2.9	23
6	A Family With A20 Haploinsufficiency Presenting With Novel Clinical Manifestations and Challenges for Treatment. Journal of Clinical Rheumatology, 2021, 27, e583-e587.	0.9	9
7	Loss-of-function mutation in IKZF2 leads to immunodeficiency with dysregulated germinal center reactions and reduction of MAIT cells. Science Immunology, 2021, 6, eabe3454.	11.9	30
8	Nagashima-type palmoplantar keratosis in Finland caused by a SERPINB7 founder mutation. Journal of the American Academy of Dermatology, 2020, 83, 643-645.	1.2	14
9	Recessive MYH3 variants cause â€œContractures, pterygia, and variable skeletal fusions syndrome 1B â€• mimicking Escobar variant multiple pterygium syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2605-2610.	1.2	5
10	Heterozygous TLR3 Mutation in Patients with Hantavirus Encephalitis. Journal of Clinical Immunology, 2020, 40, 1156-1162.	3.8	12
11	Somatic mutations and T-cell clonality in patients with immunodeficiency. Haematologica, 2020, 105, 2757-2768.	3.5	18
12	Novel Hemizygous IL2RG p.(Pro58Ser) Mutation Impairs IL-2 Receptor Complex Expression on Lymphocytes Causing X-Linked Combined Immunodeficiency. Journal of Clinical Immunology, 2020, 40, 503-514.	3.8	11
13	SLC18A3 variants lead to fetal akinesia deformation sequence early in pregnancy. American Journal of Medical Genetics, Part A, 2019, 179, 1362-1365.	1.2	5
14	A nearly fatal primary Epstein-Barr virus infection associated with low NK-cell counts in a patient receiving azathioprine: a case report and review of literature. BMC Infectious Diseases, 2019, 19, 404.	2.9	13
15	Novel TMEM173 Mutation and the Role of Disease Modifying Alleles. Frontiers in Immunology, 2019, 10, 2770.	4.8	45
16	Germline alterations in a consecutive series of acute myeloid leukemia. Leukemia, 2018, 32, 2282-2285.	7.2	24
17	ADA2 deficiency: Clonal lymphoproliferation in a subset of patients. Journal of Allergy and Clinical Immunology, 2018, 141, 1534-1537.e8.	2.9	71
18	Haploinsufficiency of A20 impairs proteinâ€“protein interactome and leads into caspase-8-dependent enhancement of NLRP3 inflammasome activation. RMD Open, 2018, 4, e000740.	3.8	26

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19	Genetic Association and Altered Gene Expression of CYBB in Multiple Sclerosis Patients. Biomedicines, 2018, 6, 117.	3.2	10
20	Diagnostics of rare disorders: whole-exome sequencing deciphering locus heterogeneity in telomere biology disorders. Orphanet Journal of Rare Diseases, 2018, 13, 139.	2.7	8
21	Damaging heterozygous mutations in NFKB1 lead to diverse immunologic phenotypes. Journal of Allergy and Clinical Immunology, 2017, 140, 782-796.	2.9	113
22	Hematopoietic stem cell transplantation rescues the hematological, immunological, and vascular phenotype in DADA2. Blood, 2017, 130, 2682-2688.	1.4	140
23	Unexpectedly High Prevalence of Common Variable Immunodeficiency in Finland. Frontiers in Immunology, 2017, 8, 1190.	4.8	49
24	Constant B cell lymphocytosis since early age in a patient with CARD11 mutation: A 20-year follow-up. Clinical Immunology, 2016, 165, 19-20.	3.2	17
25	Enrichment of rare variants in population isolates: single AICDA mutation responsible for hyper-IgM syndrome type 2 in Finland. European Journal of Human Genetics, 2016, 24, 1473-1478.	2.8	22
26	Crowdsourced assessment of common genetic contribution to predicting anti-TNF treatment response in rheumatoid arthritis. Nature Communications, 2016, 7, 12460.	12.8	73
27	Association study of MMP8 gene in osteoarthritis. Connective Tissue Research, 2016, 57, 44-52.	2.3	16
28	A Novel Homozygous CTC1 Germline Mutation Associated with Bone Marrow Failure. Blood, 2016, 128, 1508-1508.	1.4	2
29	Autoimmunity, hypogammaglobulinemia, lymphoproliferation, and mycobacterial disease in patients with activating mutations in STAT3. Blood, 2015, 125, 639-648.	1.4	229
30	Stimulating translational research: several European life science institutions put their heads together. Trends in Molecular Medicine, 2015, 21, 525-527.	6.7	5
31	Dermatologic Features of ADA2 Deficiency in Cutaneous Polyarteritis Nodosa. JAMA Dermatology, 2015, 151, 1230.	4.1	75
32	Class II HLA interactions modulate genetic risk for multiple sclerosis. Nature Genetics, 2015, 47, 1107-1113.	21.4	312
33	Large Granular Lymphocyte Infiltration in the Bone Marrow in Children and Young Adults May Suggest Primary Immune Deficiency. Blood, 2015, 126, 1024-1024.	1.4	1
34	Dominant NFKB1 Mutations Cause Antibody Deficiency and Autoinflammatory Episodes. Blood, 2015, 126, 206-206.	1.4	1
35	Functional variations modulating PRKCA expression and alternative splicing predispose to multiple sclerosis. Human Molecular Genetics, 2014, 23, 6746-6761.	2.9	32
36	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nature Genetics, 2013, 45, 1353-1360.	21.4	1,213

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37	SomaticSTAT3Mutations in Large Granular Lymphocytic Leukemia. New England Journal of Medicine, 2012, 366, 1905-1913.	27.0	681
38	High-resolution SNP array analysis of patients with developmental disorder and normal array CGH results. BMC Medical Genetics, 2012, 13, 84.	2.1	9
39	High-Throughput Ex Vivo Drug Sensitivity and Resistance Testing (DSRT) Integrated with Deep Genomic and Molecular Profiling Reveal New Therapy Options with Targeted Drugs in Subgroups of Relapsed Chemorefractory AML. Blood, 2012, 120, 288-288.	1.4	1
40	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
41	Comparison of solution-based exome capture methods for next generation sequencing. Genome Biology, 2011, 12, R94.	9.6	237
42	Recurrent Missense Mutations in the STAT3 Gene in LGL Leukemia Provide Insights to Pathogenetic Mechanisms and Suggest Potential Diagnostic and Therapeutic Applications. Blood, 2011, 118, 936-936.	1.4	6
43	PRKCA and Multiple Sclerosis: Association in Two Independent Populations. PLoS Genetics, 2006, 2, e42.	3.5	45