

Bianca Tesi

List of Publications by Year in descending order

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Version: 2024-02-01

43
papers

1,967
citations

394421

19
h-index

265206

42
g-index

45
all docs

45
docs citations

45
times ranked

3847
citing authors

#	ARTICLE	IF	CITATIONS
1	Cytomegalovirus Infection Drives Adaptive Epigenetic Diversification of NK Cells with Altered Signaling and Effector Function. <i>Immunity</i> , 2015, 42, 443-456.	14.3	650
2	The syndrome of hemophagocytic lymphohistiocytosis in primary immunodeficiencies: implications for differential diagnosis and pathogenesis. <i>Haematologica</i> , 2015, 100, 978-988.	3.5	161
3	Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. <i>Blood</i> , 2017, 129, 2266-2279.	1.4	152
4	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
5	Targeting SAMHD1 with the Vpx protein to improve cytarabine therapy for hematological malignancies. <i>Nature Medicine</i> , 2017, 23, 256-263.	30.7	102
6	ARID5B regulates metabolic programming in human adaptive NK cells. <i>Journal of Experimental Medicine</i> , 2018, 215, 2379-2395.	8.5	98
7	Hemophagocytic lymphohistiocytosis in 2 patients with underlying IFN- γ receptor deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1638-1641.e5.	2.9	69
8	Epigenetic Regulation of Adaptive NK Cell Diversification. <i>Trends in Immunology</i> , 2016, 37, 451-461.	6.8	60
9	Diversification and Functional Specialization of Human NK Cell Subsets. <i>Current Topics in Microbiology and Immunology</i> , 2015, 395, 63-93.	1.1	56
10	Nordic Guidelines for Germline Predisposition to Myeloid Neoplasms in Adults: Recommendations for Genetic Diagnosis, Clinical Management and Follow-up. <i>HemaSphere</i> , 2019, 3, e321.	2.7	51
11	Human DEF6 deficiency underlies an immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis. <i>Nature Communications</i> , 2019, 10, 3106.	12.8	48
12	Pathophysiology and spectrum of diseases caused by defects in lymphocyte cytotoxicity. <i>Experimental Cell Research</i> , 2014, 325, 10-17.	2.6	38
13	Spectrum of Atypical Clinical Presentations in Patients with Biallelic <i>PRF1</i> Missense Mutations. <i>Pediatric Blood and Cancer</i> , 2015, 62, 2094-2100.	1.5	38
14	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. <i>Genome Medicine</i> , 2015, 7, 130.	8.2	37
15	Immunomodulatory activity of commonly used drugs on Fc-receptor-mediated human natural killer cell activation. <i>Cancer Immunology, Immunotherapy</i> , 2014, 63, 627-641.	4.2	33
16	Successful Hematopoietic Stem Cell Transplantation in a Patient with LPS-Responsive Beige-Like Anchor (LRBA) Gene Mutation. <i>Journal of Clinical Immunology</i> , 2016, 36, 480-489.	3.8	30
17	Differences in Granule Morphology yet Equally Impaired Exocytosis among Cytotoxic T Cells and NK Cells from Chediak-Higashi Syndrome Patients. <i>Frontiers in Immunology</i> , 2017, 8, 426.	4.8	26
18	Unperturbed Cytotoxic Lymphocyte Phenotype and Function in Myalgic Encephalomyelitis/Chronic Fatigue Syndrome Patients. <i>Frontiers in Immunology</i> , 2017, 8, 723.	4.8	24

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19	A RAB27A 5â€² untranslated region structural variant associated with late-onset hemophagocytic lymphohistiocytosis and normal pigmentation. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 317-321.e8.	2.9	22
20	An N-Terminal Missense Mutation in STX11 Causative of FHL4 Abrogates Syntaxin-11 Binding to Munc18-2. <i>Frontiers in Immunology</i> , 2014, 4, 515.	4.8	20
21	Early activating somatic <i>PIK3CA</i> mutations promote ectopic muscle development and upper limb overgrowth. <i>Clinical Genetics</i> , 2019, 96, 118-125.	2.0	14
22	Elevated ferritin and soluble CD25 in critically ill patients are associated with parameters of (hyper) inflammation and lymphocyte cytotoxicity. <i>Minerva Anestesiologica</i> , 2019, 85, 1289-1298.	1.0	13
23	Novel STAT3 Mutation Causing Hyper-IgE Syndrome: Studies of the Clinical Course and Immunopathology. <i>Journal of Clinical Immunology</i> , 2014, 34, 469-477.	3.8	11
24	HLH: genomics illuminates pathophysiological diversity. <i>Blood</i> , 2018, 132, 5-7.	1.4	11
25	Rubella vaccineâ€“induced granulomas are a novel phenotype with incomplete penetrance of genetic defects in cytotoxicity. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 388-399.e4.	2.9	11
26	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1880.	1.2	10
27	An Indian boy with griscelli syndrome type 2: Case report and review of literature. <i>Indian Journal of Dermatology</i> , 2014, 59, 394.	0.3	9
28	Haploinsufficiency of <i>UNC13D</i> increases the risk of lymphoma. <i>Cancer</i> , 2019, 125, 1848-1854.	4.1	8
29	Patients with both Langerhans cell histiocytosis and Crohnâ€™s disease highlight a common role of interleukinâ€“23. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2021, 110, 1315-1321.	1.5	8
30	A Case of Familial Hemophagocytic Lymphohistiocytosis Type 4 With Involvement of the Central Nervous System Complicated With Infarct. <i>Journal of Pediatric Hematology/Oncology</i> , 2017, 39, e321-e324.	0.6	4
31	Microdeletion of 7p12.1p13, including <i><sc>IKZF</sc>1</i>, causes intellectual impairment, overgrowth, and susceptibility to leukaemia. <i>British Journal of Haematology</i> , 2019, 185, 354-357.	2.5	4
32	Fatal Central Nervous System Lymphocytic Vasculitis after Treatment for Burkitt Lymphoma in a Patient with a SH2D1A Mutation. <i>Pediatric Infectious Disease Journal</i> , 2019, 38, e29-e31.	2.0	4
33	A Hemophagocytic Lymphohistiocytosis Case with Newly Defined UNC13D (c.175G>C; p.Ala59Pro) Mutation and a Rare Complication. <i>Turkish Journal of Haematology</i> , 2016, 32, 355-358.	0.5	4
34	Molecular Genetics Diversity of Primary Hemophagocytic Lymphohistiocytosis among Polish Pediatric Patients. <i>Archivum Immunologiae Et Therapiae Experimentalis</i> , 2021, 69, 31.	2.3	4
35	Diagnostic challenges for a novel SH2D1A mutation associated with Xâ€“linked lymphoproliferative disease. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28184.	1.5	4
36	Heterozygous variants in <i>DCC</i>. <i>Neurology: Genetics</i> , 2020, 6, e526.	1.9	4

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37	Alternative UNC13D Promoter Encodes a Functional Munc13-4 Isoform Predominantly Expressed in Lymphocytes and Platelets. <i>Frontiers in Immunology</i> , 2020, 11, 1154.	4.8	2
38	Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. <i>Blood</i> , 2021, 138, 864-864.	1.4	2
39	Clinical and laboratory signs of haemophagocytic lymphohistiocytosis associated with pandemic influenza A (H1N1) infection in patients needing extracorporeal membrane oxygenation. <i>European Journal of Anaesthesiology</i> , 2021, 38, 692-701.	1.7	1
40	Cytopenia, Predisposition to Myelodysplastic Syndrome, Immunodeficiency, and Neurological Disease Caused By Gain-of-Function SAMD9L Mutations Is Frequently Ameliorated By Hematopoietic Revertant Mosaicism. <i>Blood</i> , 2016, 128, 4299-4299.	1.4	1
41	Adult-Onset Ataxia With Neuropathy and White Matter Abnormalities Due to a Novel SAMD9L Variant. <i>Neurology: Genetics</i> , 2021, 7, e628.	1.9	1
42	Whole Transcriptome Analysis Identifies Distinct Gene Expression Profiles between SF3B1mut and SF3B1 wt Myelodysplastic Syndrome with Ring Sideroblasts. <i>Blood</i> , 2021, 138, 3695-3695.	1.4	0
43	Childhood Kaposi sarcoma related to hypomorphic severe combined immunodeficiency caused by a novel <i>CORO1A</i> mutation. <i>Pediatric Blood and Cancer</i> , 2022, 69, e29487.	1.5	0