Eleonora Gambineri

List of Publications by Year
in descending order

71	4,888	159585 30	102487 66
papers	citations	h-index	g-index

79	79	79	6649
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. Journal of Allergy and Clinical Immunology, 2022, 149, 410-421.e7.	2.9	34
2	ALPS, FAS, and beyond: from inborn errors of immunity to acquired immunodeficiencies. Annals of Hematology, 2022, 101, 469-484.	1.8	19
3	Germline IKAROS dimerization haploinsufficiency causes hematologic cytopenias and malignancies. Blood, 2021, 137, 349-363.	1.4	32
4	IPEX Syndrome and IPEX-Related Disorders. Rare Diseases of the Immune System, 2021, , 245-278.	0.1	0
5	Atypical Presentations of IPEX: Expect the Unexpected. Frontiers in Pediatrics, 2021, 9, 643094.	1.9	25
6	Planned hematopoietic stem cell transplantation in a 17â€monthâ€old patient with highâ€risk acute myeloid leukemia and persistent <scp>SARSâ€CoV</scp>â€2 infection. Transfusion, 2021, 61, 1657-1659.	1.6	1
7	Case Report: A Novel Pathogenic Missense Mutation in FAS: A Multi-Generational Case Series of Autoimmune Lymphoproliferative Syndrome. Frontiers in Pediatrics, 2021, 9, 624116.	1.9	3
8	Initial presenting manifestations in 16,486 patients with inborn errors of immunity include infections and noninfectious manifestations. Journal of Allergy and Clinical Immunology, 2021, 148, 1332-1341.e5.	2.9	75
9	IL-2 Signaling Axis Defects: How Many Faces?. Frontiers in Pediatrics, 2021, 9, 669298.	1.9	7
10	Autoimmune Cytopenias and Dysregulated Immunophenotype Act as Warning Signs of Inborn Errors of Immunity: Results From a Prospective Study. Frontiers in Immunology, 2021, 12, 790455.	4.8	11
11	The Italian Registry for Primary Immunodeficiencies (Italian Primary Immunodeficiency Network;) Tj ETQq1 1 0.784314 rgBT /Overlock	3.8	15
12	Common presentations and diagnostic approaches. , 2020, , 3-59.		1
13	Case Report: Signal Transducer and Activator of Transcription 3 Gain-of-Function and Spectrin Deficiency: A Life-Threatening Case of Severe Hemolytic Anemia. Frontiers in Immunology, 2020, 11, 620046.	4.8	9
14	Allogeneic Hematopoietic Stem Cell Transplantation for Congenital Immune Dysregulatory Disorders. Frontiers in Pediatrics, 2019, 7, 461.	1.9	19
15	Multisystem autoimmune disease caused by increased STAT3 phosphorylation and dysregulated gene expression. Haematologica, 2019, 104, e322-e325.	3.5	15
16	CD25 deficiency: A new conformational mutation prevents the receptor expression on cell surface. Clinical Immunology, 2019, 201, 15-19.	3.2	42
17	A Case of CVID-Associated Inflammatory Bowel Disease with CTLA-4 Mutation Treated with Abatacept. Archives of Clinical and Medical Case Reports, 2019, 03, .	0.1	2
18	Long-term follow-up of IPEX syndrome patients after different therapeutic strategies: An international multicenter retrospective study. Journal of Allergy and Clinical Immunology, 2018, 141, 1036-1049.e5.	2.9	233

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19	Two male siblings with a novel LRBA mutation presenting with different findings of IPEX syndrome. JMM Case Reports, 2018, 5, e005167.	1.3	16
20	Clinical, Immunological, and Molecular Heterogeneity of 173 Patients With the Phenotype of Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome. Frontiers in Immunology, 2018, 9, 2411.	4.8	136
21	Evolution of disease activity and biomarkers on and off rapamycin in 28 patients with autoimmune lymphoproliferative syndrome. Haematologica, 2017, 102, e52-e56.	3.5	49
22	Novel molecular defects associated with very early-onset inflammatory bowel. Current Opinion in Allergy and Clinical Immunology, 2017, 17, 317-324.	2.3	6
23	Low IgE Is Insufficiently Sensitive to Guide Genetic Testing of STAT3 Gain-of-Function Mutations. Clinical Chemistry, 2017, 63, 1539-1540.	3.2	4
24	Genetic Disorders of Immune Regulation. , 2017, , 295-338.		4
25	Timely follow-up of a GATA2 deficiency patient allows successful treatment. Journal of Allergy and Clinical Immunology, 2016, 138, 1480-1483.e4.	2.9	7
26	Hyperactive mTOR pathway promotes lymphoproliferation and abnormal differentiation in autoimmune lymphoproliferative syndrome. Blood, 2016, 128, 227-238.	1.4	77
27	Wiskott-Aldrich Syndrome: A Retrospective Study on 575 Patients Analyzing the Impact of Splenectomy, Stem Cell Transplantation, or No Definitive Treatment on Frequency of Disease-Related Complications and Physician-Perceived Quality of Life. Blood, 2016, 128, 366-366.	1.4	2
28	Gut immune reconstitution in immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome after hematopoietic stem cell transplantation. Journal of Allergy and Clinical Immunology, 2015, 135, 260-262.e8.	2.9	10
29	Hyperactive mTOR Pathway Promotes Lymphoproliferation and Abnormal Differentiation in Human Autoimmune Lymphoproliferative Syndrome. Blood, 2015, 126, 1020-1020.	1.4	1
30	Single centre experience of haematopoietic SCT for patients with immunodysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. Bone Marrow Transplantation, 2014, 49, 310-312.	2.4	30
31	CACP syndrome: identification of five novel mutations and of the first case of UPD in the largest European cohort. European Journal of Human Genetics, 2014, 22, 197-201.	2.8	25
32	Late-onset of immunodysregulation, polyendocrinopathy, enteropathy, x-linked syndrome (IPEX) with intractable diarrhea. Italian Journal of Pediatrics, 2014, 40, 68.	2.6	25
33	ICON: The Early Diagnosis of Congenital Immunodeficiencies. Journal of Clinical Immunology, 2014, 34, 398-424.	3.8	34
34	The evolution of cellular deficiency in GATA2 mutation. Blood, 2014, 123, 863-874.	1.4	189
35	Langerhans cell histiocytosis in <scp>IPEX</scp> syndrome: Possible role for natural regulatory T cells?. Pediatric Allergy and Immunology, 2014, 25, 601-603.	2.6	4
36	Common Presentations and Diagnostic Approaches. , 2014, , 3-59.		2

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37	Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.	5.4	27
38	Human IL2RA null mutation mediates immunodeficiency with lymphoproliferation and autoimmunity. Clinical Immunology, 2013, 146, 248-261.	3.2	186
39	Dominant gain-of-function STAT1 mutations in FOXP3 wild-type immune dysregulationâ€“polyendocrinopathyâ€“enteropathyâ€“X-linkedâ€“like syndrome. Journal of Allergy and Clinical Immunology, 2013, 131, 1611-1623.e3.	2.9	288
40	Demethylation analysis of the FOXP3 locus shows quantitative defects of regulatory T cells in IPEX-like syndrome. Journal of Autoimmunity, 2012, 38, 49-58.	6.5	67
41	Two novel patients with Bohringâ€“Opitz syndrome caused by de novo ASXL1 mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 917-921.	1.2	38
42	Congenital and acquired neutropenias consensus guidelines on therapy and followâ€up in childhood from the Neutropenia Committee of the Marrow Failure Syndrome Group of the AIEOP (Associazione) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	4.8	80
43	New frontiers in primary immunodeficiency disorders: immunology and beyondâ€¦. Cellular and Molecular Life Sciences, 2012, 69, 1-5.	5.4	9
44	Genetic disorders with immune dysregulation. Cellular and Molecular Life Sciences, 2012, 69, 49-58.	5.4	18
45	Functional type 1 regulatory T cells develop regardless of FOXP3 mutations in patients with IPEX syndrome. European Journal of Immunology, 2011, 41, 1120-1131.	2.9	72
46	Patient-centred screening for primary immunodeficiency, a multi-stage diagnostic protocol designed for non-immunologists: 2011 update. Clinical and Experimental Immunology, 2011, 167, 108-119.	2.6	143
47	Proteomics plus genomics approaches in primary immunodeficiency: the case of immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome. Clinical and Experimental Immunology, 2011, 167, 120-128.	2.6	22
48	Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). Clinical Endocrinology, 2010, 72, 839-844.	2.4	29
49	Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). European Journal of Endocrinology, 2010, 163, 329-337.	3.7	25
50	Evans Syndrome and Antibody Deficiency: An Atypical Presentation of Chromosome 22q11.2 Deletion Syndrome. Mental Illness, 2010, 2, e13.	0.8	5
51	Point mutants of forkhead box P3 that cause immune dysregulation, polyendocrinopathy, enteropathy, X-linked have diverse abilities to reprogram T cells into regulatory T cells. Journal of Allergy and Clinical Immunology, 2010, 126, 1242-1251.	2.9	48
52	The spectrum of autoantibodies in IPEX syndrome is broad and includes anti-mitochondrial autoantibodies. Journal of Autoimmunity, 2010, 35, 265-268.	6.5	102
53	S2012 Novel Mutations and Clinical Features in Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome. Gastroenterology, 2010, 138, S-301.	1.3	0
54	Hereditary Deficiency of gp91 ^{phox} Is Associated With Enhanced Arterial Dilatation. Circulation, 2009, 120, 1616-1622.	1.6	123

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55	Th17 Transcription Factor RORC2 Is Inversely Correlated with FOXP3 Expression in the Joints of Children with Juvenile Idiopathic Arthritis. Journal of Rheumatology, 2009, 36, 2017-2024.	2.0	33
56	OR.5. The Naturally Occurring Splice Variant of FOXP3 Lacking Exon 2 is not Sufficient to Maintain Immune Homeostasis and Prevent IPEX in vivo in Humans. Clinical Immunology, 2009, 131, S7.	3.2	0
57	Clinical, Laboratory, And Molecular Evaluation Of 105 Patients With A Phenotype Of Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-linked (IPEX) Syndrome. Journal of Allergy and Clinical Immunology, 2009, 123, S148-S148.	2.9	0
58	Higher risk of hepatitis C virus perinatal transmission from drug user mothers is mediated by peripheral blood mononuclear cell infection. Journal of Medical Virology, 2008, 80, 65-71.	5.0	45
59	Clinical and molecular profile of a new series of patients with immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome: Inconsistent correlation between forkhead box protein 3 expression and disease severity. Journal of Allergy and Clinical Immunology, 2008, 122, 1105-1112.e1.	2.9	199
60	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	8.5	406
61	Other Well-Defined Immunodeficiencies. , 2008, , 251-290.		5
62	Severe Food Allergy as a Variant of IPEX Syndrome Caused by a Deletion in a Noncoding Region of the FOXP3 Gene. Gastroenterology, 2007, 132, 1705-1717.	1.3	236
63	Role of regulatory T cells and FOXP3 in human diseases. Journal of Allergy and Clinical Immunology, 2007, 120, 227-235.	2.9	228
64	IPEX, FOXP3 and regulatory T-cells: a model for autoimmunity. Immunologic Research, 2007, 38, 112-121.	2.9	164
65	Immunodeficiencies with Autoimmune Consequences. Advances in Immunology, 2006, 89, 321-370.	2.2	64
66	Defective regulatory and effector T cell functions in patients with FOXP3 mutations. Journal of Clinical Investigation, 2006, 116, 1713-1722.	8.2	462
67	A new case of IPEX receiving bone marrow transplantation. Bone Marrow Transplantation, 2005, 35, 1033-1034.	2.4	66
68	Safety and immunogenicity of measlesâ€“mumpsâ€“rubella vaccine in children with congenital immunodeficiency (DiGeorge syndrome). Vaccine, 2005, 23, 1668-1671.	3.8	41
69	Inducible CO-stimulator molecule, a candidate gene for defective isotype switching, is normal in patients with hyper-IgM syndrome of unknown molecular diagnosis. Journal of Allergy and Clinical Immunology, 2003, 112, 958-964.	2.9	21
70	Immune dysregulation, polyendocrinopathy, enteropathy, and X-linked inheritance (IPEX), a syndrome of systemic autoimmunity caused by mutations of FOXP3, a critical regulator of T-cell homeostasis. Current Opinion in Rheumatology, 2003, 15, 430-435.	4.3	502
71	Lack of transmission of TT virus through immunoglobulins. Transfusion, 2001, 41, 1505-1508.	1.6	7