Eleonora Gambineri

List of Publications by Year
in descending order

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71	4,888	159358 30	102304 66
papers	citations	h-index	g-index

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

#	Article	IF	Citations
1	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.	2.0	502
2	Defective regulatory and effector T cell functions in patients with FOXP3 mutations. Journal of Clinical Investigation, 2006, 116, 1713-1722.	3.9	462
3	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	4.2	406
4	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.	1.5	288
5	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.	0.6	236
6	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.	1.5	233
7	Role of regulatory T cells and FOXP3 in human diseases. Journal of Allergy and Clinical Immunology, 2007, 120, 227-235.	1.5	228
8	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.	1.5	199
9	The evolution of cellular deficiency in GATA2 mutation. Blood, 2014, 123, 863-874.	0.6	189
10	Human IL2RA null mutation mediates immunodeficiency with lymphoproliferation and autoimmunity. Clinical Immunology, 2013, 146, 248-261.	1.4	186
11	IPEX, FOXP3 and regulatory T-cells: a model for autoimmunity. Immunologic Research, 2007, 38, 112-121.	1.3	164
12	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.	1.1	143
13	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.	2.2	136
14	Hereditary Deficiency of gp91 ^{phox} Is Associated With Enhanced Arterial Dilatation. Circulation, 2009, 120, 1616-1622.	1.6	123
15	The spectrum of autoantibodies in IPEX syndrome is broad and includes anti-mitochondrial autoantibodies. Journal of Autoimmunity, 2010, 35, 265-268.	3.0	102
16	Hyperactive mTOR pathway promotes lymphoproliferation and abnormal differentiation in autoimmune lymphoproliferative syndrome. Blood, 2016, 128, 227-238.	0.6	77
17	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.	1.5	75
18	Functional type 1 regulatory T cells develop regardless of FOXP3 mutations in patients with IPEX syndrome. European Journal of Immunology, 2011, 41, 1120-1131.	1.6	72

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19	Demethylation analysis of the FOXP3 locus shows quantitative defects of regulatory T cells in IPEX-like syndrome. Journal of Autoimmunity, 2012, 38, 49-58.	3.0	67
20	A new case of IPEX receiving bone marrow transplantation. Bone Marrow Transplantation, 2005, 35, 1033-1034.	1.3	66
21	Immunodeficiencies with Autoimmune Consequences. Advances in Immunology, 2006, 89, 321-370.	1.1	64
22	Evolution of disease activity and biomarkers on and off rapamycin in 28 patients with autoimmune lymphoproliferative syndrome. Haematologica, 2017, 102, e52-e56.	1.7	49
23	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.	1.5	48
24	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.	2.5	45
25	CD25 deficiency: A new conformational mutation prevents the receptor expression on cell surface. Clinical Immunology, 2019, 201, 15-19.	1.4	42
26	Safety and immunogenicity of measlesâ€“mumpsâ€“rubella vaccine in children with congenital immunodeficiency (DiGeorge syndrome). Vaccine, 2005, 23, 1668-1671.	1.7	41
27	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 ETQq1 1 0.784314 rgBT /Over	2.8	39
28	Two novel patients with Bohringâ€“Opitz syndrome caused by de novo ASXL1 mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 917-921.	0.7	38
29	ICON: The Early Diagnosis of Congenital Immunodeficiencies. Journal of Clinical Immunology, 2014, 34, 398-424.	2.0	34
30	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. Journal of Allergy and Clinical Immunology, 2022, 149, 410-421.e7.	1.5	34
31	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.	1.0	33
32	Germline IKAROS dimerization haploinsufficiency causes hematologic cytopenias and malignancies. Blood, 2021, 137, 349-363.	0.6	32
33	Single centre experience of haematopoietic SCT for patients with immunodysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. Bone Marrow Transplantation, 2014, 49, 310-312.	1.3	30
34	Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). Clinical Endocrinology, 2010, 72, 839-844.	1.2	29
35	Does NADPH Oxidase Deficiency Cause Artery Dilatation in Humans?. Antioxidants and Redox Signaling, 2013, 18, 1491-1496.	2.5	27
36	Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). European Journal of Endocrinology, 2010, 163, 329-337.	1.9	25

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37	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.	1.4	25
38	Late-onset of immunodysregulation, polyendocrinopathy, enteropathy, x-linked syndrome (IPEX) with intractable diarrhea. Italian Journal of Pediatrics, 2014, 40, 68.	1.0	25
39	Atypical Presentations of IPEX: Expect the Unexpected. Frontiers in Pediatrics, 2021, 9, 643094.	0.9	25
40	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.	1.1	22
41	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.	1.5	21
42	Allogeneic Hematopoietic Stem Cell Transplantation for Congenital Immune Dysregulatory Disorders. Frontiers in Pediatrics, 2019, 7, 461.	0.9	19
43	ALPS, FAS, and beyond: from inborn errors of immunity to acquired immunodeficiencies. Annals of Hematology, 2022, 101, 469-484.	0.8	19
44	Genetic disorders with immune dysregulation. Cellular and Molecular Life Sciences, 2012, 69, 49-58.	2.4	18
45	Two male siblings with a novel LRBA mutation presenting with different findings of IPEX syndrome. JMM Case Reports, 2018, 5, e005167.	1.3	16
46	Multisystem autoimmune disease caused by increased STAT3 phosphorylation and dysregulated gene expression. Haematologica, 2019, 104, e322-e325.	1.7	15
47	The Italian Registry for Primary Immunodeficiencies (Italian Primary Immunodeficiency Network;) Tj ETQq1 1 0.784314 rgBT /Overlock	2.0	15
48	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.	2.2	11
49	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.	1.5	10
50	New frontiers in primary immunodeficiency disorders: immunology and beyondâ€¦. Cellular and Molecular Life Sciences, 2012, 69, 1-5.	2.4	9
51	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.	2.2	9
52	Lack of transmission of TT virus through immunoglobulins. Transfusion, 2001, 41, 1505-1508.	0.8	7
53	Timely follow-up of a GATA2 deficiency patient allows successful treatment. Journal of Allergy and Clinical Immunology, 2016, 138, 1480-1483.e4.	1.5	7
54	IL-2 Signaling Axis Defects: How Many Faces?. Frontiers in Pediatrics, 2021, 9, 669298.	0.9	7

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55	Novel molecular defects associated with very early-onset inflammatory bowel. Current Opinion in Allergy and Clinical Immunology, 2017, 17, 317-324.	1.1	6
56	Evans Syndrome and Antibody Deficiency: An Atypical Presentation of Chromosome 22q11.2 Deletion Syndrome. Mental Illness, 2010, 2, e13.	0.8	5
57	Other Well-Defined Immunodeficiencies. , 2008, , 251-290.		5
58	Langerhans cell histiocytosis in <scp>IPEX</scp> syndrome: Possible role for natural regulatory T cells?. Pediatric Allergy and Immunology, 2014, 25, 601-603.	1.1	4
59	Low IgE Is Insufficiently Sensitive to Guide Genetic Testing of STAT3 Gain-of-Function Mutations. Clinical Chemistry, 2017, 63, 1539-1540.	1.5	4
60	Genetic Disorders of Immune Regulation. , 2017, , 295-338.		4
61	Case Report: A Novel Pathogenic Missense Mutation in FAS: A Multi-Generational Case Series of Autoimmune Lymphoproliferative Syndrome. Frontiers in Pediatrics, 2021, 9, 624116.	0.9	3
62	Common Presentations and Diagnostic Approaches. , 2014, , 3-59.		2
63	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.	0.6	2
64	A Case of CVID-Associated Inflammatory Bowel Disease with CTLA-4 Mutation Treated with Abatacept. Archives of Clinical and Medical Case Reports, 2019, 03, .	0.0	2
65	Common presentations and diagnostic approaches. , 2020, , 3-59.		1
66	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.	0.8	1
67	Hyperactive mTOR Pathway Promotes Lymphoproliferation and Abnormal Differentiation in Human Autoimmune Lymphoproliferative Syndrome. Blood, 2015, 126, 1020-1020.	0.6	1
68	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.	1.4	0
69	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.	1.5	0
70	S2012 Novel Mutations and Clinical Features in Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome. Gastroenterology, 2010, 138, S-301.	0.6	0
71	IPEX Syndrome and IPEX-Related Disorders. Rare Diseases of the Immune System, 2021, , 245-278.	0.1	0