Gulbu Uzel

List of Publications by Year
in descending order

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45 papers	4,585 citations	236925 25 h-index	233421 45 g-index
45 all docs	45 docs citations	45 times ranked	6933 citing authors

#	Article	IF	Citations
1	Autophagy-associated immune dysregulation and hyperplasia in a patient with compound heterozygous mutations in ATG9A. Autophagy, 2023, 19, 678-691.	9.1	4
2	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
3	Nodular regenerative hyperplasia in X-linked agammaglobulinemia: An underestimated and severe complication. Journal of Allergy and Clinical Immunology, 2022, 149, 400-409.e3.	2.9	8
4	Cushing syndrome and glucocorticoids: T-cell lymphopenia, apoptosis, and rescue by IL-21. Journal of Allergy and Clinical Immunology, 2022, 149, 302-314.	2.9	4
5	Hyper-IgE Syndrome due to an Elusive Novel Intronic Homozygous Variant in DOCK8. Journal of Clinical Immunology, 2022, 42, 119-129.	3.8	4
6	Coronavirus disease 2019 in patients with inborn errors of immunity: An international study. Journal of Allergy and Clinical Immunology, 2021, 147, 520-531.	2.9	278
7	Mapping Out Autoimmunity Control in Primary Immune Regulatory Disorders. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 653-659.	3.8	3
8	Genomic Spectrum and Phenotypic Heterogeneity of Human IL-21 Receptor Deficiency. Journal of Clinical Immunology, 2021, 41, 1272-1290.	3.8	25
9	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	8.5	23
10	Recurrent Nodular Regenerative Hyperplasia Following Liver Transplantation in Common Variable Immunodeficiency. Hepatology, 2021, 74, 1698-1701.	7.3	3
11	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	8.5	32
12	Prospective Study of a Novel, Radiation-Free, Reduced-Intensity Bone Marrow Transplantation Platform for Primary Immunodeficiency Diseases. Biology of Blood and Marrow Transplantation, 2020, 26, 94-106.	2.0	28
13	Diversity of XMEN Disease: Description of 2 Novel Variants and Analysis of the Lymphocyte Phenotype. Journal of Clinical Immunology, 2020, 40, 299-309.	3.8	25
14	Activated PI3KÎ´ breaches multiple B cell tolerance checkpoints and causes autoantibody production. Journal of Experimental Medicine, 2020, 217, .	8.5	33
15	Patients With Natural Killer (NK) Cell Chronic Active Epstein-Barr Virus Have Immature NK Cells and Hyperactivation of PI3K/Akt/mTOR and STAT1 Pathways. Journal of Infectious Diseases, 2020, 222, 1170-1179.	4.0	5
16	Haploinsufficiency of immune checkpoint receptor CTLA4 induces a distinct neuroinflammatory disorder. Journal of Clinical Investigation, 2020, 130, 5551-5561.	8.2	18
17	Activating PIK3CD mutations impair human cytotoxic lymphocyte differentiation and function and EBV immunity. Journal of Allergy and Clinical Immunology, 2019, 143, 276-291.e6.	2.9	64
18	STAT1 Gain-of-Function Mutations Cause High Total STAT1 Levels With Normal Dephosphorylation. Frontiers in Immunology, 2019, 10, 1433.	4.8	41

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19	PI3K pathway defects leading to immunodeficiency and immune dysregulation. Journal of Allergy and Clinical Immunology, 2019, 143, 1676-1687.	2.9	104
20	Disseminated and Congenital Toxoplasmosis in a Mother and Child With Activated PI3-Kinase Î´ Syndrome Type 2 (APDS2): Case Report and a Literature Review of Toxoplasma Infections in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 77.	4.8	16
21	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. Journal of Allergy and Clinical Immunology, 2019, 144, 236-253.	2.9	44
22	Immune Dysregulation and Disease Pathogenesis due to Activating Mutations in PIK3CDâ€"the Goldilocksâ€™ Effect. Journal of Clinical Immunology, 2019, 39, 148-158.	3.8	26
23	Successful Bone Marrow Transplantation for XMEN: Hemorrhagic Risk Uncovered. Journal of Clinical Immunology, 2019, 39, 1-3.	3.8	29
24	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	5.0	23
25	Defective glycosylation and multisystem abnormalities characterize the primary immunodeficiency XMEN disease. Journal of Clinical Investigation, 2019, 130, 507-522.	8.2	74
26	Primary immunodeficiencies: novel genes and unusual presentations. Hematology American Society of Hematology Education Program, 2019, 2019, 443-448.	2.5	18
27	Clinical Challenges: Identification of Patients With Novel Primary Immunodeficiency Syndromes. Journal of Pediatric Hematology/Oncology, 2018, 40, e319-e322.	0.6	6
28	Germline-activating mutations in PIK3CD compromise B cell development and function. Journal of Experimental Medicine, 2018, 215, 2073-2095.	8.5	79
29	Genetic Defects in Phosphoinositide 3-Kinase Î´ Influence CD8+ T Cell Survival, Differentiation, and Function. Frontiers in Immunology, 2018, 9, 1758.	4.8	29
30	Hyperactivated PI3KÎ´ promotes self and commensal reactivity at the expense of optimal humoral immunity. Nature Immunology, 2018, 19, 986-1000.	14.5	77
31	First-in-human topical microbiome transplantation with Roseomonas mucosa for atopic dermatitis. JCI Insight, 2018, 3, .	5.0	208
32	Dedicator of cytokinesis 8â€"deficient CD4 + TÂcells are biased to a T H 2 effector fate at the expense of T H 1 and T H 17Âcells. Journal of Allergy and Clinical Immunology, 2017, 139, 933-949.	2.9	69
33	Persistent nodal histoplasmosis in nuclear factor kappa B essential modulator deficiency: Report of a case and review of infection in primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2016, 138, 903-905.	2.9	14
34	Identification of a Novel Mutation in MAGT1 and Progressive Multifocal Leucoencephalopathy in a 58-Year-Old Man with XMEN Disease. Journal of Clinical Immunology, 2015, 35, 112-118.	3.8	52
35	Common Severe Infections in Chronic Granulomatous Disease. Clinical Infectious Diseases, 2015, 60, 1176-1183.	5.8	323
36	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2015, 136, 993-1006.e1.	2.9	181

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37	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. Journal of Experimental Medicine, 2015, 212, 855-864.	8.5	70
38	Pathologic Findings in NEMO Deficiency: A Surgical and Autopsy Survey. Pediatric and Developmental Pathology, 2015, 18, 387-400.	1.0	10
39	XMEN disease: a new primary immunodeficiency affecting Mg2+ regulation of immunity against Epstein-Barr virus. Blood, 2014, 123, 2148-2152.	1.4	147
40	Immune dysregulation in human subjects with heterozygous germline mutations in CTLA4. Science, 2014, 345, 1623-1627.	12.6	745
41	Somatic reversion in dedicator of cytokinesis 8 immunodeficiency modulates disease phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1667-1675.	2.9	82
42	Mg ²⁺ Regulates Cytotoxic Functions of NK and CD8 T Cells in Chronic EBV Infection Through NKG2D. Science, 2013, 341, 186-191.	12.6	269
43	Loss-of-function of the protein kinase C Î´ (PKCÎ´) causes a B-cell lymphoproliferative syndrome in humans. Blood, 2013, 121, 3117-3125.	1.4	138
44	Second messenger role for Mg2+ revealed by human T-cell immunodeficiency. Nature, 2011, 475, 471-476.	27.8	465
45	Combined Immunodeficiency Associated withDOCK8Mutations. New England Journal of Medicine, 2009, 361, 2046-2055.	27.0	655